# SOCIOLOGICAL ANALYSIS OF RISK FACTORS OF THALASSEMIA PROPAGATION IN PUNJAB, PAKISTAN

**By**

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**INTERNATIONAL ISLAMIC UNIVERSITY, ISLAMABAD**

**2020**

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A thesis for partial fulfillment of the requirements of the
Degree of Doctor of Philosophy in Sociology

**DEPARTMENT OF SOCIOLOGY**

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**INTERNATIONAL ISLAMIC UNIVERSITY, ISLAMABAD**

**2020**

## FORWARDING SHEET

The thesis entitled, "Sociological Analysis of Risk Factors of Thalassemia Propagation in Punjab, Pakistan" submitted by Mr. Muhammad Abo ul Hassan Rashid, Reg No:19-FSS/PHDSOC/F14, in the partial fulfillment of PhD. Degree in Sociology has been completed under my guidance and supervision. I am satisfied with the quality of student's research work and allow him to submit his thesis for further processing as per IIUI rules and regulations.

Dated_____ Supervisor _____

Prof Dr. Saif-Ur-Rehman Saif Abbasi

## DECLARATION

I, Muhammad Abo ul Hassan Rashid, hereby, declare that this dissertation has been written by me entirely on the basis of my research work under the sincere and kind guidance of my supervisor, Dr. Saif ur Rehman Saif Abbasi, Professor in Sociology, International Islamic University Islamabad (IIUI). It is further declared that no portion of this research has been submitted by the researcher to any other university or educational institutional before this for the award of degree.

Dated_____ Signature _____

Muhammad Abo ul Hassan Rashid

## Acknowledgement

First of all, praises be to Allah Almighty who provided me with the strength and determination to go through the grueling task of a research undertaking of this magnitude. It would be the matter of pleasure for me, to acknowledge the efforts, advices, mentoring, guidance and president support of my research supervisor, Professor Dr. Saif ur Rehman Saif Abbasi. His vast experience in the field of sociology was a value-added aspect to complete this research. With perseverance and patience, he challenged me to learn, question, think and critically analyze which made me appreciate and light up my research.

I would like to appreciate the support and advices of my colleagues Dr. Rashida Qureshi, Dr. Muhammad Farooq Solangi and my mentor Mr. Khusro Pervaiz Khan, Head of Campus, Shaheed Zulfikar Ali Bhutto Institute of Science and Technology, Islamabad. They helped and encouraged me to complete this work with their enriched background of research and practical life. The prayers of parents and family members became lighting paths to complete my dissertation, so I am thankful to Allah Almighty for blessing me with such precious relations.

I am thankful to my wife Malik Maliha Manzoor and my son Shahmeer for bearing this painful and long journey, with patience, love and care. The moral support of my brother Ahsan Rashid Malik was another encouraging factor for me to accomplish this task.

Muhammad Abo ul Hassan Rashid

# Abstract

The chronic pervasiveness of beta thalassemia major is howbeit a genetic abnormality; due to deficiency of beta protein in human blood but it has some elemental social and cultural menaces. Blindly following the traditional and supernatural practices for the treatment and management of beta thalassemia major, worsens the quality of life and wellbeing of whole family of a sick child. Male dominancy and consanguinity do not allow women and young couples to go for career detection and as a result the infant experience a deadly disease. The aim of the present study was to sociologically analyze the risk factors of thalassemia propagation in Punjab, Pakistan. Thalassemia disease is a genetic disorder, which occurs due to the hemoglobin abnormalities of alfa or beta globin genes in human blood. It has more than 120 mutations across the world but commonly found and the life ending form is, beta thalassemia major. The significant propagation of beta thalassemia major has been ascertained in Mediterranean, African, Southeast and Northern Asiatic regions, however, countries like Iran, Bangladesh, India, Saudi Arabia and Pakistan are witnessing the subtle propagation of beta thalassemia major among all other regions of the world. Other than epidemiological explanations, there are many social, cultural and economic intuitions behind this chronic illness, that have been intensively reviewed and measured in this study. The major objective of this study was to find out the socioeconomic, cultural and disease allied risk factors and psychosocial burden of beta thalassemia major among parents of sick children. In the light of positivist research paradigm, a survey based on quantitative research approach was followed to collect data from three renowned and registered thalassemia foundations (*Sundas Foundation, Jamila Sultana Foundation and Fatimid Foundation*). A sample size of 932 was drawn by using multistage random sampling technique and data was collected from the parents of patients by using interview schedule as a tool of data collection. The study found that majority of the parents of sick children belongs to the lower income group and have diminutive formal education. Their poor socioeconomic profile has increased their vulnerability towards the poor management and effective preventive strategies of beta thalassemia major. Study found that some of the parents were still unaware about the causes and symptoms of thalassemia, though they had a thalassemia child. Majority of the parents were inclined towards the practices of pre/postnatal diagnosis of thalassemia but they lacked these preventive

measures. A strong emphasis and practice of cultural and religious values have been seen in their (respondents) social life and in terms of prevention, management and treatment of beta thalassemia major. The study found that endogamy and ethnic preferences have their influence on the lives of parents and due to repeated cousin marriages, they were unintentionally propagating beta thalassemia major in their future generations. Parents were psychosocially burdened due to the illness of their child because of finical constraints and social support. It has also been found that the psychosocial burden of beta thalassemia major may reduce the need to have appropriate knowledge of the disease.

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# List of Abbreviations

| | |
|---|---|
| WHO | World Health Organization |
| PNDs | Pre/postnatal Diagnosis |
| βeta TM | Beta Thalassemia Major |
| BMT | Bone Marrow Transplantation |
| GLM | Generalize Linear Model |
| MLR | Multiple Linear Regression |
| NIH | National Institute of Health |
| NADRA | National Database and Registration Authority |

Dedicated To

My Parents and Family

# CHAPTER 1

# 1. INTRODUCTION

## 1.1 Introduction of chapter

The chapter highlights facts, explanations and scenario of beta thalassemia major across the world and particularly in Pakistan. The variations in propagation of beta thalassemia major and its causes have been incorporated in this section of dissertation by citing relevant studies. The contextualization, existing situation and preventive efforts, implemented to reduce the propagation of beta thalassemia major, across the world has been systematically cited and explained. The chapter highlights different forms of thalassemia, evident across the world and commonly found in countries with higher propagation of beta thalassemia major. To visualize thalassemia in religious context, Islamic perspective for screening, treatment and management of the genetic disorders and particularly for beta thalassemia major, has also been incorporated. The manifestation of different social and cultural causes of the disease has been cited and explained in this section along with presenting global view of beta thalassemia major. The psychological and social aspects and impacts of the disease found by other researchers have also been explored., The chapter also highlights management and preventive measures, modelled and implemented across the world. Other than the explanation and overview of academic guidelines, the section includes rational, significance, objective, hypotheses and research questions of this study.

## 1.2 Background of the Study

Social, cultural and religious deterrents are closely entangled with thalassemia (Adly & Ebeid, 2015). These evidences can be seen in the studies conducted by research scholars and medical practitioners particularly in many developing countries in Asia (Hossain et al., 2017; Sleeboom-Faulkner, 2010; Vaz, Thakur, Banerjee, & Gangal, 2000). Thalassemia disease has become a common and amputated public health problem in developing countries (Weatherall, 2010). The significant propagation of this aliment has been witnessed in Mediterranean basins, various areas of African region, entire Middle East as well as in Indian Sub-continent, along with the South-East Asiatic regions , in Melanesia and in Pacific Islands, with the range of

2% to 25% (Agouzal, Arfaoui, Quyou, & Khattab, 2010; De Sanctis et al., 2017). It has been generally estimated that, every year 50 thousand to 1 lack children, living in the low and middle income countries die because of this chronic genetic aliment (beta thalassemia major) (Ishaq, Hasnain Abid, Akhtar, & Mahmood, 2012). However, an estimated population of 7% in the entire world is the carrier of hemoglobin disorder in the form of beta thalassemia major (Arif, Fayyaz, & Hamid, 2008).

The origin and history of thalassemia can be traced back to the Greek. The word thalassemia is taken from two words that are; *thalassa* i.e. sea and *haima,* i.e. blood. Thalassemia is generally known as a kind of anemia (having abnormalities in α- and β-globin synthesis in blood) (Liao et al., 2018). Thalassemia can be categorized into two major common types, known as alpha thalassemia and beta thalassemia and it has been estimated that around 80-90 million people cross the world are the carriers of thalassemia (Origa, 2017).

Different estimates across the world, show that among genetic disorders "thalassemia" is most common (Angastiniotis & Modell, 1998; Chatterjee, Chakravarty, Chakravarty, Chowdhury, & Sultana, 2015). Thalassemia is not confined to a certain ethic group rather it is occurring in almost all ethnic groups, in every region of the world (Pahuja, Pujani, Gupta, Chandra, & Jain, 2010). The highest frequency of Thalassemia cases occurs in Italy, Greece and Cyprus (Ward et al., 2002). In Cyprus, out of seven individuals, one is a carrier of this gene, which results in one out of forty-nine marriages between carriers out of one fifty-eight newborns, one new born is expected to have beta thalassemia major (Bozkurt, 2007). Chain of countries from west to central Asia including Turkey, Iran, Pakistan, Afghanistan, Bangladesh and India to further South East Asia retain the higher propagation of beta thalassemia major (Colah, Gorakshakar, & Nadkarni, 2010) however, thalassemia exists everywhere in the world but these regions are considered as thalassemic belts od the propagation of beta thalassemia major (Gupta, 2006).

Thalassemia disease can be seen in almost all parts of Pakistan and it has been estimated that 5000 children are born with this disease, every year (Ayyub et al., 2005). It is also evident that 5 out of 100 persons suffer while 8 million are the carrier of this disease. Several epidemiological studies have been conducted across the world

in the field of medicines as well as in health and illness and approximately 20 different mutations have been reported across the world (Najmabadi et al., 2006). It is also palpable that the patient of beta thalassemia major cannot survive after the age of 10 years (Rahim & Abromand, 2008).

### 1.3 Forms of Thalassemia

Thalassemia has various forms across the world, but two of them are very common alpha and beta (Mettananda, Gibbons, & Higgs, 2015). In any of these two forms: a certain hemoglobin part is being affected (i.e. either alpha part or beta part). Regarding thalassemia, oftenly used terms are carrier, trait, major or intermediate (Vayá et al., 2003). Since hemoglobin, that is present in blood carries oxygen to all the cells that exist in a human body (Greenburg, 2009). Hemoglobin comprises two different parts that are Alpha and Beta (Karakochuk et al., 2015), when thalassemia is called “alpha” or “beta,” this refers to the component of hemoglobin that is not being formed (Muncie & Campbell, 2009). If deficiency of alpha hemoglobin exists in human body- the disorder is recognized as Alpha Thalassemia, while in case of beta hemoglobin deficiency-it is known as Beta Thalassemia. Thalassemia (Alfa-thalassemia and beta-thalassemia) are considered as the most common congenital blood disorders in the world (Harteveld & Higgs, 2010).

The terms “trait,” “carrier”, “minor,” “intermediate,” or “major” that are frequently used in thalassemia, refer to the intensity of thalassemia. An individual who has thalassemia trait shows that he/she might not have any symptoms of thalassemia or he/she might be having mild anemia, whereas an individual with thalassemia major might have intense symptoms and he/she might require regular transfusions of blood (Muncie & Campbell, 2009). The thalassemia intermediate lies between the thalassemia minor and major, but some aspects of all the three conditions overlap sometimes, however patients of the thalassemia intermediate require occasional blood transfusions (Taher, Isma'eel, & Cappellini, 2006). Children with such disease can survive but their physical and cognitive development and growth remains retardate. The children of age 2 and 6 years are among the most affected (Zeng, Yu, & Zhang, 2018).

The differentiation of the three types of thalassemia may sometimes be difficult but certain parameters have been identified and defined. The thalassemia intermediary is identified by the level of hemoglobin i.e. around 7-10 g/dl and there are traces of changes in the skeleton such as facial bones enlargements. The thalassemia results from the defect in any of the two type of polypeptide chain i.e. alpha or beta. The difference between the beta and alpha chain cause the disease. Three different mechanisms are associated with thalassemia: inheritance of a mild or silent beta chain mutation, co inheritance of determinants linked with rise in gamma chain production and co- inheritance of alpha thalassemia (Taher et al., 2006).

### 1.4 Islamic Perspective on Thalassemia

Regarding pregnancy termination, it is totally prohibited once the soul is installed in the fetus (i.e. after 120 days of gestation), even if the ultrasound results show any defect or a disorder in fetus development (Koul & Sharma, 2018). Once the soul is breathed into the fetus, abortion is just like taking life of a child which is not permissible in any scenario likewise termination of pregnancy after a certain time of conception is not allowed. If during 120 days of gestation period, the fetus shows any serious complications and during this time if the doctor suggests pregnancy termination then mothers are permitted to get the termination of pregnancy to save their lives and to further avoid medical complications (Al-Matary & Ali, 2014).

Majority of the Muslims believe that life in fetus does not immediately start after mother's conception, rather the human life needs protection, commencement for some time duration i.e. the life in fetus starts after the development of primitive streak (Bayoumi, 2009). Consequently, reimplantation of prenatal genetic diagnosis (PGD) is encouraged wherever it is needed as it is a way to avoid termination of pregnancy (Simpson, 2001). The ethical & religious reasoning on "termination of pregnancy" has diverse justifications (García, Timmermans, & van Leeuwen, 2008).

### 1.5 Socio-cultural Causes of Beta Thalassemia Major

Like other genetic disorders, thalassemia is caused by hemoglobin disorders (alpha or beta) but these genetic disorder are the result of distinctive social, cultural and religious facets (Alswaidi et al., 2012). The reasons and risk factors for the propagation of beta thalassemia major are given in the forthcoming section of introduction of this research.

### 1.5.1 Consanguinity

Like many other genetic disorders, thalassemia transmits form parents to child by means of genetic transmission (Biswas, 2017) and creates abnormalities of either alpha or beta hemoglobin (Ali, Sinthee, Islam, & Sarwar, 2018). The propagation of this genetic disorders has been evident across the world but specially in the countries with traditional health practices and management (Abdullah, 2015). Among others, the prime cause of thalassemia disease is consanguinity (Kumar, Arya, & Agarwal, 2015). The highest rate of propagation has been reported in the countries, with utmost preferences for cousin marriages (Adly & Ebeid, 2015). Due to repeated marriages with in the families, the chances of the prorogation of beta thalassemia major increases (Roudbari, Soltani-Rad, & Roudbari, 2008). The significant propagation of beta thalassemia is noticeable in Iran, India, Bangladesh and Pakistan because of preferred cousin marriages and endogamous marriage patterns (Modell & Darr, 2002; Sullivan, 1997).

A strong emphasize on traditional and cultural practices, enforces individuals to marry within the family (Estin, 2004), which creates many genetic disorders (Hamamy, 2012) and beta thalassemia major is one of these disorders. Researchers have found the higher rate of consanguineous marriages among traditional and cultural oriented societies (Abbasi-Shavazi, McDonald, & Hosseini-Chavoshi, 2008) and explored the cause of repeated consanguineous marriages due to strong believe on cast and ethnic practices (Hussain, 1999), lack of knowledge and awareness (Alnaqeb, Hamamy, Youssef, & Al-Rubeaan, 2018) and culturally diffused practices which cannot be violated (Tadmouri et al., 2009). Countries with traditional normative practices are enormously stirred to prefer cousin marriages, in comparison to the technologically advanced nations (Abdulrazzaq et al., 1997; Bittles, 2018; Oniya, Konje, Karen, & Ahmed, 2018).

### 1.5.2 Denial of Premarital and Pre/post-natal Screening

Denial of premarital screening; due to lack of knowledge and awareness of thalassemia is another propagating factor of thalassemia syndrome (Kyriakides, 2016). It has been studied, that due to lack of access to health facilities and advance laboratory contraptions, families and couples remained ignorant of the complications

of thalassemia, if not screened (Atkin & Ahmad, 2001). It is also prudent to discuss that the disavowal of premarital screening by couples and families due to cultural and social precincts, has increased the rate of thalassemia in traditional and rural societies (Karimi et al., 2007; Rajaram & Rashidi, 1999).

Premarital screening is very important for the prevention of thalassemia, but unmarried people shows lack of interest in their blood screening and other measures of thalassemia screening and prevention (Byrd, Peterson, Chavez, & Heckert, 2004). Not only in Pakistan but propagation of beta thalassemia major can be seen across the world and especially in countries with low literacy rate, high level of health constraints and cultural restrictions for screening and genetic counseling of thalassemia (Lindau, Tomori, McCarville, & Bennett, 2001).

Pakistan is witnessing the increasing rate of beta thalassemia major (9.8 million carriers of total population) in year 2018 (Rehman, Masood, Sheikh, & Mehboob, 2019) due to lack of facilities of premarital screening and carrier counselling along with traditional practices and cultural dominancy which restrain people from choosing the screening methods (Atkin & Ahmad, 1998; Cousens, Gaff, Metcalfe, & Delatycki, 2010). Without addressing the social, cultural and ethical issues and ceilings, it is hard to implement any affective program for the prevention of beta thalassemia major in Pakistan (Suhaib Ahmed et al., 2000). It is evident form the literature, that the effective implementation of screening programs was only possible in those countries like Iran, Turkey and Saudi Arabia where the cultural, ethical and social concerns were resolved prior to the enactment (Anderson et al., 2010; Atighetchi, 2007).

### 1.5.3 Family History

Family history is another major cause of the propagation of beta thalassemia major (Usman, Moinuddin, & Ahmed, 2011). Due to the genetic transmission of beta thalassemia major, it transmits from parents to children, if not screened or diagnosed earlier (Arif et al., 2008). Pakistan is a victim of genetic disorders since its detection, because of lack of acceptance of preemptive contrivance (Basit & Shera, 2008). Family history and certain descents are other contiguous causes of genetic

abnormalities, including beta thalassemia major across the world (Leader, Mohanty, Selvan, Lum, & Giri, 2018; Merten, 2019).

### 1.5.4 Lack of Knowledge and Awareness

Low level of education and awareness about thalassemia lead families to face serious social, economic and psychological problems (Ebrahim et al., 2019). Awareness, and attitude towards screening practices for the treatment of disease depends upon the level of awareness and education of the families about the disease (Thiyagarajan, Bagavandas, & Kosalram, 2019) because, without appropriate knowledge of the causes of the disease, the prevention and effective management remains futile. Due to inadequate knowledge and understanding about thalassemia, it leads a serious confusion to differentiate between thalassemia major, minor and carriers of thalassemia. Lack of knowledge also leads to the ambiguous understanding about inheritance patterns and this causes a stern physical as well as social impact in the form of disorder(s) on affected patients and their families (Zaheer, Zaman, Iqbal, Hameed, & Wazir, 2015). Public knowledge, awareness and attitude towards the problems of thalassemia and other social aspects such as social integration have shown to be in a positive association with thalassemia, not only in the contemporary societies but also into the Greeks since 20$^{th}$ Century (Politis, Richardson, & Yfantopoulos, 1991).

The substantial prorogation of beta thalassemia major is relentlessly linked and have a strong relationship with the educational and awareness level of parents and families of sick children (Egejuru et al., 2019). The well-educated couples have less chance of having thalassemia child, because of preventive measures (genetic counseling, screening) as compared to the uneducated (Wong, George, & Tan, 2011). Beta thalassemia major required an eloquent understanding and awareness for its detection, prevention and management and due to its scientific treatment, it becomes difficult for parents and families to understand the devastating impact of this disease, with little education and awareness (Gharaibeh, Amarneh, & Zamzam, 2009).

Although parental awareness and career detection of thalassemia is available in Pakistan, but still number of thalassemia cases are on rise because of limited admittance and lack of knowledge about PNDs (Aziz, Khurshid, Shaheen, & Bscn,

2019). The foremost preventive strategy for the prevention of thalassemia is, to provide appropriate information and counseling to the families and couples-by health professionals, doctors and community workers to the general population. The risky couples (carriers of thalassemia) are required extensive information about causes, measures and preventive strategies of thalassemia for their unborn offspring (Bajwa & Basit, 2019).

### 1.6 Thalassemia a Public Health Problem: Evidences from Pakistan

Thalassemia is not only confined to developing countries, rather it is pretty much prevalent across the world but with a varied rate. In Pakistani context, the propagation and severity of beta thalassemia is high in comparison to the technologically developed shires (Alvi et al., 2016). It is estimated that 3% of the world's population is carrier of beta thalassemia major (Vo, Nguyen, Le, & Le, 2018) and around 60,000 children born every year, with beta thalassemia major across the world (Kermansaravi, Najafi, & Rigi, 2018). However, among these 60,000 affected babies around 79% are from Asia regions and the propagation of beta thalassemia major is estimated between 5 to 8% and approximately 5000 children born every year in Pakistan (Haq et al., 2016).

Thalassemia disease can be a preventable syndrome but awkwardly, it has become a serious public health problem in Pakistan (Ghani, Manji, & Ahmed, 2002; Shakeel, Arif, Rehman, & Yaseen, 2016). Due to many social and cultural factors, the propagation of beta thalassemia major is mounting. It is estimated in Pakistan that every year, approximately 5 thousand children born with thalassemia and between 6 to 7 percent of the population is the carrier of thalassemia up to 2019 (Aziz et al., 2019). A greater number of population is even unaware about the carrier detection of thalassemia because of lack of access to the facilities of carrier screening and ignorance from the importance of genetic counselling (Aslamkhan, 2015). The propagation of beta thalassemia major is alarming, and a preventable target has not been achieved by the government and non-governmental organizations because of insufficient knowledge, educational campaigns and awareness sessions (Fucharoen & Weatherall, 2016).

The immersion of social and cultural restrictions for the diagnosis and treatment of beta thalassemia major is making this disease chronic and required a serious attention of parents, families, government, policy makers and enforcement agencies to adopt the effective mechanism for the prevention of beta thalassemia major across the world (Cappellini, Porter, Viprakasit, & Taher, 2018). The thalassemic patients are the victims of hemoglobin deficiency, so they continuously need the blood transfusion in order to survive, but continuous blood transfusions require many healthy donors and mammoth amount of money which cannot be afforded by the poor people (Farrugia & Del, 2015).

Children with beta thalassemia major experience behavioral disorders, low level of acumen, psychological disorders (depression and anxiety) and physical abnormalities in Pakistan (Elzaree et al., 2018) and these children suffer from behavioral and psychological disorders along with physical abnormalities due to beta thalassemia major (Zahmatkeshan, Mobasser, & Zamanzadeh, 2016). Among all other psychosocial disorders, depression has been found very common among the families, parents and children with beta thalassemia major in Pakistan (Yasmeen & Hasnain, 2018). A range of psychological and social maladjustment have been premeditated among the parents of thalassemic children, that includes; social isolation, stigmatization, anxiety and depression (Mufti, Towell, & Cartwright, 2015).

### 1.7 A Global view on Beta Thalassemia Major

A global overview of the sickle cell disease like thalassemia generally and beta thalassemia major particularly implies that it is discernable across the world with a varied rate among different social groups, because the risk factors especially social and cultural have been studied differently and so far, the propagation of beta thalassemia major mottle across the world. A global overview of the risk factors, prevention, management and propagation of beta thalassemia major is given in impending section of this chapter.

The disease (beta-thalassemia) is widespread across the Mediterranean with an uneven distribution in Greece and Italy while it is less common at the Western end of the area, meanwhile it appeared to be very little in France expect in those of Italian

and Spanish lineage. Carriers of beta-thalassemia are found between 3-7% in North Africa (Mosawy, 2017).

### 1.7.1 Beta Thalassemia Major in Greece

Most of the countries have successfully implemented the thalassemia preventive programs by perpetrating education and obligatory screening, the protruding names are; Italy, Greece and Cyprus (Cao & Kan, 2013; Loukopoulos, 2011; Metcalfe, 2012). The areas of the Mediterranean Sea, Greece and Italy have been observed to be more affected by beta thalassemia major (Chassanidis, Boutou, Voskaridou, & Balassopoulou, 2016). A study was done in Greece to investigate the amount of awareness, the public had and how much the general public was informed about thalassemia (Tsiantis et al., 1996). The researchers found that most of the people who had heard about the diseases differed by their educational level, place of residence and age, but not by sex or marital status. So, the level of awareness increased with educational level and urbanization. The main goal of the research was to provide an extensive knowledge about the thalassemia patient in Greece so that the authorities could come up with a solution for the problem. The findings of the survey showed that education was the greatest factor of providing awareness and true knowledge regarding the disease. The awareness of the disease was highest at the age 21-44 but it varied depending on the residence.

### 1.7.2 Beta Thalassemia Major in United Arab Emirates

Thalassemia is one of the most occurring disease in the United Arab Emirates and rest of the world especially Africa. In the UAE 8.3% is the gene frequency of β-Thalassemia (Hamamy & Al-Allawi, 2013). The severe form of growth retardation and anemia has been the product of thalassemia, although it can be prevented but not cured. The low growth failure is due to the low level of hormone which acts as an intermediate in the stimulation of tissue growth by growth hormone activity (Taha, 2016). Similar problem arises in children with beta thalassemia major and it has been found that low level of amino acid concentration in this disease is the cause of decreased growth in children (Kim & Tridane, 2017).

The research found that around 63% of the thalassemia patient suffered from growth retardation in the UAE and 13 different mutations have been common due to

this disease. Despite the huge number of people suffering from the disease no major effort has been done to tackle the issue by the UAE health authorities. The reason of the disease being at high level in UAE is thought to be restraining cultural potencies and higher rate of consanguineous marriages (Baysal, 2001). It has also been figured out that the families with lower socioeconomic status are more vulnerable towards beta thalassemia major (Hazmi, Hazmi, & Warsy, 2011).

### 1.7.3 Beta Thalassemia Major in China and Malaysia

Thalassemia is one of the frequent diseases that the southern region of China is facing. An extremely high rate of the disease has been found in the region and is estimated that 24.51% of the disorder is to be found in the country. The government has introduced the prevention programs for the control of thalassemia but the program could not reach its goal because of lack of well-planned strategies (Chen et al., 2010).

In the Guilin region the most common mutations are alpha (61.37%) and overall six heterozygote alpha mutation accounts for nearly 94% of the thalassemia in the area, however in contrast, the presence of beta mutation was only 52%. The data suggested that the people of the area were at high risk of alpha and beta thalassemia. The reduction in the propagation of beta thalassemia major will only be effective with increased rate of awareness and education among the communities (Tang et al., 2015).

A cross sectional descriptive study was conducted in Malaysia to know the knowledge and attitude about thalassemia. Majority of the people have reported their knowledge level as above average and the rate of blood transfusion due to beta thalassemia major has been reported 2.1 per 1000 among Malaysian population (Haque, Puteh, Osman, Mohd Zain, & Haque, 2015). It becomes unblemished that with an average or low level of education, the propagation of beta thalassemia upsurges because couples and families fail to adopt the preventive strategies (Vasudeva Murthy, Zulkeflle, Venkateswaran, & Barua, 2015).

### 1.7.4 Beta Thalassemia Major in India

Countries with the population of diverse ethnic groups, caste system and endogamy are more vulnerable toward thalassemia; because inhabitants of these states

follow traditional norms and practices' in their social life, and they preferred the treatment of disease by adopting mock practices of health and illness (Das, 2012). Due to these factors they have common hereditary disorders; as a result the propagation of thalassemia is very viable in these areas of the world and especially in India (Kar, Phadnis, Dharmarajan, & Nakade, 2014). World Health Organization (WHO) has listed the diseases to be found in more than 60 countries around the globe and approximately 2 million cases of beta thalassemia exist in India. A highest percentage of people affected by beta thalassemia are found in Gujrat, Sindh South India and Maharashta (Mondal, Maiti, Biswas, Ghosh, & Paul, 2012).

Thalassemia disease is very conversant among the Asian communities because the distinct lineages with highest rate of thalassemia have been found among Punjabi, Bengali and Gujrati kinfolks. The rate of the propagation of beta thalassemia major has been reported that 30 million people are the carriers of thalassemia, while one hundred thousand children every year are born with the beta thalassemia major (Sachdev & Gera, 2013). Like other countries, the most common form of thalassemia among Indian communities is, beta thalassemia major (Rehman et al., 2019).

The Indian researchers, Mohanty and his colleagues unearthed that the propagation of beta thalassemia major is different in distinctive groups based on caste and religious segregation. They found that propagation of beta thalassemia major varied from 0-5 % among different castes and ethnic groups. These researchers found that beta thalassemia major is not an uncommon heredity disorder among many non-tribal and tribal population of the Indian (Mohanty et al., 2013).

The highest propagation of beta thalassemia major has been found among West Bengal region of India, due to poverty and other social factors (Muthuswamy, 2011). The state of West Bengal is not different from the rest of India; it also operates under the closed system of caste. The Bengali society underwent some social transformation where the population of the major castes was decreasing, therefore to preserve their identity, the high castes used inter caste marriage as a tool for the conservancy of distinctiveness and to increase their population. Marriage outside one's own clan is not permitted, and this led to the transfer of same diseases from one generation to another. Blood or *Rakta* in the local language is considered to be an important element of the Bengali people because it determines the purity of the people

and determines the purity of the coming generations (Dolai, Dutta, Bhattacharyya, & Ghosh, 2012).

In a society where such sort of frame of mind exists, it becomes very difficult to initiate thalassemia prevention programs. The marriage and social pressure of remaining unmarried, forces the people to avoid such programs. The failure of the prevention programs is not just because of the societal pressures but also because of lack of social awareness, knowledge and exposure through print and social media especially in the rural part of the region. The lack of education has not allowed better understanding of the terms and concept related to the disease. It becomes really difficult to reassure people to, understand scientific concept when they do not have the required education (Mendiratta, Mittal, Naaz, Singh, & Anand, 2017a) . A thalassemia carrier is socially isolated with lower martial prospects in India (Chattopadhyay, 2006; Saxena & Phadke, 2002).

### 1.7.5 Beta Thalassemia Major in Iran

Iran, being a multicultural society is composed of variegated ethnic composition, which is being influenced by migrations and annexations throughout the history (Saadatnia, Etemadifar, & Maghzi, 2007). The country has a cross border strategic position and is heavily influenced by cultural practices such as religion and consanguinity (Farhud et al., 1991; Keyfi et al., 2018). It has been reported that there are 47 distinct beta globin DNAs transformation, conscientious for beta thalassemia major in Iran. Approximately 270 million people are the carriers of thalassemia and 80 million are the fatalities of beta thalassemia major. It is also evident for the different reports that more than 60,000 children are born every year with beta thalassemia major (Rezaee, Banoei, Khalili, & Houshmand, 2012).

Iran is considered as a major center for the propagation of beta thalassemia major, among other eastern Mediterranean areas due to its strong emphasize on consanguinity (Falahati et al., 2019). More than 20,000 patients of beta thalassemia major exist in Iran along with 3 million carriers of beta thalassemia major (Kosaryan, Karami, Akbarzadeh, & Aliasghrain, 2019). Different epidemiological and social science's researchers found that; consanguinity, nutrition and infections and human

migration are considered as influential factors for the prevalence of thalassemia in Iran (Najmabadi et al., 2006).

### 1.7.6 Beta Thalassemia Major in Bangladesh

Bangladesh is among those countries, who successfully implemented the different screening programs for detection and prevention of thalassemia but still the carrier status of thalassemia is about 10 % of the total population and more than 7,000 children are born every year with thalassemia (Tahura, Selimuzzaman, & Khan, 2016). It has been found in another survey that 17.94% carriers of thalassemia are found in Bangladesh, out of which 4.02% are the carriers of beta thalassemia major (Khan, Banu, Sadiya, & Sarwardi, 2017).

Most of the academicians and health professionals have suggested a mandatory national policy for carrier screening, because effective and compulsory screening could reduce the propagation of beta thalassemia major. By reducing the marriages at risk and increasing effective mechanisms for carrier detection, genetic counseling and screening the country can prevent affected births and propagation of beta thalassemia major. Countries like Bangladesh can reduce the propagation of beta thalassemia major by public education, pre/postnatal screenings, genetic counselling and provision of advance services for diagnosis because these are the effectives ways for prevention and management of beta thalassemia major (Banu, Khan, Selimuzzaman, Sarwardi, & Sadiya, 2018; Tahura et al., 2016).

## 1.8 Impact of Beta Thalassemia Major on Children (patients)

The form of thalassemia, an individual is suffering from, dependents on which thalassemia trait has been genetically carried by a child from his parents (Fogel, Nguyen, Smink, & Sekhar, 2018). If an individual inherits a beta thalassemia trait from both his mother & father, then he/she most likely will have beta thalassemia major. Comprising thalassemia trait refers to the fact, one might not have any signs of thalassemia but parents can transfer that trait to their children and consequently increase the risk of their children to have thalassemia (Hanprasertpong et al., 2013).

Children with thalassemia face changes in their facial characteristics along with many other psychological problems (Kumar et al., 2019). They can be identified based on their skin color and shape of nose, which is s most ostensible physical feature among the thalassemic children. These changes can be seen and observed due to iron deficiency and blood transfusion that creates many other hormonal changes in the bodies of ill children. The chromosomes comprising trait of hair & body structure are transmitted from parents to their off springs (Gollo et al., 2013).

Children with thalassemia have poor academic performance (Gamayani, Lestari, Luh, Ganiem, & Panigoro, 2019), depression, anxiety, poor digestive (Toumi, Merzoug, & Boulassel, 2018) and many other physical and psychological tumults due to lack of iron and healthy protein in their blood. Not only patients but the parents also face many psycho-social sprints in their daily interaction and social life (Khurana, Katyal, & Marwaha, 2006). Children with thalassemia usually feel anxiety; depression and many other physical disorders and problems particularly body pain, due to iron and protein deficiency in their blood, so it leads to affect their school performance, physical tasks and social participation in different social events and gatherings (Shaligram, Girimaji, & Chaturvedi, 2007).

### 1.9 Impact of Beta Thalassemia Major on Parents and Families

Parents of the children, suffering from beta thalassemia major face social isolation and stigmatization by the members of their communities, because of lack of understanding about the causes of beta thalassemia major (Punaglom, Kongvattananon, & Somprasert, 2019). Mothers' of sick children are blamed by the family members in most of the traditional societies, because of false religious and supernatural believes (Antonarakis, 2019; Hakeem, Mousa, Moustafa, Mahgoob, & Hassan, 2018). Lack of knowledge and understanding of thalassemia generally and beta thalassemia major particularly create anxiety and depression among the parents and families of the patients and they become pessimistic for their future lives (Kermansaravi et al., 2018).

Regular blood transfusion and medication imposes serious economic burden on parents and families of sick children because of lower socio-economic status and expensive treatment of thalassemia (Thiyagarajan, Bhagvandas, Kosalram, &

Bhattacharya, 2019). Furthermore; parents, suffer serious emotional and psychological disorders due to difficult management of such chronic illness, which eventually upshots the quality of life of entire family of a sick child (Mainani, Dua, Mujawar, Kamal, & Rughwani, 2019).

The entire social life of a family gets affected, because the patients required consistent and exhaustive care, monitoring of their physical activities, well-timed medication and intensive dietary precautions (Moirangthem & Phadke, 2018). Due to all the rigorous management of beta thalassemia major, parents remained unable to participate in social gatherings, ceremonies and other communal events (Piyamongkol, Mongkolchaipak, & Piyamongkol, 2019; Septyana, Mardhiyah, & Widianti, 2019).

### 1.10 Psychological and Social Facets of Beta Thalassemia Major

Thalassemia is a very serious public health problem across the world and particularly in developing countries. The ample propagation of beta thalassemia major is found to be a serious threat to the middle- and low-income countries across the world (Khalid et al., 2019). The disease imposes intensive psychosocial burden on parents and children, and it has been found that parents and children with beta thalassemia major experience a mammoth psychological maladjustment due to beta thalassemia major (Joshi & Vashist, 2018; Mettananda et al., 2019).

Parents and family members of the children with beta thalassemia major feel depression and anxiety due to the illness of their children(s) (Kermansaravi et al., 2018) and it has been found that couples become pessimistic about the life of their child (Mohamadian, Bagheri, Hashemi, & Sani, 2018) because they know that beta thalassemia major is a life sacking disease. Beta thalassemia major casts a significant amount of depression, anxiety and stigmatization upon parents of thalassemic children (Moudi, Phanodi, & Vedadhir, 2019).

Due to the regular blood transfusion and therapies, parents are required to visit hospitals and blood transfusion centers on regular basis (Caocci et al., 2012; Roy & Chatterjee, 2007), which affects their (parents) social and personal life and they remained socially isolated. Another reason of the social isolation of parents is; a stern and perpetual monitoring of all physical activities of their sick child because children

with beta thalassemia major are needed prophylactic diets and restricted physical movements (Lal, Sheth, Gilbert, & Kwiatkowski, 2018).

Beta thalassemia major also creates the feelings of denial, withdrawal and lack of acceptance of the consequences of disease, among parents and families of thalassemic children (Chan et al., 2017). The disease creates emotional and psychological trepidation among parents of sick children because of its outrageous end. A strong effect, in terms of psychosocial drain of beta thalassemia major has been found on sick children by affecting their education, sports and other physical activities, school timing and nonattendance (Khanna, Prabhakaran, Patel, Ganjiwale, & Nimbalkar, 2015). Beta thalassemia major has a negative impact on sexual and physical growth of sick children because of abnormal hormonal growth and lack of healthy diet, along with iron overload due to excessive blood transfusion (Khanna et al., 2015).

### 1.11 Preemptive Approaches for Disease

The prevention and extinction of beta thalassemia major is decisive for all those societies facing an increased rate of propagation of this disease. Most of the countries (Iran, Bangladesh, Jordan, Saudi Arabia, Turkey, Italy and Cyprus) have adopted preventive mechanisms for preclusion and riddance of beta thalassemia and got a varied public response, because of social, cultural and religious aspects of these techniques (Aydınok et al., 2018; Russo et al., 2019; Sarvestani, Hasanifar, & Bagheri, 2019). The prevention of beta thalassemia major can be attained by providing education, increasing knowledge and awareness among parents and families of diagnosed children particularly and among the entire population generally (Rakhmilla, Susanah, Rohmawaty, & Effendi, 2018). Because lack of knowledge and awareness regarding carrier screening and prevention of beta thalassemia major boosted its propagation across the world (Ghafoor, 2016).

Population screening is an effective method for carrier detection of thalassemia across the world (Goonasekera, Paththinige, & Dissanayake, 2018) but it requires appropriate and updated information to facilitate the process of genetic screening and counseling. Due to lack of health education and low level of health literacy, couples and families of normal children also remain unaware for being the silent carrier of thalassemia, which increases their vulnerability towards genetic disorders, like beta

thalassemia major. It has been found that equitable education, higher awareness and grander health literacy is an imperative factor to reduce the propagation of beta thalassemia major(Carden, Newlin, Smith, & Sisler, 2016; Hassanzadeh, Mirahmadizadeh, Karimi, & Rezaeian, 2017).

### 1.12 Termination of Pregnancy and Beta Thalassemia Major

Numerous studies revealed that religion has a strong influence on the decisions of peoples, related to their lives and treatment of disease. In view of researchers and renewed religious scholars, there is no restriction of termination of pregnancy in Islam but the defined period to terminate pregnancy is before 120 days of gestation (Atkin & Ahmad, 2000; Modell, Khan, & Darlison, 2000). Though people are provided this information through print and electronic media, but it has been found that in Pakistan, out of 141 married couple 72% knew about parental diagnosis but only 56% used the available services (Badshah, 2017).

Couples and parents of children with beta thalassemia major particularly and for genetic disorders generally, remain reluctant for termination of pregnancy because of ethical and cultural concerns (Garel, Gosme-Seguret, Kaminski, & Cuttini, 2002; Mozersky et al., 2017). The effective implementation of preventive programs can be accomplished by addressing the ethical and cultural trepidations of people and communities with high propagation of beta thalassemia major. Without cooperation of educational and religious groups and institutions, the awareness level of parents cannot be increased, and implementation of preventive strategies would be futile (Mendiratta, Mittal, Naaz, Singh, & Anand, 2017b). Termination of pregnancy, premarital, pre- and post-natal screenings can reduce the risk of prevalence of disease but there are many other factors like cultural, social and religious barriers (Shenaz Ahmed, Atkin, Hewison, & Green, 2006) that restrain parents towards the adoption of these methods. Ultimately patient's families suffer from serious mental and social disorders, due to beta thalassemia major (Thiyagarajan, Bhattacharya, Sharma, Srivastava, & Dhar, 2019).

### 1.13 Management of Beta Thalassemia Major

Like other genetic disorders, beta thalassemia major also required intensive care and affluent management, which necessitates the effective knowledge, preventive

practices, and value-added counselling stratagems for parents and families (Martin & Drucilla Haines, 2016; Shah, Prescott, & Kyei-Mensah, 2018). The interplay of consanguinity, endogamy, cast and ethnic preferences for marriages are very common and persuasive factors for the propagation of beta thalassemia major in Pakistan (Mustafa, Zulfiqar, Ali, & Naseem, 2018). Beta thalassemia major is rapidly spreading in low income countries (Arif et al., 2008). Researchers, medical experts and health practitioners laid a heavy stress on social practices such as knowledge about the disease, attitude and practices of the families, cousin marriages and screening practices which can trigger thalassemia in case these are ignored (Zaman & Salahuddin, 2006).

Due to economic constraints, highly expensive treatment and agonizing treatment cost, people belonging to the rural areas and having lower socio-economic status cannot bear the curative management system (Moirangthem & Phadke, 2018; Riewpaiboon et al., 2010). While it seems possible to intend for the adaptation of preventive management system but due to low literacy rate and unavailability of information required for proper counseling and screening, thalassemia rate is prevailing in Pakistan and analogous countries (Asif & Hassan, 2016).

The first management system is known as Conservative Management System that comprise of -lab investigation, safe blood transfusion, chelation therapy and treatment with fetus hemoglobin augmenting agents (Patsali, 2018; Perrine, Faller, & Berenson, 2013). While the second way for the management of thalassemia is known as Curative Management System -bone marrow transplantation (Baronciani et al., 2016; Choi et al., 2018; Qadir & Rizvi, 2018) and third way is known as Preventive Management System of thalassemia -screening and genetic counseling (Asif & Hassan, 2016; Fucharoen & Weatherall, 2016; Ghosh, Ghosh, Agrawal, & Nadkarni, 2019). Although bone marrow transplantation is considered an effective way for the treatment of beta thalassemia major, like other genetic ailments across the world but the success rate is very low in case of beta thalassemia major (Shenoy et al., 2016; Wen et al., 2018). For effective preventive programs, micro-mapping is very important to pave the way for and bring new strategies in different national and international thalassemia control programs (Tritipsombut et al., 2012).

## 1.14 Rationale of the Study

The propagation of beta thalassemia major has been seen across the world and especially in traditional and low-income counties. The evidences of propagation of beta thalassemia in traditional societies suggest- cultural preferences in marriages, endogamy and high rate of consanguinity. While, the propagating risk factors additionally include; low level of awareness and knowledge about the diagnosis and prevention of thalassemia, lack of access and reluctance of people for premarital, pre/postnatal screenings and genetic counseling. Pakistan is witnessing grander propagation of beta thalassemia major among its denizens due to social, cultural, religious, economical and epidemiological considerations.

The greater impact of lower socio-economic status has been studied by (Safy et al., 2016; Haghpanah et al., 2013; Safdar et al., 2017; Yasmeen & Hasnain, 2018), which causes higher propagation of beta thalassemia major in Pakistan. The study intended to incorporate significant risk factors, such as-consanguinity, which has been unearthed to play a momentous role in propagation of beta thalassemia major across the world (Suhaib Ahmed, Wazir, & Qayyum, 2018; Bai, Nasir, Ahmed, Malik, & Arif, 2019; M. S. Khan, Ahmed, Khan, Mushtaq, & Wasim, 2015; Raza, Farooqi, Mubeen, Shoaib, & Jabeen, 2016; Rudra et al., 2016). The influence of religious beliefs and practices for diagnosis, management, treatment and prevention of beta thalassemia major has an evocative role, especially in traditional societies.

Like other genetic disorders, beta thalassemia major also transmits to children genetically. The treatment of this disease is possible by Bone Marrow Transplantation, but it is very expensive for Pakistani parents, because of economic destitution. Beta thalassemia major, requires an adequate knowledge for the diagnosis, prevention and management. The study amalgamated parental knowledge, awareness and education as an underlying variable of this study, as insinuated by many others (Haq et al., 2017; Manzoor & Zakar, 2019).

The possible way to prevent births of beta thalassemia major is- termination of pregnancy. But abortion is a highly sanitized measure to be suggested as a preventive measure, especially to the people with low level of education and having traditional life styles. So, keeping in view the ethical and moral values of local communities, the

scientific practices measure for the prevention of beta thalassemia major can be implied. The present study is one of the empirical corroborations, which highlights and found strong religious, cultural and social influence over the lives of parents of thalassemic children. The frequent blood transfusion, regular medications and precautionary dietetic requirements of sick children impose severe economic and psychosocial burden on parents and families of sick children.

### 1.15 Significance of the Study

Social, economic and cultural are the major risk factors of beta thalassemia major. Cross cousin marriages, different cultural beliefs and practices are the major obstacles for the prevention and management of beta thalassemia major. Meanwhile, poor economic status and financial constraints stem people to go for expensive treatment of beta thalassemia major. Hereafter, in this scenario, the responsibility lies on academician, policy makers, educationist, social activist and medical professionals to create awareness among people for prevention and management of beta thalassemia major, like other genetic disorders.

Previously, studies conducted by health experts, academician and nongovernmental organizations found that beta thalassemia major is a rapidly propagating genetic disorder in Pakistan. Most of these researches are purely epidemiological (Ahmed Kiani et al., 2016; Ansari et al., 2012; Sharif, Irshad, Tariq, Rasheed, & Tariq, 2019), some of those focused on social and economic impacts only (Ishfaq, Shabbir, Naeem, & Hussain, 2015; Mufti et al., 2015; Siddiqui et al., 2015), some tried to find out the effect of thalassemia on quality of life of patients and their families (Majid & Zafar, 2018; Sultana, Humayun, Noor, Humayun, & Zafar, 2016; Yousafzai et al., 2018). Rarely a single study conducted by (Batool, Ishfaq, & Bajwa, 2017) figured out the psychosocial burden of beta thalassemia major on parents and sick children. The present study intended to find out the all possible and yet to be reconnoitered risk factors (socio-economic, cultural, religious and disease allied), parental knowledge, practices of pre/postnatal screening and psychosocial burden of beta thalassemia major, conjointly and focus retroactively rather than figuring out the common sequels.

## 1.16 Objectives of the Study

The present study intended to address the following objectives, to sociologically analyze the risk factors of beta thalassemia major in Punjab (Pakistan)

1. To measure the knowledge and awareness of parents regarding disease and its preventive measures

2. To quantify the psychosocial burden of thalassemia on patient's parents

### 1.16.1 Specific objective of the study

1. To study Socioeconomic risk factors of thalassemia propagation among respondents (parents thalassemic children).

2. To explore cultural risk factors of thalassemia propagation among respondents (parents thalassemic children).

3. To investigate disease allied risk factors of thalassemia propagation among respondents (parents thalassemic children).

## 1.17 Hypotheses of the study

The hypotheses of the study are as follow:

### 1.17.1 Hypothetical Relationship of Risk Factors and Psychosocial Burden of Thalassemia

**H1:** Higher exposure towards the knowledge of disease is likely to decrease the psychosocial burden of disease among parents of thalassemic children.

**H2:** Higher exposure towards the practices of pre/postnatal diagnosis of disease is likely to decrease the psychosocial burden of disease among parents of thalassemic children.

**H3:** Higher exposure towards socio-economic risk factors is likely to increase the psychosocial burden of disease among parents of thalassemic children.

**H4:** Higher exposure towards cultural risk factors is likely to increase the psychosocial burden of disease among parents of thalassemic children.

**H5:** Higher exposure towards disease allied risk factors is likely to increase the psychosocial burden of disease among parents of thalassemic children.

**H6:** There is a significant relationship between socio-economic profile of patient's parents and risk factors of beta thalassemia major.

**H7:** There is a significant relationship between socio-economic profile of patient's parents and psychosocial burden of beta thalassemia major.

### 1.17.2 Hypothetical Differences of Risk Factors and Psychosocial Burden of Thalassemia

**H8:** There is a significant difference of risk factors, parental knowledge, practices of pre/postal diagnosis and psychosocial burden of beta thalassemia major between male and female children

**H9:** There is a significant difference of risk factors, parental knowledge, practices of pre/postal diagnosis and psychosocial burden of beta thalassemia between rural and urban people

**H10:** There is a significant difference of risk factors, parental knowledge, practices of pre/postal diagnosis and psychosocial burden of beta thalassemia between consanguineous and non-consanguineous marriages.

**H11:** There is a significant difference of risk factors, parental knowledge, practices of pre/postal diagnosis and psychosocial burden of beta thalassemia among different ethnic groups.

**H12:** There is a significant difference of risk factors, parental knowledge, practices of pre/postal diagnosis and psychosocial burden of beta thalassemia major, based on different family structures.

**H13:** There is a significant difference of risk factors, parental knowledge, practices of pre/postal diagnosis and psychosocial burden of beta thalassemia based on occupational taxonomies.

## 1.18 Research Questions

1. What is the knowledge of parents regarding causes, management and prevention of beta thalassemia major?

2. To what extent, the parents are aware about genetic disorders, counseling and pre/postnatal screening?

3. What is the status of socioeconomic profile of the respondents?

4. What kind of treatment practices are being followed by patients' families for disease?

5. What are the social and economic risk factors of beta thalassemia major for parents of sick children, living in Punjab Province?

6. What are the cultural risk factors of beta thalassemia major for parents of sick children, living in Punjab Province?

7. What are the disease allied risk factors of beta thalassemia major for parents of sick children, living in Punjab Province?

8. What is the extent of psychological burden of the disease on patient's parent due to beta thalassemia major?

## 1.19 Chapter Summary

The chapter summarizes the findings and vindications of studies, conducted by researchers and presented an overview of beta thalassemia major across the world. The propagation of beta thalassemia major has been seen prominent in developing countries especially, where traditionalists beliefs dominants like in Pakistan, India, Bangladesh, Iran and Saudi Arabia. The chapter included background of the study by highlighting the social causes and importance of beta thalassemia major, because the epidemiologists remain inept to study the social, cultural and religious factors of a disease within the perspectives of social settings. The chapter explored the narratives and arguments of Muslim Scholars regarding treatment and prevention of beta thalassemia major and it has been found that termination of pregnancy in case of any genetic disease, considered life threatening, is allowed but in a given period of time (120 days). The chapter also incorporated the importance of practices of pre/postnatal diagnosis and lack of acceptances by couples and families because of cultural influence, lack of education and access of these preventive practices. The impact of beta thalassemia major on sick children and their parents has been conceptualized in this chapter and it has been clinched that besides physical abnormalities and medical complications, the disease has a chronic social and psychological impact on patients, their parents and families. The effective management of beta thalassemia major has been found trifling among rural, illiterate and traditional communities.

# CHAPTER 2

# 2. REVIEW OF LITERATURE

## 2.1 Introduction of chapter

This chapter of dissertation is segmented into two parts; the first part is based on works of authors, researchers and organizations in developing countries and among those geographical location such as India, Iran, Bangladesh, Saudi Arabia, Malaysia and Pakistan, where propagation of beta thalassemia major is researched intensively. The chapter includes studies, related to risk factors (social, cultural, economic, disease allied) and psychosocial burden of beta thalassemia major, on sick children and their parents, as well as on the families. Furthermore, the section highlights significant role of consanguinity, prenatal, postnatal and premarital screening and parental knowledge of beta thalassemia major across the world and particularly in Pakistan. The pocket of evidences has also been exposed to discuss, management and prevention of beta thalassemia major in this section. The second part of this chapter is based on models of health and illness, the social construction and conceptualization of beta thalassemia major, under the guiding principles of sociology of health and illness. The detailed overview of all possible models of health and illness have been discussed and a strong emphasize is laid to explain biopsychosocial model of health because the present study aimed to borrow the theoretical orientations of this model.

## 2.2 Risk Factors of Beta Thalassemia Major

By the expansion of human knowledge and efforts to effect environment, the conception of health risk factors has been changed from determinants of natural environment (i.e. flood and earth quake) to human exertions in the form of producing pollution and global warming. These changes welcome sociological explanation of health and illness and enable researchers to identify risk factors of a disease in the context of sociology. A major shift in researches and analysis of health experts is observed as researchers and policy makers moved from environmental to social factors because evidently it was found that instead of focusing on agent host model

and ecological aspects related to health and illness, focus should be on structure agency debate. Because most of the health problems are caused by the individual themselves i.e. smoking and obesity.

Theoretically, health is the combination of life chances and life choices, and to be "at risk" is the amalgamation of social and individual factors. Epidemiologically, researchers believe that risks are required to be assessed statistically, however a lay-man approach remains stick to the knowledge and experiences that the acquired by their own experience or in a tacit way. Understanding of risk factors is required to be based on empirical evidences and the professional approaches for the assessment, so that the findings can be conveyed to the laymen and healthy practices can be diffused easily in their lives (Naidoo & Wills, 1998).

### 2.2.1 Socio-economic Risk Factors of Beta Thalassemia Major

Researchers indicated that major risk factors of thalassemia propagation are low socio-economic status and socio-economic constraints for the treatment and prevention of beta thalassemia major (Alkinani, Abbas, Faraj, & Jumaa, 2017; Jameel, Suliman, & Rehman, 2016; Sabbah et al., 2017). Children belong to the poor economic status are more vulnerable to thalassemia and families tie with traditional values and norms where consanguinity is an imperative, are at high risk to have thalassemia major (Gul, Wazir, & Rehman, 2017; Singh & Negi, 2019; Vali, Seyednezhad, Farahmandinia, Mirzai, & Abdi, 2017).

Studies conducted in Pakistan are focused to highlight the problems faced by families and parents of sick children i.e., socioeconomic problems (Rahman & Lodhi, 2004), and association between thalassemia and education (Baig et al., 2006) and explored the awareness level regarding thalassemia (Arif, Fayyaz, & Hamid, 2008). Mostly the researchers (Lomas, 1998; McEwen & Wills, 2017; Moghavvemi et al., 2017) and health practitioners belong to the biomedical sciences and their focus remained on biological aspects of disease and on epidemiological postures rather than finding the social, cultural, and economic factors associated with thalassemia major (Hossain et al., 2017; Smith & Praetorius).

Due to regular blood transfusion, medicines and precautionary diet, families suffer rigorous economic burden and the families having low socio-economic status

suffer a lot because, expenditures of hospitals and medicines are not affordable. A study was conducted by (Sattari et al., 2012) in Iran on financial and social impact of thalassemia and the entire beta-thalassemia patents were involved in the study to know the impact (financial and social) on patients and their families. The information was drawn to find the direct and indirect expenditure of the thalassemia's treatment and researchers found that patients have a very enormous economic burden for the treatment of disease. Studies conducted by (Al Sabbah et al., 2017; Grewal, Sodhi, & Sobti, 2017; Jameel et al., 2016) identified that propagation of thalassemia is mainly allied with lower socio-economic status, lack of awareness among families (Ghafoor, 2016; Goyal, Hpapani, & Gagiya, 2015; Hossain et al., 2017).

Parental education plays a vital role in normalizing their behavior and to avoid the stigmatization due to illness of a thalassemic child (Punaglom, Kongvattananon, & Somprasert, 2019; Suzanah, Zulaiha, Faszrul, & Kamaruzaman, 2011) because adequate education and knowledge enables them to be acquainted with the causes of disease and don't blame their fate, while contradictory insinuations are seen in traditional societies and among the illiterate couples because their low level of education detain them to go against their cultural values, believes and practices (Ebrahim et al., 2019). Parental knowledge and awareness about thalassemia and its preventive measures has a significant importance for the propagation of thalassemia. Parents with little knowledge about the preventive measures and causes of thalassemia make their children to be exposed to the disease (Tahura, Selimuzzaman, & Khan, 2016).

Understanding of thalassemia can be enhanced by increasing health literacy of families and caregivers, because understanding of basic knowledge of health practices and precautions of thalassemia are the result of increased health literacy of families (Sananreangsak, Lapvongwatana, Virutsetazin, Vatanasomboon, & Gaylord, 2012). Like many other genetic abnormalities, thalassemia also required a strong decision-making ability of couples and families, especially for genetic screening, counseling (Fan et al., 2018; Stevens et al., 2019) and in many cases the termination of pregnancy also (Mustafa, Zulfiqar, Ali, & Naseem, 2018). It has been found in many studies that low level of health literacy, outcomes a poor health and is considered a major barrier for self-management, active participation in communal activates and low self-esteem

of any individual (Nair & Ibrahim, 2015; Nikam, Dama, Patil, & Dama, 2012; Sananreangsak et al., 2012).

A greater deal of literature suggested that educational and awareness programs are very important to assist and facilitate parents and families of thalassemic children for the management and prevention of beta thalassemia major. Researchers (Aycicek, Koc, Bayram, & Abuhandan, 2016; A. Maheri et al., 2016) considered education a prime tool that can reduce the negative impact of thalassemia as well as social, economic, psychological and emotional disorders over the lives of parents and families of children suffering from beta thalassemia major (Atkin & Ahmad, 2000; Prasomsuk, Jetsrisuparp, Ratanasiri, & Ratanasiri, 2007); Sadiq, Eigel, and Horst (2001).

Like many other chronic illnesses, patients of thalassemia and their families required meticulous support from relatives, health professionals and from rest of the community members to manage their psychological and social maladjustment. It has been investigated by researchers (Patel et al., 2019) that family is believed to be the primary and most important source of social support for patients and their parents to mitigate the psychological and social burden caused by thalassemia. Patients of thalassemia are required medical as well as social support from their families, medical professionals and other community members for their social adjustment and confrontation against thalassemia. The effective management and social support also required general knowledge about the disease, so the general population of any area required to gain at least the basic (Kelsey, 2015).

Researchers (Piel & Weatherall, 2014; Radke, Paulukonis, Hulihan, & Feuchtbaum, 2019) found that lack of knowledge about disease, manifestation of disorder, rate of survival, availability of treatment and health facilities (medicalization and counseling), psychological and cultural issues may cause barriers to ideal health care including thalassemia. For all hemoglobin disorders generally and thalassemia particularly, awareness, attitude, perception and adaptation of screening towards the disease is very important to reduce the vulnerability and susceptibility of the couples towards thalassemia.

It has been found by (Pouraboli, Abedi, Abbaszadeh, & Kazemi, 2017) that patients of thalassemia are required a very ardent attention, care and psycho social support to mainstream themselves into social activities and to help them in a significant hale and hearty lives. Parents have many challenges to handle the emotional disturbances of their children due to thalassemia because it is very difficult for children to understand the chronologies of thalassemia due to excessive physical movement.

A study was conducted by (Ishfaq, Dia, Ali, Fayyaz, & Batool, 2018) revealed that 63 % of the patient's with thalassemia major expressed, that their disease affected their academics. While 54% of the respondents in mentioned study were unable to participate in any kind of indoor sports activities due to their illness. Blood demands and financial assistance required from friends resulted a laidback response because almost 70% of the respondents were facing ignorance of relatives and friends due to the blood demands. It seems difficult for parents to manage an excessive amount of blood on regular basis for transfusion because many thalassemia foundations working in Pakistan are facing serious hurdles to arrange blood for the patients (Tahir, Shahid, & Mahmood, 2011).

### 2.2.2 Cultural Risk Factors of Beta Thalassemia Major

Due to many cultural risk factors, propagation of thalassemia is found to be projecting across the world and especially in those countries where family values are constricted with culture. Marriages within the families are prominent and termination of pregnancy and prenatal screening is considered as unethical and religiously excluded. An effervescent sway of cultural and religious factors has also been patent for the treatment and management of beta thalassemia major among Pakistani parents (Maheen, Malik, Siddique, & Qidwai, 2015).

The review of studies, steered by (Furnham, 2015; Tokur-Kesgin, Kocoglu-Tanyer, & Demir, 2019) suggested that family attitude, role and understanding is mainly determined by social, cultural and religious factors, especially in rural and traditional societies. These factors determine the patterns of living and life styles of residents of practicing communities, where individuals are bound to follow traditional ways for disease management and coping strategies. However, if individuals follow

humanitarian practices, then religion could be the main source of social support for parents and families to manage sickle cell disease like thalassemia (Chong et al., 2019).

Considering consanguinity, a major factor of thalassemia propagation across the world and especially in those countries where cultural dominancy and family preferences are apex phenomenon (Waheed, Fisher, Awofeso, & Stanley, 2016). It has been found that close family linkages and traditional family ties destined individuals to fuse with their familial customs and consanguinity is one of those traditional factors (Abdulhadi, 2018). As a result among other genetic disorders, thalassemia is also very common in these vicinities where cousin marriages are preferred (Nadkarni, Phanasgaonkar, Colah, Mohanty, & Ghosh, 2008). Birth incidences of many genetic disorders, including thalassemia occur due to consanguineous marriages. The growing rate of these cases is difficult to end because cousin marriages are encouraged by local cultural and traditional patterns. Substantial persuasion of endogamy, caste system, sectarian preferences and aboriginal customs impel peoples to prefer consanguine marriages; that result in many chronic genetic abnormalities like thalassemia (Premawardhena et al., 2019).

A study was conducted by (Agouzal, Arfaoui, Quyou, & Khattab, 2010) on consanguineous marriages in Morocco, and found that the overall prevalence of thalassemia was 66.22 % among consanguineous marriages while the rate among non-consanguineous was 47 %. The prevalence of beta thalassemia is especially high in those countries where there are close family marriages and people follow their cultural and religious values more extremely (Chawla, Singh, Lakkakula, & Vadlamudi, 2017; Hosoya, 2017).

Genetic disorders due to consanguineous marriages are not new, these have been studied and sought for many decades and were found momentous for inherited disease like thalassemia, mental abnormalities, down syndrome and intellectual disabilities (Al Arrayed, 1999; Al-Gazali, Hamamy, & Al-Arrayad, 2006). Studies conducted by (Abu-Libdeh & Teebi, 2010) in West Bank and Gaza included 130 families to find out the genetic aspects of beta thalassemia and it was found that 77.3 % of the couples having cousin marriages are found to be the carrier of same mutation. Another study conducted by (Ashfaq, Amanullah, Ashfaq, & Ormond, 2013), focusing the effects of

cousin marriages on genetic disorders among Qatari inhabitants, found a very significant relationship between adult genetic disorders and the cousin marriages.

It has been found by the researchers (Burki, Qayum, & Siddiqui, 1998; Siong, Au, & Leung, 2019) that parental consanguinity increased thalassemia twofold in the children. There is a probability that if thalassemic couples are found to be cousin, then recessive disorder has almost double tendency to occur in infants. The more frequent chances of having thalassemia have been reported among first cousins and then to the second cousins and relative. Plentiful empirical evidences are available to reveal the importance of consanguinity in prompting thalassemia, but a very alarming situation has been seen among the first cousins (M. S. Khan, Ahmed, Khan, Mushtaq, & Wasim, 2015).

Numerous studies (Cremonini, Westerheijden, & Enders, 2009; Katz, Lazarsfeld, & Roper, 2017; Nutini & Bell, 2019) included cultural and religious aspects as major variables to reckon their influence on individual's choices and decisions about his life. It has been found by (Adly & Ebeid, 2015) that these factors (religious and social) have a greater influence on individual choices and preferences of medication and treatment of beta thalassemia major. Although, Islam allows abortion with certain conditions (if any disease can cause death of the mother or neonatal) and Muslim community can follow the certain instructions, that are very clear in *Fatwa* (before 120 days of gestation, a fetus can be terminated in case of having any chronic illness, that might cause death or any other serious complication for mother or new born) (Iqbal, Habib, & Amer, 2019). Having clear religious instructions, nobody should blame the true spirit of any religion including Islam, for the treatment of any chronic disease. These are only myths and synthesis that blame religious true spirit and retain individuals to choose scientific ways to handle such chronic ailments (Alkali, binti Mohd, Hak, & Soh, 2015).

It is evident from different studies (Gulzar et al., 2019; Sher, Bussmann, Hart, & de Boer, 2016) that in Pakistan, majority of the people living in rural areas and following meticulously their traditional norms and beliefs in every sphere of life generally and in treatment of diseases particularly. It has been found by (Arif et al., 2008) that majority of the people are following the same kind of religious practices and customs in the treatment of their thalassemic child Several studies have been

conducted to find out the factors linked with the attitude of couples regarding prenatal diagnosis of thalassemia.

Researchers (Cope, Garrett, Gregory, & Ashley-Koch, 2015) found that prenatal diagnosis are strongly related to religious convictions. Muslim couples have been shown reluctant to opt and follow prenatal diagnosis on the basis of religious grounds (S. Ahmed, Atkin, Hewison, & Green, 2006; Tan et al., 2010). Similarly, termination of pregnancy is also considered a taboo among families and couples due to many religious, moral, cultural and traditional values, generally followed by the families and communities where these couples are residing, or they are the members of (Arousell & Carlbom, 2016).

It has been found by canvassers (Ebrahim et al., 2019), that attitude of couples and families toward the termination of pregnancies with beta thalassemia major or minor is highly associated with different beliefs and practices. Due to religious constrains and ignorance of the consequences of the disease, people stick themselves to the notion that termination of the fetus will result the curse of God or even it would be a murder. Though these kinds of emotional beliefs and practices are difficult to exterminate from the lives of people but with the implementation of different educational and awareness programs people can be secured to stop the carriers of thalassemia along with the other chronic diseases (Kim & Tridane, 2017).

Many individuals and collective decrees -related to treatment, understanding, antidotes and consequences of thalassemia produce different social, cultural and religious practices, which later on these practices become the risk factors of thalassemia (Zhong et al., 2018). Families handling dexterities, disease management and adaptation approaches are shaped by social, cultural and religious practices (Ishfaq, 2015). As found by (Chong et al., 2019), support provided by religion, society and community members, including family, relatives and friends is the main factor, that helps parents to assuage the problems of sickle cell disease or thalassemia. The perceived importance of social, cultural and religious factors altering people's opinion regarding treatment of thalassemia in not universal and it varies for countries and families based on indigenous practices and acquaintance. The importance of cultural and religious factors cannot be ignored in diagnosis and treatment of

thalassemia especially in traditional societies, where families and couples are more ardent toward their religious beliefs (Hossain et al., 2017).

Distinctive studies have been conducted by (Moudi, Phanodi, Ansari, & Zohour, 2018; Renani, Dashtbozorgi, Papi, Navah, & Latifi, 2016) on socio-cultural and moral aspects of beta thalassemia major, and it has been found that these aspects are vital to ensure the universal standards of genetic services and women health reproduction, but they become leading snags when followed traditional and strict norms. Albeit, these factors are evident to explore the chronic impact of thalassemia over the lives of patients and their families but there are rare evidences to explore the effect of beta thalassemia major on decision making, gender roles and health literacy at the time of marriages, pregnancies and prenatal screening (Seven, Paşalak, Sahin, & Akyuz, 2019). Fear of stigmatization and social exclusion are significant associated factors and results of beta thalassemia and require rigorous attention to be overpowered (Kyriakides, 2016).

In case of Pakistan the higher ratio of the disease is found in children due to cousin marriages and large population size (Uddin et al., 2017). It is estimated that, 400 cases of blood transfusion dependents are born per year in Pakistan and the regions identified are Sindh, Baluchistan and KPK. The reason identified behind the presence of such a huge number of patients in these regions is low incom, cousin marriages and lack of awareness. As (Afroze et al., 2016) conducted a caste-based analysis of the disease and the results showed that the highest percentage was seen in *Rajputs, Arians, Sheiks, Pathans, Jats, Khokars* and lastly in *Baloachs*. A common contributor for all the castes is the tradition of consanguineous marriages.

### 2.2.3 Disease Allied Risk Factors of Beta Thalassemia Major

Medical negligence, lack of professional support from hospital staff, doctors and blood donation are all factors for which thalassemia is propagating worldwide and in Pakistan. As reported by (Surapon, 2011), during clinical practices doctors face numerous breaches to manage the negligence of supporting staff, basic health units and specifically the parents, who don't care about the premarital and prenatal measures of thalassemia's diagnosis. Among others; one major challenge is to decide

about the patient's care and management of disease because like other sickle cell diseases, thalassemia also required a strenuous management (Benz & Angelucci, 2018; Giusti, Pinto, Forni, & Pilotto, 2016). The second highlighted challenge is the availability of technologies, medicines and therapeutic apparatuses because most of these elements are out of the reach of families of developing countries (Sultana et al., 2016). The third challenge is very chronic because its negligence leads to serious troubles for patients and their families and that is the knowledge of doctors and health practitioners (Heidari, Ahmadi, Solati, & Habibian, 2018b).

Along with medical and paramedical staff, parents and families are required to have information about the carrier detection, propagation and treatment of thalassemia. It has been found in heath literature, that enough information related to disease management and patient's care is required by the parents and families (Ahmed et al., 2019; Hummelinck & Pollock, 2006). There is an unplumbed need to educate caregivers and provide them enough support, because it is directly related to patient's health (Hisam, 2018). This support includes technical knowledge and information as well social, financial, psychological and moral support to the patients and their families (Baraz, Miladinia, & Mosavinouri, 2016).

Thalassemia engrains because it requires an effective and expensive management mechanism, in the form of blood transfusion and bone marrow transplantation (Faulkner, 2018), while these methods a very expensive and required a reflective procedural knowledge and skills. Other than social and psychological therapies and interventions in the form of counselling, guidance and awareness, these epidemiological measures are obligatory to cure and manage thalassemia (Tanveer, Masud, & Butt, 2018).

The study conducted by Khan and his colleagues in 2017, concluded that thalassemia carriers are also linked with other diseases and deficiencies, for instance, many patients showed vitamin D deficiency and many thalassemia patients were linked with thyroid dysfunctions, which are the signs of poor health structure in a society. Socio-economic status of people plays a major role because everyone in Pakistan has no access to a better health care (Khan, Hamzulla, Shah, & Khan, 2017).

## 2.3 Psychosocial Burden of Beta Thalassemia Major

The expenses and the management of the patients go beyond the control of the standards of the therapeutic care and the incapability to deal with the issue has also increased the social and psychological problems (Khanna, Prabhakaran, Patel, Ganjiwale, & Nimbalkar, 2015). Due to the illness of a single child, whole family faces stigmatization and social isolation, especially in illiterate and traditional areas of the Asian region. Illness of child creates anxiety and depression, not only for the parents but the whole family suffers such kind of circumstances (Koutelekos & Haliasos, 2013).

In many rural areas of Pakistan, due to traditional cultural practices, blind religious beliefs, ignorance and pressure from family and society, parents of thalassemic child face many problems especially in the treatment of their sick child (Aziz, Sadaf, & Kanwal, 2012). Most of the time, it has been reported that couple divorced and stigmatized women due to the illness of a thalassemic child/children (Dadipoor et al., 2015). Beside medical and professional support, these people required awareness, acceptance, recognition and removal of social pressure to manage the disease and deal with these decisive circumstances.

Thalassemic children face substantial loss of their education because of regular blood transfusion and hospitalization meanwhile it becomes essential for parents to fall back on their families, friends and relatives for economic, social and psychological support. Previous studies have shown that social capital and strong ties among communities have a positive impact on children's health, development and wellbeing (Palanisamy, Kosalram, & Gopichandran, 2017). In case of thalassemia these strong ties and enriched social capital is very helpful to manage the disease very effectively which can be seen among rural communities of Pakistan but parents of thalassemic children are facing stigmatization and social isolation because of lack of education and awareness of causes and propagation of thalassemia (Hussein, Weng, Kai, Kleijnen, & Qureshi, 2018).

Researchers (Pouraboli et al., 2017) identified two important factors which parents of thalassemic children experience, one is concealment and another is censure because parents of thalassemic children suffer from cultural poverty of the society

(Chattopadhyay, 2006). People usually treat them unwisely because of the false beliefs and superstitions about the causes and propagation of thalassemia which results in social stigmatization and social disconnection of parents with normal community and even, they face isolation in their interaction with relatives and friends.

As (Fung, 2010) found that, due to illness such as thalassemia and dwindling health with such kind of a disease, most of the people become distrait for their future lives. Lack of decision making and mental health problems i.e. depression, anxiety and sleeplessness has also been seen very common among patients and their families, (especially parents). A close attention of researchers, policy makers and health experts is required to highlight these psychological and emotional problems faced by the parents and families and the responsibility lies on psychologists and psycho-therapists to provide knowledge and counseling to the sufferers. It has been seen that teenagers required more psychological attention, moral support and care to meet the persistent abnormalities due to genetic deriders like thalassemia (Koutelekos & Haliasos, 2013). These disturbances lead further to the problems, aligned with social interaction, participation in social activities and exclusively the problems related to psychological malfunctioning, as an impact of thalassemia (Zakiyah, Mediani, & Mardiah, 2018; Zani, Di Palma, & Vullo, 1995).

In their study (Hakeem, Mousa, Moustafa, Mahgoob, & Hassan, 2018) found that likewise other genetic disorders, thalassemia also affects the entire life of a patient because it damages the intellectual abilities, creative skills and physical workout. Due to restrictions of body movements, a patient is bound for certain activities and this results in a severe loss of educational activities, mental capabilities, mental disorders and de-functionalization of psychological, social, emotional and physiological adjustment (Sadeghloo, Shamsaee, Hesari, Akhondzadeh, & Hojjati, 2019).

In another study, conducted by (Alizadeh, Chehrzad, Mirzaee, & Leyli, 2019) it has been found that due to thalassemia; the life of patient's parents also writhes because parents are required to provide intensive care, appropriate medication and consistent hospitalization to their children (s). This charge them a substantial financial encumbrance and as a result they suffer from mental stress (Paria, Halder, Nayek, Mukhopadhyay, & Ghosh, 2016), psychological disorder (Anum & Dasti,

2016), emotional burden (Mohamed et al., 2017), social pressure (Zaheer et al., 2016), stigmatization and social isolation (Kumar et al., 2019), emotional shocks and severe frustration (Yohani et al., 2019). All these factors have an analogous effect on the personalities of sick children and the quality of life of rest of their siblings (Pelentsov, Laws, & Esterman, 2015). It has also been found that severity of all these factors is high in traditional societies as compared to urban areas because of low level of education, health literacy and awareness of thalassemia. Meanwhile, the propagation of thalassemia is also extreme in traditional and less educated areas (Adam, Afifi, Thomas, Magdy, & El-Kamah, 2017). All these social and psychological problems emerged due to non-scientific understanding of thalassemia and domination of traditional religious beliefs, spiritual myths and indigenous fables related to the cause and treatment of thalassemia (Zhong et al., 2018).

Children with beta thalassemia major exhibited the problems of low self-esteem and self-image, social and psychological maladjustment (Khamoushi et al., 2015; Mettananda et al., 2019) and excruciation due to their physical anomalies (Tomaj et al., 2016). The literature reviewed showed that due to their social isolation and school truancies these children feel worthless (Inamdar, Inamdar, & Gangrade, 2015), pessimistic (Gan, Lum, Wakefield, Nandakumar, & Fardell, 2017) and a deep-rooted sense of powerlessness has also been observed then. But these situations vary within families across the world because of diverse sociocultural norms and familial practices. In the context of Pakistan, the previous studies revealed psychosocial maladjustment of thalassemic children and parents also face the burden of social and psychological adjustment along with monetary problems (Saqib & Ansar, 2017; Zaheer et al., 2016). It has been seen by (Gamayani et al., 2017; Kumar, Singh, Khullar, & Arora, 2018) that school performance of a child can strongly be affected by thalassemia, because blood transfusion and medical checkup led them to the school absenteeism. Frequent visits to the doctors and medical centers are considered to be the main barrier for their (children) school performance and this also causes frustration among them (Liem, Gilgour, Pelligra, Mason, & Thompson, 2011).

Studies of (Khanna et al., 2015; Sharma, Seth, Jawade, Ingale, & Setia, 2017) investigated that, patients of beta thalassemia major endure serious problems of sociopsychological disorders which result in low quality of life of the entire family.

Furthermore, it was reviewed by (Safdar et al., 2017; Suwanthol, Sangpaypan, Naknum, & Sanpakit, 2017) that excessive economic burden on parents and families due to regular medications, blood transfusions and therapies results in poor quality of life as compare to other members of their communities. A persistent effect of thalassemia had also been reported by (Mehr et al., 2019) on individual identities of patients and their families and therefore, the issue such as identity crisis, stigmatization and lethargy are common among these sufferers. These problems further lead to the cultural differences and complications in the stages of development of personality and adulthood (Bafna, Bafna, Sampagar, & Rupavataram, 2018). Like other chronic genetic disorders, thalassemia imposes the financial burden on families and parents must manage the expenses of regular treatment of their children. Consequently, they (parents) seek help from their relatives, friends and community members and sacrifice their self-esteem and self-image for not gaining any supportive response (Renani et al., 2016).

In their study (Liem et al., 2011) found that among Asian families, living in United State of America, the risk factors of thalassemia are associated with low quality of life while, emotional and social wellbeing was also aggravated due to thalassemia. As suggested by these researchers, it is requisite to include social, cultural and economic factors as major risk factors for the propagation of thalassemia because lower socio-economic status is strongly associated with thalassemia (Mediani, Tiara, & Mardhiyah, 2019; Ogaz, 2017).

Continual academic studies of (Caocci et al., 2012; Mednick et al., 2010) have anticipated demographic factors, such age and gender to expose the disparities and impact of thalassemia on social and psychological wellbeing of patients. It was found by the authors (Huda Gharaibeh, Amarneh, & Zamzam, 2009) that negative feelings, emotional disruptions, depression and anxiety is significantly high in male patients as compared to females. Due to absence of social, emotional and moral support for patients of thalassemia and their families these deriders are frequent in developing countries (Raman, Prakash, & D'Souza, 2019). Furthermore, review of literature explored that thalassemia imposes physical, emotional, psychological and social burden on caregivers (Saqib & Ansar, 2017) and that further creates high level of

apprehension and depression among patients and caregivers (Shamsi, Amiri, Ebadi, & Ghaderi, 2017).

There exists a considerable body of literature on social and psychological problems, associated with thalassemia and the psychosocial burden of beta thalassemia major (Adam et al., 2017; Kumaravel, Jagannathan, Balaji, Karthick, & Pugalendhiraja, 2016; Shahraki-vahed, Firouzkouhi, Abdollahimohammad, & Ghalgaie, 2017). It was found by these researchers, that parents, caregivers and children by themselves are envisioned to face psychological disorders, economic and social pressure and financial crisis due to thalassemia because it heavily cost financial burden along with psychological and social disorders and maladjustment of the parents, families and children among the society. An exploratory study was conducted by (Tsiantis et al., 1996) and they highlighted the problems of social and psychological adjustment among children and families of beta thalassemia major. Their assessment was based on problems of adjustment and prevalence of thalassemia based on father's educational level and medical complications. Furthermore, these researchers found that poor family adjustment and high level of prevalence of thalassemia is because of low education and poor medical facilities. Along with many other problems, it had also been uncovered; that patients of beta thalassemia major experience memory stumps (Lubis & Yunir, 2018) and attention span maladies (Elhabiby, ElSalakawy, Khalil, Hassan, & Hjislam, 2016). Different studies found that thalassemic children have insufficiencies of conceptions (Centauri et al., 2017) and unable to visualize and construct the artisan ideas (Mohamadian, Bagheri, Hashemi, & Sani, 2018).

A study was conducted by (Shaligram, Girimaji, & Chaturvedi, 2007) in India and revealed that while measuring the psychosocial effect of thalassemia on parents of sick children, majority of the respondents were not getting social and psychological support from their family members, resulting their social withdrawal in community participation and lack of family management. Researchers revealed that parents were facing anxiety in India. These problems lead to the chronic psychological illness and a permanent social seclusion. Due to long term complications of disease, not only the parents and children face psychological and emotional stress but the whole family

suffers the immense social, psychological and financial pressure (Khanna et al., 2015).

It is evident that due to thalassemia, parents and children suffer from increased level of psychological, social, mental and behavioural problems, because thalassemia affects very enduringly the entire life of a child and his/her parents. A study was conducted by (Aydinok, Erermis, Bukusoglu, Yilmaz, & Solak, 2005) and they found that patients and their parents, having worse experience of thalassemia are required instantaneous psychiatric support and therapies to cope up their mental stress, disorder and behavioural complications.

### 2.4 Consanguinity and Beta Thalassemia Major

Recurrent analyses of genetic disorders revealed that consanguineous marriages are the main factor that ensures vulnerability to many genetic diseases (Bai, Nasir, Ahmed, Malik, & Arif, 2019; Thiyagarajan, Bhattacharya, Sharma, Srivastava, & Dhar, 2019) and due to repeated cousin marriages, Pakistani population is at risk to increase the tendency of chronic genetic abnormalities including thalassemia (Khalid, Noreen, Qureshi, & Mahesar, 2019). As compared to many developed countries the genetic disorders due to consanguinity are rare while the situation in developing countries and in traditional societies is inverse (Romdhane et al., 2019). Researchers (Mokhtar, Adly, Alfy, Tawfik, & Khairy, 2010) have found that along with consanguinity, there are many other factors such as; socio-economic status, migration, pre and post-natal services which also prompt genetic disorder in low income countries.

A collaborative research was conducted by Pakistani researchers (Munir, Iqbal, Jamil, & Muhammad, 2013) in an industrial city of the country (Faisalabad), to measure the connotation of thalassemia and cousin marriages and they found that rate of beta thalassemia is recorded as highest in first cousins (69.66%) suggested that among very significant factors that increases the frequency of thalassemia, two are very important 1. low income status and 2. lack of awareness (Sengupta, 2008). Among other triggering social factors, consanguinity and marriages within the same

cast and ethnicity are very imperative to increase the frequency of thalassemia (S. Khan, 2018; Zafar et al., 2018).

Researchers (Zamani, Khazaei, & Rezaeian, 2015) found that a general assumption set by many healthcare professionals as well as documented literature that focuses on cousin marriages and gives direction to determine the rate and frequency of carriers of thalassemia is true but a very important factor, particularly in the context of South Asian women is often neglected by the academicians and professionals. The major contributory factor is the poorer quality of antenatal care that results in poor birth outcome and this causes many deficiencies in the protein level which is necessary for healthy women and their babies. As a result, these women face many chronic diseases, such as anemia and the babies are more vulnerable to the genetic disorders like thalassemia.

Consanguinity is playing a vital role in propagation of thalassemia, especially in those countries, where people sternly follow endogamous marriage practices (Hamali & Saboor, 2019; Safdar et al., 2017). Caste, ethnic practices and different cultural restrictions also compel people to marry with cousins (Chordiya, Katewa, Sharma, Deopa, & Katewa, 2018). Consanguinity and thalassemia nexus are not new or emerging reason, people only need awareness and education to know the importance of screening, counseling and results of repeated marriages within the families because many studies have been conducted across the world and researchers found a very significant role of consanguinity to propagate thalassemia (Bittles, 2001; Laghmich et al., 2019; Mansour et al., 2010).

### 2.5 Prenatal Diagnosis and Premarital Screening

Population screening programs aim to identify the carriers and vulnerability of individuals to any genetic disorder. The diminution of reproductive issues and many genetic disorders including thalassemia is possible, by providing and adopting the methods of prenatal, postnatal and premarital screening. Among others, the two countries like Saudi Arabia and Iran have obligation for couples to go for carrier screening (Al-Suliman, 2006; AlHamdan, AlMazrou, AlSwaidi, & Choudhry, 2007; Hashemizadeh & Noori, 2013), while ensuring the confidentiality, people are required

and in case of carrier detection, they are given choices to continue their relations or not. Prenatal, post-natal and pre-marriage screening programs are the effective ways to reduce genetic abnormalities across the world and especially in developing countries (Mukhopadhyay et al., 2015).

Many countries like Iran, Cyprus, Greek and Italy started campaigns and conducted awareness sessions to raise the level of cognizance among families and couples about prevention and expansion of thalassemia. In Pakistan, there is an appalling need to start such programs in collaboration with educational and religious institutions, media and NGOs (Arif et al., 2008; Asif & Hassan, 2016). The Beta thalassemia presence can be seen at a large scale in the Pakistanis families, thus to provide with the genetic counseling and prenatal diagnosis during early pregnancy it is important to analyze the relevant changes in the DNA (Ikram, 2019).

It has been considered by (Kanwal, Bukhari, & Perveen, 2017) that in Pakistan, there is not so much attention given to the prenatal diagnosis (PND), exclusively of beta-thalassemia major. This is because of many of social issues within the society along with the financial issues of the families. A significant number of the couples ignore the diagnosis because it's not free of cost. But as the news spread of the free of cost prenatal diagnosis (PND) services, more couples are registering for the test (Naseem, Ghazanfar, & Rashid, 2016).

According to (Rahgoi, Sojoodi, Khoshknab, Rahgozar, & Shahshahani, 2019), parent's education, carrier counseling and awareness sessions are considered as helpful techniques to overawe the prorogation of thalassemia but families and parents of thalassemic children are reluctant to follow these effective strategies to control thalassemia propagation in Pakistan (Ishfaq, 2015). Without parental consent and intention, it is difficult to prevent thalassemia across the world because families have their own choices and practices of marriages. In this regard the effective ways for thalassemia prevention are; awareness, genetic counseling, blood screening and parental education regarding pre and post-natal screening (Rudra et al., 2016).

Parents need to get aware of thalassemia and its prevalence through genes and how marriage is a factor. Thalassemia transmits through genes and it has been reported by (Khodaei, Farbod, Zarif, Nateghi, & Saeidi, 2013) that nearly 3% of the

entire world's population contains the genes of beta thalassemia. However, in Pakistan around 7% thalassemia homozygotes are born every year (Hassan et al., 2019). The prevention and the treatment of such kind of disease is necessary all around the world. A study was conducted by (Kosaryan et al., 2018) in Iran to compare the treatment and prevention cost of beta thalassemia major, through National Thalassemia Prevention Program and they found that cost of the disease rises every year to ensure the reduce prevalence of beta thalassemia major. The National Thalassemia Prevention Program in Iran demonstrates that if the prevention of the diseases can be happened in the early stages then a lot of finance can be saved.

It has been found by (Biswas et al., 2019) that, educational programs and awareness sessions are important tools for the prevention of beta thalassemia major, because the prevention strategies mainly consist the different educational and awareness programs that include the right information for the screening of the affected families. The appropriate screening and detection of the disease and timely counseling need serious attention of parents and families (Lou et al., 2017). These sessions can be planned for vulnerable couples and families (carriers of thalassemia and families with high rate of cousin marriages) to reduce the morbidities of beta thalassemia major (Akers, Howard, & Ford, 2017; Henneman et al., 2016).

Studies conducted by (Ghafoor, 2016; Raza, Farooqi, Mubeen, Shoaib, & Jabeen, 2016; Saleem, Ghafoor, Anwar, & Saleem, 2016) in Pakistan and it had been reconnoitered that, proper prevention of the thalassemia and the counseling can save a lot at the early stages. Although there is no cure of the disease but prevention on time is possible. All around the world mostly the thalassemia affected parents are from the low-income groups. The mentioned study (Saleem et al., 2016) was conducted in the thalassemia center where the parents belong to the low-income group come and avail free medicines, therapies and health awareness sessions, which helps in the prevention.

### 2.6 Parental Knowledge and Education Regarding Beta Thalassemia Major

Knowledge, education, awareness about causes and information about the preventive measures of thalassemia play a significant role for the propagation and control of the disease (Zaman & Salahuddin, 2006) measured a connotation between

the level of education of parents and caregivers and the prevalence of thalassemia and it had been found that educated parents and caregivers tend to worry more about career screening and keen to learn about genetic counselling as compared to the respondents with low level of education.

In their study, (Arif et al., 2008), detailed that in metropolitan and densely populated Karachi city, only 15% of the respondents were aware of the causes of beta thalassemia major. They found that only 12 % of the parents had a knowledge about consanguinity being the prominent risk factor of thalassemia. These figures demonstrate that families having little knowledge about risk factors and propagation of thalassemia are more susceptible to the disease. Parental knowledge and education of thalassemia, its propagation and prevention need to be clear and vibrant because this could be the possible way to reduce social and cultural risk factors and eventually propagation of thalassemia will be reduced (Thiyagarajan, Bagavandas, & Kosalram, 2019).

Majority of the Pakistani parents are living in rural areas where educational and awareness seminars or sessions are difficult to be provided by educationist and health experts, furthermore, low level of education about thalassemia, increased their vulnerability to such chronic disorder (Ishaq, Hasnain Abid, Akhtar, & Mahmood, 2012; Majeed, Akhter, Nayyar, Riaz, & Mannan, 2013; Riaz et al., 2011). It has been premeditated by (Coifman, Ross, Kleinert, & Giardina, 2014) that higher literacy rate, poses a stronger impact on parental behavior to cope and manage thalassemia. Increased literacy rate has also been found to have a positive impact on prenatal diagnosis and premarital screening, which enables the couple to effectively manage thalassemia and to opt the preventive methods, such as termination of pregnancy.

Parents can cope up their behavioural and mental problems and can reduce their stress level by increasing their level of knowledge and awareness. As suggested and found by (Ali & Malik, 2015), that educated and well aware parents can better understand and manage the emotional and psycho-social burden of thalassemia. Increased knowledge of parents is very helpful in detection and implantation of preventive measures of thalassemia, because the diagnosis and prevention of disease highly depends upon parental practices. It has been found by (Nikam, SB, Dama, & Saraf, 2012) that educated parents took preventive measures more seriously as

compared to uneducated parents, together with the high rate of illiteracy in country like Pakistan majority of the people (82.4%) are not aware of the preventive measures of thalassemia and many other health problems (W. Ahmed, Shaikh, Soomro, Qazi, & Soomro, 2018).

Distinctive educational programs and counselling sessions with couples and families are required, to physique a family centered enablement model to control and prevent thalassemia, and to enrich parental knowledge, sense of control over the disease, emotional disorders and adjustment of psychological malfunctioning (Shawkat, Jwaid, & Awad, 2019). Furthermore, these strategies develop the ability among the couples to make decisions about the lives of the sick children and families and to provide care. Researches have proven that family functioning and family empowerment are the two major blocks, that enable couples, parents and families to build social, psychological and emotional fortes to effectively allay thalassemia (Borimnejad, Parvizy, Haghaani, & Sheibani, 2018).

### 2.7 Treatment of Beta Thalassemia Major

Treatment of beta thalassemia requires massive economic resources in the form of medicalization, hospitalization and transfusion of blood on regular basis that results in economic burden on patient's families (Moirangthem & Phadke, 2018). Poor families are more vulnerable to the economic crises because of costly treatment of the disease. Like other genetic abnormalities, beta thalassemia major also required a laborious financial tussle, especially for low income families because of regular blood transfusion and medication. An average 100 USD cost, for the treatment of thalassemia have been estimated by (Ullah et al., 2016) in Pakistan. The expenses on blood transfusion and chelation therapies are not manageable by the parents, without the support of NGOs, welfare organizations, blood donors, volunteers and the government as well.

Despite several options for treatment, there is no clear guideline for its management. Therefore, there is a need for further study of the disease to better understand its causes and options to cure it. For the present time the two main issues have been the managing of thalassemia in adults and developments of complication in

the youth. For adults; with thalassemia following recommendations have been put forward.

- Transfusion at early stage and iron chelation therapy
- Regular follow up
- Regular follow up of liver iron concentration
- Avoid smoking, and use of oral contraceptives (Taher, Isma'eel, & Cappellini, 2006).

Thalassemic children are required to have proper vaccinations against other serious diseases such as; hepatitis A and B, especially when they are receiving blood transfusion as a part of their treatment. Parents and caregivers of thalassemic children are required to be persistently active and up to date with the vaccination schedule of children because it is an important part of thalassemia treatment. Children with thalassemia are considered at high risk for certain infections, mainly if they have had their spleen removed (Surani, Shah, & Sinha, 2018).

Treatment of beta thalassemia major includes a regular cost and it is not easy to bear hospital expenditure especially for the parents living with low income and have limited resources (Roy & Chatterjee, 2007). In their study (Ward et al., 2002) uncovered the direct and indirect financial expenditures of thalassemia and its effects on parental lives. They found that due to huge financial burden, families and parents face psychological anguishes, physical and social problems of adjustment and compatibilities.

Another study was conducted by (HF Gharaibeh & Gharaibeh, 2012) to find out the link between wellbeing and school performance of students, along with quality of life of thalassemic children. Those researchers found that due to dejected quality of life of thalassemia children, there is a negative effect on their (children's) physical wellbeing and on school performance too.

It has been found by the researchers (Raja & Janjua, 2008) that in Pakistan, approximately 60 percent of the population lives in rural area with unviability of health facilities including doctors, medicines and laboratories. These rural areas lack modern technologies and training facilities for doctors and paramedical staff. Due to

the absence of basic health facilities, and quick access to latest clinical information sources, families and couples are left over to find out the identification and remedial measures of thalassemia. In their research (Bishop et al., 2008) pointed out that most of the studies carried out on doctor's information, have ignored the concepts of unrecognized needs and even the doctors working in remote areas are not keenly interested to find out the innovative ideas and technologies in the field of health and illness. (Samarbafzadeh, Makvandi, Zandian, & Pedram, 2007).

### 2.8 Management of Beta Thalassemia Major

Beta thalassemia major is complicated by the iron excess (which causes damage of cellular and tissues, that ultimately affects major organs of the body e.g. heart, liver and the endocrine glands) (Gupta, Singh, Utreja, & Verma, 2016). Thalassemia major's patient does not survive beyond 5-10 years and in some case, it went to the age of 13 or above, resulting the death of patients (Unissa et al., 2018). The effective management of thalassemia is imperative because this not only improve the quality of life of parents and family members of thalassemic child but enable their parents to manage the social anticipations, related to education and employment of the parents (Sulastri, Gatot, Rustina, & Darmawan, 2018).

Management of thalassemia is very important in the form of blood transfusion and chelation therapy because patients are required pertinent medical care and monitoring on regular basis, from a hematologist (a medical specialist who treats disorders of the blood) or a doctor who is specialized in treating the patients with thalassemia (Cappellini, Porter, Viprakasit, & Taher, 2018). Similarly, parents ought to be more cognizant about diet and exercise of their thalassemic children because they (children) need very precautionary diets and exercises and parents need to be alert for their daily activities (Malako, Teshome, & Belachew, 2018). Parents are required genetic counseling because this is considered as an effective process of communication for providing information and support to individuals and families (Alderfer et al., 2017). In case of Pakistan due to cultural sensitization for genetic counseling, majority of the people ignored it (Zhong et al., 2018). While genetic counselling must be and is important at diagnosis, during adolescence, prior to and after any genetic testing and prior to pregnancy or earlier in the pregnancy (Theodoridou et al., 2018).

Many thalassemia foundations and centers are working in Pakistan for blood provision and working on screening, counseling and creating awareness among the common people (Tanveer et al., 2018) but people living in rural areas of Pakistan, are facing serious problems regarding the treatment of disease because of low income, unavailability of services and lack of knowledge. Different factors such as: low level of literacy, lack of knowledge, traditional health care practices, lack of financial resources are considered to be the major factors for identification and treatment of thalassemia in Pakistan (Arif et al., 2008). Unavailability of health facilities, for instance; screening facilities, genetic counseling are causing higher propagation of thalassemia in Pakistan (Ghaffar, Zaidi, Qureshi, & Hafeez, 2013). Family caregivers often lack the information and advanced effective treatment, so it becomes challenging for the management of thalassemia (M. Maheri, Rohban, Sadeghi, & Joveini, 2019). Deficient information from health professionals and community services providers, to the families is another propagating factor of thalassemia (Heidari, Ahmadi, Solati, & Habibian, 2018a).

It has been found by (Lal et al., 2018), that the effective management and prevention of beta thalassemia required standardization of preventive practices especially for genetic counselling (literature should be up to date and conceptual clarity for people, with low level of education) and for laboratories as well. It is also evident that the disavowal of couples, for screening and counselling is highly influenced by ethical and moral traits (Chakravorty & Dick, 2019), so there is an obligation of moral standardization for the effective management and implantation of preventive programs (Moudi, Phanodi, & Vedadhir, 2019).

It has been extracted from the glossary of World Health Organization that, health education is not reduced to the propagation of basic information and healthy practices but too fostering the motivations, skills and confidence of individuals as well (Smith, Tang, & Nutbeam, 2006). Public is required to be provided with the information about social, economic and developmental conditions for effective management of thalassemia (Manglani & Kini, 2017).

Screening is very important for the prevention of thalassemia, whatever the categorization of thalassemia might be (mass screening/target screening), it has a very persuasive importance in prevention and management of thalassemia (Cao, Rosatelli, Monni, & Galanello, 2002). Low level of knowledge and education of masses/individuals about the importance of thalassemia screening is the major risk factor of disease. Prospective screening is more important as compared to the retrospective screening, particularly in case of thalassemia and many other genetic diseases (Cao & Kan, 2013). In many parts of the world, research and innovation in the field of medicine and technology has enabled people to effectively manage thalassemia and to live a relaxed life in comparison to the developing world, because of global health disparities in healthcare and in adaptation of technology (Levine, 2018).

Health literacy is the major outcome of health education and it strongly depends on basic education and knowledge of healthy practices to avoid the illness (Caruso et al., 2018). According to the Pakistan Bureau of Statistics, 1/3 of the total population has attained primary education while only 20% attained middle school education (Khattak, Iqbal, Abdullah, & Chowhan, 2018; Nishtar et al., 2013; Sabzwari, 2017). The data of literacy revealed that in terms of percentages, Pakistani women lagged behind men (Sabzwari, 2017) .

# Review of literature (Theoretical Elucidation)

This segment of research focuses on specific models of health and illness, posed by many researchers across the world. The sequential variations and critic of each model is summed up by the researcher and the relevant theoretical orientations of beta thalassemia major have been tried to incorporate in this part of the study.

## 2.9 Models of Health and Illness

A number of health models are developed by sociologist, psychologist and health professionals to expose the cause and consequences of health and illness. These theorists and researchers intended to explain social (Hughes & Paterson, 2006; Shakespeare & Watson, 1997), cultural (Jegede, 2002; Wong & King, 2007), psychological, ecological (Coreil, 2010; Murray & Chamberlain, 1999; Willig, 2000), as well as biomedical explanations of health and illness (Farre & Rapley, 2017).

The prominent approaches in the field of health and illness gained a significant influence of biomedical and social vindications. Although these two approaches have significant differences with each other in terms of their examination, explanation and interpretation of disease and health. According to World Health Organization (WHO), the state of health is defined as *exhibiting physical fitness along with mental reasoning* (Organization, 2005). In views of (Blaxter, 2004),

> *if a person can easily perform his/her daily activities e.g. performing household tasks and managing and participating in social gatherings, the state of robustness reveals he/she is healthy* (p.122).

The most commonly found scientific and theoretical constructions of health are listed below.

### 2.9.1 Biomedical Model of Health and Illness

From most of the earlier explanations of health and illness in medical sociology; biomedical model is instigator (Alonso, 2004). The model remained highly appreciated and followed by social and medical scientists during 1900s in Western Societies. In the view of biomedical model of health and illness, the complete functioning and malfunctioning of a body depends upon the biological explanations. The estimation of this model suggested that fixing biological issues by treating people

through surgeries, using medicines and drugs for different therapies can ensure health and diminish illness (Minaire, 1992). Later on, with the emergence of different environmental, social and psychological explanations of health and illness, this model was criticized by many researchers (Adibi, 2014; Fertonani, de Pires, Biff, & dos Anjos Scherer, 2015; Smart, 2006).

In many of the researchers' view, the absence of illness is defined as health (Calnan, 1987; Hughner & Kleine, 2004; R. Williams, 1983), however, many others perceive health as a relative phenomenon, that truly depends on the experiences and evaluations made by individuals and considered the state as health or not by means of their age, gender, social status and medical conditions. For those, who measured health on these lay perceptions, conceptualized health as only the absence of any physical problems which interfere their daily life activities (Bury, 2005).

The explanation made by (Baggott, 2004) about biomedical model depends on the physical functioning of an individual, if an individual is physically performing well he/she must be considered as healthy and the explanation of bad health can be described as the presence of any disease or illness, resulting physical malfunctioning due to any injury or infection. Bio-medical model ignores the social and psychological factors and keeps its focus on medical and biological changes which can be measured and defined in isolation. These biological and medical factors are failed to explain the complete state of health or illness because their elucidation depends on dysfunction of the organ or tissue of the body instated of overall condition of the patient (quality of life, wellbeing, adjustment and other social, psychological and mental aspects of health). The core of biomedical model entails a specific etiology e.g. every disease is/can be caused by the germs, bacteria or any kind of parasite (Seale, 2003).

An important illustration about biomedical model is based on its generic nature, which explains that each disease either viral, genetic or by any mean is based on the participles of universalism (Fischer, 2009). The approach ignores the cultural and social explanation and construction of health by believing on the biological agentic nature of health (Farmer et al., 2012). Denigration of this explanation is based on the idea of cultural variations to explain health, an illness and the changing definition of any disease with the passage of time. Usually, it can be comprehended

that what is seen as illness in any particular society, at the given period of time, depends on social and cultural contextualization (Naidoo & Wills, 2016).

Another approach of biomedicine is supported by the spat of measurable biotic variables and perceived illness; as a deviation from normal range. The psychological abnormalities and impacts are the counter arguments for this explanation because of varying nature of mental and psychological factors of emotional and psychological disorders (Engel, 1978). It is also believed by health sociologists and psychologists that; mental abnormalities and emotional disorders often remain unclear and cannot be generalized on the principles of normality. Medical scientists have realized that human behaviors are not universal, and illness or health cannot be perceived universally (Cockerham & Cockerham, 2014). It has also been realized by many researches of medical sciences, that human organism diverges in terms of structural and functional patterns and even the genetic variations remain un clear (Heylighen, 1999).

Having a strong belief on neutrality and objectivism, medical theories perceived individual as a product of biological progression. According to the biological naturalistic and objectivist; individuals have little control over their lives and on the construction of health (Crawford, 2006). The critics have carped this argument of neutrality because there are various dictating forces in terms of social, cultural and political conducts which shape and affect the human health and illness. Being unhealthy could be the result of these (social, political/administrative and cultural) factors and practices (Harley et al., 2011; S. J. Williams, 1995).

In contemporary debates of biomedicine and biomedical, it has been acknowledged by health experts and researchers that there are multiple approaches for the interpretation of health and illness, instead of having a robust deem on biological determinism and value of neutrality. These multiple approaches include; social and psychological causes, anxiety, depression, cultural practices and different environmental factors (Noar, Chabot, & Zimmerman, 2008). Current debates have incorporated the multidimensional approaches and their interrelatedness for the explanation and investigation of health and illness (Chenier, 1997). Biomedical model focuses on the quantification and treatment of any particular disease by means of only medications and clinical trials and due to this narrow and reductionist slant, it has

been highly criticized by the social and environmental scientists because this approach perceives health as only the absence of any indisposition (Alberts, Kirschner, Tilghman, & Varmus, 2014; Engel, 1977).

### 2.9.2 Social Model of Health

After the dominant criticism over biomedical model in 21st Century, the literature drew attention of health experts and medical scientists to integrate social causes for the exoneration of health and illness. Social narratives highlighted the cultural, religious, communal and familial practices, which shape the health of any individual and the practices to avoid ill health. The social conceptions included health and hygiene, marriage practices, psychological aspects and health disparities, other than focusing on medicines and clinical experiments, which were dominant in earlier model. The social model included the social and cultural variations for the construction of health and explained health-as a continuous process of individual, cultural and social practices (Fried et al., 2004). From Weberian explanation of *life chances and choices* (Cockerham, Abel, & Lüschen, 1993) to *sick role* of (Parsons, 1951), health and illness have been conceptualized differently across the world (Larson, 1999) and the universal agreement on the definition of health is still a debatable phenomenon (Zautra, Hall, & Murray, 2010) because the social and cultural practices and understandings are not identical.

The social model interprets biological mechanisms in terms of social causes and construct heath or being unhealthy is based on social explanations. The model contextualizes health as an amalgamation of social factors including socio-economic status, cultural practices, sanitation and hygiene. The proponent of social model (Anastasiou & Kauffman, 2011; Ghaemi, 2009) has accentuated to include social aspects e.g. poverty, awareness, parental practices, ethnic and cultural practices and religious beliefs to be cuddled for the understanding of health and illness. Social model also allowed mental and psychological factors to be given share in its core domain because these (mental and psychological) aspects either become causes or consequences of any disease (Kawachi & Berkman, 2001).

### 2.9.3 Bio-psychosocial Model

Another major theoretical and practical contribution by the researcher (Engel, 1978, 1981, 1982) in sociology of health and illness is bio-psychosocial model of

heath and illness. Bio-psychosocial model is known as the extension of the former model (biomedical). This model is also known as a multi-factorial approach of health and illness because it includes biological facets of health, along with psychological and social, incriminated with the health status of a patient (Alonso, 2004; Havelka, Despot Lučanin, & Lučanin, 2009). This multidimensional approach of health and illness is characterized based on; 1. biological factors (genes and chromosomes), 2. behavioral/psychological factors (life styles, stress management practices, health believes etc.) and 3. social factors (cultural, religious life histories, social relations and social support etc.). It includes; social, psychological and biological factors that influence health unlike biomedical model that only theorize, and measure health, illness and practices based on genetic and biological aspects.

### 2.9.4 Web of Causation

The major contribution by epidemiologist was to develop the web of causation approach also known as agent host model, which views health or illness as a direct result of an intricate set-up of interviewing risk factors between the agents, the host and the environment (Norell, 1984). This model includes the elements of environment such as; unhygienic living conditions, pollution, problems of sanitations, contaminated water and food and industrial chemicals which are responsible to spread the virus of any disease and the individual becomes recipient for being carless or unaware of heath standards (MacMahon, Pugh, & Ipsen, 1960). The model emphasizes on healthy environment and precautionary individual's practices by protecting and cleaning their own surroundings. The ideal health standards can be ensured by avoiding the unhealthy practices, protecting environment and ensuring the standardized practices of quality of life by the individual.

### 2.9.5 Ecological model

The ecological model of health and illness is imitative from the field of human ecology (Meade, 1977). The focus of ecological model remains on interrelationship of human interaction, social organization and particularly on environment, to study health (Clarke, 1991). The model correlates health and illness with ecological development at a certain level of population (Thurston & Vissandjée, 2005), in terms of the aspects of human settlement (Hyman, Guruge, & Mason, 2008), industrial and technological development (Dustin, Bricker, & Schwab, 2009) and culture (Lohrmann, 2008). Ecological model of health has a similarity with agent host

model, because both include the environment as an element to ensure health state of any individual (Reifsnider, Gallagher, & Forgione, 2005; Sallis, Owen, & Fisher, 2015).

### 2.9.6 New public health

New public model is a contemporary approach in the field of health sociology (Baum, 1998)., which unlike other approaches includes the social, cultural, behavioural and political aspects of health (Nutbeam, 2000) along with the traditional health traits i.e., hygienic practices, clean air and sanitation (Armstrong, 1993; Barnes, 2007). It directs attention to the prevention of illness trough community participation and social reforms that address living and working conditions (Graham, 2004; Nutbeam, 2000). Instead of merely focusing on individual factors, the new public health model indicates a broader range of social, political, historical and economic forces of health's production and distribution (Frenk, 1993). The model focuses on policies and behaviors which change the health practices of communities (Fielding, Teutsch, & Breslow, 2010).

## 2.10 The Limits of Medicine

Although biomedical model of health and illness significantly contributed in the field of health and illness especially in the treatment, causes and prevention of diseases. Whereas, a very significant criticism can be found on bio-medical model with reference to the point of view of social and psychological researchers and philosophers. They pointed out that biomedical model ignored social, behavioral and psychological parameters and aspects of health and illness. Neglecting these factors, an individual cannot understand the complexities of health and illness. One of the major criticisms on bio-medical, pointed out that, the causation of disease does not always mean the interaction (Dubos, 1987). The prevalence of disease is based on multiple factors, that can be physical, psychological (stress) and related to practices (food and nutrition). Another major criticism on bio-medical model is reductionism (Mehta, 2011).

In the view of many researchers (Daniels, 1989; Gifford, 2016), bio-medical model focuses on smaller units (e.g. cells, biological organism) and completely ignores the general and holistic aspects such as social and psychological. Reductionism also lead to biological determinism and according to this point of view,

determinism and explanation of health and illness is based on the biologically determined factors (chromosomes, hormones and genes), while many aspects of life, such as; happiness, psychological and emotional disorders, anxiety, depression and even many genetic disorders are based on social and cultural practices rather than solely depending on biological factors (Germov, 2014).

In summing up, biomedical model is highlighted by its nature of victim blaming. As it creates culpability for any sick individual among other members of his/her community by considering the individual a sole cause of his/her disease and ignore all other social facets by which a disease can prevail. Because of many social, economic, religious and cultural practices, attitudes and beliefs, biomedical model faced a very strong criticism by social researchers (Broom & Adams, 2016; Geist & Dreyer, 1993; Lyman, 1989). Due to the widespread criticism on biomedical model of health and illness, medical and social researchers developed varieties of new models and frameworks to explain and contextualize health and illness.

### 2.11 Social Construction of Health and Illness

It has been observed that in the field of health sociology the, effect of social aspects such as; caste, class, ethnicities, marriage patterns, socio-economic status, family systems and socio-political conditions of any society play a vital role (Germov & Hornosty, 2016; Sharf & Vanderford, 2003). In the discussion of health sociology, sociologists have tried to incorporate the aspects of social pathologies, health seeking behaviours, individual and collective communal practices, to understand any disease (Cohn, 2014). It also offers to undercover the ways as an elucidation of an illness (Nettleton, 2006).

In the view of (Fleming & Parker, 2015), *health or absence of health is not merely the attribute of biological or natural aspects* (p,27), social researches found the contribution of socio-economic status, cultural beliefs and practices (consanguinity, stigmatization and marriage patterns) and the values (cast system, ethnic preferences etc.) have a greater influence over the assertion of being healthy or not. In the diverse definitions of Sociology of health, (Mattlin, 2018) focused on three major domains in the field of health and illness; first one is the conceptualization of health, second is the study of measurement and social distribution of health and third is known as the justification of the patterns in health and illness. Furthermore, there is

a significant difference between medical sociology (that focuses on epidemiological demeanors, health institutions and dexterities of medical practitioners) and sociology of health and illness (Nettleton, 2006) because *sociology of health and illness includes social, cultural, economic and political facets, which influence the health practices, statuses and focus on underlying causes of any disease* (p,13).

Sociology of health and illness swathes the ecological and environmental causes of an illness and attitudes and perceptions of individuals towards the understanding and precautions of any disease. It also focuses on psychological and emotional problems, allied with the jounce of any mental or psychological malady. Unlike studying or researching the specific extent of any disease, sociology of health and illness have a holistic view to study the collective effects and cardinal geneses (social, psychological, cultural, political and emotional) of any scrupulous disease (McKee, 1988). Conceptualization of health and illness varies from the point of view of different schools, the conventional approaches to health and illness focus on biological and behavioural explanations and construction of health and illness (Arber, 1987), however the formation of health and illness is based on man social factors, e.g. class differences, awareness, knowledge, attitudes, perceptions, practices, socio-economic status, gender, race and ethnicity (Merrild, Vedsted, & Andersen, 2016). Because of all these social aspects, researchers focused to find out the social causes and consequences of many chronic diseases and genetic disorders. The individualistic approaches tend to focus on and alter the life styles and practices which are prone to the propagation of many genetic disorders i.e. thalassemia.

## 2.12 Theoretical Construction of Beta Thalassemia Major

As identified by (Goffman, 1963), *a person become stigmatized based on an attribute, that negatively affects his/her social interaction with rest of the community or society* (p,138). In the view of (Goffman, 1986), stigma is classified into three major forms, which include; any kind of physical *disability of body image* while the second explanation of stigma is, any kind of *individual characteristic* (e.g. mental and psychological abnormality) and lastly, he proposed that *tribal factors* such as race, gender, cast, religion and ethnicity also endorse stigma.

It has been explored by many researchers (Anum & Dasti, 2016) that patients and families of thalassemia are usually pilloried, particularly because of tribal factors.

Mostly these individuals (patients of thalassemia disease) are called and pronounced as *thalassemic* (being labeled based on their disease). Social isolation and exclusion from communal activities are hard facts, often faced by the parents and families of sick children due to their disease. In many cases, mothers undergo severe mental and psychological stress and face social pressure due to the illness of their child (Arbabisarjou, Karimzaei, & Jamalzaei, 2015).

In Pakistan, where the patriarchal family structure is excursed overpoweringly, women have faced a strong level of criticism and stigmatization for giving birth to a child with any genetic disorder like thalassemia. The ideas of victim blaming and self-blaming (reference) become germane in the context of thalassemia because couple and parents of thalassemic children indict themselves for being the cause of thalassemia (Murphy, 2005).

Theoretical framework for the present study is bio-psychosocial model of health and illness. Biopsychosocial model is an annex of biomedical model. It has been known as a multiple approach in the field of health generally and health sociology particularly, which incorporates social, psychological and biological factors together to elucidate the health status, or state of being unhealthy. The bio-psychosocial model conceptualizes disease in terms of measurable biological variables that indicate the presence or absence of disease or disease severity and social understanding as well as psychological factors and their intervention (Engel, 1977).

The application of bio-psychosocial model in thalassemia can interpret the biological factors related to the thalassemia in the context of genetic vulnerability, disability and physical health while social factors include; awareness and knowledge about the disease and cultural practices for the understanding and propagation of thalassemia. The present study used deductive approach and extract biological and social risk factors of thalassemia without including psychological factors as risk intrusions. Reviewing the literature showed that psychological aspects are only measured as impact of thalassemia on patients and their families. A very clear manifestation of psychological impact can be seen in literature; however, genetic and social factors are major risk factors of thalassemia's propagation.

This study aimed to include bio-psychosocial model as a theoretical framework to identify the social, cultural, economic and therapeutic risk factors of beta thalassemia major among children, while the pomological factors are considered as an impact on patients and their families because these factors do not prompt thalassemia, yet impose a significant effect on quality of life, social adjustment and wellbeing of parents and patients. It is also evident that even though the social and psychological narrations have been widely accepted by the researches and experts still there is a debate and a negative argument prevails against these psychosocial explanations of health and illness.

## 2.13 Sociological Analysis of Beta Thalassemia Major

Over the period, medical sociology morphed into the specific discipline known as sociology of health and illness and social scientists considered medicines too restrictive as an indicator of the sociological interest in health realm (Bloom et al., 2002; Conrad, 2005). A shift in the field of medicines and then into medical sociology can be seen by the critics on epidemiologist and medical sociologist who were researching and analyzing health and illness on solely clinical parameters. Ignoring social, psychological and cultural factors allied with health and illness caused seriousness in failure of finding the risk factors and basis of many chronic genetic disorders, including thalassemia. Traditionally, the biological attributes and natural conditions remained influential in explaining the state of being healthy or not (Goldenberg, 2006). Sociologists have demonstrated that the spread of diseases is heavily influenced by the socioeconomic status of individuals (Link & Phelan, 1995), ethnic traditions or beliefs (Angel & Thoits, 1987) and other cultural factors (Lupton, 2012; Nayak & Geroge, 2012). Where medical research might gather statistics on a disease, a sociological perspective of an illness would provide insight on what external factors caused the demographics that contracted the disease to become ill. The remarkable efforts of social scientists in determining the social factors as underpinning with health and illness develop independent discipline- *Sociology of Health and Illness*.

Perceiving health and illness with the perspective of sociology became more distinctive after the contribution of an American Sociologist Charles Wright Mills (1916–62), who used a very idiosyncratic term "*Sociological Imagination*" to describe any social aspect of human life with the lens of sociology. This method provides an

in-depth and a very comprehensive understanding of social life. The perspective enables the researcher to understand social problems, their possible causes and proposed solution to avoid future incongruities. Whereas thalassemia as a major public health problem across the globe and particularly in Pakistan, required social understanding, remedies and solution rather than focusing on biomedical, because it has more to deal with social aspects of life.

The Australian sociologist Evan Willis (1993; 2011) suggests that the sociological analysis consists of four interrelated parts. The intension, to apply these aspects to gain understanding of thalassemia as a public health problem, existing across the world can be traced and explored with the lens of sociology.

1. Historical factors: How the past influences the present (family history of the patients and inheritance leads to the higher propagation of thalassemia)
2. Cultural factors: How culture impacts on our lives (consanguinity, religiousity, stigmatization and termination of pregnancy)
3. Structural factors: How particular forms of social organizations affect our lives (treatment cost, hospital expenditures, blood transfusion)
4. Critical factors: How we can improve our social environment (awareness level of the respondents about prenatal and post marriage screening, knowledge about the disease and its preventive measures can reduce the prevailing condition of thalassemia across the world and especially in Pakistan)

## 2.14 Conceptual Framework

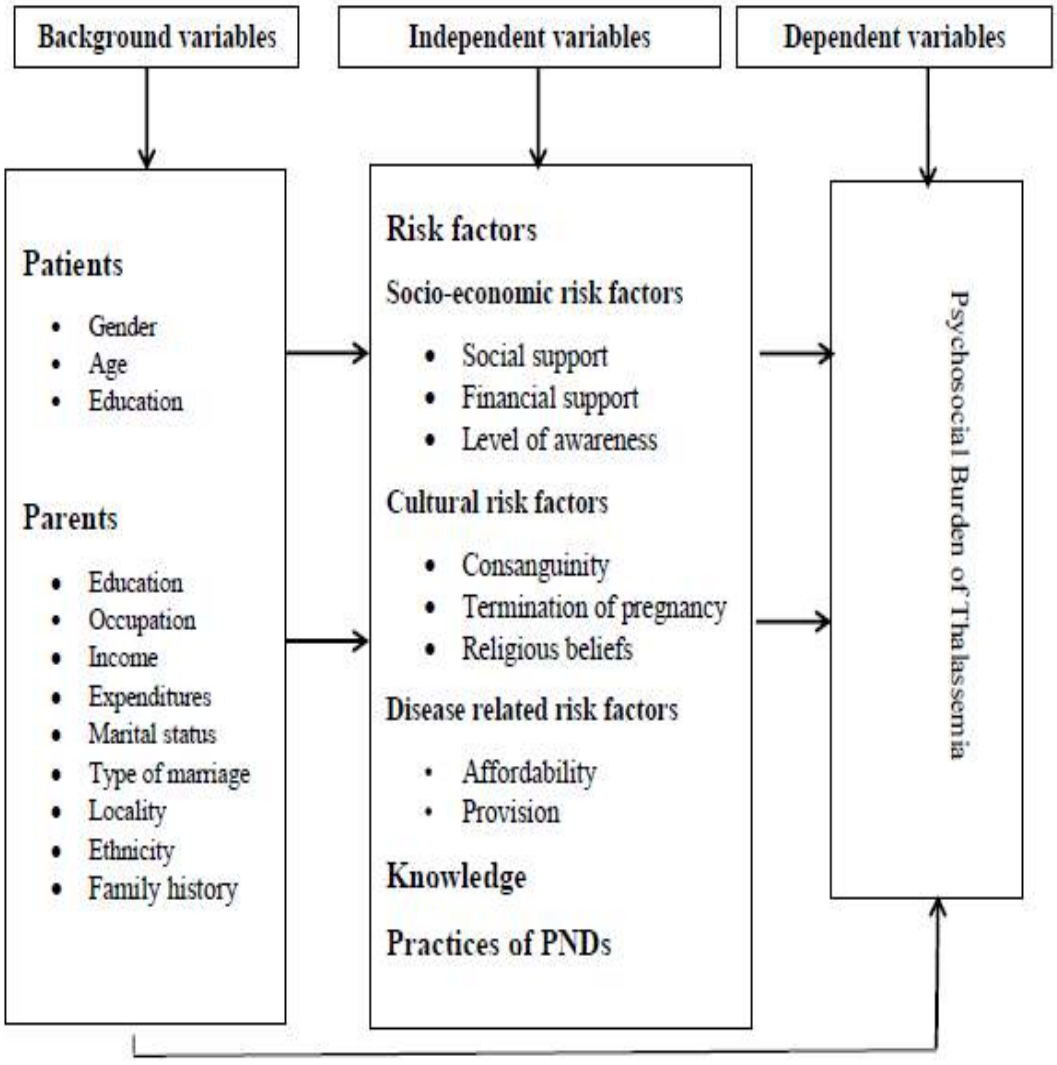



*Figure 2.1 Conceptual Framework of the Study*

## 2.15 Chapter Summary

The chapter highlighted contribution of previous researchers to determine the socioeconomic, cultural and disease allied risk factors of beta thalassemia major across the world which special focus on Pakistan. Review of the literature showed that beta thalassemia major puts a significant economic burden on parents and families of sick children due to expensive treatment and regular blood transfusion. Parents of sick children required social and financial support from their relatives, friends and family members for the treatment and management of disease but they often lack to gain social, financial and psychological support. The chapter included

studies on cultural risk factors of beta thalassemia major and found that consanguineous marriages, religious and moral restriction regarding carrier counseling and genetic screening contributed in the propagation of beta thalassemia major across the world and especially in developing countries. The professional attitude of medical staff and health activists play an evenhanded role for the management and prevention of disease. Studies conducted in Iran, Bangladesh, India and Pakistan highlighted the problems of psychological and social adjustment of parents and families of sick children due the beta thalassemia major. Parents and sick children face weak social interaction, social isolation, stigmatization, depression and anxiety due to this chronic illness. The second part of this chapter is based on theoretical orientation of health and illness and incorporated theoretical models of health and illness in the field of sociology. The theoretical construction of beta thalassemia major incorporates biopsychosocial model of health and illness. The model has a multidimensional view about the construction of health and illness and focuses on social, psychological and biological aspects of health and illness, unlike biological determinism and reductionism, it does not ignore the social and psychological causes of any disease.

# CHAPTER 3

# 3. RESEARCH METHODOLOGY

## 3.1 Chapter Introduction

A complete picture of research techniques which have been applied to conduct this research have been discussed in this chapter. This chapter includes area of the study, selection of foundations and respondents, sampling techniques, sample size determination, tools for data collection, data entry and analysis, research design, ethical considerations and limitations of the study.

## 3.2 Area of the Study

The present study was conducted in Punjab Province of Pakistan to find out the risk factors of thalassemia propagation in the Province. The selection of Province is based on its assorted terrestrial characteristics. The province is expected to have the demographic, socio-cultural and economic gen, required to meet the objectives of this study. Punjab Province has a vast geographical area (205,344 $km^2$) and (110 million) population size out of them 70 million are living in rural areas whereas, 40 million live in urban areas of the selected province[1], with multiple casts, ethnicities and having different socio-economic status and underlying practices of social life. The higher rate of consanguinity, male dominancy, low level of literacy and cultural dominancy are some of the prominent features of provinces and the same were required to ample this empirical testimony. The selection of this province is also based on the criteria of having higher number of extended families, among which the frequency of beta thalassemia major was also noticeably higher (more than 60 %) (Aqueel & Anjum, 2019).

## 3.3 Selected Foundations for Data Collection

The data was collected from three major foundations of thalassemia (Jamila Sultana Foundation, Sundas Foundation (five centers), Fatimid Foundation), working in study area and across the country. However, these foundations are in Punjab

[1] Source: Pakistan Bureau of Statistics Census Results 2017

Province and registered patients also belong to the same territory. The complete list of Thalassemia Foundations, registered with Pakistan Thalassemia Foundation for Punjab Province is given in (ANXEURE-A).

### 3.3.1 Jamila Sultana Foundation- Rawalpindi

Jamila Sultana Foundation is voluntary organization, working as a blood transfusion center in Rawalpindi City. The foundation was established in 2004 and up till now it has become one of the major blood bank and transfusion center which provides free blood and medications to the children with thalassemia. The foundation is affiliated with International Federation of Thalassemia, registered with Thalassemia Federation of Pakistan and working under the license by Punjab Blood Bank Authority. The foundation has a list of 800 registered patients of beta thalassemia major.

### 3.3.2 Sundas Foundation

The second foundation selected as a study site is Sundas Foundation. The network of thalassemia centers of Sundas Foundation is comparatively bigger than other voluntary organizations. The foundation has five mega centers of blood transfusion in Punjab Province, with total number of 3297 registered patients of beta thalassemia major. The foundation was established in 1998 as one of the forerunner blood transfusion centers in Punjab Province. The foundation is registered with Government of Punjab and affiliated with International Thalassemia Federation. The selected centers of Sundas Foundation were; Sundas Foundation Gujrat, Sundas Foundation Gujranwala, Sundas Foundation Sialkot, Sundas Foundation Lahore and Sundas Foundation Faisalabad.

### 3.3.3 Fatimid Foundation- Multan

Fatimid Foundation is a non-governmental charitable organization and it is known to be the trailblazer of the frat of blood transfusion services in Pakistan. The foundation was established in 1978 in Karachi City and up till now, is working within the entire country. The center of Fatimid Foundation located in Multan City with 1500 registered patients of beta thalassemia major was selected for the present study.

Reason for selecting only these Foundations was;

1. These organizations have updated and rich data of the patients

2. Patient's frequency of visits is high in these organizations

3. Patient's registration was a replication of different small centers and organizations and during pre-fields visits, this replication was found substantial.

### 3.4 Selection of the Respondents

The study population consisted of the parents of those children who were sufferer of Beta Thalassemia Major.

#### 3.4.1 Inclusion Criteria

Selection of the respondents was based on the following criteria:

1. Parents of the patients were selected from Punjab Province

2. Parents of those children who were diagnosed with Beta Thalassemia major

3. Parents of those children, who were not suffering from any other genetic disorder

4. Parents of those children, who were not suffering from any other illness

#### 3.4.2 Exclusion Criteria

Exclusion of the people by considering them unrepresentative members was based on the following directions:

1. Parents of any child who was not the patient of beta thalassemia major

2. Parents of a child who was suffering from any other genetic disorder

3. Any patient whose parents were not available to collect required information

4. Parents who belong to any province of Pakistan, other than Punjab

### 3.5 Sampling Technique

Multistage random sampling technique was employed to select the representative of the population for present study. The respondents consist of parents of beta thalassemia major's children. This sampling technique was also applied by many (Alshamrani et al., 2017; Farmakis, Giakoumis, Aessopos, & Polymeropoulos, 2003; Manzoor & Zakar, 2019) to study beta thalassemia major, across the world. The selection of each stage is mentioned below;

#### 3.5.1 Selection of Blood Transfusion Centers

At first stage; the blood transfusion centers were selected by using random sampling technique because the desired study area of this research was Punjab Province and the located blood transfusion centers/thalassemia foundation were searched and found suitable to acquire data.

#### 3.5.2 Selection of the Study Respondents

At second stage the equal proportion of inferred sample was obtained by applying proportionate sampling technique for selected seven centers/foundations of thalassemia. The third stage which was employed to select the respondents of this study, was random sampling. The random selection is based on the inclusion criteria and the list of registered patients at thalassemia foundations.

### 3.6 Sample Size Determination

Prior to decide the sample size of this study, the field visits were planned and executed to obtain the lists of registered patients and to specify the thalassemia foundations for data collection. Data was collected from seven different centers of blood transfusion by selecting 932 parents of thalassemic children with the help of Taro Yamane formula (1973:258) for known population. It provides a simplified way

to calculate sample sizes and is commonly used in the fields of social sciences (Eckhardt & Ermann, 1977; Songkram, Khlaisang, Puthaseranee, & Likhitdamrongkiat, 2015; Yamane, 1973).

$$n = \frac{N}{1 + N(e^2)}$$

$n$ = Sample size

$N$= Total population- number of patients (mother or father of a patient was selected based on patients).

$e$= Margin of error

The total population of this study was 5597 patients which is denoted by $N$, while $e$ is known as margin of the error and commonly used as 0.05. The margin of error used in this study was 0.03 which is less than the common maximum range. The sample size is denoted by $n$, was 932, which was finally selected by planned methodology of this study.

### 3.6.1 Proportional Allocation

To assign proportion of the respondents (parents of thalassemic children) from the population of present study, the formula is given below:

$$n_i = n\frac{N_i}{N}$$

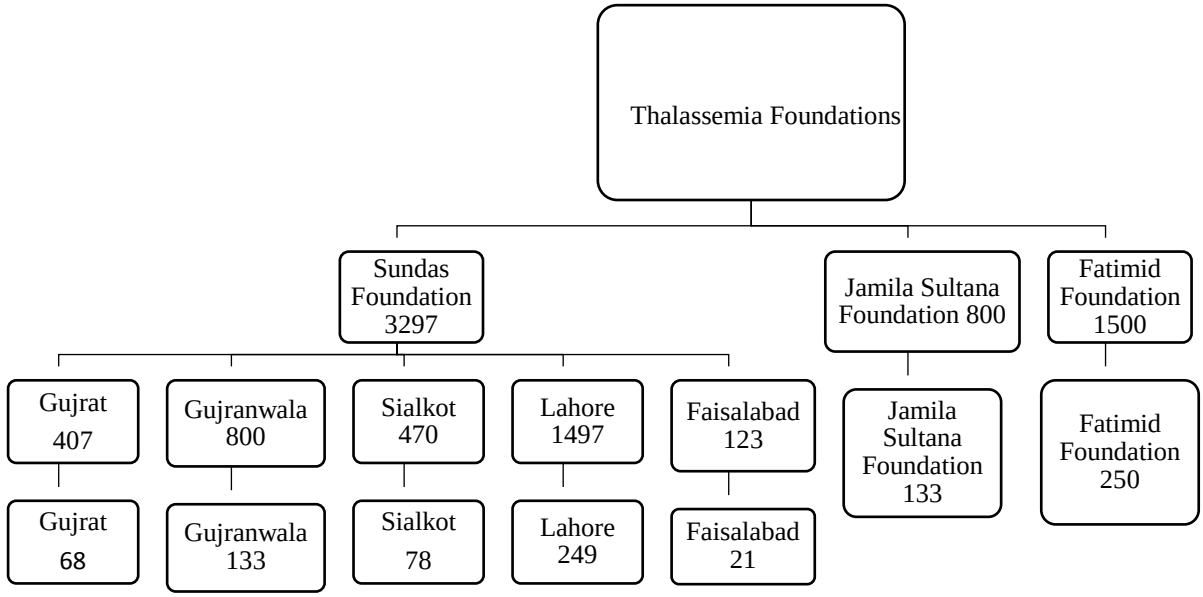
Where, $i$=1, 2, 3...

$n_i$= Selected proportion of parents from each thalassemia foundation

$n$= Sample size of the given population

$N_i$= Total population of each thalassemia foundation

$N$= Total population of selected thalassemia foundations




*Figures 3.1 Flow Chart of Sample Selection*

### 3.7 Tools of Data Collection

The researcher used two tools (interview schedules) to collect data for the present study. The first interview schedule was used for the patient's parents to analyze their socioeconomic status and social, economic, cultural and disease allied risk factors of thalassemia. Prior to the development of interview schedule, the researcher conducted three focus group discussions (FGDs) with parents of thalassemic children, health experts and representatives of thalassemia foundations at *Jamila Sultana Foundation, Sundas Foundation-Gujrat and Fatimid Foundation-Multan,* including 12 participants (doctors, health experts, representatives of thalassemia foundations and parents of thalassemic children) in each focus group session. By using thematic analysis for interpretations of responses, themes were developed and classified into socioeconomic, cultural and disease allied risk factors of beta thalassemia major (see section 3.7.1).

The statements of interview schedule were developed and rated on 5-point Likert Scale, which included the agreements and disagreement along with the situation of being undecided regarding the statements for response. Further, to quantify these risk factors, to meet the aim of this study (ANXEURE-B). The interview guide was classified as demographic and socioeconomic delineations of patients- which includes, gender, age and educational level of thalassemic children, while parental demographic information includes marital status (married, widow or divorced) of both parents, locality, ethnicity, age, educational level, income and occupation.

The second part of the study's tool includes information regarding thalassemia, which was gathered from the respondents (parents of thalassemic children). The intension of asking these questions was; to know the family history of beta thalassemia major, parental awareness of genetic disorders and counseling, as well as recording the blood groups of mothers', fathers' and the sick child. The third section of study's tool is based on the information, related to treatment of beta thalassemia major followed by parental knowledge and their practices of pre/postnatal diagnosis. The specific statements narrated to find out the socioeconomic, cultural and disease allied risk factors were also incorporated in interview schedule.

The second type of tool for the measurement of psychosocial burden of thalassemia on patient's families was also interview schedule "psychosocial burden of thalassemia" developed by (Canatan, Ratip, Kaptan, & Cosan, 2003) which was adapted by the researcher. The interview schedule consisted of total 16 items in which 9 items measure the burden (psychosocial) on patient's parent while 7 are related to the children. The interview schedule was also measured on 5-point Likert Scale (ANXEURE-C).

### 3.7.1 Tool Development Procedure (Development of Interview Schedule)

The following section highlights the emerged themes from three focus group discussions, conducted by researcher at three blood transfusion centers, to develop an interview schedule for the measurement of risk factors, parental knowledge of the disease and their practices of pre/postnasal diagnoses.

***Classification of the participants***

The focus group discussions (FGDs) were conducted at *Jamila Sultana Foundation, Sundas Foundation-Gujrat and Fatimid Foundation-Multan,* including 12 participants (representatives of thalassemia foundations and parents of thalassemic children) in each focus group session. In each session, other than representatives of foundations, parents were ranging from illiterate to holding university education. Their professions and natures of jobs were house wives, labourer, small entrepreneurs, private jobs and government employees and they had a diverse ethnic background; eight were Punjabi, five were Pashtun and three were Kashmiri and two form Sindhi ethnic groups, living in the Punjab Province.

***Personal feelings of the participants***

With a very warm and gratified attitude, almost all parents appreciated the contribution and aid provided by the thalassemia foundations. They were much obliged by the timely and proactive response of these centers for the provision of blood and chelation therapies. They were of the view that these organizations are helping us beyond their expectations- "*the organization is the only hope for survival of our kid- I was hopeless but this organization exhibited a beam of life for my child"*. The centers were managing blood through their camps, door to door collections, providing seasonal clothes to the poor families and arranging recreational trips for thalassemic children. "*I was unable to manage blood because my family members, relatives and friends stop their social, moral and financial support, then I came to the organization and I am receiving blood and medicines for my kid, free of cost"*.

***Self-blaming***

The results of study revealed that the parents blindly followed social and cultural practices, generally followed in health, marriages and for family planning due to lack of knowledge and awareness. They were of the view that if they considered the aftereffects of cousin marriages and denial of screening, they could be able to save the life of their kid. "*I never thought that due to consanguinity, my child could have thalassemia"*. These parents realized the importance of blood screening, genetic counseling, prenatal and post-natal precautions after their children became thalassemic and they said that everyone should go for these precautions to save the

lives of future children. "*I ignored the importance of screening and counseling, which resulted my child to suffer the chronic disease"*.

***Consanguinity and cultural mythologies***

Some of the parents were living in joint and extended families and they all had cousin marriages. Due to endogamy, cast, ethnic preferences and traditional normative practices, these participants opted cousin marriages. "*In our family we cannot wish to marry out of cast"*. Another participant said that "*my parents did the same as I because we are not allowed to marry out of family"*. Their marriage decisions hardly include their personal choices and due to repeated cousin marriages, their children were suffering from this chronic genetic disorder. "*When my child born with thalassemia and I came to know that this happens due to cousin marriages, so I requested my family to stop this tradition of intentional killing of children"*.

***Religious fallacies***

False concept narrated by religious preachers and local people misguided parents for the identification and treatment of thalassemia. Participant believed that it is the will of God that our children had this chronic disease otherwise religion does not restrain couple to go for abortion-but within a particular time of fetus maturity-, "*although this is a disfavoured and unlawful act but Islam allow Muslims to abort, in case of having fear of death of mother or child within 120 days of fetus maturity"*. This is evident that due to delayed identification of disease, it becomes chronic and allied many other severe health problems such as cardiac, diabetes, anemia, insomnia, hemophilia, body pain, mental stress and other physical issues. The reasons for delayed identification were reported by the parents that; "*I initially went to the spiritual healers and religious celebrants for the treatment of thalassemia because I was unaware of its cause and consequences, later I came to know that there is no other way but blood transfusion to battle this disease"*.

***Patriarchy***

Due to male dominancy in traditional societies, women are usually not allowed to take part in decision making, even related to their personal lives (i.e.

education, career choices, marriage and family planning). "*I was not asked before marriage, either I want to marry or not"*. Pre-marital and post-natal screenings are compulsory for career detection, but male dominancy does not allow women to go for it especially for premarital screening and they even deny for themselves by considering a humiliation of their self-esteem. "*I never recognized the importance of screening which resulted the pervasiveness of chronic disease"*. The study found that along with female respondents, the male members were also viewed patriarchal structure as a hinderance for carrier detection, genetic counselling and adaptation of preventive measures (birth control and termination of pregnancy).

***Problems of blood management***

Majority of the parents viewed that regular blood transfusion required a plenty of blood at transfusion centers, but they often face shortage of blood and they must wait until they receive call from transfusion center. In this regard, the representatives of thalassemia foundation shared their views that this happens because "*we don't have regular donors and general population shows reluctance to donate blood"*. They shared their experiences that local people usually perceive them negative change agents for their society and call police by filling FIR against the representatives of thalassemia foundation.

### 3.8 Data Collection Procedure

Erstwhile to the collection of data, the researcher visited all selected centers of thalassemia foundations and decided to plan the time and date for data collection, with the help of management of the foundations. In every thalassemia foundation the number of patients depend upon availability of blood, the decided date was planned accordingly to collect maximum data on given dates. Researcher employed skilled data enumerators for a shorter period because it was not possible for the researcher to collect data individually, due to rutted dates and days of blood transfusion (parents visit these centers, when their child required blood or at the next given date by health practitioner of the center). With the consent and permission of management of thalassemia foundations, parents were briefed about the nature and purpose of present study. The respondents were ensured about the confidentiality of their information and a consent form was signed by them to willingly reveal the required information

about their demographic and socio-economic profiles, their knowledge and practices of pre/postnatal diagnosis, their levels of agreement or disagreement with the statements of risk factors and psychosocial burden of disease.

### 3.9 Process of Data Entry and Analysis

Before starting the procure of data entry, the filled forms were edited by the researcher with the help of data enumerators and technical experts. The half-filled 30 forms were screened out for further progression of data entry. The collected data were then entered into SPSS v.20 at first step. After complete entries, the reliability of data was checked, using Cronbach's alpha and the recorded value was 0.694, the detail of tool reliability is given in (ANXEURE-D). Data was analyzed statistically and the researcher applied descriptive statistical techniques to measure Frequencies, Percentages, Mean and Standard Deviation as well as inferential statistical techniques to measure One Way ANOVA, Independent sample t-test, Pearson Correlation and Generalized Linear Regression, Multinomial Logistic Regression and Multivariate Linear Regression.

The analysis route was classified into three preceding sections

#### 3.9.1 Univariate Analysis

The researcher employed descriptive statistics for each selected variable in terms of calculating percentages (%) and frequencies (f) of all statements of research tool by using SPSS v.20. Furthermore, the means and standard deviations along with minimum and maximum values were also recorded in the section of results and discussion (chapter 4). Descriptive statistics is appropriate method for data understanding and for the generalizations of results (Shahravan, Ghassemi, & Baneshi, 2015) and the technique is commonly used in the fields of academia. Descriptive modes of analysis simplify the complex information by tabulating data into a distinct scaffold and describes the basic features of data in a study.

#### 3.9.2 Bivariate Analysis

The second statistical technique applied by the researcher, for the analysis of data is inertial statistics-bivariate analysis. The method of analysis is also significantly

used to predict and measure the differences and relationships between two variables (Bartholomew, Steele, & Moustaki, 2008). The researcher applied One Sample t-test and One-way ANOVA along with the Pearson's Correlation to measure the differences and relationship between risk factors, parental knowledge, practices of prenatal diagnosis, risk factors and psychosocial burden of disease, based on demographic and socio-economic profiles of the respondents.

### 3.9.3 Multivariate Analysis

The third method of data analysis is inferential statistics-multivariate analysis. The methods of multivariate analysis are known to be the powerful ways to signify the relationships and effects between independent and dependent variables (Var, 1998). Researcher applied *Generalized Linear Regression* for two independent variables; consanguinity and ethnicity and selected dependent variables; parental knowledge, practices of pre/postnatal diagnosis, risk factors (socioeconomic, cultural, disease allied) and psychosocial burden of beta thalassemia major on parents of thalassemic children.

*Generalized Linear Regression* is an extension of linear multiple regression technique but unlike other linear models, including logistic regression- generalized linear model allows researchers to use a non-normally distributed dependent variable. Furthermore, generalized linear regression is commonly used in the field of sociology, psychology and health and illness. It measures the interaction effect of two independent variables on a single dependent variable. The reason for applying generalized linear region was to find out the interaction effect of consanguinity and different ethnicities on selected dependent variables. Researchers found a plethora of researches that revealed, consanguinity and ethnicities are significantly related to the propagation of beta thalassemia major and have found to be in an association with parental knowledge, practices of PNDs, risk factors and psychosocial burden of beta thalassemia major. The interaction effect is signified based on *p* value, if less than 0.05 or 0.01 or greater and the level of effect; either small, medium or high is based on the explanations of Cohen 1988, criteria. However, the pictorial illustration depicts estimated marginal means of dependent variables for each independent group (consanguinity and ethnicity).

The second technique applied by the researcher was *Multinomial Logistic Regression*, to find out the odd ratios of consanguinities ($1^{st}$, $2^{nd}$, $3^{rd}$ cousins and distant relatives) and significant predictors of psychosocial burden of beta thalassemia major. For this purpose, the dependent variable (psychosocial burden) was indexed into three levels (mild, moderate and high).

The researcher also applied *Multivariate Linear Regression* (MLR) to predict the significant independent variables; risk factors (socioeconomic, cultural, disease allied), parental knowledge of disease and practices of pre/postnatal diagnosis (PNDs) as variance explainers of dependent variable; psychosocial burden of beta thalassemia major.

### 3.10 Research Paradigm

Following the quantitative loom of research methodology, the present research implies Positivism as a paradigm of this research.

As believed by August Comte (1975)

> *Observations and reasons are the paramount modes to understand the behaviours of humans, because true knowledge is based on experiences of senses* (p.292) *and can be obtained by observation and experiment* (Comte, 1975). *Ontologically, Positivism assumes that -reality is objectively given and is measured by using research tools and instruments; independent of the researcher* (p.381).

It is the believe of positivists that, knowledge is objective and quantifiable (Arghode, 2012; Bahari, 2010) because positivist thinkers and researchers believe on quantification of facts, by adopting scientific methods to describe and predict the relationship and measure effects of parameters scientifically (Martin, 2003; Tafreshi, Slaney, & Neufeld, 2016).

Epidemiologically, the positivist researchers and thinkers are be certain to consider social science is an organized method- which implies deductive nomological propositions (Della Porta & Keating, 2008; Kuhn & Boulding, 1974)- in order to predict general patterns of human behaviors and activities (Fuchs, 1993). In the view of positivists, social phenomena are governed by causal connections and the

relationship between cause and effect can be quantified by using statistical techniques. The paradigm believed on manipulation of natural phenomenon, through empirical and experimental means (Weinberg, 2013).

### 3.11 Ethical Considerations

In present study, researcher collected the data from parents of sick children at selected blood transfusion centers, by prior consent and they were assured that, it is a pure academic research. A letter of authorization for data collection was taken signed by the supervisor and International Islamic University Islamabad to make it authentic that researcher is a PhD scholar and this research is the part of dissertation. Meanwhile, transfusion centers and organizations were taken in to confidence that there will not be any misuse of the information regarding the list of registered patients and their families. Furthermore, parents were assured that they are not required to share their personal information and familial disputes, the required information is related to find out the risk factors and psychosocial burden of disease for academic purpose, instead of any commercial use.

### 3.12 Limitations of the study

The aim of the present research was, to sociologically analyze the risk factors and propagation of beta thalassemia major in Punjab province. The first limitation of this study is its geographical constraint due to limited resources, the researcher was unbale to collect data form entire country and meanwhile, the higher propagation of beta thalassemia was also evident in selected provinces. Although, many small-scale thalassemia foundations and blood transfusion centers are working in Pakistan, but the researcher selected those foundations, who have data of registered patients and provide services for blood transfusion and this is the second limitation of this study. The third constraint for this research is selection of the true respondents because the timing and dates of blood transfusion were not same for all registered patients, so their selection was on random basis, without considering systematic way. Lastly, the results of this study do not allow researcher to make generalization for entire Pakistani population because the measured variables such as income, expenditures, marriage trends, parental knowledge, practices of pre/postnatal diagnosis (PNDs) and risk factors are not same as can be observed for normal population.

# CHAPTER 4

# 4. RESULTS AND DISCUSSION

The chapter of dissertation presents statistical analysis of collected data from the parents of sick children (patients of beta thalassemia major) from seven Thalassemia Foundations, located in Punjab Province of Pakistan.

The chapter is cataloged into two major portions descriptive and inferential statistics, which are further classified into three sections

a) Univariate Analysis
b) Bivariate Analysis
c) Multivariate Analysis

**Univariate Analysis**

Univariate Analysis is an approach that comprises descriptive statistical analysis e.g. frequencies, percentages, means, minimum and maximum values of a quantifiable data (Gaur & Gaur, 2006). Descriptive statistics is the best method for gaining clear understanding about the nature of any particular problem and for the unit of analysis (Pinsonneault & Kraemer, 1993). The univariate section comprised frequencies and percentages of the respondents for demographic profiles, socio-economic status, parental knowledge about beta thalassemia major, prenatal and postal natal practices, social and cultural along with disease allied risk factors of beta thalassemia major and psychosocial burden of the disease on respondents and their sick children.

### 4.1 Demographic Information of Patients

Demographic information of the patients includes their gender, age at the time of beta thalassemia diagnosed (in months), age at the time of data collection (in completed years) and educational level (in completed years) of thalassemic children.

Table 4.1

*Demographic Information of the Patients*

| Variables | Frequency (f) | Percent (%) |
|---|---|---|
| Age at the time of thalassemia diagnosed (in months) | | |
| 1-4 | 658 | 70.6 |
| 5-8 | 248 | 26.6 |
| 9-12 | 19 | 2.0 |
| 13-16 | 7 | .8 |
| Age now (in completed years) | | |
| <5 | 381 | 40.9 |
| 5-10 | 452 | 48.5 |
| 11-15 | 96 | 10.3 |
| >15 | 3 | .3 |
| Education (in completed years) | | |
| No education | 807 | 86.6 |
| 1-5 | 103 | 11.1 |
| 6-10 | 22 | 2.4 |

Male=486, Female=446

According to the gender of the patients, 486 were male children while with a minor difference 446 were female patients. The minimum age at the time of beta thalassemia diagnosed was one month and maximum age was reported as 2.5 years. Less than half of the patients (40.9) aged below five year, while 48.5% of selected patients belonged to the age group of 5 to 10 years in study area. It was found that only few (0.3%) were above 15 years. According to their educational level, very high majority (86.6%) of the thalassemic children were not going to school, regardless of having an appropriate age of a school going child.

## 4.2 Demographic and Social Characteristics of Respondents

The respondents (Parents of sick children) were categorized on the basis of their social and demographic profiles. The classification is based on their relationship with child (either being mother or father) and marital status (widow, divorced and living together). Respondents were selected based on their residential status (rural or urban) as well, to observe the propagation of beta thalassemia among children, whose parents belong to different ethnic groups. The respondents were classified into five major ethnic categories including *Punjabi, Pashtun, Sindhi, Balochi* and *Kashmiri*. Furthermore, family types and occupations of family heads were also recorded in table 4.2.

Table 4.2 (a)

*Demographic and Social Characteristics of the Respondents (N=932)*

| Variables | | Frequency (f) | Percent (%) |
|---|---|---|---|
| Relation with child | | | |
| | Father | 375 | 40.2 |
| | Mother | 557 | 59.8 |
| Marital Status | | | |
| | Widow | 41 | 4.4 |
| | Divorce | 25 | 2.7 |
| | Living together | 866 | 92.9 |
| Area of residence | | | |
| | Rural | 663 | 71.1 |
| | Urban | 269 | 28.9 |
| Father's Ethnicity | | | |
| | Punjabi | 763 | 81.9 |
| | Pashtun | 105 | 11.3 |
| | Sindhi | 25 | 2.7 |
| | Balochi | 18 | 1.9 |
| | Kashmiri | 21 | 2.3 |
| Mother's Ethnicity | | | |
| | Punjabi | 768 | 82.4 |
| | Pashtun | 107 | 11.5 |
| | Sindhi | 18 | 1.9 |
| | Balochi | 18 | 1.9 |
| | Kashmiri | 21 | 2.3 |
| Family type | | | |
| | Nuclear | 445 | 47.7 |
| | Joint | 450 | 48.3 |
| | Extended | 37 | 4.0 |
| Occupation of the family head | | | |
| | Self-employee | 315 | 33.8 |
| | Private job | 148 | 15.9 |
| | Government job | 213 | 22.9 |
| | Laborers | 256 | 27.5 |

Out of total number of the respondents, 375 were fathers and 557 were mothers of sick children. The selection requirement was either being father or mother of thalassemic child so the researcher can acquire exact information about the disease. According to the marital status of the respondents, 92.9%, were living together, while 4.4%, were widows and 2.7%, were divorced and they (respondents) were females.

Table 4.2 (a) contains information of the residential status of the respondents, in which 71.1% of the respondents of the current study had their rural residential locality (villages) in Punjab whereas 28.9% of the respondent's belonged to urban regions (cities) of Punjab Province. The respondents were classified into different ethnic groups in which the ethnic background of parents was recorded separately. Data demonstrated that majority of the respondents 81.9% Male and 82.4%, Females had *Punjabi* ethnic background. The second major ethic group of the respondents was *Pashtun* 11.3%, Male and 11.5%, Females, then *Sindhi* 2.7% Males; 1.9% Females and *Kashmiri* ethnic group comprises 2.3%. The lowest numbers of the respondents belong to *Balochi* 1.9%, same for both sexes. Majority of the parents (respondents) had joint and nuclear family structure 48.3% and 47.7% respectively along with only 0.4 % belonged to the extended family system. It is also evident from the data, that mainstream (33.8%) of the family head's occupation was self-employment and few (15.9%) were doing private jobs. Many of them (27.5%) were working as laborers, with a very little earning (see table 4.2 a).

Table 4.2 (b)

*A Territorial Overview of Demographic and Social Characteristics the Respondents*

| Variables | | Upper Punjab N=133 | | Southern Punjab N=250 | | Central Punjab N=549 | |
|---|---|---|---|---|---|---|---|
| | | Frequency (f) | Percent (%) | Frequency (f) | Percent (%) | Frequency (f) | Percent (%) |
| Relation with child | | | | | | | |
| | Father | 54 | 40.2 | 175 | 70.0 | 112 | 20.0 |
| | Mother | 79 | 59.8 | 75 | 30.0 | 447 | 80.0 |
| Marital Status | | | | | | | |
| | Widow | 13 | 9.7 | 33 | 13.2 | 40 | 7.3 |
| | Divorce | 20 | 15.0 | 155 | 62.0 | 9 | 1.7 |
| | Living together | 100 | 75.3 | 62 | 24.8 | 500 | 91 |
| Area of residence | | | | | | | |
| | Rural | 26 | 19.6 | 235 | 94.0 | 470 | 85.6 |
| | Urban | 107 | 80.4 | 15 | 6.0 | 79 | 14.4 |
| Family type | | | | | | | |
| | Nuclear | 10 | 7.5 | 125 | 50.0 | 32 | 5.8 |
| | Joint | 63 | 47.3 | 50 | 20.0 | 339 | 61.7 |
| | Extended | 60 | 45.2 | 75 | 30.0 | 178 | 32.5 |
| Occupation of the family head | | | | | | | |
| | Self-employee | 25 | 19.0 | 36 | 14.4 | 60 | 11.0 |
| | Private job | 40 | 30.0 | 45 | 18.0 | 36 | 6.5 |
| | Government job | 36 | 27.0 | 40 | 16.0 | 75 | 13.7 |
| | Laborers | 32 | 24.0 | 129 | 51.6 | 378 | 68.8 |

**Note** f=number of parents, %=percentage

Table 4.2 (b) revealed a territorial overview of demographic and social characteristics of the respondents. The study area (Punjab Province) was classified into three regions; Upper Punjab, Southern Punjab and Central Punjab and respondents were selected from blood transfusion centers, located in these three regions. The data revealed that 40.2% of the respondents (fathers) and 59.8% (mothers) belonged to the Upper Punjab, while 70.0% fathers and 30.0% mothers belonged to the Southern Punjab. However, 20.0% fathers and 80.0% mother respondents belonged to the Central Punjab region of this study.

The data also revealed that 9.7% of the respondents were widows, 15.0% were divorced and majority of them (75.3%) were living together in Central Punjab. According to the marital status of the respondents, 13.2% were widows, 62.0% were divorced and 24.8% were living together in Southern Punjab. However, 7.3% were widows, only 1.7% was divorced and a very high majority (91%) was living together in Central Punjab.

According to the residential status of the respondents, 19.6% belonged to rural and 80.4% of the respondents belonged to urban areas of Upper Punjab, while a very high majority (94.0%) belonged to the rural areas and only few (6.0%) were living in urban areas of Southern Punjab. The data also revealed that, 85.6% were living in rural areas and 14.4% in urban areas of Central Punjab.

The Present study found that, according to the family system of the respondents, 7.5% belonged to the nuclear families, 47.3% belonged to joint and 45.2% belonged to extended family system of Upper Punjab region. It is also reported that half of the respondents (50.0%) had nuclear family system, 20.0% had joint and 30% had extended family system in Southern Punjab. However, the study reported that from Central Punjab region only 5.8% had nuclear family system while, 61.7% and 32.5% were living joint and extended families respectively.

The respondents of this study were also classified according to the occupation of family head, 19.0% of the respondents revealed that their family heads are self-employed, 30.0% were doing private jobs, 27.0% were doing government jobs and 24.0% were working as laborers in Upper Punjab. Similarly, the data revealed that

14.4% were self-employed, 18.0% were doing private jobs, 16.0% were government employed and 51.6% were laborer in Southern Punjab. The respondents belonged to the Central Punjab region were 11.0% self-employed, 6.5% and 13.7% were doing private and government jobs respectively and majority of them (68.8%) were working as laborer.

### 4.3 Socio-economic Status of the Respondents

Socio-economic status includes age, educational level, monthly family income and expenditures (Baker, 2014). The present study measured the ages (now and at the time of their marriages) of mother(s) and father(s) along with educational level, monthly income of entire family and expenditures, along with the expenditures on the treatment of beta thalassemia major.

Table 4.3.1 (a)

*Descriptive Statistics of Age and Educational Status of the Respondents*

| Variables | Father | Mother |
|---|---|---|
| | %(f) | %(f) |
| Age at the time of interview (in completed years) | | |
| <25 | 17.7(66) | 30.3(169) |
| 25-40 | 57.7(216) | 54.7(305) |
| >40 | 24.6(92) | 14.9(83) |
| | M=35.82 | M=32.50 |
| | Mini=20 | Mini=18 |
| | Maxi= 60 | Maxi= 62 |
| Age at marriage | | |
| <18 | 17.5(65) | 30.4(169) |
| 18-30 | 72.3(272) | 68.9(384) |
| >30 | 10.2(38) | 0.8(4) |
| | M=23.92 | M=21.7 |
| | Mini=14 | Mini=12 |
| | Maxi= 35 | Maxi= 35 |
| Education | | |
| Illiterate | 16.1(60) | 40.1(223) |
| Primary | 11.9(45) | 14.9(83) |
| Middle | 12.0(45) | 5.6(31) |
| Matriculation | 47.1(177) | 25.1(140) |
| Intermediate | 10.1(38) | 9.8(55) |
| Graduation | 0.6(2) | 2.1(12) |
| Masters and above | 2.1(8) | 2.4(13) |
| | M=7.65 | M=5.64 |
| | Mini=0 | Mini=0 |
| | Maxi= 18 | Maxi= 16 |

**Note** f=number of parents, %=percentage, M=Mean, Mini=Minimum, Maxi=Maximum

Age is a very important variable because the comprehension and understanding of social life potently based on experiences, skills and knowledge which boosts with growing age. The understanding and management of beta

thalassemia major has been studied and explored by many other researchers (Heydarnejad & Hasanpour-Dehkordi, 2008) and they found a significant relationship between parental age and knowledge of genetic disorder.

The data characterized in table 4.3.1 (a) revealed that more than half of the fathers of sick children (57.7%) had their age ranging between of 25-40 years while 24.6% were, above 40 years and 17.7% of the respondent's belonged to below 25 years. The average age of fathers was 35.82 minimum age was 18 years, while maximum age was recorded as 60 years. The data also recorded same information of mothers' age and it was found that 54.7% of the mothers belonged to age group of 25-40 years, 30.4% were less than 25 years and 14.9% were, above 40 years. The average age of mothers 32.50 years, while minimum recorded age was 18 years and maximum age was 62 years.

The second classification of parental age at the time of their marriage (in completed years) is also noted in table 4.3.1 (a), which shows that a great majority of the fathers (72.3%) were between 18-30 years and 17.5% reported less than 18 years, while 10.2% of the parents had their age, above 30 years at the time of their marriage. The average age of fathers of sick children was 23.92 years, minimum age was 14 years and maximum age was 35 years at the time of marriage. The data also revealed that majority (68.9%) of the mothers of thalassemic children belonged to 18-30 years age group at the time of marriage (in completed years) while 30.4% were below 18 years and only few (0.8%) reported above 30 years at the time of marriage. The average age of this classification was 21.7 years, maximum age was 35 years and minimum age of mothers was 12 years. Considering lowest age group, as an evidence of early marriages in Pakistan which affects the heath of mothers and neonates (Nasrullah, 2015).

Education is another important variable of socioeconomic status because level of education is significantly related to the quality of life of entire family and higher socioeconomic status. The study found that 16.1% of the fathers of sick children were illiterate in study area, while 11.9% had primary education and 12.0% had secondary education. The data further reveals that 47.1% of the fathers had matriculation, 10.1% had intermediate and only .6% completed graduation level and

very few (2.1%) got university education. The average score of father's educations was 7.65, which is equal to the middle schooling and even less than male literacy rate of middle level (15.2%)[1] and maximum level of education was 18 years.

The data also figured out mother's educational level by highlighting illiterate (40.1%), 14.9% had primary education. 5.6% had middle schooling, 25.1% had matriculation level and 9.8% had intermediate certificate. Only few mothers (2.1%) completed graduation and 2.4% post-graduation in the study area. The average score of mother's educations was 5.64, which is equal to the primary level of education.

[1] Pakistan. Demographic and Health Survey. 2017-18

Table 4.3.1 (b)

*A Territorial Overview of Descriptive Statistics of Age and Educational Status of the Respondents*

| | Upper Punjab N=133 | | Southern Punjab N=250 | | Central Punjab N=549 | |
|---|---|---|---|---|---|---|
| Variables | Father %(f) | Mother %(f) | Father %(f) | Mother %(f) | Father %(f) | Mother %(f) |
| Age at the time of interview (in completed years) | | | | | | |
| <25 | 27.8(15) | 38.0(30) | 22.8(40) | 26.7(20) | 56.2(63) | 5.7(70) |
| 25-40 | 37.0(20) | 40.5(32) | 14.2(25) | 20.0(15) | 40.2(45) | 12.3(55) |
| >40 | 35.2(19) | 21.5(17) | 63.0(110) | 53.3(40) | 3.6(4) | 72.0(322) |
| Age at marriage | | | | | | |
| <18 | 3.7(2) | 7.5(6) | 2.3(4) | 2.7(2) | 10.7(12) | 1.8(8) |
| 18-30 | 77.8( 42) | 63.3(50) | 70.2(123) | 38.7(29) | 75.9(85) | 49.5(221) |
| >30 | 18.5(10) | 29.2(23) | 27.5(48) | 58.6(44) | 13.4(15) | 48.7(218) |
| Education | | | | | | |
| Illiterate | 11.2(6) | 22.8(18) | 42.9(75) | 57.3(43) | 20.5(23) | 30.0(136) |
| Primary | 18.5(10) | 31.7(25) | 20.0(35) | 33.4(25) | 34.0(38) | 8.0(40) |
| Middle | 9.2(5) | 45.5(36) | 8.6(15) | 0 | 11.0(12) | 38.0(160) |
| Matriculation | 22.3(12) | 0 | 5.8(10) | 0 | 19.6(22) | 4.0(18) |
| Intermediate | 14.8(8) | 0 | 4.5(8) | 0 | 13.3(15) | 2.0(10) |
| Graduation | 24.0(13) | 0 | 18.2(32) | 0 | 0 | 3.0(15) |
| Masters and above | 0 | 0 | 0 | 9.3(7) | 1.6(2) | 15.0(68) |

**Note** f=number of parents, %=percentage

The data recorded in table 4.3.1 (b) revealed a territorial descriptive analysis of age and educational status of the respondents of the study. The study found that from three regions of Punjab Province (Upper, Southern and Central) 27.8% of fathers and 38.0% mothers from Upper Punjab belonged to the age group less than 25 years at the time of interview. While, 37.0% fathers and 40.5% mothers belonged to the age group 25-40 years and 35.2% fathers and 21.5% mothers belonged to the age group >40 years at the time of interview. From Southern Punjab region, 22.8% fathers and 26.7% mothers had their age <25 years, 14.2% fathers and 20.0% mothers had age between 25-40 years and 63.0% fathers and 53.3% mothers had their age >40 years at the time of interview. From third region (Central Punjab), 56.2% fathers and 5.7% mothers had their age >25 years while, 40.2% fathers and 12.3% mothers had their age between 25-40 years and only few (3.6%) fathers but majority of the mothers (72.0%) had their age >40 years at the time of interview.

The data also revealed that 3.7% fathers and 7.5% mothers from Upper Punjab belonged to the age group <18 years at the time of their marriage. Furthermore, 77.8% fathers and 63.3% mothers belonged to the age group 18-30 years and 18.5% fathers and 29.2% mothers belonged to the age group >30 years when they married. From Southern Punjab, 2.3% fathers and 2.7% mothers belonged to the age group <18 years while, 70.2% fathers and 38.7% mothers belonged to the age group 18-30 years and 27.5% fathers and 58.6% mothers had their age >40 years when they married. The respondents (10.7 % fathers and 1.8% mothers) belonged to the Central Punjab region had their age <18 years at the time of their marriage. Furthermore, the data revealed that 75.9% fathers and 49.5% mothers had their age 18-30 years and 13.4% fathers and 48.7% mothers had their age >30 years when they married and living in Central Punjab region.

The data recorded in table 4.3.1 (b) also revealed information regarding educational status of the respondents and from 133 respondents selected from Upper Punjab region, 11.2% fathers and 22.8% mothers were found illiterate, 18.5% fathers and 31.7% mothers had primary level of education, 45.5% mothers reached till middle level of education and it was the maximum level of their (mothers') educational status. Although, 22.3% fathers completed matriculation, 14.8% intermediate and 24.0% reached till graduation. From Southern Punjab region 57.3% of the mothers

were illiterate, 33.4% had primary education and only few (9.3%) completed their master's degree. In contrary, 42.9% fathers were illiterate, 20.0% had primary, 8.6% middle, and 4.5% intermediate while, 18.2% had completed graduation level. The study also found that from central Punjab region, 20.5% fathers and 30.0% mothers were liberate, 34.0% fathers and only few (8.0%) mothers had primary education, 11.0% fathers and 38.0% mothers had middle schooling, only few (1.6%) fathers and 15.0% mothers go university education.

Table 4.3.2 (a)

*Descriptive Statistics of Income and Expenditures of the Respondents (N=932)*

| Variables | Frequency(f) | Percentage (%) |
|---|---|---|
| Monthly Income | | |
| <10,000 | 49 | 5.3 |
| 10,000-30,000 | 584 | 62.7 |
| 30,001-40,000 | 112 | 12.0 |
| 40,001-50,000 | 111 | 11.9 |
| >50,000 | 76 | 8.2 |
| | M=29,482.83, Mini=5,000, Maxi= 100,000 | |
| Monthly expenditures | | |
| <10,000 | 123 | 13.2 |
| 10,000-30,000 | 705 | 75.6 |
| 30,001-40,000 | 46 | 4.9 |
| 40,001-50,000 | 45 | 4.8 |
| >50,000 | 13 | 1.4 |
| | M=22,633.05, Mini=2,000, Maxi= 80,000 | |
| Monthly expenditures on treatment of disease | | |
| <100,0 | 163 | 17.5 |
| 100,0-500,0 | 394 | 42.3 |
| 500,1-10,000 | 326 | 35.0 |
| >10,000 | 49 | 5.3 |
| | M=5336.37, Mini=500, Maxi=20,000 | |

**Note** f=number of parents, %=percentage, M=Mean, Mini=Minimum, Maxi=Maximum

Data recorded in table 4.3.2 (a) revealed that 62.7% of the respondents of the present study had average income between 10 thousands to 30,000 (PKRs), while only few (8.2%) had income level more than 50,000. The average monthly income of a family was 29,482.83 (PKRs), which is equal to 188 USD approximately. The maximum level of recorded family income was 100,000 and minimum level of income was 5000. The study also found that 75.6% of the respondents had monthly family expenditures between 10 thousands to 30,000 (PKRs). Very few of them (1.4%) had monthly family expenditures above 50 thousand and the average score of household expenditures was 22,633.83. The study intended to estimate the monthly expenditures of parents for medications and treatment of their sick child. The data recorded in table 4.3.2 (a) revealed that parents on average spent 5336 (PKRs) for the treatment of their children. Although they were provided free blood and medicines form thalassemia foundations, but their expenditures included; travelling and consultancy fees of medical practitioners.

Table 4.3.2 (b)

*A Territorial Overview of Descriptive Statistics of Income and Expenditures of the Respondents*

| | Upper Punjab N=133 | | Southern Punjab N=250 | | Central Punjab N=549 | |
|---|---|---|---|---|---|---|
| Variables | Frequency(f) | Percentage (%) | Frequency(f) | Percentage (%) | Frequency(f) | Percentage (%) |
| Monthly Income | | | | | | |
| <10,000 | 65 | 48.8 | 70 | 28.0 | 312 | 56.8 |
| 10,000-30,000 | 20 | 15.0 | 32 | 12.8 | 55 | 10.0 |
| 30,001-40,000 | 30 | 22.6 | 90 | 36.0 | 165 | 30.0 |
| 40,001-50,000 | 10 | 7.6 | 20 | 8.0 | 10 | 1.9 |
| >50,000 | 8 | 6.0 | 38 | 15.2 | 7 | 1.3 |
| Monthly expenditures | | | | | | |
| <10,000 | 20 | 15.0 | 30 | 12.0 | 70 | 12.8 |
| 10,000-30,000 | 75 | 56.4 | 45 | 18.0 | 130 | 23.7 |
| 30,001-40,000 | 32 | 24.0 | 122 | 48.8 | 125 | 22.8 |
| 40,001-50,000 | 6 | 4.6 | 30 | 12.0 | 179 | 32.6 |
| >50,000 | 0 | 0 | 23 | 9.2 | 45 | 8.1 |
| Monthly expenditures on treatment of disease | | | | | | |
| <100,0 | 45 | 34.0 | 70 | 28.0 | 211 | 38.4 |
| 100,0-500,0 | 36 | 27.0 | 38 | 15.2 | 302 | 55.0 |
| 500,1-10,000 | 40 | 30.0 | 132 | 52.8 | 20 | 3.6 |
| >10,000 | 12 | 9.0 | 10 | 4.0 | 16 | 3.0 |

**Note** f=number of parents, %=percentage

Table 4.3.2 (b) revealed a territorial overview of income and expenditures of the respondents. The study found that from Upper Punjab region, 48.8% of the respondents had their income less than 10 thousands (PKRs), 15.0% has between 10 thousands to 3o thousands (PKRs) and only few (6.0%) of the respondents had their income above 50 thousands (PKRs). From southern Punjab region, 48.8% of the respondents had their income between 10 thousands to 40 thousands (PKRs) and 15.2% of the respondents had their income above 50 thousands (PKRs). Data collected from Central Punjab region revealed that, 56.8% of the respondents had their monthly income less than 10 thousands (PKRs) and only few of them (1.3%) had their income above 50 thousands (PKRs).

The monthly expenditures of the respondents from Upper Punjab region were reported as; 15.0% had less than 10 thousands (PKRs), 80.4% between 10 thousands to 30 thousands (PKRs) and only 4.6% of them had their monthly expenditures above 40 thousands (PKRs). From Southern Punjab, 12.0% had less than 10 thousands (PKRs), a great majority (78.8%) had their monthly expenditures between 10 thousands to 50 thousands (PKRs) and only few (9.2%) had above 50 thousands (PKRs). From third region of Punjab Province (Central Punjab), 12.8% of the respondents had their monthly expenditures less than 10 thousands (PKRs), a great majority (70.1%) had their monthly expenditures between 10 thousands to 50 thousands (PKRs) and only few (8.1%) had monthly family expenditures above 50 thousand (PKRs).

With reference to the monthly expenditures of families on treatment and medication of their sick children, 34.0% of the respondents of Upper Punjab region had less than 1 thousand (PKRs), 57% had their medical expenditures between 1 thousand to 10 thousands (PKRs) and only few (9.0%) reported their monthly medical expenditures as above 10 thousands (PKRs). The data also revealed that 28.0% of the respondents from Southern Punjab and 38.4% of the respondents from Central Punjab had their monthly medical expenditures less than 1 thousands. Furthermore, only few (4.0%) of the respondents from Southern Punjab and 3.0% of the respondents of Central Punjab had their monthly medical expenditures above 10 thousand (PKRs).

## 4.4 Background Information of Beta Thalassemia Major

Background information of beta thalassemia major includes family history of the respondents, symptoms of the disease, genetic disorder and counseling and information about blood groups of patients and their parents (respondents'). The data was collected from 932 parents of those beta thalassemic children in Punjab Province of Pakistan.

Table 4.4.1

*Family history of the Respondents (N=932)*

| Statement | | Frequency (f) | Percent (%) |
|---|---|---|---|
| No. of children suffering from thalassemia | | | |
| | One | 835 | 89.6 |
| | Two | 97 | 10.4 |
| Any other patient of thalassemia | | | |
| | No | 544 | 58.4 |
| | Yes | 388 | 41.6 |

**Note** f=number of parents, %=percentage

Data reported in table 4.4.1 contains the information about family history of respondents. Majority of the people (89.6%) had a one beta thalassemic child but few (10.4%) had two children with beta thalassemia major. The researcher also found that in the family history of the families of sick children that, 41.6% of the people have another patient of beta thalassemia major (either in the family or any other immediate relative) while 58.4% of the respondents do not have other individual suffering from beta thalassemia major in their family or within relatives. The similar findings have been found in a study conducted by (Uddin et al., 2017) on beta thalassemia major, in Karachi city in 2015 and these researchers found that 56% of the respondents of their study had patients of beta thalassemia major in their families. The risk of propagation of beta thalassemia major prompts due to family history and repeated cousin marriages.

Table 4.4.2

Initial Symptoms of Beta Thalassemia Major *(N=932)*

| Symptoms | Frequency (f) | Percent (%) |
|---|---|---|
| Fatigue | 471 | 50.5 |
| Weakness | 71 | 7.6 |
| Pale or yellow skin | 99 | 10.6 |
| Facial bone deformities | 52 | 5.6 |
| Slow growth | 83 | 8.9 |
| Abdominal swelling | 15 | 1.6 |
| Dark urine | 1 | 0.1 |
| Fever | 140 | 15.0 |

**Note** f=number of parents, %=percentage

Parents were asked to report the initial symptoms of their child disease, prior to the diagnosis of beta thalassemia (table 4.4.2). It has been found that half of the children showed fatigue (50.5%) and 7.6% reported weakness. This study found that 10.6% of the children with beta thalassemia flaunted pale yellow skin while 5.6% were brought to the physician because of facial bone deformities, before the diagnosis of beta thalassemia major. Only few (0.1%) and (1.6%) reported dark urine and abdominal swelling respectively, while 15.0% suffered fever and 8.9% showed slow physical growth as compared to their siblings and other children.

Table 4.4.3

*Awareness of Parents about Genetic Disorder and Counseling (N=932)*

| Statement | | Frequency(f) | Percent (%) |
|---|---|---|---|
| Cousin marriages causes genetic disorders | | | |
| | No | 261 | 28.0 |
| | Yes | 671 | 72.0 |
| Normal children of cousin couple can be carrier of genetic diseases | | | |
| | No | 442 | 47.4 |
| | Yes | 490 | 52.6 |
| Acquaintance of genetic counseling | | | |
| | No | 66 | 7.1 |
| | Yes | 866 | 92.9 |

**Note** f=number of parents, %=percentage

Data mentioned in table 4.4.3 encloses the information about parental (respondent's) retorts regarding genetic disorder and counseling. It was found that majority of people (72%) were aware that, cousin marriages are one of the main causes for genetic disorders. The study found that some of the respondents (28%) were of the view that cousin marriages are not one of the causes of genetic disorders.

It has also been found in this study that, respondents have lack of scientific and technical knowledge regarding genetic disorders because almost half of the respondents (47.4%) repudiated that normal children of cousin couples may be the carrier of a genetic disease and the almost same (52.6%) favored this prerogative. Majority of the parents (92.9%) of the thalassemic children were aware of the genetic counseling while few of them (7.1%) were unacquainted about genetic counseling.

The comprehensive information regarding genetic disorder, its cause and preventive strategies is decisive for couples and families especially with higher rate of consanguineous marriages (Zlotogora, Carmi, Lev, & Shalev, 2009). In Pakistan, majority of the people are unaware about the causes of genetic disorders and the importance of genetic counseling. The similar findings have been obtained by (Naseem, Ahmed, & Vahidy, 2008), they found that out of 149 couples, 90 (60%) did not request prenatal diagnosis, 23% of the respondents were illiterate.

Table 4.4.5

*Information of Blood Groups (N=932)*

| Blood group | Sick Child | Mother | Father |
|---|---|---|---|
| | %(f) | %(f) | %(f) |
| A+ | 31.8(296) | 31.7(295) | 18.8(175) |
| A- | 5.4(50) | 9.3(87) | 9.3(87) |
| B+ | 19.7(184) | 19.6(183) | 22.5(210) |
| B- | 4.0(37) | 7.7(72) | 8.7(81) |
| O+ | 24.1(225) | 24.4(227) | 15.8(147) |
| O- | 4.3(40) | 3.3(31) | 17.4(162) |
| AB+ | 8.3(77) | 3.8(35) | 1.5(14) |
| AB- | 2.5(23) | .2(2) | 6.0(56) |

**Note** f=number of parents, %=percentage

Majority of the patients (31.8%) with beta thalassemia major had A+ blood group (table 4.4.5) while the second major category of blood group was O+ (24.1%). Less than one fourth (19.7%) patients with beta thalassemia major had B+ blood group, AB+ (8.3%), A- (5.4%), B- (4.0%), O- (4.3%) and lastly 2.5% had blood group AB-. Additional information regarding blood groups of mothers(s) of sick children are recorded in table 4.4.5. It has been found that resembling the blood groups of children (patients of beta thalassemia major), their mothers were greater in majority (31.7%) with A+ blood group and O+ (24.4%). Same as the patients, their mother's blood group B+ had almost similar values (19.6%). The least number of mothers had blood group AB- (0.2%), after AB+ (3.8%) and O- (3.3%) respectively. The study also valued the data regarding blood groups of the fathers of sick children (table 4.4.5). It has been found that majority of the patient's fathers had B+ blood group (22.5%), O- (17.4%) and O+ (15.8%) accordingly. The least number was reported with AB+ (1.5%) and AB- (6.0%). Some of them were having A- (9.3%) as well as B- (8.7%).

### 4.5 Treatment of Beta Thalassemia Major

The data was collected from the parents of sick children due to beta thalassemia major by inquiring about the first step took by the parents (respondents) after the sickness of their child. Parents were also asked about the ways of treatment, frequency and sources of blood for their children.

Table 4.5

*Treatment of Beta Thalassemia Major (N=932)*

| Statements | Frequency(f) | Percent (%) |
|---|---|---|
| First step taken for the treatment of child | | |
| Govt. hospital | 419 | 45.0 |
| Basic health unit | 138 | 14.8 |
| Private hospital / clinic | 339 | 36.4 |
| Family welfare center | 16 | 1.7 |
| Hakim | 12 | 1.3 |
| Religious healing | 8 | 0.9 |
| Kind of treatment | | |
| Blood transfusion | 850 | 91.2 |
| Chelation therapy | 82 | 8.8 |
| Frequency of blood transfusion (monthly) | | |
| Once | 340 | 36.5 |
| Twice | 450 | 48.3 |
| Thrice | 118 | 12.7 |
| Four times | 18 | 1.9 |
| Five times | 6 | 0.6 |
| Source of blood for transfusion | | |
| Govt. Institute | 22 | 2.4 |
| Private | 91 | 9.8 |
| With the help of relatives | 23 | 2.5 |
| Source of family | 82 | 8.8 |
| Thalassemia Foundation | 714 | 76.6 |

**Note** f=number of parents, %=percentage

Data recorded in table 4.5 revealed the information regarding first step taken by the parents for treatment of beta thalassemia major. Majority of the parents took their sick children to government hospitals (45.0%) and 36.4% to private clinics. Data revealed that only few (1.7%), (1.3%) and (0.9%) went to the family welfare center, *hakim* (local physician) and for religious healing respectively.

The most common ways for the treatment of beta thalassemia major are blood transfusion and chelation therapies (Rachmilewitz & Giardina, 2011). Majority of the patients (91.2%) were depending upon blood transfusion along with chelation therapies (because both ways remain abreast) while 8.8% were only depending on chelation therapies with minor frequency of blood transfusion. The frequency of blood transfusion is associated with the age and severity of the disease (Sarnaik, 2005). It has been evident that with growing age, the patients of beta thalassemia major required vaster frequency of blood transfusion (Drasar et al., 2011). Data recorded in table 4.5 revealed that, majority of the patients (36.5%) and (48.3%) were being transfused once and twice in a month respectively. The present study also found that 12.7% were being transfused three times in a month, while only few (1.9%) and (0.6%) were transfused four and five times on monthly basis respectively.

Arrangement of blood for the patients of beta a very serious challenge for parents and families of the patient (Nagaraj, Umashree, Devarhubli, & Shankara, 2011). Without the support of voluntary organizations and thalassemia foundation, it becomes impossible for parents to collect and manage the considerable amount of blood for the survival of a patient with such a chronic genetic disease (Weatherall & Clegg, 2008). The taxonomies mentioned in table 4.5 are supported with number of parental categories, according to their sources of blood. Majority of the parents (76.6%) were dependent upon the thalassemia foundation (*Sundas Foundation* (all centers), *Jamila Sultana Foundation and Fatimid Foundation*) for the arrangement of blood. Only few respondents (2.4%) were dependent upon the sources provided by the government of Pakistan (government hospitals and blood transfusion centers). Some (9.8%) were managing blood from private transfusion centers and 8.8% relied on donations from their family members. It has been found that only few (2.5%) were getting blood from their relatives.

### 4.6 Parental Knowledge

Data recorded in table 4.6 is about the knowledge of parents regarding thalassemia. The information was drawn based on correct or incorrect responses. Majority of the respondents (89.7%) marked the statement correct, that disease (thalassemia) can be identified by blood examination. In response to the statement related to the other conditions of general illness i.e. fainting, fever, diarrhea and

vomiting can worsen the disease (thalassemia), 73.6% of the parents marked it correct while 26.4% marked incorrect. There were an equal number of parents 51.7% and 48.3% marked incorrect and correct respectively regarding the statement that a patient of thalassemia can spend normal life with appropriate treatment. Majority of the parents (72.4%) were of the view that thalassemia can only be treated with medication.

Table 4.6

*Parental Knowledge of Thalassemia (N=932)*

| Statements | Incorrect %(f) | Correct %(f) | References |
|---|---|---|---|
| Thalassemia can be identified through blood test | 10.3(96) | 89.7(836) | **Correct;** (Cheung, Goldberg, & Kan, 1996; Li et al., 2006; Setsirichok et al., 2012; Silvestroni & Bianco, 1983; Verma, Choudhry, & Jain, 1992) |
| Conditions of general illness i.e., fainting, fever and diarrhea worsen thalassemia major | 26.4(246) | 73.6(686) | **Correct;** (Maheen, Malik, Siddique, & Qidwai, 2015; Wei, Yang, Cheng, & Lo, 2011) |
| A person suffering from beta thalassemia major, lives normal lives with appropriate treatment | 51.7(482) | 48.3(450) | **Correct;** (NIENHUIS et al., 1979; Piomelli & Loew, 1991) |
| Thalassemia can only be treated with medications | 27.6(257) | 72.4(675) | **Correct;** (Di Maggio & Maggio, 2017; Karnon et al., 1999; Rund, 2016; Schrier & Angelucci, 2005) |
| Marriages within the family are significant risk factor for the | 38.3(357) | 61.7(575) | **Correct;** (Akers, Howard, & Ford, 2017; Muhammad, Shakeel, |

| | | | |
|---|---|---|---|
| propagation thalassemia | | | Rehman, & Lodhi, 2017; Okyay, Çelenk, Nazlıcan, & Akbaba, 2016) |
| A person suffering from thalassemia minor lives a normal life | 28.4(265) | 71.6(667) | **Correct;** (Bajwa & Basit, 2019; Haq et al., 2017; Thiyagarajan, Bhattacharya, Sharma, Srivastava, & Dhar, 2019) |
| If one parent (mother/father) is a carrier of thalassemia minor, the chance of having a child with thalassemia disease increases | 37.1(346) | 62.9(586) | **Correct;** (De, 2016; Tahura, Selimuzzaman, & Khan, 2016) |
| Thalassemia is preventable disease | 31.7(295) | 68.3(637) | **Correct;** (Mardhiyah & Sriati, 2018; Rerkswattavorn, Sirachainan, Songdej, Kadegasem, & Chuansumrit, 2018; Ruangvutilert, 2017) |
| Thalassemia is an inherited disorder | 33.6(313) | 66.4(619) | **Correct;** (Husna et al., 2017; Thein, 2018; Weatherall, 2018) |
| Surgery is an effective way to treat thalassemia | 72.7(678) | 27.3(254) | **Correct;** (Haq et al., 2017; Pignatti, Zanella, & Borgna-Pignatti, 2017; Qadir & Rizvi, 2018) |
| Thalassemia passes from parents to | 25.2(235) | 74.8(697) | **Correct;** |

| | | | |
|---|---|---|---|
| children through genes | | | (Kalokairinou, 2007; Lewis, 2012; McGann, Nero, & Ware, 2017) |
| There is a cure for thalassemia major | 70.9(661) | 29.1(271) | **Correct;** (Lin & Lin, 1992; Lucarelli, Andreani, & Angelucci, 2002; Mavrogeni et al., 2018; Srivastava & Shaji, 2017) |
| Thalassemia (any kind) transmits sexually | 15.7(146) | 84.3(786) | **Correct;** (Abdelmawla, Moemen, Darwish, & Mowafy, 2019; Santarone et al., 2017; Xu et al., 2019) |
| Thalassemia minor increases risk of general illnesses | 15.9(148) | 84.1(784) | **Correct;** (Sarnaik, 2005; Stein, Berg, Jones, & Detter, 1984; Vanichsetakul, 2014) |
| Thalassemia can be detected during pregnancy | 19.0(177) | 81.0(755) | **Correct;** (Barrett, Saminathan, & Choolani, 2017; Leung, Lau, & Chung, 2005; Taher, Isma'eel, & Cappellini, 2006; Traeger-Synodinos & Harteveld, 2017) |
| Parents (both mother & father) having thalassemia minor increases the chances of a thalassemic child | 17.7(165) | 82.3(767) | **Correct;** (Barrett et al., 2017; Cebrian, Flores, Álvarez, Salinas, & Iturrate, 2016; Maheen et al., 2015) |
| After identification of thalassemic, child parents should have family planning | 24.5(228) | 75.5(704) | **Correct;** (Gee, Piercy, & Machaczek, 2017; Mardhiyah & Sriati, 2018; Zaheer, Zaman, Iqbal, Hameed, & Wazir, 2015) |

A greater number (61.7%) viewed that marriages within the family are significant risk factor for the propagation thalassemia. A person suffering from thalassemia minor lives a normal life was marked correct by majority of the parents (71.6%). Most of the respondents (62.9%) viewed it correct that, if one parent (mother/father) is a carrier of thalassemia minor, the chance of having a child with thalassemia disease increases, while (33.6) marked it incorrect to be an inherited genetic disorder because of low level of education (see table 4.3.1). Mostly (68.3%) of the parents' marked correct that, thalassemia is preventable disease. Thalassemia can be treated with surgery was viewed as incorrect by most of the parents (72.7%) and almost the same (70.9%) viewed the statement incorrect that there is a cure for thalassemia major. Majority of the respondents (74.8%) marked the statement correct that thalassemia can pass on to the child through a gene and a greater majority (84.3%) was marked correct, that thalassemia is a sexually transmitted disease. Most of the parents (81.0%) were well known that thalassemia can be detected during pregnancy and if parents are having thalassemia minor, it increases the chances of a thalassemic child (82.3% correct). Parents of thalassemic children were of the view that after identification of thalassemic child, parents should have family planning (75.5% correct). The variation in responses of the parents is based on their health literacy and awareness level of beta thalassemia major. It has been found by (Uddin et al., 2017) that 63% respondents of their study were unaware about the practices of treatment and prevention of beta thalassemia major, even after having second child with beta thalassemia.

### 4.7 Prenatal Diagnosis of the Disease (PNDs)

It has been intensively discussed by (Kia, Karami, Mohamadian, & Malehi, 2018; Kohli, 2016; Sadick, 2019) that practices of pre/postnatal diagnosis cannot be ignored for the prevention of beta thalassemia major.

Table 4.7

*Prenatal Diagnosis (PNDs) of the Disease (N=932)*

| Statement | SD | D | U | A | SA |
|---|---|---|---|---|---|
| | %(f) | %(f) | %(f) | %(f) | %(f) |
| Screening before marriage is helpful in identification of thalassemia | 9.5(89) | 1.7(16) | 2.4(22) | 29.8(278) | 56.5(527) |
| Pre-natal screening is helpful in identification of thalassemia | 8.6(80) | 5.7(53) | 16.6(155) | 23.0(214) | 46.1(430) |
| Post-natal screening is helpful in identification of thalassemia | 8.6(80) | 5.7(53) | 16.6(155) | 23.0(214) | 46.1(430) |
| Pregnancy with thalassemia major should be terminated | 8.6(80) | 5.7(53) | 16.6(155) | 23.0(214) | 46.1(430) |
| Parental education regarding thalassemia prevention is important | 2.8(26) | 1.7(16) | 18.2(170) | 27.(256) | 49.(464) |
| Counseling and awareness sessions are important for identification and prevention of thalassemia | 5.5(51) | 1.8(17) | 2.8(26) | 22.6(211) | 67.3(627) |

**Note** f=number of parents, %=percentage, SD= Strongly Disagree, D=Disagree, U=Undecided, A=Agree, SA=Strongly Agree

The data mentioned in table 4.7 exposed that majority of the parents (SA=56.5% & A=29.8) were strongly agreed that there should be the compulsory screening before marriage to detect the carriers of thalassemia. The present study found A greater number of parents (SA=46.1% & A=23.0%), were in a strong favor that government should impose, and couples and families should adopt the pre/post-natal screening mechanisms to prevent beta thalassemia major. The same were in the state that the pregnancy with thalassemia generally and with beta thalassemia major particularly should be terminated

after neonatal detection. Majority of the parents (SA=49.8% & A=27.5%) agreed that parental education regarding thalassemia prevention is important to eradicate thalassemia while only 2.8% were strongly disagreed with the importance of parental education. The importance of educational seminars and health awareness sessions especially, the awareness and counseling for identification and prevention of thalassemia was agreed and strongly agreed by 22.6% and 67.3% respectively. Although the practices of prenatal diagnosis are encouraged by the respondents but a wider gapes regarding knowledge and practices of pre/postnatal diagnosis has been reported by (Naseem, Ghazanfar, & Rashid, 2016). These researchers found that 88.6% of their research participants had knowledge about prenatal diagnosis while only 31.5% were practicing pre/postnatal diagnosis. They also found 40% of their respondents encouraged religious beliefs for pre/postnatal diagnosis.

### 4.8 Socioeconomic Risk Factors

Apart from low financial status, the financial management, social support, regular blood transfusion and medication are very crucial for the management of thalassemia (Mediani, Tiara, & Mardhiyah, 2019). Due to lack of social and technical support from community members and blood donators, the management of beta thalassemia major propagates to its worse end (Arian, Mirmohammadkhani, Ghorbani, & Soleimani, 2019).

The measurement of socioeconomic risk factors of beta thalassemia major (table 4.8) includes; financial constraints, social support, attitude of relatives, family members and doctor, along with social participation of parents. However, the last five statements of socioeconomic risk factors; 5. Thalassemia foundation support you for treatment of the disease, 6. Your friends pay for your child's treatment, 7. Your relatives pay for your treatment, 8. Attitude of doctors is sympathetic during treatment, 9. Attitude of nurses and other medical staff is sympathetic during treatment- were reverse coded for theoretical justification for bivariate and multivariate analysis.

Table 4.8

*Socioeconomic Risk Factors of Beta Thalassemia Major (N=932)*

| Statements | SD | D | U | A | SA |
|---|---|---|---|---|---|
| | %(f) | %(f) | %(f) | %(f) | %(f) |
| Low financial condition is the main hurdle for treatment of thalassemia | 11.4(106) | 4.2(39) | 14.6(136) | 26.0(242) | 43.9(409) |
| Pre-marriage consultation is the only way to prevent the thalassemia incidence | 17.6(164) | 18.6(173) | 16.7(156) | 23.8(222) | 23.3(217) |
| Financial management of the expenditure, for treatment of thalassemia is a problem for parents and families | 7.9(74) | 8.5(79) | 15.2(142) | 26.9(251) | 41.4(386) |
| Your child loses very important days of her/his education during the treatment of disease | 6.2(58) | 4.6(43) | 15.9(148) | 32.3(301) | 41.0(382) |
| Thalassemia foundation support you for treatment of the disease | 3.8(35) | 1.0(9) | 5.5(51) | 24.0(224) | 65.8(613) |
| Your friends pay for your child's treatment | 24.4(227) | 49.8(464) | 2.7(25) | 3.9(36) | 19.3(180) |
| Your relatives pay for your treatment | 39.7(370) | 28.3(264) | 4.7(44) | 14.8(138) | 12.4(116) |
| Attitude of doctors is sympathetic during treatment | 10.6(99) | 5.9(55) | 3.5(33) | 16.8(157) | 63.1(588) |
| Attitude of nurses and other medical staff is sympathetic during treatment | 10.6(99) | 5.9(55) | 3.5(33) | 16.8(157) | 63.1(588) |
| It is difficult to participate in family/community gatherings and events due to the illness of your child | 10.6(99) | 5.9(55) | 3.5(33) | 16.8(157) | 63.1(588) |

**Note** f=number of parents, %=percentage, SD= Strongly Disagree, D=Disagree, U=Undecided, A=Agree, SA=Strongly Agree

The data mentioned in table 4.8 revealed that less than half of the parents of thalassemic children (43.9%) were strongly agreed and 26.0% of them were agreed that low financial condition is the main hurdle for the treatment of beta thalassemia major. They (parents) considered financial management of the disease a very serious problem because of their lower socio-economic status (SA=41.4% & A=26.9 %), while only 7.9% were strongly disagreed and did not consider the financial management as a serious problem. Data recorded in table 4.6 also revealed that due to beta thalassemia major, the child loses very important days of his/her education, due to regular treatment and blood transfusion (SA=41.0% & A=32.3%). This is the reason that majority of the children are out of school (see table 4.1). Majority of the respondents (SA=65.8% & A=24.0%) revealed that thalassemia foundations (Sundas (all centers), Fatimid and Jamila Sultana Foundation) support them for the treatment of their child's disease. Parents of thalassemic children were not getting any considerable financial support from their friends and relatives (SD=24.4% & D=49.8%) and (SD=39.7% & D=28.3%) respectively. It was found in the present study that majority of the respondents were strongly agreed (63.1%), while only (10.6%) were strongly disagreed, that attitude of doctors, nurses and other paramedical staff was sympathetic towards them. The same data was noted in response to their social participation in different communal activities and gatherings.

### 4.9 Consanguinity and Beta Thalassemia Major

The propagation of beta thalassemia major is highly associated with cousin marriages across the world and particularly in Pakistan(S. S. Khan, 2018). It is evident that countries with higher rate of consanguineous marriages are facing alarming propagation of beta thalassemia major (Masilamani et al., 2018). Studies found that repeated cousin marriages increase the chance of beta thalassemia major among children (Saima Ali & Safiullah, 2015; Balobaid, Qari, & Al-Zaidan, 2016; Waheed, Fisher, Awofeso, & Stanley, 2016).

Table 4.9.1

*Respondent's Consanguinity*

| Statement | Frequency (f) | Percent (%) |
|---|---|---|
| Are you married with your cousin | | |
| No | 197 | 21.1 |
| Yes | 735 | 78.9 |

Due to a strong emphasize on endogamy and traditional cultural practices in Pakistan, majority of the respondents of the present study (78.9%) were married with cousins, while only (21.1%) were married out of their family (table 4.9.1). The similar findings have been revealed in the study of (M. S. Khan, Ahmed, Khan, Mushtaq, & Wasim, 2015), that out of 180 respondents, 133 (74%) were married with their cousins and due to the higher rate of cousin marriages, beta thalassemia major is alarmingly propagating in Pakistan. Another study conducted by researchers, (Ishaq, Hasnain Abid, Akhtar, & Mahmood, 2012) in Lahore city found that 81.7% of their respondents were married with their first cousins while, 18.3% were non consanguineous marriages in the mentioned study.

Table 4.9.2

*Nature of Respondent's Consanguinity*

| Nature of Consanguinity | Respondent | Husband's Parents | Husband's Grandparents | Wife's Parents | Wife's Grandparents |
|---|---|---|---|---|---|
| | %(f) | %(f) | %(f) | %(f) | %(f) |
| No information | 21.1(197) | 18.8(175) | 32.7(305) | 27.9(260) | 37.0(345) |
| 1st cousin | 49.4(460) | 37.7(351) | 17.6(164) | 31.4(293) | 12.4(116) |
| 2nd cousin | 10.6(99) | 11.4(106) | 10.8(101) | 10.3(96) | 6.3(59) |
| 3rd cousin | 11.4(106) | 1.8(17) | 7.0(65) | 6.7(62) | 4.0(37) |
| Distant relative | 6.7(62) | 16.3(152) | 11.6(108) | 9.7(90) | 21.0(196) |
| *Baradari* | .9(8) | 14.1(131) | 20.3(189) | 14.1(131) | 19.2(179) |

The detailed distribution of nature of cousin marriages among parents of beta thalassemia major patients is given in table 4.9.2, in which it is evident that majority of the parents were married with their first cousins (49.9%), while (10.6%) were married with second cousin and (11.4%) with third cousin. Data demonstrates that (6.7%) of the respondents were married with their distant relatives and only (.9%) within their *Baradari*[2]. Data recorded reveals that most (37.7%) of the husband's parents were married with their first cousins, 11.4% were married with second cousin while 16.3% were married with distant relatives. The information of 175 cases was not available due to lack of respondent's knowledge about husband's parental consanguinity. It is evident that 14.1% of the husband's parents were married within their *Baradari*, without having any immediate relationship with their spouse. The percentage of first cousin marriage among wife's parents. was 17.6%, with second cousin 10.8%, third cousin was only 7 % while rest of the members 11.6% with distant relatives and 20.3% were married within their *Baradari* respectively. The detailed information about the consanguinity of husband's grandparents is given in table 4.9.2. The data revealed that majority of the people (31.4%) were married with their first cousins, 10.3% were married with second cousin and only 6.7% were married with their third cousin. Meanwhile, 9.7 percent of the people were married with distant relatives, while 14.1% were married within their *Baradari*. Data given in table 4.9.2 elucidated about the nature of the consanguinity of wife's grandparents. Although a very less significant number of people (12.4%, 6.3% & 4.0%) were married with 1$^{st}$, 2$^{nd}$ and 3$^{rd}$ cousins respectively, while 21.0% were married with distant relatives and 19.2% within their *Baradari*.

## 4.10 Cultural Risk Factors of Thalassemia Propagation

The present research was intended to find out the risk factors of beta thalassemia major in Pakistan. Table 4.10 figured out the cultural risk factors of beta thalassemia major in the study area. It has been found by many researchers (Adly & Ebeid, 2015; HF Gharaibeh & Gharaibeh, 2012; Roy & Chatterjee, 2007) that role of culture always

[2] *Brotherhood* originating from the Persian word meaning "Brother". In Pakistan and India, it is used to denote several social strata among South Asian Muslims

remain significant in diagnosis, prevention and management of beta thalassemia major across the world and especially in traditional societies.

Table 4.10

*Cultural Risk Factors of Beta Thalassemia Major (N=932)*

| Statement | SD | D | U | A | SA |
|---|---|---|---|---|---|
| | %(f) | %(f) | %(f) | %(f) | %(f) |
| Religious beliefs play an important role regarding prevention and control of thalassemia | 8.3(77) | 10.1(94) | 14.8(138) | 25.1(234) | 41.7(389) |
| Your religion encourages cousin marriage | 8.5(79) | 27.0(252) | 8.8(82) | 33.0(308) | 22.6(211) |
| Your religion restrains termination of pregnancy | 13.8(129) | 28.3(264) | 11.3(105) | 20.7(193) | 25.9(241) |
| Your religion restrains blood screening | 16.3(152) | 45.7(426) | 10.2(95) | 10.5(98) | 17.3(161) |
| You believe in unnatural forces, unseen powers causing disease | 33.8(315) | 24.7(230) | 7.5(70) | 15.0(140) | 19.0(177) |
| You feel stigmatization in your society due to your thalassemic child | 9.2(86) | 32.8(306) | 6.0(56) | 35.6(332) | 16.3(152) |
| Relationship of a married couple break-up after knowing that they have a thalassemic child | 22.9(213) | 30.7(286) | 2.5(23) | 22.5(210) | 21.5(200) |
| Patriarchy plays an important role regarding treatment and screening of thalassemia | 13.6(127) | 10.9(102) | 4.7(44) | 37.6(350) | 33.2(309) |

**Note** f=number of parents, %=percentage, SD= Strongly Disagree, D=Disagree, U=Undecided, A=Agree, SA=Strongly Agree

Table 4.10 also revealed that majority of the parents (SA=41.7% & A=25.1%) of children with beta thalassemia major in Punjab Province were agreed that religious beliefs play an important role regarding prevention and control of thalassemia (SD=8.3% & D=10.1%). Similarly, the parents were of the view that their religion encourage cousin marriages (SA=22.6 & A=33.0) but a significant number of the respondents 27.0% were also disagreed with this statement. The religious restrictions regarding termination of pregnancy (if fetus is found thalassemic) was almost equally agreed (SA=25.9% & A=20.7%) and disagreed (SD=13.8% & D=28.3%) by the parents. The study found that religion does not restrain blood screening because the facts are clear about the responses of parents (SD=16.3%, D=45.7%, SA=17.3%, A=10.5%). Majority of the respondents of the present study believed that unseen and aberrant forces cannot cause beta thalassemia major (SD=33.8% & D=24.7%). An equal number of the respondents felt stigmatized due to their thalassemic child and the other way around (D=32.8% & A=35.6%). The data also revealed that most of the parents did not believe that relationship of a married couple break-up after knowing that they have a thalassemic child (SD=22.9 & D=30.7%). The study also found that patriarchy plays a significant role regarding treatment, screening and management of beta thalassemia major in Pakistan (SA=33.2% & A=37.6%).

The similar findings have been reported by (Ishaq et al., 2012), that 40% of the respondents did not consider termination of pregnancy as a religiously acceptable way. The role of patriarchy is very evident for diagnosis, counseling and screening of thalassemia (Atkin & Ahmad, 1998; Culley, Rapport, Katbamna, Johnson, & Hudson, 2004; Kromberg, 2018). The results of present study revealed that majority of the respondents were agreed that religious beliefs play an important role regarding prevention and control of thalassemia and more than half of the them accepted that their religion encourages cousin marriage, the same viewed that their religion restrains termination of pregnancy for the prevention of beta thalassemia major in Punjab Province.

Without addressing religious influences and ensuring ethical standards, the preventive measures such as termination of pregnancy, cannot be an effective prevention of beta

thalassemia major. Due to lack of knowledge and awareness, regarding causes of beta thalassemia major, people still believe that supernatural powers and unseen forces can cause this genetic disorder. This also led them to feel stigmatization and encourage the self-denial and pessimism, which further leads to increase psychological and social burden on their lives.

### 4.11 Disease Allied Risk Factors of Beta Thalassemia Major

There are some significant clinical factors which play a vital role for the propagation of beta thalassemia major across the world. The present study figured out the responses of parents of thalassemic children regarding provision of blood, treatment cost, travelling and medication expenditures along with adequacy of blood supply for their children. Researchers have found that families and parents, with low income status face problems of blood transfusion (Karlson et al., 2012) and unable to meet the treatment cost of beta thalassemia major (Amid, Saliba, Taher, & Klaassen, 2015).

Table 4.11

*Disease Allied Risk Factors of Beta Thalassemia Major (N=932)*

| Statements | SD | D | U | A | SA |
|---|---|---|---|---|---|
| | %(f) | %(f) | %(f) | %(f) | %(f) |
| Treatment cost of your thalassemia is affordable | 46.9(437) | 44.8(418) | 0.3(3) | 0.9(8) | 7.1(66) |
| Travelling expenses for treatment of your child are affordable | 46.9(437) | 44.8(418) | 0.3(3) | 0.9(8) | 7.1(66) |
| Hospital expenses for treatment of your child are affordable | 46.9(437) | 44.8(418) | 0.3(3) | 0.9(8) | 7.1(66) |
| Therapeutic facilities are affordable for the prevention of thalassemia | 46.9(437) | 44.8(418) | 4.6(43) | 0.9(8) | 2.8(26) |
| Blood supplies are adequate at the center where your child is transfused | 51.7(482) | 15.7(146) | 16.2(151) | 5.3(49) | 11.2(104) |

**Note** f=number of parents, %=percentage, SD= Strongly Disagree, D=Disagree, U=Undecided, A=Agree, SA=Strongly Agree

Table 4.11 revealed that a greater majority of the parents (SD=46.9% & D=44.8%) were of the view that they cannot bear the treatment cost, hospital expenditures and cost of therapeutic facilities individually. The same number of the parent was of the view that they are unable to manage the travelling expenditures for blood transfusion and regular checkups of their children. This is because of low financial status of the parents (see table 4.3.2). The study found that a very smaller number of parents (SA=7.1% & A=0.9%) considered them to be able to manage the treatment cost of thalassemia, travelling expenditures as well as medication and hospital operating cost. The study also found that majority of the parents of sick children were facing difficulties to manage blood for their children because (SD=51.7%) were strongly disagreed and (D=15.7%) were disagreed about the adequacy of blood at transfusion centers. While only few of them revealed that

blood is adequate at transfusion centers (SA=11.2%), (A=5.3%) strongly agreed and agreed respectively.

## 4.12 Psychosocial Burden of Beta Thalassemia Major

Beta thalassemia major leaves an extensive amount of psychosocial burden on patients and parents (Mazzone, Battaglia, Andreozzi, Romeo, & Mazzone, 2009). Numerous studies have been conducted across the world (Chordiya, Katewa, Sharma, Deopa, & Katewa, 2018; Shahraki-vahed, Firouzkouhi, Abdollahimohammad, & Ghalgaie, 2017; Shosha, 2014) and especially in Pakistan (Ammad, Mubeen, Shah, & Mansoor, 2011; Zaheer et al., 2016) and found the negative impact of beta thalassemia major on psychosocial adjustment of patients and their parents by affecting their minimal standards of life (Shazia Ali, Sabih, Jehan, Anwar, & Javed, 2012; Baraz, Miladinia, & Mosavinouri, 2016). Previous studies measured psychosocial burden of beta thalassemia major on patients, in terms of school absenteeism(Schwartz, Radcliffe, & Barakat, 2009), lack of participation in sports (Haghpanah et al., 2013), feeling of difference (Hajibeigi, Azarkeyvan, Alavian, Lankarani, & Assari, 2009) and lack of social integration (Koutelekos & Haliasos, 2013).

Similarly, the significant amount of burden has been considered on parents of thalassemic children (Canatan, Ratip, Kaptan, & Cosan, 2003), by adding the components such as anxiety (Yalçn et al., 2007), lack of social support (Mashayekhi, Jozdani, Chamak, & Mehni, 2016), weak family interaction (Khurana, Katyal, & Marwaha, 2006), stigmatization, confusion, sense of guilt and a significant effect on family size(Gamberini, Canella, Lucci, Vullo, & Barrai, 1991).

Table 4.12

*Psychosocial Burden of Beta Thalassemia Major (N=932)*

| Statements | SD | D | U | A | SA |
|---|---|---|---|---|---|
| | %(f) | %(f) | %(f) | %(f) | %(f) |
| Thalassemia is affecting education of your child | 6.3(59) | 11.9(111) | 17.7(165) | 24.9(232) | 39.2(365) |
| There is a significant effect of thalassemia on school timing of your child | 8.2(76) | 14.5(135) | 34.5(322) | 20.9(195) | 21.9(204) |
| Your child can participate in sport | 30.6(285) | 38.0(354) | 26.0(242) | 3.3(31) | 2.1(20) |
| You usually feel a certain level of anxiety due to the bad health of your child | 5.5(51) | 10.2(95) | 2.9(27) | 42.6(397) | 38.8(362) |
| You have weak family interactions due to your thalassemic child | 1.1(10) | 35.4(330) | 2.8(26) | 18.8(175) | 42.0(391) |
| You feel social isolation due to your thalassemic child | 3.4(32) | 1.4(13) | 20.2(188) | 30.7(286) | 44.3(413) |
| Thalassemia have a significant effect on your social life | 1.1(10) | 35.4(330) | 2.8(26) | 18.8(175) | 42.0(391) |
| Your child can have the feelings of difference due his/her disease | 1.1(10) | 35.4(330) | 2.8(26) | 18.8(175) | 42.0(391) |
| Your child can have social stigmatization due to thalassemia | 1.1(10) | 35.4(330) | 2.8(26) | 18.8(175) | 42.0(391) |
| Your child has a high-level social integration | 11.2(104) | 0.9(8) | 66.7(622) | 13.2(123) | 8.0(75) |
| Your child is facing bad | 5.5(51) | 10.2(95) | 2.9(27) | 42.6(397) | 38.8(362) |

| | | | | | |
|---|---|---|---|---|---|
| expression of self-image | | | | | |
| You have the feelings of denial | 5.5(51) | 10.2(95) | 2.9(27) | 42.6(397) | 38.8(362) |
| You feel confusion | 5.5(51) | 10.2(95) | 2.9(27) | 42.6(397) | 38.8(362) |
| You have feelings of guilt | 5.5(51) | 10.2(95) | 2.9(27) | 42.6(397) | 38.8(362) |
| You have weak social integration | 10.4(97) | 19.3(180) | 3.3(31) | 34.1(318) | 32.8(306) |
| Thalassemia effected/effect your family size | 2.5(23) | 3.4(32) | 6.7(62) | 39.1(451) | 39.1(364) |

**Note** f=number of parents, %=percentage, SD= Strongly Disagree, D=Disagree, U=Undecided, A=Agree, SA=Strongly Agree

Table 4.12 revealed the data regarding psychosocial burden of beta thalassemia major on parents of sick children in Pakistan and found that thalassemia is affecting education of their child (SA=39.2% & A=24.9%), while (U=17.7%) as most of the children were not going to school because of their disease or not reached at the age to be enrolled for school education (see table 4.1). In the meantime, only few (SD=6.3%, D=11.9%) were not agreed. Beta thalassemia major significantly affects the school timings of sick children (SA=21.9% & A=20.9%), while most of the parents were not sure (U=34.5%) about the effect of disease on school timing because their child has not yet reached the school age. Only few were disagreed (SD=8.2% & D=14.5%) that their children's school timing is not being affected due to beta thalassemia major.

The present study also found that majority (SD= 30.6% & D=38.0%) of the parents of sick children, were not agreed that their children can participate in sport. The results are also supported by the findings of (Huda Gharaibeh, Amarneh, & Zamzam, 2009), because the mentioned research also found that majority (66.0%) children were unable to actively participate in sports and other physical activities due to their sickness. Majority of the parents were agreed (SA=38.8% & A=42.6%) that they feel anxiety due to the sickness of their child, because of intensive care required by sick children, parents remain unable to perform creative and analytical tasks in their professional lives.

Findings of the research conducted by (Canatan et al., 2003) also reported that 82% of their selected parents feel anxiety due to the sickness of their child. This study also comprehends that majority of parents (SA=42.0% & A=18.8%) were agreed, that they have weak family interactions due to their thalassemic child while a considerable number was also disagreed (D=35.4%). The week family interaction could also be the result of nuclear family system because an equal proportion of the respondents were living in joint as well as in nuclear families. The strength of a joint family system is to promote strong social ties (Komter & Knijn, 2006) and in such families, couples are not left alone to face the social and economic challenges, due to any reason. Data mentioned in table 4.12 revealed that majority of the parents feel social isolation due to their thalassemic child (SA= 44.3% & A= 30.7%) whereas only few (SD=3.4% & D=1.4%) disagreed with this statement.

Previous studies of (Mallik et al., 2010) also found that 20.7% parents feel social isolation in their daily life due to the illness of their child because they remained restricted to actively participate in community gatherings and family functions. Another study conducted by (Siddiqui, Ishtiaq, Sajid, & Sajid, 2014) in Karachi City to measure the quality of life of parents and children with beta thalassemia major found that majority of the parents (69.3%) lack social support from their friends and feel social isolation due to their beta thalassemic children.

The overall social life of the parents was also significantly affected by beta thalassemia major (SA=42.0% & A=18.8%) whereas some of the parents were disagreed (D=35.4%). The same figures were computed for feeling of stigmatization of the parents and either the sick child feel different as compare to their normal children. Parental stigmatization has been measured by (Gorakshakar & Colah, 2009) and these scholars also found almost same numeral (20%) in their study. The respondents of this study were unable to make sure about social integration of their child (U=66.7%) either because of low age of their child or the child is being treated kindheartedly by family members.

This study found the same response (SA=38.8%, A=42.6%, D=10.2% & SD=5.5%) for sense of guilt and feeling of parental confusion due to the sickness of their child as

well as the feeling of bad self-expressions of sick children. Whereas majority of the parents were agreed about sense of weak level of social integration (SA=32.8 & A=34.1%) but few of them were disagreed (SD=10.4% % D=19.3%). It has been found that beta thalassemia major significantly affects the family size of those families who have thalassemic children(s) (SA=39.1% & A=39.1%). Whereas only few parents were disagreed (D=3.4% & SD=2.5%) by saying that thalassemia did not affect their family size.

### 4.13 Summary of Univariate Analysis

The analysis of the section included descriptive statistics of demographic characteristics, socioeconomic profiles, background information of genetic disease, treatment practices of beta thalassemia major, parental knowledge, prenatal practices, risk factors (socioeconomic, cultural, disease allied) and psychosocial burden of beta thalassemia major on parents of sick children. The findings revealed that average age of thalassemic children is 8.5 years and majority of the children (86.6%) were out of schools due to their illness. According to the ethnic distribution, majority of the parents belonged to *Punjabi* ethnic groups. The parents have nuclear and joint families and almost equal in percentages meanwhile, most of them were working as a laborer or being self-employed. The study found that 163 mothers of sick children were married below the legal age limit for females in Pakistan (16 years)[3]. Majority of the respondents were living in a low-income stratum and the cost of disease was additional charging in their monthly expenditures. The significant blood group of a majority of the sick children was A+ and the only way of treatment was blood transfusion and chelation therapies.

The empirical evidences found in this study revealed that more than half of the respondents (58.4%) have family history of beta thalassemia major. The study found that majority of the parents of thalassemic children were aware about genetic counselling and they were agreed with the importance of prenatal/postnatal practices of thalassemia. It has been found that although parents emphasized on pre/postnatal diagnosis but their

[3] Child Marriage Restraint Act 1929 (No XIX) Section 2

knowledge about beta thalassemia slanted their practices because 33.6% of the respondents did not consider thalassemia, a genetic disorder and 68.3% viewed that the disease is not preventable. It has also been found that some of them are still not aware that thalassemia can be detected during pregnancy and believed not to terminate the pregnancy if the carrier has been detected during screening.

It has also been found that due to lack of income and resources, parents acknowledged the financial burden of beta thalassemia major due to traveling and hospital expenditures. Albeit, the parents admired thalassemia foundations for their efforts of blood transfusion and lifesavings of their children but the adequate amount of blood for their children often lacks. The scantiness of blood at transfusion centers is based on the less frequent support of community members and blood donors because mainly, thalassemia foundations depend upon academic institutes (universities and colleges) for blood while during summer vacations and examination periods, the closure of these institutes result in shortage of blood for transfusion centers.

The propagation of beta thalassemia major has been found significant among cousin marriages (78.9%) and especially among $1^{st}$ cousins (49.4%). Along with patriarchy and cultural dominancy, role of religious perceptions was also found, because 41.7% were of the view that-religion restrains termination of pregnancy. An intensive amount of psychological and social burden has also been found among parents because majority of them were feeling confusion and denial, along with sense of guilt due to sickness of their child. The studies also found that majority of the parents were feeling anxiety and weak social integration because of their sick child.

**Bivariate Analysis**

Bivariate analysis is a statistical way to analyze and interpret the relationship and differences of two variables. It measures the level of strength and association based on binomial connection of two variables. This is the common way to explain and interpret the results of dependent and independent variables based on demographic variables such as age, gender, residential status, family structure, income and educational level of desired population.

The present study implied some significant and commonly used statistical techniques in the field of social sciences i.e., one sample t-test, Pearson's correlation r and one-way analysis of variance, to understand and figure out the differences and relationship between selected demographic variables and independent variables (parental knowledge, practices of prenatal diagnosis and risk factors of beta thalassemia major) along with the dependent variable (psychosocial burden of the disease on parents of sick children).

The independent t test was applied to check the differences of parental knowledge of beta thalassemia major, practices of pre/postnatal diagnosis, risk factors (socioeconomic, cultural and disease allied) along with psychosocial burden of beta thalassemia major was also examined by applying t test to find out the differences based on gender, area of residence, background information of genetic disorder and consanguinity. The values of means, standard deviations and significance level at 0.05 and 0.001 were reported for each variable.

Furthermore, one-way analysis of variance was also applied for parental knowledge of the disease, practices of pre/postnatal diagnosis risk factors and psychosocial burden of beta thalassemia major for parental ethnicities, family structure of the respondents and occupations of family head.

Table 4.13

*Results of Means, Standard Deviations and t-test for Parental Knowledge of Beta Thalassemia Major by Demographic Variables, Background Information of Genetic Disease and Consanguinity (N=932)*

| Groups | *Parental Knowledge* | | | |
|---|---|---|---|---|
| | N | Mean | SD | *t* |
| Gender | | | | |
| Male | 375 | 11.397 | 3.035 | -1.490 |
| Female | 557 | 11.682 | 2.794 | |
| Locality | | | | |
| Rural | 663 | 11.849 | 2.635 | 4.710** |
| Urban | 269 | 10.755 | 3.422 | |
| BIGD-No. of thalassemic Children | | | | |
| One | 835 | 11.548 | 3.045 | .782 |
| Two | 97 | 11.402 | 1.525 | |
| BIGD-Any other patient of thalassemia | | | | |
| No | 544 | 11.671 | 3.125 | 1.756 |
| Yes | 388 | 11.340 | 2.607 | |
| BIGD-Carriers in cousin marriages | | | | |
| No | 261 | 10.804 | 3.095 | -4.598** |
| Yes | 671 | 11.816 | 2.806 | |
| BIGD-Carriers in non-cousin marriages | | | | |
| No | 442 | 10.463 | 2.961 | -11.212** |
| Yes | 490 | 12.498 | 2.530 | |
| BIGD-Awareness of genetic counseling | | | | |
| No | 66 | 11.757 | 3.018 | .628 |
| Yes | 866 | 11.516 | 2.918 | |
| Consanguinity | | | | |
| No | 197 | 11.934 | 2.924 | 2.167* |
| Yes | 735 | 11.425 | 2.917 | |

**Note** *$p$<.05, **$p$<.001, SD= Standard Deviation, BIGD= Background Information of Genetic Disease

The independent t test was applied to check the differences of parental knowledge of beta thalassemia major regarding gender of the respondents, area of the residence, background information of the genetic disorder and consanguinity.

The respondents of the present study belong to both rural and urban areas of Punjab Province. Level of education and literacy rate of rural communities differ from urban localities, due to lack of access and institutional development (Munawar & Akhter, 2017). The study found substantial differentiations among both communities with regards to knowledge (M=11.849, 10.755 & SD=2.635, 3.422). The study also found significant differences of knowledge on the basis of parental information about carriers of consanguineous marriages (M=10.804, 11.816 & SD=3.095, 2.806) and the existence of thalassemia carriers in non-cousin marriages (M=10.463, 12.498 & SD=2.961, 2.530) along with the classification of respondents, having cousin marriages or not (M=11.934, 11.425 & SD=2.924, 2.917) in study area ($p$<0.05). It has been found that there is no significant difference of knowledge ($p$>0.05) by gender, number of thalassemic children, having any other patient of thalassemia in their (respondent's) family and awareness of genetic counseling (table 4.13).

Table 4.14

*Results of Means, Standard Deviations and t-test for Prenatal Diagnosis of Beta Thalassemia Major by Demographic Variables, Background Information of Genetic Disease and Consanguinity (N=932)*

| Groups | *Prenatal Diagnosis* | | | |
|---|---|---|---|---|
| | N | Mean | SD | *t* |
| Gender | | | | |
| Male | 375 | 24.599 | 4.747 | -.233 |
| Female | 557 | 24.673 | 4.926 | |
| Locality | | | | |
| Rural | 663 | 24.965 | 4.739 | 3.238** |
| Urban | 269 | 23.818 | 4.966 | |
| BIGD-No. of thalassemic Children | | | | |
| One | 835 | 24.893 | 4.829 | 5.382** |
| Two | 97 | 22.402 | 4.251 | |
| BIGD-Any other patient of thalassemia | | | | |
| No | 544 | 25.862 | 3.812 | 9.071** |
| Yes | 388 | 22.912 | 5.537 | |
| BIGD-Carriers in cousin marriages | | | | |
| No | 261 | 24.789 | 5.007 | .598 |
| Yes | 671 | 24.573 | 4.763 | |
| BIGD-Carriers in non-cousin marriages | | | | |
| No | 442 | 23.850 | 5.151 | -4.719** |
| Yes | 490 | 25.340 | 4.409 | |
| BIGD-Awareness of genetic counseling | | | | |
| No | 66 | 23.409 | 3.798 | -2.657** |
| Yes | 866 | 24.727 | 4.889 | |
| Consanguinity | | | | |
| No | 197 | 24.533 | 4.740 | -.335 |
| Yes | 735 | 24.661 | 4.857 | |

**Note ****p*<.05, ***p*<.001, SD= Standard Deviation, BIGD= Background Information of Genetic Disease

The independent t test was applied to check the differences of parental practices of pre/postnatal diagnosis for beta thalassemia major on the basis of gender

of the respondents, area of the residence, background information of the genetic disorder and consanguinity.

The study found significant differences ($p$<0.05) of prenatal diagnosis (table 4.14), on the basis of residential status of the respondents (rural, urban) (M=24.965, 23.818 & SD=4.739, 4.966), number of thalassemic children (M=24.893, 22.402 & SD=4.829, 4.251), any other patient with thalassemia in their (respondent's) family (M=25.862, 22.912 & SD=3.812, 5.537), carrier existence in non-cousin marriages (M=23.850, 25.340 & SD=5.151, 4.409) and parental awareness regarding genetic counseling (M=23.409, 24.727 & SD=3.798, 4.889). While all other variables (gender, carrier's existence in cousin marriages and their consanguinity) were found to be non-significant in terms of prenatal practices of beta thalassemia diagnosis ($p$>0.05).

Table 4.15

*Results of Means, Standard Deviations and t-test for Socio-economic Risk Factors of Beta Thalassemia Major by Demographic Variables, Background Information of Genetic Disease and Consanguinity (N=932)*

| Groups | *Socio-economic Risk Factors* | | | |
|---|---|---|---|---|
| | N | Mean | SD | *t* |
| Gender | | | | |
| Male | 375 | 36.080 | 7.731 | -2.043* |
| Female | 557 | 37.099 | 7.484 | |
| Locality | | | | |
| Rural | 663 | 36.692 | 7.473 | .761 |
| Urban | 269 | 36.260 | 7.999 | |
| BIGD-No. of thalassemic Children | | | | |
| One | 835 | 36.655 | 7.752 | 1.191 |
| Two | 97 | 35.814 | 6.432 | |
| BIGD-Any other patient of thalassemia | | | | |
| No | 544 | 39.156 | 5.560 | 12.503** |
| Yes | 388 | 32.938 | 8.597 | |
| BIGD-Carriers in cousin marriages | | | | |
| No | 261 | 38.034 | 7.277 | 3.777** |
| Yes | 671 | 35.997 | 7.688 | |
| BIGD-Carriers in non-cousin marriages | | | | |
| No | 442 | 36.708 | 7.928 | .532 |
| Yes | 490 | 36.440 | 7.349 | |
| BIGD-Awareness of genetic counseling | | | | |
| No | 66 | 38.409 | 6.773 | 2.269* |
| Yes | 866 | 36.427 | 7.673 | |
| Consanguinity | | | | |
| No | 197 | 39.126 | 5.685 | 6.495** |
| Yes | 735 | 35.881 | 7.931 | |

**Note** *$p$<.05, **$p$<.001, SD= Standard Deviation, BIGD= Background Information of Genetic Disease

The independent t test was applied to check the differences of socioeconomic risk factors of beta thalassemia major on the basis of gender of the respondents, area

of the residence, background information of the genetic disorder and on the basis of consanguinity.

Table 41.5 revealed that respondents (parents of male and female children with beta thalassemia major) have significant differences of socio-economic risk factors, on the basis of their gender (M=36.08, SD=7.731 & M=37.099, SD=7.484), having any other patient of thalassemia in their family (M=39.156, 32.938 & SD=5.560, 8.597), carrier's existence in children of cousin couples (M=38.034, 35.997 & SD=7.277, 7.688), awareness of genetic counseling (M=38.409, 36.427 & SD=6.773, 7.673) and on the basis of their cousin marriages (M=39.126, 35.881 & SD=5.685, 7.931). It has been found that there is no significant difference of socio-economic risk factors of beta thalassemia major based on localities, number of thalassemic children and existence of thalassemia carriers in non-cousin marriages ($p$>0.05).

Table 4.16

*Results of Means, Standard Deviations and t-test for Cultural Risk Factors of Beta Thalassemia Major by Demographic Variables, Background Information of Genetic Disease and Consanguinity (N=932)*

| Groups | *Cultural Risk Factors* | | | |
|---|---|---|---|---|
| | N | Mean | SD | *t* |
| Gender | | | | |
| Male | 375 | 25.780 | 6.026 | 2.563* |
| Female | 557 | 24.816 | 5.452 | |
| Locality | | | | |
| Rural | 663 | 25.813 | 5.756 | 4.170** |
| Urban | 269 | 24.100 | 5.650 | |
| BIGD-No. of thalassemic Children | | | | |
| One | 835 | 25.027 | 5.560 | -3.846** |
| Two | 97 | 27.824 | 6.908 | |
| BIGD-Any other patient of thalassemia | | | | |
| No | 544 | 24.084 | 4.919 | -7.641** |
| Yes | 388 | 27.049 | 6.413 | |
| BIGD-Carriers in cousin marriages | | | | |
| No | 261 | 25.770 | 5.449 | 1.541 |
| Yes | 671 | 25.143 | 5.891 | |
| BIGD-Carriers in non-cousin marriages | | | | |
| No | 442 | 25.884 | 5.350 | 2.871** |
| Yes | 490 | 24.808 | 6.093 | |
| BIGD-Awareness of genetic counseling | | | | |
| No | 66 | 23.303 | 4.158 | -3.950** |
| Yes | 866 | 25.472 | 5.853 | |
| Consanguinity | | | | |
| No | 197 | 24.502 | 6.077 | -2.152* |
| Yes | 735 | 25.537 | 5.675 | |

**Note** *$p$<.05, **$p$<.001, SD= Standard Deviation, BIGD= Background Information of Genetic Disease

The independent t test was applied to check the differences of cultural risk factors of the disease (beta thalassemia major) based on of gender of the respondents,

area of the residence, background information of the genetic disorder and on the basis of consanguinity.

Data labeled in table 4.16 revealed that respondents of this study had a significant difference ($p<0.05$, $p<0.01$) of cultural risk factors of beta thalassemia major on the basis of their gender (M=25.780, SD=6.02), localities (M=25.813, 24.100 & SD=5.756, 5.650), number of thalassemic children (M=25.027, 27.824 & SD=5.560, 6.908), having any other patient of thalassemia in their family M= 24.084, 27.049 & SD=4.919, 6.413), awareness of genetic counseling (M=23.303, 25.472 & SD=4.158, 5.853), carrier existence in non-cousin marriages (M=25.884, 24.808 & SD=5.350, 6.093), their cousin marriages (M=24.502, 25.537 & SD=6.077, 5.675). The data depicts that there is no significant difference ($p>0.05$) of cultural risk factors of thalassemia propagation, based on groups (carrier existence of genetic disorders in cousin marriages).

Culture influences rural and urban peoples differently because of their life styles, access to the health facilities and their exposure to manage genetic disorders like beta thalassemia major. The marriage patterns also vary among people of urban and rural areas because the severe higher rate of cousin marriages has been found in rural areas, as compared to urban localities of Pakistan (Khan & Mazhar, 2018).

Table 4.17

*Results of Means, Standard Deviations and t-test for Disease Allied Risk Factors of Beta Thalassemia Major by Demographic Variables, Background Information of Genetic Disease and Consanguinity (N=932)*

| Groups | *Disease Allied Risk Factors* | | | |
|---|---|---|---|---|
| | N | Mean | SD | *t* |
| Gender | | | | |
| Male | 375 | 9.284 | 4.192 | 1.867 |
| Female | 557 | 8.805 | 3.636 | |
| Locality | | | | |
| Rural | 663 | 9.163 | 3.998 | 1.346 |
| Urban | 269 | 8.788 | 3.792 | |
| BIGD-No. of thalassemic Children | | | | |
| One | 835 | 8.925 | 3.903 | -2.829** |
| Two | 97 | 10.164 | 4.104 | |
| BIGD-Any other patient of thalassemia | | | | |
| No | 544 | 8.536 | 3.090 | -4.484** |
| Yes | 388 | 9.780 | 4.802 | |
| BIGD-Carriers in cousin marriages | | | | |
| No | 261 | 9.743 | 4.200 | 3.203** |
| Yes | 671 | 8.786 | 3.805 | |
| BIGD-Carriers in non-cousin marriages | | | | |
| No | 442 | 9.153 | 3.863 | .731 |
| Yes | 490 | 8.965 | 4.011 | |
| BIGD-Awareness of genetic counseling | | | | |
| No | 66 | 8.803 | 3.903 | -.543 |
| Yes | 866 | 9.073 | 3.945 | |
| Consanguinity | | | | |
| No | 197 | 8.538 | 2.807 | -2.593* |
| Yes | 735 | 9.193 | 4.183 | |

**Note** *$p$<.05, **$p$<.001, SD= Standard Deviation, BIGD= Background Information of Genetic Disease

The independent t test was applied to check the differences of disease allied risk factors of beta thalassemia major on the basis of gender of the respondents, area

of the residence, background information of the genetic disorder and on the basis of consanguinity.

The data recorded in table 4.17 revealed, that there are significant dissimilarities of disease allied risk factors of beta thalassemia major, among the respondents of present research, on the basis of their number of thalassemic children (M=8.925, 10.164 & SD=3.903, 4.104), any other patient of thalassemia in their family (M=8.536, 9.780 & SD=3.090, 4.802) existence of genetic disorder among cousin marriages (M=9.743, 8.786 & SD=4.200, 3.805) and on the basis of their cousin marriages (M=8.538, 9.193 & SD=2.807, 4.183). The data revealed that all other groups have no significant differences regarding disease allied risk factors of beta thalassemia major in Pakistan ($p$>0.05).

Respondents' number of thalassemic children (either one or two) and existence of any other patient of thalassemia in their families are the components of family history in this study. It has been found in this study that disease allied risk factors have significant difference based on these two aspects (see table 4.17). Since parents of sick children become more cognizant and gain more scientific knowledge about the causes and prevention of beta thalassemia major after having second child with thalassemia. The differences exist because parents become able to find out the multiple sources of blood transfusion and they can get help from different blood transfusion centers, on the strength of their enhanced social capital.

Table 4.18

*Results of Means, Standard Deviations and t-test for Psychosocial Burden of Beta Thalassemia Major by Demographic Variables, Background Information of Genetic Disease and Consanguinity (N=932)*

| Groups | *Psychosocial Burden* | | | |
|---|---|---|---|---|
| | N | Mean | SD | *t* |
| Gender | | | | |
| Male | 375 | 58.837 | 9.410 | .383 |
| Female | 557 | 58.596 | 9.751 | |
| Locality | | | | |
| Rural | 663 | 59.710 | 9.426 | 4.996** |
| Urban | 269 | 56.286 | 9.505 | |
| BIGD-No. of thalassemic Children | | | | |
| One | 835 | 58.116 | 9.579 | -6.805** |
| Two | 97 | 63.938 | 7.767 | |
| BIGD-Any other patient of thalassemia | | | | |
| No | 544 | 59.358 | 8.295 | 2.299* |
| Yes | 388 | 57.829 | 11.064 | |
| BIGD-Carriers in cousin marriages | | | | |
| No | 261 | 59.942 | 8.772 | 2.559* |
| Yes | 671 | 58.247 | 9.828 | |
| BIGD-Carriers in non-cousin marriages | | | | |
| No | 442 | 58.798 | 9.211 | .233 |
| Yes | 490 | 58.653 | 9.891 | |
| BIGD-Awareness of genetic counseling | | | | |
| No | 66 | 62.530 | 7.045 | 4.419** |
| Yes | 866 | 58.431 | 9.677 | |
| Consanguinity | | | | |
| No | 197 | 63.1066 | 8.385 | 8.020** |
| Yes | 735 | 57.5469 | 9.531 | |

**Note** *$p$<.05, **$p$<.001, SD= Standard Deviation, BIGD= Background Information of Genetic Disease

One of the major intentions of this study was, to find out the differences among respondents of the study for psychosocial burden of beta thalassemia major.

The independent t test was applied to check the differences of psychosocial burden of beta thalassemia major on the basis of gender of the respondents, area of the residence, background information of the genetic disorder and on the basis of consanguinity.

The study found significant differences of psychosocial burden of beta thalassemia major, on the basis of residential area (rural, urban) (M=59.710, 56.286 & SD=9.426, 9.505), number of thalassemic children (M=58.116, 63.938 & SD=9.579, 7.767) and respondents having any other patient of thalassemia in his/her family (M=59.358, 57.829 & SD=8.295, 11.064). The data mentioned in table 4.18 also reveals that there are significant differences of psychosocial burden of beta thalassemia major on parents and respondents, on the basis of their groups e.g. existence of genetic disorder in cousin marriages (M=59.942, 58.247 & SD=8.772, 9.828), awareness of genetic counseling (M=62.530, 58.431& SD=7.045, 9.677) and on the basis of consanguinity (M=63.1066, 57.5469 & SD=8.385, 9.531). Furthermore, it has also been found that there is no significant difference of psychosocial burden of beta thalassemia major, based on gender of the respondents and their grouping regarding existence of genetic disorder in non-cousin marriages ($p$>0.05).

Table 4.19

*One-Way Analysis of Variance of Parental Knowledge, Prenatal Diagnosis, Risk Factors and Psychosocial Burden of Beta Thalassemia Major by Father's Ethnicity (N=932)*

| Variables | Sources | SS | df | MS | *F* |
|---|---|---|---|---|---|
| | Between Groups | 267.659 | 4 | 66.915 | 8.062** |
| Knowledge | Within Groups | 7694.310 | 927 | 8.300 | |
| | Total | 7961.969 | 931 | | |
| | Between Groups | 305.908 | 4 | 76.477 | 3.309* |
| PND | Within Groups | 21422.327 | 927 | 23.109 | |
| | Total | 21728.235 | 931 | | |
| | Between Groups | 1611.895 | 4 | 402.974 | 7.110** |
| SERF | Within Groups | 52540.846 | 927 | 56.678 | |
| | Total | 54152.741 | 931 | | |
| | Between Groups | 549.629 | 4 | 137.407 | 4.176** |
| CRF | Within Groups | 30502.727 | 927 | 32.905 | |
| | Total | 31052.355 | 931 | | |
| | Between Groups | 253.911 | 4 | 63.478 | 4.142** |
| DARF | Within Groups | 14206.299 | 927 | 15.325 | |
| | Total | 14460.209 | 931 | | |
| | Between Groups | 1002.177 | 4 | 250.544 | 2.756* |
| PSB | Within Groups | 84266.848 | 927 | 90.903 | |
| | Total | 85269.025 | 931 | | |

**Note** *$p$<.05, **$p$<.001, PND=Prenatal Diagnosis, SERF=Socio-economic Risk Factors, CRF=Cultural Risk Factors, DARF= Disease Allied Risk Factors, PSBT=Psychosocial Burden of Beta Thalassemia, SS=Sum of Squares, MS= Mean Square

Table 4.19 revealed the results of one-way analysis of variance for parental knowledge, Prenatal diagnosis, risk factors and psychosocial burden of beta thalassemia major by different ethnicities of father(s). It has been found that there are highly significant average differences of mean among ethnicities with regard to the parental knowledge of beta thalassemia major $F$ (4,927) =8.062, socioeconomic risk factors $F$ (4,927) =7.110, cultural risk factors $F$ (4,927) =4.176 and disease allied risk factors $F$ (4,927) =4.142 because $p$<0.01. It has also been found in this study, that

ethnic groups of fathers, significantly differ in practices of prenatal diagnosis $F$ (4, 927) = 3.309 and in psychosocial burden of beta thalassemia major $F$ (4, (27) = 2.756, $p$<0.05.

This shows that due to differences of social and cultural patterns in different ethnic groups of Punjab, the risk factors, preventive strategies and management of beta thalassemia major vary. All these ethnic groups have their own behavioral, cultural and social patterns of living, so their understanding and management of beta thalassemia major may also vary. The ethnic variations include marriage practices, religious slants, as well as the understanding and treatment of genetic disorders like beta thalassemia major. The differences among ethnic groups, in terms of knowledge and practices of beta thalassemia major have been found by (Maheen, Malik, Siddique, & Qidwai, 2015). The selected ethnic groups of mentioned study were Urdu speaking groups, Saraiki and Pathan, the results revealed there were significant differences of knowledge scores of thalassemia among all groups.

Table 4.20

*One-Way Analysis of Variance of Parental Knowledge, Prenatal Diagnosis, Risk Factors and Psychosocial Burden of Beta Thalassemia Major by Mother's Ethnicity (N=932)*

| Variables | Source | SS | df | MS | *F* |
|---|---|---|---|---|---|
| | Between Groups | 147.926 | 4 | 36.981 | 4.387** |
| Knowledge | Within Groups | 7814.043 | 927 | 8.429 | |
| | Total | 7961.969 | 931 | | |
| | Between Groups | 340.103 | 4 | 85.026 | 3.685** |
| PND | Within Groups | 21388.132 | 927 | 23.072 | |
| | Total | 21728.235 | 931 | | |
| | Between Groups | 1722.550 | 4 | 430.638 | 7.614** |
| SERF | Within Groups | 52430.191 | 927 | 56.559 | |
| | Total | 54152.741 | 931 | | |
| | Between Groups | 581.955 | 4 | 145.489 | 4.426** |
| CRF | Within Groups | 30470.400 | 927 | 32.870 | |
| | Total | 31052.355 | 931 | | |
| | Between Groups | 163.839 | 4 | 40.960 | 2.656* |
| DARF | Within Groups | 14296.370 | 927 | 15.422 | |
| | Total | 14460.209 | 931 | | |
| | Between Groups | 1253.340 | 4 | 313.335 | 3.457** |
| PSB | Within Groups | 84015.685 | 927 | 90.632 | |
| | Total | 85269.025 | 931 | | |

**Note** *$p$<.05, **$p$<.001, PND=Prenatal Diagnosis, SERF=Socio-economic Risk Factors, CRF=Cultural Risk Factors, DARF= Disease Allied Risk Factors, PSBT=Psychosocial Burden of Beta Thalassemia, SS=Sum of Squares, MS= Mean Square

Data recorded in table 4.20 revealed the average difference of mean for parental knowledge, prenatal practices, risk factors and psychosocial burden of beta thalassemia major, among different ethnicities of mothers of sick children. The data reveals that parental knowledge $F$ (4,927) = 4.378, prenatal diagnosis $F$ (4, 927) = 3.685, socioeconomic risk factors $F$ (4, 927) = 7.614, cultural risk factors $F$ (4, 927) = 4.426 and psychosocial burden of beta thalassemia major $F$ (4, 927) = 3.457 have highly significant differences ($p$<0.01) on the basis of mothers' ethnicities in study

area. The study also found significant differences of disease allied risk factors of beta thalassemia major F (4, 927) = 2.656, $p$<0.05.

Similarly, as the results of one-way ANOVA reported in table 4.19, for fathers' ethnicities, the mean difference of mothers' ethnicities was also evident because of social and cultural backgrounds. The influence of endogamy and marriages with same casts and ethnicities are not same for all ethnic groups of selected population for the present study. The differences of knowledge and practices for the prevention of beta thalassemia major, among different ethnic groups have also been found by (Chawla, Singh, Lakkakula, & Vadlamudi, 2017) in India and these researchers also found significant influence of religious and cultural aspects on management and propagation of beta thalassemia in selected study area.

Table 4.21

*One-Way Analysis of Variance of Parental Knowledge, Prenatal Diagnosis, Risk Factors and Psychosocial Burden of Beta Thalassemia Major by Family Structure of the Respondents (N=932)*

| Variables | Source | SS | df | MS | *F* |
|---|---|---|---|---|---|
| | Between Groups | 87.984 | 2 | 43.992 | 5.190** |
| Knowledge | Within Groups | 7873.985 | 929 | 8.476 | |
| | Total | 7961.969 | 931 | | |
| | Between Groups | 470.450 | 2 | 235.225 | 10.280** |
| PND | Within Groups | 21257.785 | 929 | 22.882 | |
| | Total | 21728.235 | 931 | | |
| | Between Groups | 145.194 | 2 | 72.597 | 1.249 |
| SERF | Within Groups | 54007.548 | 929 | 58.135 | |
| | Total | 54152.741 | 931 | | |
| | Between Groups | 269.328 | 2 | 134.664 | 4.064* |
| CRF | Within Groups | 30783.028 | 929 | 33.136 | |
| | Total | 31052.355 | 931 | | |
| | Between Groups | 87.521 | 2 | 43.760 | 2.829 |
| DARF | Within Groups | 14372.689 | 929 | 15.471 | |
| | Total | 14460.209 | 931 | | |
| | Between Groups | 3440.139 | 2 | 1720.070 | 19.528** |
| PSB | Within Groups | 81828.886 | 929 | 88.083 | |
| | Total | 85269.025 | 931 | | |

**Note** *$p$<.05, **$p$<.001, PND=Prenatal Diagnosis, SERF=Socio-economic Risk Factors, CRF=Cultural Risk Factors, DARF= Disease Allied Risk Factors, PSBT=Psychosocial Burden of Beta Thalassemia, SS=Sum of Squares, MS= Mean Square

Table 4.21 revealed the results of one-way analysis of variance of parental knowledge of the disease, practices of pre/postnatal diagnosis, socioeconomic risk factors, cultural risk factors, disease allied risk factors and psychosocial burden, the study found significant differences of knowledge $F$ (2, 929) = 5.190, prenatal diagnosis $F$ (2, 929) = 10.280, Psychosocial burden $F$ (2, 929) = 19.528 and cultural risk factors of beta thalassemia major $F$ (2, 929) = 4.064 as $p$<0.01 and $p$<0.05 accordingly. However, the data divulged that there is no significant difference of

socioeconomic and disease allied risk factors of beta thalassemia major, based on family structure of the respondents.

The importance of family structure for the management and propagation of thalassemia has been found very significant in the study of (Kumar et al., 2019). Parents and couples of joint and extended families have different life styles, level of education and practices of prenatal diagnosis, as compared to the nuclear families. Meanwhile, culture plays a significant role to shape our lives and influence the practices and decision-making abilities of its partakers. The other studies also found that couples belong to joint, nuclear and extended families differ in having knowledge about beta thalassemia major (Biswas et al., 2019), they also have difference of pre/post-natal practices for screening and diagnosis of beta thalassemia major. It has been found by (Anum & Dasti, 2016), that couples belong to nuclear families, significantly differs with other family structures (joint/extended) in terms perceiving psychosocial burden of beta thalassemia major.

Table 4.22

*One-Way Analysis of Variance of Parental Knowledge, Prenatal Diagnosis, Risk Factors and Psychosocial Burden of Beta Thalassemia Major by Occupation of the Family Head (N=932)*

| Variables | Source | Sum of Squares | df | Mean Square | *F* |
|---|---|---|---|---|---|
| | Between Groups | 194.382 | 3 | 64.794 | 7.741** |
| Knowledge | Within Groups | 7767.587 | 928 | 8.370 | |
| | Total | 7961.969 | 931 | | |
| | Between Groups | 1675.363 | 3 | 558.454 | 25.844** |
| PND | Within Groups | 20052.872 | 928 | 21.609 | |
| | Total | 21728.235 | 931 | | |
| | Between Groups | 13852.232 | 3 | 4617.411 | 16.325** |
| SERF | Within Groups | 40300.509 | 928 | 43.427 | |
| | Total | 54152.741 | 931 | | |
| | Between Groups | 6318.908 | 3 | 2106.303 | 79.029** |
| CRF | Within Groups | 24733.447 | 928 | 26.652 | |
| | Total | 31052.355 | 931 | | |
| | Between Groups | 295.747 | 3 | 98.582 | 6.459** |
| DARF | Within Groups | 14164.462 | 928 | 15.263 | |
| | Total | 14460.209 | 931 | | |
| | Between Groups | 3052.328 | 3 | 1017.443 | 11.484** |
| PSB | Within Groups | 82216.697 | 928 | 88.596 | |
| | Total | 85269.025 | 931 | | |

**Note** *$p<.05$, **$p<.001$, PND=Prenatal Diagnosis, SERF=Socio-economic Risk Factors, CRF=Cultural Risk Factors, DARF= Disease Allied Risk Factors, PSBT=Psychosocial Burden of Beta Thalassemia, SS=Sum of Squares, MS= Mean Square

Table 4.22 enclosed the results of one-way analysis of variance of parental knowledge of the disease, practices of pre/postnatal diagnosis, risk factors and psychosocial burden of beta thalassemia major. The study found highly significant differences ($p<0.01$) of means in parental knowledge $F$ (3, 928) = 7.741, practices of prenatal/postnatal diagnosis $F$ (3, 928) = 25.844, socio-economic risk factors $F$ (3, 928) = 16.325, cultural risk factors $F$ (3, 928) = 79.029, disease allied risk factors $F$

(3,928) = 6.459 and psychosocial burden of beta thalassemia major $F$ (3, 928) = 11.484 among different occupational groups (see table 4.22). The reported significant differences have also been supported by the findings of (Basu, 2015), because the researcher also found that based on different occupation, the parents of thalassemic children significantly differ in knowledge, practices, quality of life and other aspects of the disease ($p$<0.05). The respondents of this study have different occupations (see table 4.2) and due to diverse socioeconomic background, their understanding and experiences of the risk factors and psychosocial burden of beta thalassemia major vary.

Table 4.23

*Pearson's r Correlation between Demographic Variables, Parental Knowledge, Prenatal Diagnosis, Risk Factors and Psychosocial Burden of Beta Thalassemia Major (N=932)*

| Demographic Variables | Knowledge | PND | SERF | CRF | DARF | PSB |
|---|---|---|---|---|---|---|
| Father's Age | .013 | -.010 | .016 | .005 | .032 | .050 |
| Mother's Age | -.031 | .020 | .017 | .129** | .080* | .380* |
| FATM | .078* | .016 | .041 | -.014 | -.013 | -.284* |
| MATM | .025 | .040 | .040 | .039 | -.022 | -.103** |
| Father's Education | .072* | .158** | .149** | .002 | -.047 | .048 |
| Mother's Education | .071* | .263** | .318** | -.244** | -.187** | -.458** |
| MIF | .042 | .126** | .014 | .068* | .020 | -.658** |
| MEF | -.016 | .088** | .006 | .028 | .055 | .374* |
| MEM | .085* | .056 | .039 | .089** | .020 | .012 |

**Note** *$p$<.05, **$p$<.001, PND=Prenatal Diagnosis, SERF=Socio-economic Risk Factors, CRF=Cultural Risk Factors, DARF= Disease Allied Risk Factors, PSB=Psychosocial Burden of Beta Thalassemia, FATM=Father's age at the time of marriage, MATM=Mother's age at the time marriage, MIF=Monthly income of family (PKRs), MEF=Monthly expenditure of family (PKRs), MEM=Monthly expenditure of medication (PKRs).

The results recorded in table 4.23 revealed analysis of Pearson's Correlation between demographic variables, parental knowledge, prenatal diagnosis, risk factors (socio-economic, cultural, disease allied) and psychosocial burden of beta thalassemia major and show that there is a significant positive relationship between parental knowledge of beta thalassemia major and father's age at the time of marriage (in completed years) ($r$=.078, $p$<0.05), which shows a positive relationship between two variables. Father(s) of sick children having higher age at the time of their marriages have greater knowledge regarding beta thalassemia major. The parental knowledge of beta thalassemia major has been significantly positively associated with education of fathers' ($r$=.072, $p$<0.05) and mothers', ($r$=.071, $p$<0.05).

The data shows that parents of sick children, having higher education have higher scores of knowledge in the study area. It has also been found by (Hashemi-Soteh et al., 2019), that parental knowledge has a significant positive relationship with educational level of parents and care givers of children with beta thalassemia major. Monthly expenditures on medications of sick children has also been found in highly positive and significant relationship with parental knowledge ($r$=.085, $p$<0.01). This shows that parents with appropriate knowledge are spending more on the medication and treatment of their children (see table 4.23).

Data recorded in table 4.23 also revealed that parental practices of prenatal diagnosis are highly significantly and positively linked with fathers' ($r$=.158, p<0.01) and mothers' education ($r$=.263, p<0.01). Based on these findings, it can be stated that parents, with higher level of education are strongly linked with effective preventive practices of beta thalassemia major.

The similar findings had been reported by (Al Sabbah et al., 2017) in Palestine, they found a significant relationship (p<0.05) between mother's education and practices of prenatal diagnosis, especially termination of pregnancy. These scholars found that there is a direct and positive relationship between education of parents and practices of prenatal diagnosis.

The study also found a significantly highly and positive association between monthly expenditures of a family and prenatal diagnosis of beta thalassemia major ($r$=.088,

*p*<0.01). It means that the effective and standardize prenatal practices are showing up in financial expenditures of the families. The data also revealed that parental education is significantly positively allied with socio-economic risk factors of beta thalassemia major (*p*<0.01), because fathers' and mothers' education have higher value of Pearson correlation (*r*=.149 and .318) respectively. While the all other demographic variables have no significant relationship with socio-economic risk factors of the disease. The study found that cultural risk factors of the disease are highly significantly and positively linked with mothers' age (*r*=.129, *p*<0.01). Mothers with growing age are more likely to follow cultural restrictions and values for the management and prevention of beta thalassemia major.

It has been found that there was a significant negative association between mothers' education and cultural risk factors (r=-.244, p<0.01). The relationship is strong but negative in its natures, which shows that higher educational level of mothers reduces the endorsement of cultural beliefs, values and restrictions for the prevention and management of beta thalassemia major (Widayanti et al., 2011). The screening and prevention of beta thalassemia major is highly sensitized by cultural values and ethical concerns of peoples so, without addressing these apprehensions, the effective implementation of any preventive measure of thalassemia remains vague and unsuccessful. The successful and effective prevention of beta thalassemia major can be effectuated by educating the mothers of a community. Data mentioned in table 4.23 also revealed that monthly income of a family has a significant positive relationship with cultural risk factors of beta thalassemia major (*r*=.068, *p*<0.05). These factors have highly significant and positive relationship with monthly expenditures of a family, on medications and treatment of sick children (*r*=0.89, *p*<0.01).

This study also found that disease allied risk factors have a significant positive relationship with mothers' age (*r*=.080, *p*<0.05), while strong and negative relationship with education of mothers (*r*=-.187, *p*<0.01). These relationships are very strong in their nature. With growing age, mothers become aware about the management of beta thalassemia major and can have better understating of the attitude of doctors and paramedical staffs. However, after having higher level of

education, they become less concerned to gain sympathies and can confiscate their dependencies on nurses and doctors.

The result of Pearson's correlation revealed that psychosocial burden of beta thalassemia major has a significant positive relationship with mothers' age ($r$=.380, $p$<0.05). Moreover, significant negative relationship with mothers' age at the time of marriages ($r$=-.103, $p$<0.01) and fathers' age at the time of marriage ($r$=-.284, $p$<0.05) respectively (see table 4.23). The present study also found that was a significantly negative association between the education of mothers of thalassemic children and psychosocial burden of the disease ($r$=-.458, $p$<0.01).

The similar findings have been reported by (Maheri et al., 2018) in Iran, particularly focusing the two major components (depression and anxiety) of psychosocial burden of beta thalassemia major. These researchers found that higher level of parental education resulted in lower level of selected components.

The data of this study also revealed, that monthly income of a family has a significant negative ($r$=-.658, $p$<0.01) but expenditures have a significant positive relationship ($r$=.374, $p$<0.05) with psychological and social burden of the disease. The psychological and social burden of beta thalassemia major may increase for the women with old age because of higher level of social commitments and sense of realization about the causes and consequences of this genetic disorder. Young people are less likely to feel the burden in terms of social and psychological aspects of their lives because they have more abilities to understand and manage this chronic illness. It is also evident from the results of this study, that the psychological and social burden of the disease can be reduced by increasing level of education for parents and sick children. The direct and positive connection of psychosocial burden of beta thalassemia major and financial expenditures has also been found in this study.

### 4.14. Summary of Bivariate Analysis

The present study found that there are significant differences of knowledge; practices of pre/postnatal diagnosis and psychosocial burden of beta thalassemia major based on residential status of the respondents (see results of independent sample t-test). The other significant differences of prenatal diagnosis, cultural risk factors, disease allied risk factors and psychosocial burden of beta thalassemia major

do exit based on family history of thalassemia ($p$<0.01, 0.05). The study also found significant differences ($p$<.0.1, 0.05) of parental knowledge, prenatal practices, risk factors and psychosocial burden of beta thalassemia major among parents of sick children, by applying one-way analysis of variance for different ethnic groups *(Punjabi, Pashtun, Sindhi, Balochi and Kashmiri)*. The ANOVA also measured significant differences ($p$<.0.1, 0.05) of knowledge, prenatal practices, cultural risk factors and psychological and social burden among the desired population of this study, due to the sickness of their children. Respondents were selected from seven different blood transfusion centers of Punjab Province, with diverse occupational taxonomy. Based on different occupational categories (Self-employee, Private Job, Government employment and working as a Laborers), the significant differences ($p$<.0.1, 0.05) between independent variables and dependent variable has been measured by their mean differences. The study found that there exists a significantly positive association between parental knowledge of the disease and father's age at the time of marriage (in completed years), father's education, mother's education and monthly expenditures on medications of sick children ($p$<0.05). The empirical evidences of this study also revealed that practices of prenatal diagnosis (PNDs) are significantly positively associated with father's and mother's education, monthly income and expenditures of a family ($p$<0.01). The findings of this study also revealed that mothers age and monthly expenditures on medication of a child are significantly positively associated with cultural risk factors of beta thalassemia major, while mother's education is in significantly negative association with cultural and disease allied risk factors of beta thalassemia major ($p$<0.01). The data also revealed that monthly income of a family has a positive relationship with cultural risk factors of beta thalassemia major ($p$<0.05). The results of this study found that mother's age at the time of marriage and her education, along with monthly income of a family has a significantly negative relationship with psychosocial burden of beta thalassemia major ($p$<0.01).

**Multivariate Analysis**

Multivariate analysis is based on the statistical procedures to find out the effect of two or more independent variables on one or more dependent variables (Timm, 1975). This section of inferential statistics is based on the analysis of *Generalized Linear Model* for two independent variables; consanguinity and ethnicity and selected dependent variables; parental knowledge, practices of pre/postnatal diagnosis, risk factors (socioeconomic, cultural, disease allied) and psychological and social burden of beta thalassemia major on the respondents.

The second technique of multivariate analysis was *Multinomial Logistic Regression,* which was applied to find out the probabilities of having moderate and high psychosocial burden of beta thalassemia major among respondents, married with $1^{st}$ cousins, $2^{nd}$ cousins, $3^{rd}$ cousins and with their distant relatives, with reference to those who were married with non-cousins.

The third technique, which was used for multivariate analysis is *Multivariate Linear Regression* to find out the measurement of variance explained by independent variables; socioeconomic risk factors, cultural risk factors, disease allied risk factors, parental knowledge of thalassemia and practices of pre/postnatal diagnosis for the identification of disease for dependent variable; psychological and social burden of the disease (beta thalassemia major). Forward selection method in multiple linear regression (MLR) was chosen to find out the best combination of predictors of variance of dependent variable.

In a patriarchal and gender based segregated society, the women's involvement in decision making and marriage preferences is bitterly snubbed by cultural and social environment (Bourque & Warren, 2010) as a result families and parents unintentionally increase the risk factors of thalassemia (Raffa, 2019). Due to lack of knowledge and awareness of premarital and genetic screening and counseling, thalassemia occurs repeatedly among the extended families and all other communities where cast, ethnic and cultural preferences dominate for decision making regarding marriage (Shaw, 2009). Misperceptions about religious beliefs and practices also increase the vulnerability of thalassemia (Cappellini, Cohen, Porter, Taher, & Viprakasit, 2014) because lack of education and empowerment, especially, among

the women limit them to raise their voices against tradition, detracted and misleading concepts related to culture and religion (Fullwiley, 2011).

Repeated cousin marriages have been reported as the major cause of thalassemia disease (Aqueel & Anjum, 2019). In many countries including Iran, India, Bangladesh and Pakistan where traditional beliefs and practices are dominant over decision making about marriages, genetic counseling, premarital and prenatal screenings, it is difficult and takes a long time to create awareness among families and alarm them about the consequences of consanguinity (Battu, Mallipatna, Elackatt, Schouten, & Webers, 2018). Among other social and cultural factors, low level of awareness and health literacy increases the risk of thalassemia (Javadzade, Mahmoodi, Hajivandi, Ghaedi, & Reisi, 2019). In many traditional societies, Pakistani couples or families also believed in cultural practices and preferred cousin marriages; which resulted in higher rate of thalassemia (11% of the population is career of thalassemia) (Tanveer, Masud, & Butt, 2018) especially in rural areas.

### 4.15 Generalized Linear Model

Table 4.24

*Generalized linear model for Ethnicities, Consanguinity and Parental Knowledge of Beta Thalassemia Major*

| Source | Type III SS | df | MS | *F-test* | *p* | $\eta_p^2$ |
|---|---|---|---|---|---|---|
| Corrected Model | 1277.079[a] | 9 | 141.898 | 51.208 | .000 | .333 |
| Intercept | 5865.934 | 1 | 5865.934 | 2116.910 | .000 | .697 |
| Ethnicities | 57.578 | 4 | 14.394 | 5.195 | .000 | .022 |
| Consanguinity | 230.211 | 1 | 230.211 | 83.079 | .000 | .083 |
| Ethnicities* Consanguinity | 45.970 | 4 | 11.492 | 4.147 | .002 | .135 |
| Error | 2554.852 | 922 | 2.771 | | | |
| Total | 49388.000 | 932 | | | | |
| Corrected Total | 3831.931 | 931 | | | | |

a. $R^2$ = .333 (Adjusted $R^2$ = .327)

Table 4.24 consisted of the results of generalized linear model of ethnicities, consanguinity and parental knowledge of beta thalassemia major in Punjab. The $R^2$ value of the model showed that the predictors explained 33.3% of the variance in the

outcome variable. Further, the analysis of the variance showed ethnicities, $F$ (4, 932) =5.195, $\eta_p^2$= 0.022, $p$<.001, consanguinity, $F$ (1,932) =83.1, $\eta_p^2$= 0.022, $p$<.001, and interaction of ethnicity and consanguinity, $F$ (4, 932) =4.147, $\eta_p^2$= 0.022, $p$<.001, are significantly different by parental knowledge of beta thalassemia major.

Furthermore, the estimated marginal mean of dependent variable is illustrated in figure 4.1. Kashmir cousins and non-cousin couples are more inclined towards acquiring the knowledge of beta thalassemia major, as compared to all other ethnic groups. However, the knowledge acquisition in non-cousin Sindhi ethnic communities is low as compared to Pashtun, Punjabi and Balochi ethnic groups. The reason behind higher preference of knowledge acquisition of beta thalassemia of Kashmiri parents is their higher educational and awareness level regarding beta thalassemia major. Similar study conducted by (Ahmed et al., 2019) in Azad Jammu and Kashmir also found that their selected community has sufficient knowledge regarding prevention and management of beta thalassemia major, as 76.2% people were aware about management of beta thalassemia major. The second ethnic group with higher preferences of parental knowledge of beta thalassemia major was Punjabi (see figure 4.1). As the present study found least preferences of Sindhi ethnicities towards knowledge of beta thalassemia major, so it can be predicted based on data that this group required greater attention for knowledge acquisition of the disease for prevention.to prevention. Two years back, a study was conducted by (Kandhro, Prachayasittikul, Isarankura Na-Ayudhya, & Nuchnoi, 2017) in Sindhi ethnic groups and they found that 56% carrier rate of beta thalassemia major was existing in selected community.

The variations of parental knowledge, regarding beta thalassemia major is based on two aspects; the first is higher number of cousin couples, as compared to non-cousin couples and the second reason is their sensitization about consanguineous marriages, which has been seen by the researcher during data collection.

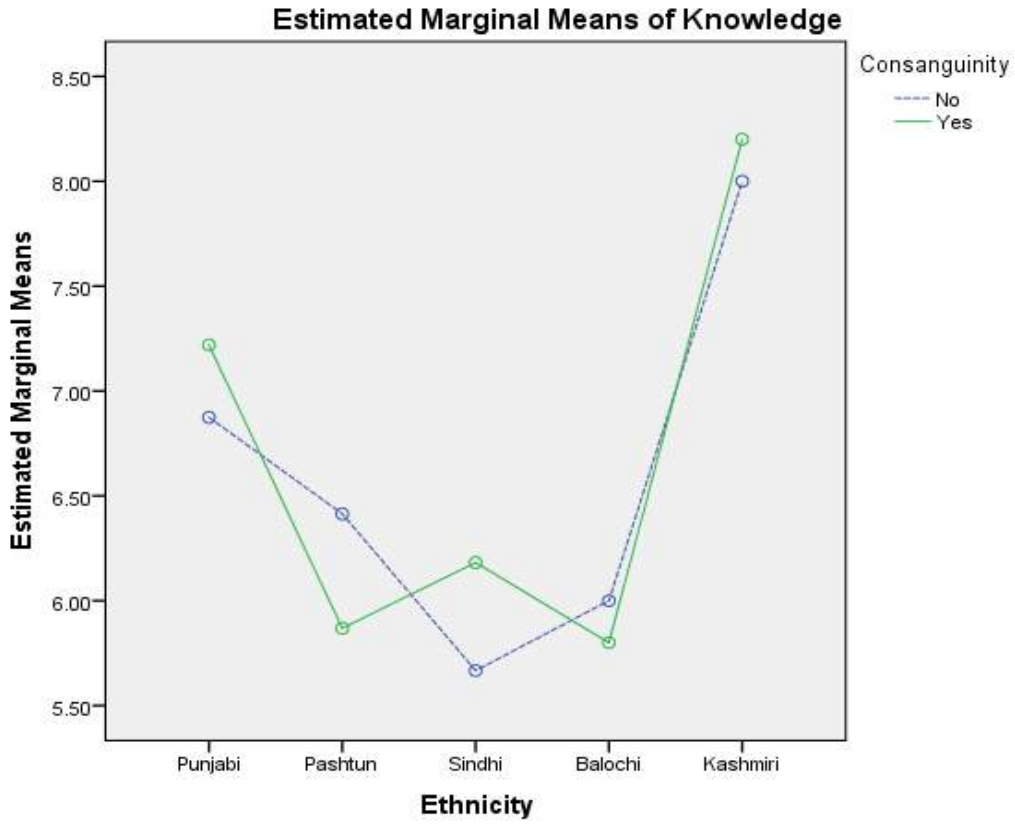



Figure 4.1 *Mean plot of parental knowledge of beta thalassemia major by consanguinity in ethnicities*

Table 4.25

*Generalized linear regression model for Ethnicity, Consanguinity and Pre/post-natal practices of Parental Diagnosis of Beta Thalassemia Major*

| Source | Type III SS | df | MS | *F-test* | *p* | $\eta_p^2$ |
|---|---|---|---|---|---|---|
| Corrected Model | 4972.602[a] | 9 | 552.511 | 19.533 | .000 | .160 |
| Intercept | 94640.461 | 1 | 94640.461 | 3345.833 | .000 | .784 |
| Ethnicities | 4228.591 | 4 | 1057.148 | 37.373 | .000 | .140 |
| Consanguinity | 93.590 | 1 | 93.590 | 3.309 | .069 | .004 |
| Ethnicities * Consanguinity | 492.965 | 4 | 123.241 | 4.357 | .002 | .138 |
| Error | 26079.753 | 922 | 28.286 | | | |
| Total | 628497.000 | 932 | | | | |
| Corrected Total | 31052.355 | 931 | | | | |

a. $R^2$ = .160 (Adjusted $R^2$= .152)

It has been suggested by (Vichinsky, 2016), that effective management of beta thalassemia major can be ensured by screening practices of pre/postnatal diagnosis but cultural factors play an important role for these preventive and management measures. In most of the developing countries including Pakistan, people follow conservative management practices for beta thalassemia major (Arif, Fayyaz, & Hamid, 2008). Table 4.25 revealed the results of generalized linear model of ethnicities, consanguinity and practices of pre/postnatal diagnosis. The value of $R^2$ model showed that 16% variation in outcome variable has been explained by ethnicities and consanguinity. Furthermore, the analysis of variance showed ethnicities $F$ (4, 932) = 37.373, $\eta_p^2$ = 140, $p$<.001 are significantly different by practices of pre/postnatal diagnosis. However, ethnicities were not significantly different by practices of pre/postnatal diagnosis $p$>.05. The present study found a significant interaction effect $F$ (4, 932) = 4.357, $p$<0.05, $\eta_p^2$=.13 of consanguinity and ethnicities. The study also reveals estimated marginal mean of practices of pre/postnatal diagnosis among consanguinity or non-consanguinity in different ethnicities (see figure 4.2).

The present study found that Balochi and Sindhi ethnicities have least preferences towards PNDs, and this happened due to lack of facilities, low educational level and strict cultural preferences, that restrain individuals to adopt the scientific and modern management and preventive practices like practices of pre/postnatal diagnosis. It has been seen that Balochi cousin couples have least preferences for practices of pre/postnatal diagnosis (see fig 4.2), because consanguinity is a culturally preferred practice (Agha, 2016; Wahab & Ahmad, 2005) and the higher rate of cousin marriages have been reported by (Hussain, 2005) in Balochi ethnicities. It has been found that among Punjabi ethnic groups, cousin couples have higher preferences of practices for pre/postnatal diagnosis while Pashtun non-cousins are more inclined towards practices of pre/postnatal diagnosis. The figure 4.2 also reveals that in Kashmiri ethnicities, both couples have higher presences for practices of pre/postnatal diagnosis for their collective scrutiny.

The countries once known for their high level of thalassemia cases are now using various prevention strategies which include carriers' identification, genetic

counseling and diagnostic tests of the parents. All these initiatives have almost eliminated the risk of homozygous thalassemia in new generation (Marioni et al., 2015). But in case of Pakistan, a strong resistance for all these preventive measures has been reported by researches (Ishfaq, 2015; Khan, 2018; Saeed & Piracha, 2016), which leads this disease to become a chronic aliment (Yasmeen & Hasnain, 2019).

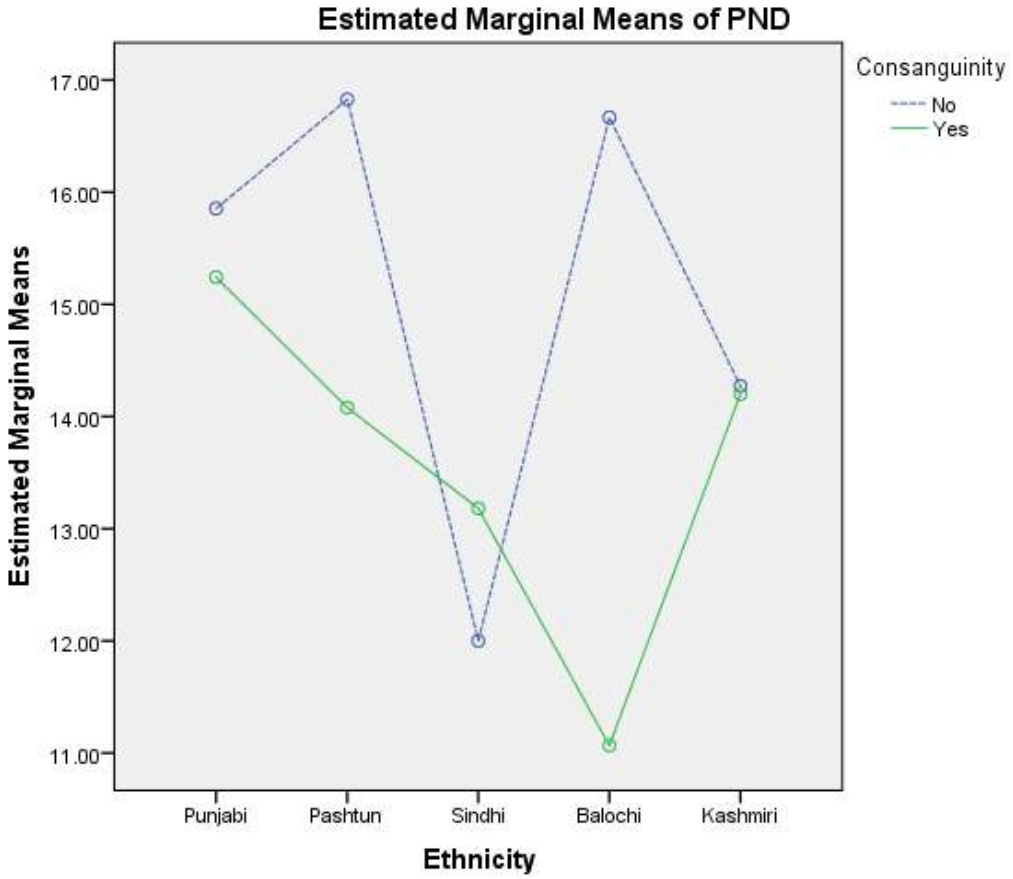



Figure 4.2 *Mean plot of parental practices of prenatal diagnosis of beta thalassemia major by consanguinity in different ethnicities*

Table 4.26

*Generalized linear regression model for Ethnicity, Consanguinity and Socioeconomic Risk Factors of Beta Thalassemia Major*

| Source | Type III SS | df | MS | *F-test* | *p* | $\eta_p^2$ |
|---|---|---|---|---|---|---|
| Corrected Model | 2400.724[a] | 9 | 266.747 | 44.224 | .000 | .302 |
| Intercept | 16288.570 | 1 | 16288.570 | 2700.485 | .000 | .745 |
| Ethnicities | 64.539 | 4 | 16.135 | 2.675 | .031 | .011 |
| Consanguinity | 302.774 | 1 | 302.774 | 50.197 | .000 | .052 |
| Ethnicities * Consanguinity | 195.690 | 4 | 48.923 | 8.111 | .000 | .185 |
| Error | 5561.245 | 922 | 6.032 | | | |
| Total | 131933.000 | 932 | | | | |
| Corrected Total | 7961.969 | 931 | | | | |

a. $R^2$ = .302 (Adjusted $R^2$ = .295)

Generalized linear regression was applied to find out the interaction effect of different ethnic groups and consanguineous marriages on socioeconomic risk factors of beta thalassemia major in the study area. Data recorded in table 4.26 revealed the value of $R^2$ = .302, which showed that the amount of variance explained by ethnicities and consanguinity was 30.2%. The analysis of variance revealed that ethnicities $F$ (4, 932) = 2.675, $\eta_p^2$ =.011, $p$<.05, consanguinity $F$ (1, 932) = 50.197, $\eta_p^2$ = .052, $p$<.001 and the interaction effect of ethnicities and consanguinity $F$ (4, 932) = 8.111, $p$<0.05, $\eta_p^2$ = .185 are significantly different by socioeconomic risk factors of beta thalassemia major. Results indicated that on the basis of partial eta square value the combine effect of independent variables is large as suggested by (Cohen, 1988)[1].

Furthermore, the graphical representation of the data (figure 4.3) depicts that Kashmiri non-cousin couples are more likely to have socioeconomic risk factors of beta thalassemia major as compared to other ethnic groups. These parents were perceived to have less social support form thalassemia foundation and they were considering it difficult to participate in family/community gatherings and events due to the illness of their child. The financial management, for the treatment of

[1] The value of $\eta_p^2$ = 0.01 (small), 0.06 (medium), 0.14 (large).

thalassemia is their major problem and they were seemed dissatisfied with the social and financial support of their relatives and friends. The illustration is also clear that Sindhi non-cousin couples are less likely to socioeconomic risk factors of beta thalassemia major which includes social support, financial support and level of awareness. The higher propagation and low level of social support among Sindhi ethnicities is also evident in Asian region, not only in Pakistan. As it has been found by (Saldanha, 2015), in a study conducted in Bangalore that Sindhi ethnic groups has 25% higher risk of beta thalassemia major and facing low level of social support, as compared to other ethnicities and research area.

Lack of social support from the society, relatives and friends; creates a massive social burden, emotional disorder and lack of self-efficacy among parents of thalassemic children (Pouraboli, Abedi, Abbaszadeh, & Kazemi, 2017), which ultimately affects the social wellbeing and quality of life of parents and children (Mikael & Al-Allawi, 2018). It becomes challenging for the parents to bear the psychological and emotional pain due to physical abnormalities of their children due to thalassemia(Shahraki-vahed, Firouzkouhi, Abdollahimohammad, & Ghalgaie, 2017).

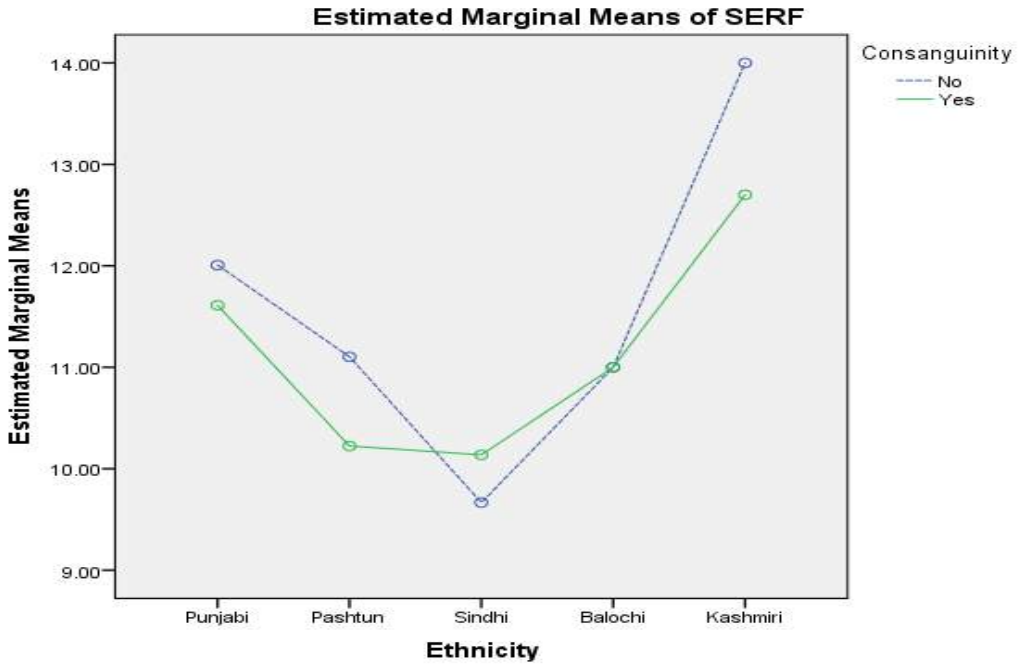



Figure 4.3 *Mean plot of socioeconomic risk factors of beta thalassemia major by consanguinity in different ethnicities*

Table 4.27

*Generalized linear regression model for Ethnicity, Consanguinity and Cultural Risk Factors of Beta Thalassemia Major*

| Source | Type III SS | df | MS | *F-test* | *p* | $\eta_p^2$ |
|---|---|---|---|---|---|---|
| Corrected Model | 19787.260[a] | 9 | 2198.584 | 34.568 | .000 | .252 |
| Intercept | 57926.013 | 1 | 57926.013 | 910.773 | .000 | .497 |
| Ethnicities | 16922.411 | 4 | 4230.603 | 66.518 | .000 | .224 |
| Consanguinity | 108.799 | 1 | 108.799 | 1.711 | .191 | .002 |
| Ethnicities * Consanguinity | 1228.834 | 4 | 307.208 | 4.830 | .001 | .145 |
| Error | 58640.044 | 922 | 63.601 | | | |
| Total | 409882.000 | 932 | | | | |
| Corrected Total | 78427.305 | 931 | | | | |

a. $R^2$ = .252 (Adjusted $R^2$= .245)

Cultural factors play an important role for the prevention and management of the disease-beta thalassemia major (Mendiratta, Mittal, Naaz, Singh, & Anand, 2017) . The present study aimed to find out the cultural risk factors; in terms of ethical and religious beliefs regarding blood transfusion, screening, termination of pregnancy and marriage preferences. Table 4.27 revealed the results of generalized linear model for ethnicities, consanguinity and cultural risk factors in Punjab Province. The $R^2$ value of the model showed that the predictors explained 25.2% of the variance in the outcome variable. Further, the analysis of the variance showed ethnicities, $F$ (4, 932) =66.518, $\eta_p^2$= .224, $p$<.001, and interaction of ethnicity and consanguinity, $F$ (4, 932) =4.830, $\eta_p^2$= .145, $p$<.05, are significantly different by cultural risk factors of beta thalassemia major.

Furthermore, the estimated marginal mean of dependent variable is illustrated in figure 4.4. Data revealed that Balochi and Pashtun non-cousin couples are more likely to towards cultural restrictions of beta thalassemia major, as compared to cousin couple (see fig 4.4). However, Kashmiri cousin couples have higher preferences towards religious and cultural beliefs of beta thalassemia major as compared to all others and this is an evidence to have higher rate of beta thalassemia major (Ajaz, 2013; Darr, 1991). Repeated cousin marriages are the products of

cultural practices and the preferences of cousin couples, towards cultural risk factors is an inveterate for higher propagation of beta thalassemia major (Ashfaq, Amanullah, Ashfaq, & Ormond, 2013). The propagation of beta thalassemia major becomes evident, when couples are more inclined towards cultural values and beliefs, rather than focusing scientific practices such as PNDs, screening and counselling for the management and prevention of thalassemia. It has been intensively found in the literature review, that strong emphasize on ethnicities, consanguinity, and religious beliefs propagate beta thalassemia major, across the world and the present study found so, in Pakistan.

The cultural implications in Pakistan involve restrictions for premarital screening, genetic and career counseling (Mirza et al., 2013). Termination of pregnancy is considered unethical and it has religious constraints allied with thalassemia (Ngim, Lai, & Ibrahim, 2013). Due to ethical and traditional curbs, thalassemia is treated conservatively with all its innate complications, which eventually lead to the death of the patients (Drakonaki et al., 2005). The only solution to minimize the mortality rate due to thalassemia is; by making the general public aware about the ailment but keeping their religious and cultural implications into the consideration (Hassan, Aslam, & Ikram, 2002).

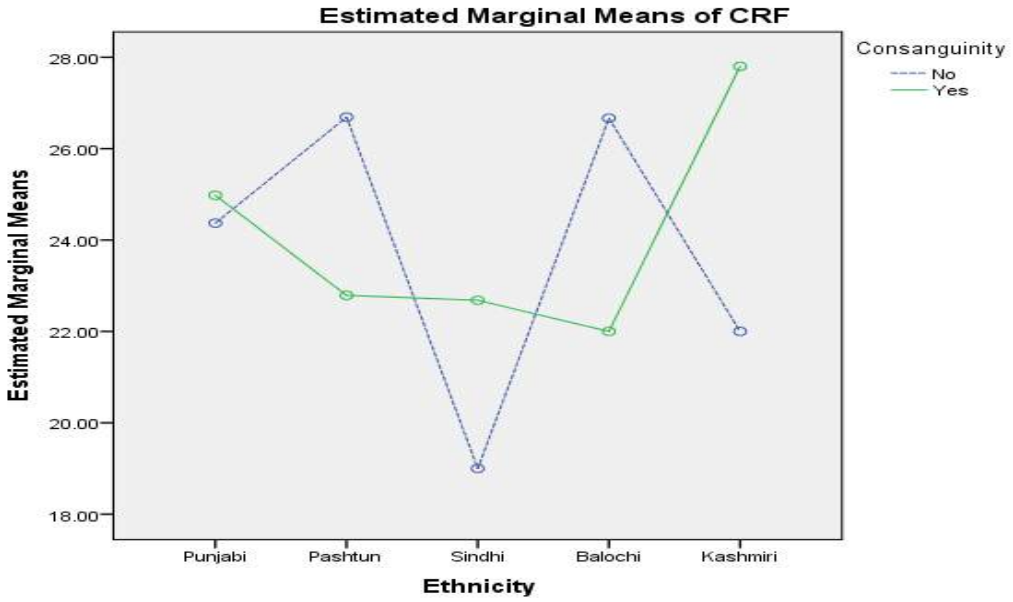



Figure 4.4 *Mean plot of cultural risk factors of beta thalassemia major by consanguinity in ethnicities*

Table 4.28

*Generalized linear regression model for Ethnicity, Consanguinity and Disease Allied Risk Factors of Beta Thalassemia Major*

| Source | Type III SS | df | MS | *F-test* | *p* | $\eta_p^2$ |
|---|---|---|---|---|---|---|
| Corrected Model | 930.941[a] | 9 | 116.368 | 37.024 | .000 | .243 |
| Intercept | 4541.636 | 1 | 4541.636 | 1444.999 | .000 | .610 |
| Ethnicities | 45.798 | 4 | 11.449 | 3.643 | .006 | .016 |
| Consanguinity | 268.248 | 1 | 268.248 | 85.348 | .000 | .085 |
| Ethnicities * Consanguinity | 36.697 | 4 | 12.232 | 3.892 | .009 | .110 |
| Error | 2900.991 | 922 | 3.143 | | | |
| Total | 49388.000 | 932 | | | | |
| Corrected Total | 3831.931 | 931 | | | | |

a. $R^2$= .243 (Adjusted $R^2$= .236)

Table 4.28 revealed the results of generalized linear model for ethnicities, consanguinity and disease allied risk factors in Punjab Province. The $R^2$ value of the model showed that the predictors explained 24.3% of the variance in the outcome variable. Further, the analysis of the variance showed ethnicities, $F$ (4, 932) =3.643, $\eta_p^2$= 0.016, $p$<.05, consanguinity $F$ (1,932) = 85.348, $\eta_p^2$= 0.085, $p$<.001, and interaction of ethnicity and consanguinity, $F$ (4, 932) =3.892, $\eta_p^2$= .110, $p$<.05, are significantly different by disease allied risk factors of beta thalassemia major. Disease allied risk factors were measured by the constructs; affordability of travelling and hospitalization expenditures, therapeutic facilities and provision of blood at transfusion centers.

Furthermore, fig 4.5 depicts that Sindhi and Kashmiri non-cousin couples have least predilections of disease allied risk factors as compared to other non-cousin couples in the study area. However, the high presence of disease allied risk factors has been seen in Balochi non-cousins and Punjabi cousins as well as non-cousin marriages. As majority of people in the study area belonged to Punjabi ethnicities and the selected centers were also located in Punjab Province so it becomes vivid that the dominant ethnic group is more likely to have inclinations towards disease allied risk factors of beta thalassemia major.

Insufficient healthcare facilities, exorbitant medical treatments and financial constraints, are the major obstacles for effective management of thalassemia (Punaglom, Kongvattananon, & Somprasert, 2019), since most of the vulnerable families and communities living in rural areas of Pakistan with limited financial resources and having low level of education cannot be screened by the government, nongovernmental organizations and blood donation organizations.

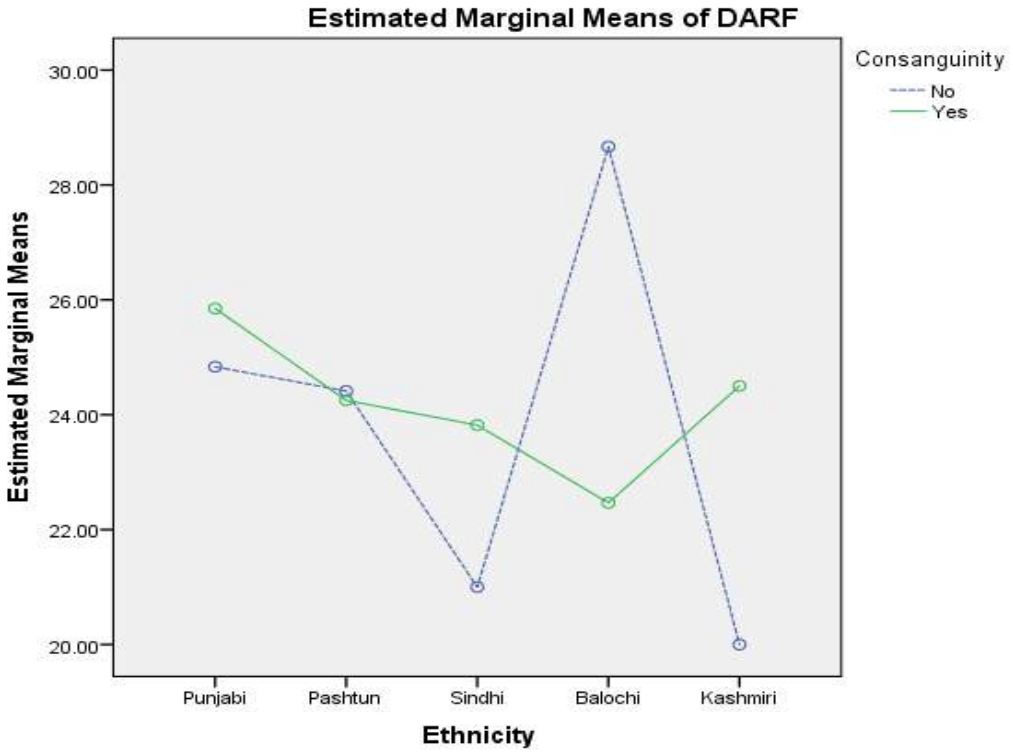



Figure 4.5 *Mean plot of disease allied risk factors of beta thalassemia major by consanguinity in ethnicities*

Table 4.29

*Generalized linear regression model for Ethnicity, Consanguinity and Psychosocial Burden of Beta Thalassemia Major*

| Source | Type III SS | df | MS | *F-test* | *p* | $\eta_p^2$ |
|---|---|---|---|---|---|---|
| Corrected Model | 22447.354[a] | 9 | 2805.919 | 46.264 | .000 | .286 |
| Intercept | 42847.010 | 1 | 42847.010 | 706.463 | .000 | .434 |
| Ethnicities | 11512.381 | 4 | 2878.095 | 47.454 | .000 | .171 |
| Consanguinity | 703.663 | 1 | 703.663 | 11.602 | .001 | .012 |
| Ethnicities * Consanguinity | 357.817 | 4 | 119.272 | 1.967 | .117 | .078 |
| Error | 55979.951 | 922 | 60.650 | | | |
| Total | 409882.000 | 932 | | | | |
| Corrected Total | 78427.305 | 931 | | | | |

a. $R^2$ = .286 (Adjusted $R^2$ = .280)

Table 4.29 consisted of the results of generalized linear model of ethnicities, consanguinity and psychosocial burden of beta thalassemia major in Punjab Province. The $R^2$ value of the model showed that the predictors explained 28.6% of the variance in the outcome variable. Further, the analysis of the variance showed ethnicities, $F$ (4, 932) =47.454, $\eta_p^2$= .171, $p$<.001 and consanguinity, $F$ (1,932) =11.602, $\eta_p^2$= 0.012, $p$<.05, are significantly different by psychosocial burden of beta thalassemia major. However, data recorded in table 4.29 revealed that there is no interaction effect of consanguinity and ethnicities on psychosocial burden of beta thalassemia major $F$ (4, 932) = 1.967, $p$>0.05, $\eta_p^2$ = .078. Further, fig 4.6 revealed that both cousins and non-cousin couples of Punjabi ethnic groups are more likely to face psychosocial burden of beta thalassemia major because of two reasons; first due to their greater number (see table 4.2) and second because of higher propagation of beta thalassemia major in this ethnic group, as previously found by (Hafeez, Aslam, Ali, Rashid, & Jafri, 2007) in their study (60.7%) as compared to other ethnicities. The least predilections of psychosocial burden of beta thalassemia major have been seen in Kashmiri cousins and non-cousin couples as compared to other ethnicities.

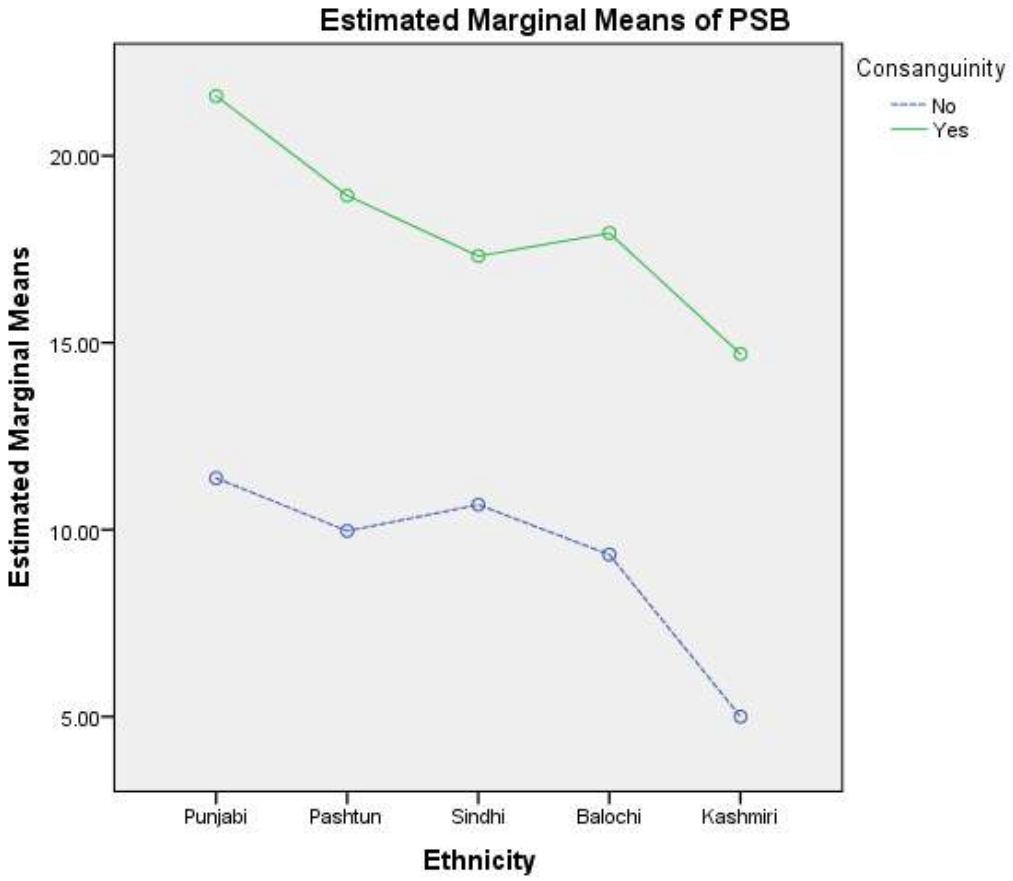



Figure 4.6 *Mean plot of psychosocial burden of beta thalassemia major by consanguinity in different father's ethnicities*

### 4.16 Multinomial Logistic Regression

Table 4.30

*Predictors' Contribution in the Model of Multinomial Logistic Regression (N=932)*

| Predictors | $\chi^2$ | df | *p* |
|---|---|---|---|
| SERF | 98.431 | 4 | .000** |
| CRF | 64.404 | 4 | .000** |
| DARF | 10.040 | 4 | .040* |
| Parents' Consanguinity | 84.842 | 4 | .000** |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, *$p$<.05, **$p$<.001

**Model Fit Information:** $\chi^2$ (20) = 315.605**, $R^2$= .287 (Cox and Snell), .308 (Nagelkerke)

To model a relationship between the risk factors, consanguinities of parents of patients across 1st, 2nd, 3rd cousin and distant relative, the researcher applied multinomial logistic regression, while the reference category was non-consanguineous couples for psychosocial burden of beta thalassemia major (being moderate or high, while the reference category was mild psychosocial burden of beta thalassemia major). The data labeled in table 4.30 showed that socioeconomic, cultural and disease allied risk factors and parents' consanguinities are significant predictors of the model. The model fit information also revealed that addition of predictors to the model contains significant intercept of the model and data $\chi^2$ (20) = 315.6, Nagelkerke $R^2$= .308, $p$<.001.

Table 4.30.1

*Parameters Estimate Contrasting the Non-consanguineous Groups (Parents of Children) Versus Each of the Other Groups (N=932)*

| Variables | Moderate PSB | | High PSB | |
|---|---|---|---|---|
| | B(SE) | OR | B(SE) | OR |
| SERF | .113(031) ** | 1.120 | .253(.078) ** | 1.288 |
| CRF | .025(.026) | 1.025 | .124(.039) ** | 1.131 |
| DARF | .006(.018) | 1.006 | .048(.025) * | 1.049 |
| 1st cousin | -.222(.027) ** | .801 | -.230(.039) ** | .795 |
| 2nd cousin | .543(.203) ** | 1.722 | -.741(.284) ** | .476 |
| 3rd cousin | -.150(.030) ** | .861 | -.152(.032) ** | .859 |
| Distant Relative | .201(.037) ** | 1.222 | .246(.044) ** | 1.279 |
| Non-cousins | Reference category | | | |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, PSB=Psychosocial Burden, *$p$<.05, **$p$<.001

Table 4.30.1 showed that socioeconomic risk factors were significantly positively predicted moderate and high level of psychological and social burden among the respondents with reference to the mild level of psychological and social burden among the respondents B (.113) and B (.253), $p$<.001 respectively. Furthermore, cultural risk factors were significantly positive predictors of high psychosocial burden of beta thalassemia major B (.124), $p$<.001. Similarly, the data

revealed that disease allied risk factors significantly positively predicted high psychosocial burden of beta thalassemia major B (.048), $p<.05$.

The results of multinomial logistic regression, recorded in table 4.30.1 also revealed that parents of sick children married with their 1st cousins were less likely to face moderate OR= .801and high level of psychosocial burden of beta thalassemia major OR=.795, $p<.001$ as compared to the non-cousin couples. It has been found that parents of sick children, married with their 2nd cousins were more likely to face moderate psychosocial burden OR= 1.722, $p<.001$ while less likely to face high level of psychosocial burden of beta thalassemia major OR=.476, $p<.001$ as compared to the non-cousin couples. The study also found that parents of sick children, married with their 3rd cousins were less likely to face moderate OR=.861 and high psychosocial burden of beta thalassemia major OR=.859, $p<.001$ with refence to those who were married with non-cousins. However, it has been found that couples married with their distant relatives were more likely to face moderate OR=1.222 and high psychosocial burden of beta thalassemia major OR= 1.279, $p<.001$ as compared to the non-cousin couples.

Table 4.31

*Predictors' Contribution in the Model of Multinomial Logistic Regression (N=932)*

| Predictors | $\chi^2$ | df | *p* |
|---|---|---|---|
| SERF | 98.782 | 4 | .000** |
| CRF | 65.510 | 4 | .000** |
| DARF | 9.913 | 4 | .042* |
| Husband's Parents Consanguinity | 25.394 | 8 | .001* |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, *$p<.05$, **$p<.001$

**Model Fit Information:** $\chi^2$ (20) = 309.040**, $R^2$= .282 (Cox and Snell), .302 (Nagelkerke)

To model a relationship between the risk factors, consanguinities of husband's parents (if the respondent was father) or father/mother in laws (if the respondent was mother) of patients across 1st, 2nd, 3rd cousin and distant relative, the researcher applied multinomial logistic regression, while the reference category was non-consanguineous couples for psychosocial burden of beta thalassemia major (being

moderate or high, while the reference category was mild psychosocial burden of beta thalassemia major). The data labeled in table 4.31 showed that socioeconomic, cultural and disease allied risk factors and husbands' parents' consanguinities are significant predictors of the model. The model fit information also revealed that addition of predictors to the model contains significant intercept of the model and data $\chi^2$ (20) = 309.0, Nagelkerke $R^2$= .302, $p$<.001.

Table 4.31.1

*Parameters Estimate Contrasting the Non-consanguineous Groups (Husbands' Parents) Versus Each of the Other Groups (N=932)*

| Variables | Moderate PSB | | High PSB | |
|---|---|---|---|---|
| | B(SE) | OR | B(SE) | OR |
| SERF | .224(.031)** | 1.251 | .148(.036)** | 1.159 |
| CRF | .029(.026) | 1.029 | .124(.039)** | 1.131 |
| DARF | .005(.018) | 1.005 | .048(.025) | 1.050 |
| 1st cousin | -.208(.027)** | .812 | -.244(.040)** | .783 |
| 2nd cousin | -.272(.458) | .762 | 1.075(.893) | 2.931 |
| 3rd cousin | .158(.462) | 1.171 | 1.112(.888) | 3.040 |
| Distant Relative | .209(.038)** | 1.301 | .535(.670) | 1.674 |
| Non-cousins | Reference category | | | |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, PSB=Psychosocial Burden, *$p$<.05, **$p$<.001

Table 4.31.1 showed that socioeconomic risk factors were significantly positive predictors of moderate and high psychosocial burden of beta thalassemia major B (.224) and B (.148), $p$<.001. The data also revealed that cultural risk factors were only significantly positive predictors of high psychosocial burden of beta thalassemia major B (124), $p$<.001.

Data recorded in table 4.31.1 also revealed that husbands' parents married with 1st cousins were significantly less likely to face moderate OR= .812, $p$<.001 and high psychosocial burden of beta thalassemia major OR= .783, $p$<.001 as compared to the those who had non-consanguineous marriages. However, it has been found that only distant relatives were significantly more likely to face moderate psychosocial

burden of beta thalassemia major OR= 1.301, $p<.001$, with refence to the non-cousins. The data revealed that 2nd and 3rd cousin marriages of husbands' parents did not show significant odds towards either moderate or high psychosocial burden of beta thalassemia major, $p>.05$.

Table 4.32

*Predictors' Contribution in the Model of Multinomial Logistic Regression (N=932)*

| Predictors | $\chi^2$ | df | *p* |
|---|---|---|---|
| SERF | 28.105 | 4 | .000** |
| CRF | 38.276 | 4 | .000** |
| DARF | 10.917 | 4 | .028* |
| Husband's Grandparents Consanguinity | 84.805 | 4 | .000** |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, *$p<.05$, **$p<.001$

**Model Fit Information:** $\chi^2(20) = 470.527^{**}$, $R^2 = .396$ (Cox and Snell), .425 (Nagelkerke)

To model a relationship between the risk factors, consanguinities of husband's grandparents (if the respondent was father) or parents of father/mother in laws (if the respondent was mother) of patients across 1st, 2nd, 3rd cousin and distant relative, the researcher applied multinomial logistic regression, while the reference category was non-consanguineous couples for psychosocial burden of beta thalassemia major (being moderate or high, while the reference category was mild psychosocial burden of beta thalassemia major). The data labeled in table 4.32 showed that socioeconomic, cultural and disease allied risk factors and husbands' parents' consanguinities are significant predictors of the model. The model fit information also revealed that addition of predictors to the model contains significant intercept of the model and data $\chi^2(20) = 470.5$, Nagelkerke $R^2 = .425$, $p<.001$.

Table 4.32.1

*Parameters Estimate Contrasting the Non-consanguineous Groups (Husbands' Grandparents) Versus Each of the Other Groups (N=932)*

| Variables | Moderate PSB | | High PSB | |
|---|---|---|---|---|
| | B(SE) | OR | B(SE) | OR |
| SERF | .318 (.047) ** | 1.375 | .337 (.047) ** | 1.401 |
| CRF | .042(.026) | 1.042 | .071(.041) | 1.073 |
| DARF | .002(.018) | 1.002 | .052(.025) * | 1.053 |
| 1st cousin | .639(.216) ** | 1.895 | -2.118(.349) ** | .120 |
| 2nd cousin | .181(.039) ** | 1.198 | .231(.047) ** | 1.260 |
| 3rd cousin | .048(.025) | 1.049 | -.014(.029) | .986 |
| Distant Relative | -1.731(.299) ** | .177 | -2.275(.410) ** | .103 |
| Non-cousins | Reference category | | | |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, PSB=Psychosocial Burden, *$p$<.05, **$p$<.001

Data recorded in table 4.32.1 revealed that socioeconomic risk factors were significantly positive predictors of moderate B (.318) and high psychosocial burden of beta thalassemia major B (.337), $p$<.001. It has also been found that disease allied risk factors significantly positively predicted high psychosocial burden B (.052), $p$<.05.

Table 4.32.1 also revealed that those respondents (if father then his grandparents, or in case of mother then parents of his mother/father in law) who were married with their 1st cousins were more likely to face moderate psychosocial burden OR= 1.895, $p$<.001. While less likely to face high psychosocial burden of the disease OR=.120, with refence to those, who were married with non-cousins. The study also found that 2nd cousins were more likely to face moderate OR=1.198 and high psychosocial burden of beta thalassemia major OR=1.260, $p$<.001as compared to the non-cousin couples. However, married with distant relative were less likely to face moderate OR=.177 and high psychosocial burden of the disease OR=.103, $p$<.001 with reference to the no-cousin couples.

Table 4.33

*Predictors' Contribution in the Model of Multinomial Logistic Regression (N=932)*

| Predictors | $\chi^2$ | df | *p* |
|---|---|---|---|
| SERF | 85.770 | 4 | .000** |
| CRF | 121.105 | 4 | .000** |
| DARF | 43.629 | 4 | .000** |
| Wife's Parents Consanguinity | 19.135 | 4 | .001* |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, *$p$<.05, **$p$<.001

**Model Fit Information:** $\chi^2$ (20) = 404.685**, $R^2$= .352 (Cox and Snell), .377 (Nagelkerke)

To model a relationship between the risk factors and consanguinities of wife's parents (if the respondent was mother) or father/mother in laws (if the respondent was father) of patients across 1st, 2nd, 3rd cousin and distant relative, the researcher applied multinomial logistic regression, while the reference category was non-consanguineous couples for psychosocial burden of beta thalassemia major (being moderate or high, while the reference category was mild psychosocial burden of beta thalassemia major). The data labeled in table 4.33 showed that socioeconomic, cultural and disease allied risk factors and husbands' parents' consanguinities are significant predictors of the model. The model fit information also revealed that addition of predictors to the model contains significant intercept of the model and data $\chi^2$ (20) = 404.7, Nagelkerke $R^2$= .377, $p$<.001.

Table 4.33.1

*Parameters Estimate Contrasting the Non-consanguineous Groups (Wives' Parents) Versus Each of the Other Groups (N=932)*

| Variables | Moderate PSB | | High PSB | |
|---|---|---|---|---|
| | B(SE) | OR | B(SE) | OR |
| SERF | .103 (.044) ** | 1.108 | .289 (.033) ** | 1.335 |
| CRF | .108(.031) * | 1.114 | .032(.037) | 1.033 |
| DARF | .008(.021) | 1.008 | .012(.025) | 1.013 |
| 1st cousin | .236 (.080) * | 1.266 | .308 (.034) ** | 1.360 |
| 2nd cousin | -.294(.032) ** | .745 | -.116(.038) * | .890 |
| 3rd cousin | .858(.220) ** | 2.358 | .197(.266) | 1.218 |
| Distant Relative | .227 (.065) * | 1.255 | -.062 (.022) * | .940 |
| Non-cousins | Reference category | | | |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, PSB=Psychosocial Burden, *$p$<.05, **$p$<.001

Table 4.33.1 divulged the information related to predictors of moderate and high psychosocial burden of beta thalassemia major and odd ratios of wives' parents' consanguinities. It has been found that socioeconomic risk factors were significantly positive predictors of moderate B (.103) and high psychosocial burden B (.289), $p$<.001. Whereas, cultural risk factors only predicted moderate psychosocial burden B (.108), $p$<.05 significantly.

The data also revealed that respondents (if mother of sick child then her parents and in case of father then his mother/father in law) married with 1st cousins were more likely to face moderate OR= 1.266, $p$<.05 and high psychosocial burden of beta thalassemia major OR= 1.360, $p$<.001 with reference to the non-cousins. However, 2nd cousins were less likely to face moderate OR= .745, $p$<.001 and high psychosocial burden of the disease OR=.890, $p$<.05 as compared to the non-cousins. Table 4.33.1 also revealed that marriages with 3rd cousins and distant relatives increased the vulnerabilities of the respondents to face moderate psychosocial burden of beta thalassemia major OR= 2.358, $p$<.001 and OR=1.255, $p$<.05 respectively. Whereas, the posteriors were significantly less likely to face high psychosocial burden of the disease OR=.940, $p$<.05 as compared to the reference category.

Table 4.34

*Predictors' Contribution in the Model of Multinomial Logistic Regression (N=932)*

| Predictors | $\chi^2$ | df | *p* |
|---|---|---|---|
| SERF | 89.945 | 4 | .000** |
| CRF | 16.323 | 4 | .000** |
| DARF | 44.700 | 4 | .000** |
| Wife's Grandparents Consanguinity | 61.812 | 4 | .000** |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, *$p$<.05, **$p$<.001

**Model Fit Information:** $\chi^2$ (20) = 447.362**, $R^2$= .381 (Cox and Snell), .408 (Nagelkerke)

To model a relationship between the risk factors, consanguinities of wife's grandparents (if the respondent was mother) or parents of father/mother in law (if the respondent was father) across 1st, 2nd, 3rd cousin and distant relative, the researcher applied multinomial logistic regression, while the reference category was non-consanguineous couples for psychosocial burden of beta thalassemia major (being moderate or high, while the reference category was mild psychosocial burden of beta thalassemia major). The data in table 4.34 showed that socioeconomic, cultural and disease allied risk factors and husbands parents' consanguinities are significant predictors of the model. The model fit information also revealed that addition of predictors to the model contains significant intercept of the model and data $\chi^2$ (20) = 447.4, Nagelkerke $R^2$= .408, $p$<.001.

Table 4.34.1

*Parameters Estimate Contrasting the Non-consanguineous Groups (Wives' Grandparents) Versus Each of the Other Groups (N=932)*

| Variables | Moderate PSB | | High PSB | |
|---|---|---|---|---|
| | B(SE) | OR | B(SE) | OR |
| SERF | .255 (.046) ** | 1.290 | -.129(.035) | .879 |
| CRF | .114(.031) ** | 1.120 | .026(.038) | 1.026 |
| DARF | .007(.021) | 1.007 | .011(.026) | 1.011 |
| 1st cousin | .030(.037) * | 1.031 | 1.484(1.093) * | 4.411 |
| 2nd cousin | .010(.025) | 1.010 | .721(1.107) | 2.056 |
| 3rd cousin | .206(.065) ** | 1.229 | -.071(.022) | .931 |
| Distant Relative | .241(.076) ** | 1.272 | .299(.034) | 1.348 |
| Non-cousins | Reference category | | | |

**Note:** SERF=Socioeconomic Risk Factors, CRF=Cultural Risk Factors, DARF=Disease Allied Risk Factors, PSB=Psychosocial Burden, *$p$<.05, **$p$<.001

The information recorded in table 4.34.1 revealed that socioeconomic risk factors significantly positively predicted moderate psychosocial burden of beta thalassemia major B (.255), $p$<.001 among wives' grandparents (if respondent was a mother then her grandparents and it father then parents of his mother/father in laws). Similarly, cultural risk factors also significantly positively predicted moderate psychosocial burden of the disease B (.114), $p$<.001.

The study also found that 1st cousins were more likely to face moderate OR=1.031 and high psychosocial burden of beta thalassemia major OR=4.411, $p$<.05 as compared to the non-cousin couples. The moderate psychosocial burden of beta thalassemia major was also significantly higher among 3rd cousins OR=1.229 and distant relatives OR=1.272, $p$<.001, as compared to the non-cousins.

## 4.17 Multiple Linear Regression

Table 4.35

*Predictors of Psychosocial Burden of Beta-Thalassemia Major in Non-Cousin Marriage (M9)*

| Model | Unstandardized Coefficients | | t | *p* |
|---|---|---|---|---|
| | B | SE | | |
| (Constant) | 20.003 | .736 | 27.177 | .000 |
| Parental Knowledge | -.181 | .019 | -9.672 | .000 |
| MB | .272 | .094 | 2.910 | .004 |
| BUF | .449 | .104 | 4.305 | .000 |
| Stigmatization | .479 | .109 | 4.375 | .000 |
| Social support | -.333 | .105 | -3.164 | .002 |
| RRTP | .610 | .100 | 6.072 | .000 |
| RBP | .482 | .109 | 4.415 | .000 |
| RECM | -.222 | .104 | -2.128 | .034 |
| $R^2$ (.394) | | | | |
| Adj $R^2$ (.291) | | | | |
| $\Delta R^2$ (.004) | | | | |
| F (18.879)$^{**}$ | | | | |
| $\Delta$F (4.530) | | | | |

Note: $^{*}p<0.05$, $^{**}p<0.001$, MB= Marital break-up, BUF= Believe in unnatural forces, RRTP= Religious restriction regarding termination of pregnancy, RBP= Religious believes regarding prevention, RECM= Religion encouragement regarding cousin marriages

Table 4.35 reveals the data of predictors of psychosocial burden of beta thalassemia major in non-cousin couples (respondents). Using stepwise (forwards method) multiple linear regression technique, the final model (M9) figured out eight significant predictors of outcome variable (psychosocial burden). Data reveals that 39.4% variation of outcome variable has been explained by the eight significant predictors ($R^2$=.394). Furthermore, the study found that one unit increase in parental knowledge of disease resulted 18.1% decrease in psychosocial burden among non-cousin couples (B=-.181)**. Furthermore, it has been found one unit increase in parent's belief regarding marital break-up resulted 27.2% increase in dependent variable (B=.272)*, while belief in unnatural forces to be the cause of beta thalassemia major resulted 44.9% increase in dependent variables (psychosocial burden) (B=.449)**. The study also found that parental feelings of stigmatization contributes 47.9% to increase psychosocial burden among non-cousins (B=.479)**, while increasing one unit in social support, the psychological and social burden decreased to 33.3%. The results detailed in table 4.35 depicted that religious restrictions regarding termination of pregnancy significantly positively increased psychosocial burden among parents (married with non-cousins) (B=.610)**. While religious beliefs regarding prevention of the disease (B=.482)** significantly increasing the burden of the disease in terms of psychological and social facets among non-cousin couples. The study also found that religious encouragement regarding cousin marriages significantly decreases the burden in non-cousin couples (B=-.222)*.

Table 4.36

*Predictors of Psychosocial Burden of Beta Thalassemia Major in Cousin Marriage (M10)*

| Model | Unstandardized Coefficients | | t | *p* |
|---|---|---|---|---|
| | B | SE | | |
| (Constant) | 20.545 | .891 | 23.052 | .000 |
| Parental Knowledge | -.123 | .029 | -4.194 | .000 |
| MB | .347 | .109 | 3.170 | .002 |
| BUF | .413 | .129 | 3.192 | .001 |
| Stigmatization | .409 | .128 | 3.202 | .001 |
| Social support | -.496 | .132 | -3.750 | .000 |
| Patriarchy | .186 | .134 | 1.391 | .165 |
| RRTP | .499 | .125 | 3.987 | .000 |
| RBP | .366 | .137 | 2.678 | .008 |
| RECM | -.315 | .131 | -2.398 | .017 |
| $R^2$ (.326) | | | | |
| Adj $R^2$ (.280) | | | | |
| $\Delta R^2$ (.007) | | | | |
| F (8.635)$^{**}$ | | | | |
| $\Delta$F (5.752) | | | | |

Note: $^{*}p<0.05$, $^{**}p<0.001$, MB= Marital break-up, BUF= Believe in unnatural forces, RRTP= Religious restriction regarding termination of pregnancy, RBP= Religious believes regarding prevention, RECM= Religion encouragement regarding cousin marriages

Data enclosed in table 4.36 reveals the predictors of psychosocial burden of beta thalassemia major among cousin couples (respondents). Using stepwise (forwards method) multiple linear regression technique, the final model (M10) figured out nine significant predictors of outcome variable (psychosocial burden). Data shows that 32.6% variation of psychosocial burden of beta thalassemia major, among cousin couples has been explained by these nine predictors. Furthermore, it has been found that one unit increase in parental knowledge regarding disease decreased 12.3% psychosocial burden of beta thalassemia major among cousin couples (B=-.123)$^{**}$. Similarly, increasing social support (B=-.496)$^{*}$ and religious encouragement regarding termination of pregnancy (B=-.315)$^{*}$ decreased psychosocial burden to 49.6% and 31.5% respectively. However, one unit increase in couple's belief regarding marital break-ups (B=.347)$^{*}$, beliefs in unnatural forces (B=.413)$^{*}$, stigmatization (B=.409)$^{*}$, patriarchy (B=-.186)$^{*}$, religious restrictions regarding termination of pregnancy (B=.499)$^{**}$ and religious beliefs regarding prevention of disease (B=.366)$^{*}$ significantly increases psychosocial burden of beta thalassemia major among cousin couples.

The previous studies conducted by (Alswaidi et al., 2012; Memish & Saeedi, 2011) focused on social and religious factors which propagate beta thalassemia major and caused serious economic, social and psychological burden on parents of sick children (Prasomsuk, Jetsrisuparp, Ratanasiri, & Ratanasiri, 2007). It has also been intensively studied that cousin marriage is the major reason behind beta thalassemia major (Ayub et al., 2017; Faizan-ul-Haq et al., 2016). The researchers (Ishfaq, Shabbir, Naeem, & Hussain, 2015; Muhammad, Shakeel, Rehman, & Lodhi, 2017) focused rigorously to study consanguinities and beta thalassemia major but none of them figured out the effect of social, cultural and religious factors of psychosocial burden of beta thalassemia major on cousins and non-cousin couples distinctively.

The study found that patriarchy is a significant factor among cousin couples and it was found to be positively affecting the psychosocial burden of beta thalassemia major among cousin couples. It has been found that parental knowledge of disease has a significant effect on psychosocial burden of beta thalassemia major among both cousin and non-cousins couples. The effective management and prevention of thalassemia requires comprehensive understanding and knowledge

about beta thalassemia major (Elewa & Elkattan, 2017; Rund & Rachmilewitz, 2005). Lack of knowledge and awareness regarding causes and management of beta thalassemia major, not only worsen the disease but has a strong impact on social and psychological adjustment of parents of sick children (Abu Shosha & Al Kalaldeh, 2018).

Parental maladjustment due to continuous stress and social pressure imposes a negative effect on their quality of life (Mettananda et al., 2019; Tomaj et al., 2016) and social adjustment (Inamdar, Inamdar, & Gangrade, 2015). Strong emphasize on endogamy and cultural practices for marriages restrain couples and families to follow precise management practices for the treatment of beta thalassemia major (Bener, Al-Mulla, & Clarke, 2019) and they avoid pre/postnatal diagnosis and genetic screening methods (Antonarakis, 2019). As a result, the genetic abnormality (beta thalassemia major) prompts over the large scale across the world. It has been generally estimated that, every year 50 thousand to 1 lack children, living in the low and middle income countries die because of this chronic genetic aliment (beta thalassemia major), however, an estimated population of 7% in the entire world is the carrier of hemoglobin disorder in the form of beta thalassemia major (Arif et al., 2008).

Lack of social support is also a significant factor, to increase psychosocial burden of the disease (Palanisamy, Kosalram, & Gopichandran, 2017). Like many other chronic illnesses, patients of thalassemia and their families required meticulous support from relatives, health professionals and from rest of the community members to manage their psychological and social maladjustment. It has been investigated by researchers (Patel et al., 2019) that family is believed to be the primary and most important source of social support for patients and their parents to mitigate the psychological and social burden caused by thalassemia. Patients of thalassemia are required medical as well as social support from their families, medical professionals and other community members for their social adjustment and confrontation against thalassemia which requires moral, social and medical support to the children and their families. The effective management and social support also required general knowledge about the disease, so the general population of any area at least know the basic (Kelsey, 2015).

Table 4.38

*Predictors of Psychosocial Burden of Beta Thalassemia Major*

| | Unstandardized Coefficients | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Models | 1 | | | 2 | | | 3 | | |
| Predictors | B(SE) | t | *p* | B(SE) | t | *p* | B(SE) | t | *p* |
| SERF | .591(.026) | 22.309 | .000 | .589(.026) | 22.342 | .000 | .593(.026) | 22.488 | .000 |
| CRF | | | | .062(.021) | 2.909 | .004 | .067(.021) | 3.119 | .002 |
| Knowledge | | | | | | | -.102(045) | -2.273 | .023 |
| F | | 247.6** | | | 139.8** | | | 101.9** | |
| $R^2$ | | .210 | | | .231 | | | .248 | |
| $\Delta R^2$ | | | | | .021 | | | .017 | |
| $\Delta F$ | | | | | 25.4 | | | 20.4 | |

**Note** *$p$<.05, **$p$<0.001, $\Delta R^2$= R square changes, $\Delta F$= F changes, N=932

**Predictors;** SERF=Socio-economic Risk Factors, CRF=Cultural Risk Factors, Parental Knowledge

**VIF Considering Last MLR Model**; SERF=1.02, CRF=1.4, Parental Knowledge=1.434

**Durbin-Watson;** 1.779

**Excluded Variables in for Last MLR Model**; PND=Practices of Pre/postnatal Diagnosis, DARF=Disease Allied Risk Factors

A multiple linear regression (MLR) was applied, using forward selection method to predict that socioeconomic risk factors, cultural risk factors and parental knowledge could significantly explain the variance of psychosocial burden of beta thalassemia major on parents of sick children. Before elucidation of multivariate linear regression model, researcher assured the assumptions of multicollinearity by using VIF (variance inflation factors) and found a non-significant value for SERF=1.02, CRF= 1.4 and parental knowledge= 1.434. The second assumption was satisfied for the concern of independent error and value of Durbin-Watson test revealed 1.779, which was also satisfactory for the assumption. The third assumption of regression analysis was based on linearity and the chart of normal distribution and scattered plots are attached in (ANXEURE-E).

The data mentioned in table 4.38 reveals that socioeconomic risk factors, cultural risk factors and parental knowledge of the disease (thalassemia) are significant predictors of psychosocial burden of beta thalassemia major on parents of sick children ($F$=101.9, $p$<.001). The findings of present study revealed, that 24.8% of the variance had been explained by three independent variables (socioeconomic risk factors, cultural risk factors and knowledge). The data revealed that one unit increase in socioeconomic risk factors (B=.593)** and cultural risk factors (B= .067)** increased 59.3% and 6% psychosocial burden of beta thalassemia major among parents of sick children. However, it has been seen that parental knowledge significantly decreased the psychosocial burden of the disease (B=-.102)** on parents of sick children.

Researches (Aziz, Sadaf, & Kanwal, 2012) and (Hakeem, Mousa, Moustafa, Mahgoob, & Hassan, 2018) found that social, cultural and economic factors impose a rigorous social and psychological burden on patients and their parents and it has been intensively reckoned by (Zaheer et al., 2016) that parents face social pressure due to beta thalassemia major. It is also evident from the findings of (Neha Kumar et al., 2019), that parents face social isolation and stigmatization due to the sickness of their children. Researchers, (Gamayani et al., 2017; Naresh Kumar, Singh, Khullar, & Arora, 2018) also found that school performance and higher absenteeism of children is associated with their disease. Many of the researchers (Antonarakis, 2019; Hakeem

et al., 2018) also highlighted social and cultural factors which significantly contribute in stigmatization of mothers of sick children. The present study, tried to incorporate the significant social and cultural factors, such as social and financial support along with the level of awareness to build a significant variable for the prediction of psychosocial burden of beta thalassemia major.

Religious beliefs and practices for the management of beta thalassemia major have been studied by Pakistani researchers (Maheen, Malik, Siddique, & Qidwai, 2015) and many others (Cremonini, Westerheijden, & Enders, 2009; Katz, Lazarsfeld, & Roper, 2017; Nutini & Bell, 2019) at a global level and found to be the significantly associated with propagation and imposition of psychosocial burden of beta thalassemia major. In present study, the researchers tried to incorporate religious and cultural beliefs and practices for the estimation of variance in dependent variables. The disease allied risk factors; problems of blood transfusion and management have been incorporated in this study as predictors of psychosocial burden of beta thalassemia major. The practices of pre/postnatal diagnosis and parental knowledge of beta thalassemia major have also been incorporated as the predictors of psychosocial burden of beta thalassemia major on parents of sick children.

The empirical evidences of this research did not validate disease allied risk factors and practices of pre/postnatal diagnosis as variance explanatory variables. However, the variation being unexplained by socioeconomic and cultural risk factors, along with the parental knowledge of the disease was almost 75%. The unexplained variation remained evident because of some other factors, which need to be incorporated and required extensive attention for quantitate measurement of psychosocial burden of beta thalassemia major. The justification of these variables is also significant in the sections of univariate and bivariate analysis of this research.

The unexplained variance of psychosocial burden of beta thalassemia major, in terms of child's education and school performance is based on their percentages of ages and on the level of education, because 40.9% of the sick children were not yet reached to the approbative school age (less than 5 years) to measure their effective

school performance. Similarly, majority of the children (86.6%) were having no education, so parents were unable to be sure about their educational performance. The study also highlighted in the previous findings that there are many demographic and socioeconomic factors such as; residential status, (71.1% from rural areas), parental education (16% mothers and 40% fathers were illiterate) and low level of monthly income (10 to 30 thousand PKRs) which can cause psychosocial burden of beta thalassemia, other than the predictors employed in the model of study.

The study also found in the bivariate analysis section that, there is a significant difference between rural and urban people to contextualize psychosocial burden of beta thalassemia major (see table 4.10). Furthermore, the ethnic differences are also significant in terms of psychosocial burden of the disease (see table 4.19 and 4.20). In most of the developing countries including Pakistan, rural people have lack of education, low level of income (Asghar, 2018; Subhani, Yaseen, Khan, Jeelani, & Fatima, 2015) and they remain under huge psychological and social pressure (Lodhi et al., 2019; Waqas et al., 2015) so it assures that factors can be optimum variance explanator of psychosocial burden of beta thalassemia major.

The other contributory factors may also tend to be religious beliefs and practices, which required distinct and in-depth analysis because the study also found that most of the parents did not confirm the termination of pregnancy as encouraging religious belief (46.6%) and a considerable number (42%) were feeling no stigmatization as well as (53.6%) denied that couples' relationship broke due to disease (see table 4.10). Parental factors such as mothers' education and monthly income of family has a significant negative relationship with psychosocial burden of beta thalassemia major. However, the monthly expenditures were in a significantly negative relationship with psychosocial burden of the disease (see table 4.23).

### 4.18 Summary of Multivariate Analysis

Summarizing the results of multivariate analysis, researcher found that based on generalized linear regression model, ethnicities and consanguinity have a significant interaction effect on socioeconomic, cultural and disease allied risk factors, parental

knowledge and practices of pre/postnatal diagnosis of beta thalassemia major ($p<0.05$). However, it has been found that there is no significant interaction effect of two independent variables on psychosocial burden of beta thalassemia major ($p>0.05$).

The researcher also estimated marginal means of risk factors, knowledge, practices of pre/postnatal diagnosis and psychosocial burden of beta thalassemia major among cousin/non-cousin couples for five selected ethnic groups (Punjabi, Pashtun, Sindhi, Balochi and Kashmiri). The study found that Kashmiri and Panjabi ethnic groups having cousin and non-cousin marriages are more likely towards socioeconomic risk factors of beta thalassemia major. While the least preferences of socioeconomic risk factors have been seen between both (cousin, non-cousin) couples of Sindhi ethnic groups. It has also been found that cousin and non-cousin couples among Punjabi ethnicities have higher predilections towards cultural risk factors of beta thalassemia major. While the least preferring groups were Kashmiri and Sindhi non-cousin couples. Punjabi cousin and non-cousin couples were also more likely towards disease allied risk factors however, the least preferences have been seen among Kashmiri and Sindhi non-cousin couples.

The study also found that cousin as well as non-cousin couples-among Kashmiri and Punjabi ethnicities have higher means of parental knowledge regarding beta thalassemia major. The lowest mean score of knowledge has been seen among Sindhi non-cousin couples. Practices of pre and postnatal diagnosis have been explicitly higher among Pashtun and Balochi non-cousin parents and between both parents of Punjabi ethnicity. The study found that Punjabi parents, having cousin and non-cousin marriages have higher proportion of psychosocial burden of beta thalassemia major contrary to the both parents of Kashmiri ethnicities, who have least effect of psychosocial burden of beta thalassemia major.

It has been found that parental knowledge of the disease, social support, stigmatization, marital breakups (due to disease), belief in unnatural forces and religious beliefs regarding (termination of pregnancy, prevention of thalassemia and cousin marriages) were significant predictors of psychosocial burden of beta

thalassemia major among non-cousins and cousin couples. Additionally, patriarchy was only significant predictors of outcome variable among cousin couples. The study also found that the significant predictors of psychosocial burden of beta thalassemia major are (for all respondents); socioeconomic and cultural risk factors along with the parental knowledge of beta thalassemia major.

# CHAPTER 5

# 5. MAJOR FINDINGS, RECOMMENDATIONS AND CONCLUSION

This section of the present study summarizes and highlights the major findings of empirical data and provides suggestions for policy makers for effective management and prevention of beta thalassemia major. It has been found and explored in the first two chapters introduction (chapter one) and literature review (chapter two), that the propagation of beta thalassemia major is evident across the world, and especially, in developing counties because of lack of education and awareness for effective management of beta thalassemia major. Pakistan is also witnessing an alarming propagation of beta thalassemia major in almost all regions, due to significant social, cultural, religious and epidemiological factors. The major objective of the present study was, to sociologically analyze the risk factors (socioeconomic, cultural and disease allied) and psychosocial burden of beta thalassemia major on parents of sick children in Punjab Province. This chapter includes major findings of the study, followed by conclusion and some practical as well as theoretical recommendations.

## 5.1. Major Findings

- According to the gender of patients, (52 %) were male children while (47.9%) were female patients in the study area. The findings revealed that average age of thalassemic children was 8.5 years and majority of the children (86.6%) was out of schools due to their illness. The respondents of this study were, 375 fathers and 557 mothers of sick children. According to the percentage distribution of residential status of the respondents, 71.1% were the residents of rural areas of Punjab Province whereas 28.9% were the residents of urban areas of Punjab Province.

- Analysis revealed that half of the fathers of sick children (57.7%) were reported to belong the age group of 25-40 years while 24.6% were, above 40 years and 17.7% were reported to belong to the age group of below 25 years. The classifications of mothers' age revealed that 54.7% of the mothers of thalassemic children were

reported to belong to age group of 25-40 years, 30.4% were less than 25 years and 14.9% were above 40 years. The average age of fathers of sick children at the time of marriage was 23.92 years, while the average age of mother in this classification was 21.7 years and maximum age was 35 years, but minimum age of mothers was 12 years.

- The study found that 16.1% of the fathers of sick children were illiterate in study area and the average score of father's educations was 7.65. The mother's educational level revealed that (40.1%) were illiterate and only few mothers (2.1%) completed graduation and 2.4% post-graduation in study area. The average score of mother's educations was 5.64. Majority of the respondents (62.7%) had average income between 10,000 to 30,000 (PKRs), while only few (8.2%) had income level more than 50,000. The average monthly income of a family was 29,482.83 (PKRs), which is equal to 188 USD approximately. While the average measure of monthly household expenditures was 22,633.83 PKRs.

- It has been found that half of the children showed fatigue (50.5%) and (7.6%) reported weakness as an initial symptom of their disease. While (10.6%) of the children with beta thalassemia flaunted pale yellow skin and (5.6%) were brought to the physician because of facial bone deformities, before the diagnosis of beta thalassemia major. Fifteen percent suffered fever and (8.9%) showed slow physical growth as compared to their siblings and other children.

- It was found that 72% respondents were aware that, cousin marriages are one of the main causes for genetic disorders and a very high majority of the respondents were aware of the genetic counseling (92.9%) while few of them (7.1%) were uninformed about genetic counseling.

- The study found that 45% parents consulted government hospital at first step and after blood screening they came to know that their child was sick due to thalassemia, while 36.4% consulted private clinics.

- The monthly blood transfusion procedure was frequent among 35.6%, while bimonthly transfusion was reported by 48.3% in the study area. Majority of the parents (76.6%) were managing blood from selected thalassemia foundations, while only few (9.8%) were managing privately, 8.8% were depending on family

members and only 2.5% were managing from their friends and relatives and consulting thalassemia centers for transfusion.

- Regarding parental knowledge of the disease, the study found that majority of the respondents (89.7%) marked the statement correct; that thalassemia can be identified by blood test and (72.4%) were of the view that thalassemia can only be treated with medication. A greater number (61.7%) viewed that intermarriages are important risk factors for the propagation of thalassemia and most of the respondents (62.9%) viewed it correct that, if one parent has thalassemia minor (is a carrier), the couple has a chance of having a child with thalassemia disease, while (33.6) marked it incorrect to be an inherited genetic disorder.

- Majority of the respondents (74.8%) marked the statement correct that thalassemia can pass on to the child through a gene and a greater majority (84.3%) was familiar, that thalassemia is a sexually transmitted disease. Most of the parents (81.0%) were well known that thalassemia can be detected during pregnancy and if parents have thalassemia minor, it increases the chances of a thalassemic child marked by 82.3% respondents as correct. It has also been found that 75.5% of the parents were familiar with the importance of family planning after carrier detection and emphasized for compulsory birth control practices.

- The present study also revealed the views and practices of pre/postnatal diagnosis among parents of the sick children and found that a greater number of parents (69.1%), were in a strong favor that government should impose, and couples and families should adopt the pre/post-natal screening mechanisms to prevent beta thalassemia major.

- With respect to the socioeconomic risk factors, the study found that 69.9% of the parents were agreed that low financial condition is the main hurdle for the treatment of beta thalassemia major and 68.3% of the parents considered financial management of the disease a very serious problem because of their lower socio-economic. It was found in the study that 63.1% respondents were strongly agreed, while only (10.6%) were strongly disagreed, that attitude of doctors, nurses and other paramedical staff was sympathetic towards them and level of their social participation in different communal activities and gatherings.

- The study found that due to a strong emphasize on endogamy and traditional cultural practices in Pakistan, majority of the respondents (78.9%) was married to cousins, while only (21.1%) were married out of their family. The study found that majority of the parents were married to their first cousins (49.9%), while (10.6%) were married to second cousin and (11.4%) to third cousin.

- With respect to the cultural and religious risk factors of disease, it has been found in the present research that 66.8% of respondents were agreed with the significance of religious beliefs and instructions regarding prevention and control of thalassemia. The study also found that 55.6% of the respondents have a firm belief that their religion encourages cousin marriages while almost an equal number did not support the statement. It has also been found that an equal percentage of the respondents (55.6% agreed and 55.6% disagreed) about the religious consent for abortion in case of carrier's detection.

- More than half of the respondents of the present study believed that unseen and aberrant forces cannot cause beta thalassemia major (58.5%). The data also revealed that most of the parents do not believed that relationship of a married couple break-up after knowing that they have a thalassemic child (53.6%). The study also found that patriarchy plays a significant role regarding treatment, screening and management of beta thalassemia major in Pakistan (70.8%).

- In the measurement of disease allied risk factors, the study found that a greater majority of the parents (91.7%) was of the view that they cannot bear the treatment cost, hospital expenditures, cost of therapeutic facilities and they were unable to manage the travelling expenditures for blood transfusion and regular checkups of their children.

- The study found a significant amount of psychosocial burden of beta thalassemia major on the parents of sick children because majority of the parents were agreed (81.4%) that they feel anxiety due to the sickness of their child, because of intensive care required by sick children, parents remain unable to perform creative and analytical tasks in their professional lives.

- This study also comprehends that 61% of parents were agreed, that they have weak family interactions due to their thalassemic child while a considerable

number was also disagreed (35.4%) because of joint family system and strong social ties. The study also revealed that majority of the parents were feeling social isolation due to their thalassemic child (75%).

- This study found that majority (81.4%) of the parents viewed that they have a sense of guilt and feeling of confusion due to the sickness of their child as well as the feeling of bad self-expressions of sick children, because the disease significantly affects the physical features and body images of a child due to slow growth and health complications.

- The study found substantial differences among rural and urban parents with regards to knowledge (M=11.849, 10.755 & SD=2.635, 3.422). The study also found significant differences of knowledge on the basis of parental information about carriers of consanguineous marriages (M=10.804, 11.816 & SD=3.095, 2.806) and the existence of thalassemia carriers in non-cousin marriages (M=10.463, 12.498 & SD=2.961, 2.530) along with the classification of the respondents, having cousin marriages or not (M=11.934, 11.425 & SD=2.924, 2.917) in study area ($p<0.05$).

- The study found significant differences ($p<0.05$) of PNDs, on the basis of residential status of the respondents (rural, urban) (M=24.965, 23.818 & SD=4.739, 4.966), number of thalassemic children (M=24.893, 22.402 & SD=4.829, 4.251), any other patient with thalassemia in their (respondent's) family (M=25.862, 22.912 & SD=3.812, 5.537), carrier existence in non-cousin marriages (M=23.850, 25.340 & SD=5.151, 4.409) and parental awareness regarding genetic counseling (M=23.409, 24.727 & SD=3.798, 4.889).

- Study found that respondents had significant differences of socio-economic risk factors, on the basis of their gender (M=36.08, SD=7.731 & M=37.099, SD=7.484), having any other patient of thalassemia in their family (M=39.156, 32.938 & SD=5.560, 8.597), awareness of genetic counseling (M=38.409, 36.427 & SD=6.773, 7.673) cousin marriages (M=39.126, 35.881 & SD=5.685, 7.931), $p<0.05$.

- Data of this study also revealed that respondents had significant differences of cultural risk factors of beta thalassemia major on the basis of their gender

(M=25.780, SD=6.02), localities (M=25.813, 24.100 & SD=5.756, 5.650), number of thalassemic children (M=25.027, 27.824 & SD=5.560, 6.908), having any other patient of thalassemia in their family M= 24.084, 27.049 & SD=4.919, 6.413), awareness of genetic counseling (M=23.303, 25.472 & SD=4.158, 5.853) their consanguinity (M=24.502, 25.537 & SD=6.077, 5.675), $p<0.05$.

- Study also found that there are significant differences of disease allied risk factors of beta thalassemia major, among the respondents of present research, on the basis of their number of thalassemic children; either one or two (M=8.925, 10.164 & SD=3.903, 4.104), any other patient of thalassemia in their family (M=8.536, 9.780 & SD=3.090, 4.802) and on the basis of their cousin marriages (M=8.538, 9.193 & SD=2.807, 4.183).

- The study found significant differences of psychosocial burden of beta thalassemia major, on the basis of residential area (rural, urban) (M=59.710, 56.286 & SD=9.426, 9.505), number of thalassemic children (M=58.116, 63.938 & SD=9.579, 7.767) and on the basis of consanguinity (M=63.1066, 57.5469 & SD=8.385, 9.531).

- It has been found that there are significant differences among different ethnicities of children's fathers with regard to the parental knowledge of beta thalassemia major $F$ (4,927) =8.062, socioeconomic risk factors $F$ (4,927) =7.110, cultural risk factors $F$ (4,927) =4.176 and disease allied risk factors $F$ (4,927) =4.142 of beta thalassemia major because $p<0.01$.

- The study found highly significant differences of parental knowledge, prenatal practices, risk factors and psychosocial burden of beta thalassemia major, among different ethnicities of mothers of sick children have been found in this study and according to the data; parental knowledge $F$ (4,927) = 4.378, prenatal diagnosis $F$ (4, 927) = 3.685, socioeconomic risk factors $F$ (4, 927) = 7.614, cultural risk factors $F$ (4, 927) = 4.426 and psychosocial burden of beta thalassemia major $F$ (4, 927) = 3.457.

- The study also found that, there are significant differences of parental knowledge $F$ (2, 929) = 5.190, prenatal diagnosis $F$ (2, 929) = 10.280, psychosocial burden $F$

(2, 929) = 19.528 and cultural risk factors of beta thalassemia major $F$ (2, 929) = 4.064, based on family structure of the respondents.

- The study found highly significant differences ($p$<0.01) of means in parental knowledge $F$ (3, 928) = 7.741, practices of prenatal/postnatal diagnosis $F$ (3, 928) = 25.844, socio-economic risk factors $F$ (3, 928) = 16.325, cultural risk factors $F$ (3, 928) = 79.029, disease allied risk factors $F$ (3,928) = 6.459 and psychosocial burden of beta thalassemia major $F$ (3, 928) = 11.484 among different occupational groups.

- The study found that there exist a significant and positive relationship between parental knowledge of the disease and father's age at the time of marriage, father's education, mother's education and monthly expenditures on medications of sick children ($p$<0.05). The empirical evidences of this study also revealed that practices of prenatal diagnosis (PNDs) are significantly positively associated with father's and mother's education, monthly income and expenditures of a family ($p$<0.01). The findings of this study highlighted that education of parents has a significantly positive relationship with socioeconomic risk factors of beta thalassemia major.

- The results of this study found that mother's age at the time of marriage and her education, along with monthly income of a family has a significantly negative relationship with psychosocial burden of beta thalassemia major ($p$<0.01). However, mother's age and monthly expenditures of family has a significant positive relationship with psychosocial burden of beta thalassemia major ($p$<0.05).

- The results of generalized linear regression revealed a significant strong interaction effect of ethnicity and consanguinity on parental knowledge of beta thalassemia major in $F$ (4, 932) = 4.147, $p$<0.05, $\eta_p^2$ =.135 and found that, Kashmir cousins and non-cousin couples were more inclined towards the knowledge of beta thalassemia major, as compared to all other ethnic groups. However, the knowledge acquisition in non-cousin Sindhi ethnic communities was low as compared to Pashtun, Punjabi and Balochi ethnic groups.

- The present study found a significant interaction effect of consanguinity and ethnicities $F$ (4, 932) = 4.357, $p$<0.05, $\eta_p^2$=.13 and found that Balochi and Sindhi ethnicities had least preferences towards PNDs. While, Punjabi ethnic groups, cousin couples had higher preferences of PNDs while Pashtun non-cousins were more inclined towards practices of pre/postnatal diagnosis. For collective analysis of preferences towards PNDs between cousins and non-cousin parents among Kashmiri ethnicities, both couples had higher presences of practices of pre/postnatal diagnosis.

- The present study also revealed that there is a significant interaction effect of ethnicities and consanguinity $F$ (4, 932) = 8.111, $p$<0.05, $\eta_p^2$ = .185 on socioeconomic risk factors of beta thalassemia major, as Kashmiri non-cousin couples were more likely towards socioeconomic risk factors of beta thalassemia major as compared to other ethnic groups.

- It has been found that the consanguinity and ethnicities have a significant interaction effect on cultural risk factors of beta thalassemia major $F$ (4, 032) = 4.830, $p$<0.05, $\eta_p^2$ = .145. Data reveals that Balochi and Pashtun non-cousin couples were more likely to follow cultural restrictions of beta thalassemia major, as compared to cousin couple. However, Kashmiri cousin couples have higher preferences towards religious and cultural beliefs of beta thalassemia major as compared to all other groups.

- Findings of this study revealed that there is a significant interaction effect of consanguinity and ethnicity on disease allied risk factors of beta thalassemia major $F$ (4, 932) = 3.892, $p$<0.05, $\eta_p^2$= .110 and Sindhi and Kashmiri non-cousin couples had least predilections of disease allied risk factors as compared to other non-cousin couples in the study area. However, the higher presences of disease allied risk factors have been seen in Balochi non-cousins and Punjabi cousins as well as non-cousin marriages.

- The results of multinomial logistic regression revealed that parents of sick children married to their 1$^{st}$ cousins were less likely to face moderate OR= .801and high level of psychosocial burden of beta thalassemia major OR=.795, $p$<.001 as compared to the non-cousin couples. It has been found that parents of

sick children, married to their 2nd cousins were more likely to face moderate psychosocial burden OR= 1.722, *p*<.001 while less likely to face high level of psychosocial burden of beta thalassemia major OR=.476, *p*<.001 as compared to the non-cousin couples.

- The study also revealed that husbands' parents married to 1st cousins were significantly less likely to face moderate OR= .812, *p*<.001 and high psychosocial burden of beta thalassemia major OR= .783, *p*<.001 as compared to the those who had non-consanguineous marriages. However, it has been found that only distant relatives were significantly more likely to face moderate psychosocial burden of beta thalassemia major OR= 1.301, *p*<.001, with refence to the non-cousins.

- The results also revealed that those respondents (if father then his grandparents, or in case of mother then parents of his mother/father in law) who were married to their 1st cousins were more likely to face moderate psychosocial burden OR= 1.895, *p*<.001. While less likely to face high psychosocial burden of the disease OR=.120, with refence to those, who were married to non-cousins.

- The data also revealed that respondents (if mother of sick child then her parents and in case of father then his mother/father in law) married to 1st cousins were more likely to face moderate OR= 1.266, *p*<.05 and high psychosocial burden of beta thalassemia major OR= 1.360, *p*<.001 with reference to the non-cousins. However, 2nd cousins were less likely to face moderate OR= .745, *p*<.001 and high psychosocial burden of the disease OR=.890, *p*<.05 as compared to the non-cousins.

- It has been found that parental knowledge of the disease, social support, stigmatization, marital breakups (due to disease), beliefs in unnatural forces and religious beliefs regarding (termination of pregnancy, prevention of thalassemia and cousin marriages) were significant predictors of psychosocial burden of beta thalassemia major among non-cousins and cousin couples. Additionally, patriarchy was only significant predictors of outcome variable among cousin couples.

- The study found that socioeconomic risk factors, cultural risk factors and parental knowledge of the thalassemia are significant predictors of psychosocial burden of

beta thalassemia major on parents of sick children (F=101.9, $p$<0.001). The findings revealed that the amount of variance explained by three variables is 24.8%. The data revealed that socioeconomic risk factors and cultural risk factors are the significantly positive predictors of psychosocial burden of beta thalassemia major. However, it has been seen that parental knowledge is a significant but negative predictor of psychosocial burden of beta thalassemia major, on parents of sick children.

## 5.2. Recommendations

Based on findings of this study and inferred from the literature gap, the researcher intends to propose some theoretical and practical persuasions for academic researchers, policy makers, epidemiologists, professionals, change agents, social activists and community mobilizers for the management and prevention of beta thalassemia major. According to the prescribed format of IIUI, the section (recommendations) appears after conclusion in dissertation but as suggested by the foreign examiner the section (conclusion) should be placed at the end of dissertation.

### 5.2.1. Theoretical recommendations

- The prominent effect of socioeconomic, cultural factors, along with the parental knowledge of disease in increasing psychosocial burden of beta thalassemia major validated the theoretical model (bio-psychosocial model of health). However, the researcher builds an argument based on priory works and results of present study, that the model is an appropriate selection to study beta thalassemia major, but-psychological aspects should be measured as an impact, rather than cause of beta thalassemia major.
- Researcher found that in Pakistan and among all countries with higher propagation of beta thalassemia major, researchers did best to conceptualize and measure beta thalassemia generally and beta thalassemia major particularly but none of them provided theoretically validated constructions for disease. Researcher suggests, that based on applied theoretical (bio-psychosocial) and empirically validated model

and findings of this study, a comprehensive model can be built for operationalization of beta thalassemia major.

- The findings of epidemiologists and health experts are though important and based on pure clinical trials, so their validity cannot be challenged but they do not incorporate social, cultural and religious beliefs and practices such as; marriages practices, religious preaching's and patriarchy as propagating factors of beta thalassemia major, which are socially constructed observable phenomena and play a vital role for the prevalence and prevention of beta thalassemia major.
- Analysis and measurement of beta thalassemia major should also incorporate the theories of cultural and psychological poverty, other than the pandemic measurement of socioeconomic status and analysis of income and expenditures of people.
- It would be fruitful to conduct action researches to improve the parental knowledge of thalassemia and their practices of pre/postnatal diagnosis of beta thalassemia major. Especially focusing the vulnerable communities and disadvantage groups.
- Based on the findings of this research, the researcher suggests, integrating religious and cultural practices with modern health practices in more scientific way for moral sensitization of the parents and families of rural areas along with the epidemiological methods of carrier detection and counseling.

### 5.2.2. Policy recommendations

- In this study, the researcher found that most people belong to the rural areas of province, with little income and low level of formal education, which are the main reasons for lack of knowledge of the disease. To address the issue, government and non-governmental organizations may be actively involved to provide comprehensive knowledge and awareness about beta thalassemia major.
- The counseling and awareness sessions should be conducted regularly, especially with the vulnerable groups (peoples of rural areas, having more traditional beliefs

and practices for the preventive and management practices, with strict endogamous marriages patterns) and for general masses at regular interval.

- Pakistan is lacking the legal measures to control the propagation of beta thalassemia major through strict and firm legislative actions, as taken by the other countries (Turkey, Saudi Arabia, Iran, Jordan and Bangladesh). The only act which can be seen as a measure to control thalassemia is known as "Sindh Prevention and Control of Thalassemia Act, 2013" (ANXEURE-F), which was passed by the Sindh Provisional assembly but not successfully implemented. It should be implemented in a practical way and all provincial government should take such measures through legislation and implement their policies with the help of governmental and non-governmental organizations.
- Government of Pakistan should maintain complete database for thalassemia, by differentiating its all mutations and it should be linked with national record management systems i.e. The National Database and Registration Authority (NADRA) and Pakistan Bureau of Statistics and should be figured out in every Pakistan Demographic and Health Survey (PDHS).
- Government should establish the blood transfusion centers in foremost areas by keeping in view the travelling expenditures and financial burden of disadvantaged groups, as the present study found- majority of the people facing problems to manage blood for their thalassemic children
- It is also the responsibility of religious institutes and groups to work for moral correction of people in a scientific way and should not encourage the traditional values and beliefs for screening and counseling of all genetic disorders and especially beta thalassemia major.
- Health experts and epidemiologist should look beyond the clinical investigation of beta thalassemia major and incorporate the cultural, social, religious and psychological aspects of beta thalassemia major, in collaboration with social scientists for effective control of this deadly disease.
- The voluntary efforts of all thalassemia foundations (Sundas Foundation, Jamila Sultana Foundation, Fatimid Foundation) required more financial and social support, so it becomes the responsibility of government, bilateral donors and other charitable organizations to provide them more resources for advance treatment procedure, like Bone Marrow Transplantation.

### 5.2.3. Recommendations for further studies

- Using a multidisciplinary approach, a team of health experts, epidemiologists and sociologists are required to conduct further study for the analysis of consanguinity and religious factors in more detail to address the social, cultural and religious root cause analysis of beta thalassemia major.

Table 5.1

*Strategic Plan to Prevent Beta Thalassemia Major*

| Category | Strategy | Implementing Agency |
|---|---|---|
| Short term | • Religious sensitization of the couples and families to adopt screening and preventive measures | • Formal and informal educational institutes, Council of Islamic Ideology, and All Pakistan Ulema Council can play an effective role to sensitize masses regarding cousin marriages, termination of pregnancy and other morally allowed preventive measures. |
| | • Access to pre/postnatal diagnostic services are required to be provided to the masses | • District Headquarters Hospitals can provide access to the pre/postnatal diagnosis services for screening and diagnosis of beta thalassemia major. |
| | • Provide genetic counseling | • Community based organizations can create awareness among couples and families to realize the importance of genetic counseling through seminars and counseling sessions. |
| Medium term | • Provision of adequate health facilities | • Provision of adequate health facilities by local government and well equipped basic health units especially in remote areas can reduce the number of infected births. |
| | • Initiation of media campaigns for awareness and advocacy | • Print, electronic and social media can run different campaigns, awareness sessions and telecast short films on causes and consequences of beta thalassemia major. |
| | • To organize thalassemia weeks for capacity building | • Educational institutes can arrange seminars and sessions with the collaboration of NGOs CBOs on thalassemia weeks to encourage students for blood donation and volunteerism. |
| Long term | • Legislative measures for carrier detection | • Prior to the marriage, Government can execute compulsory screening for couples and in case of being carrier; they should not be allowed to marry. |
| | • To maintain a data bank for monitoring and evaluation | • In collaboration of Pakistan Bureau of Statistics, National Institute of Population Studies, NADRA and NIH a data bank can be maintained for confirmed cases along with their complete bio-data to monitor the prevalence of disease. |
| | • Bone marrow transplantation centers and establishment of blood banks | Subsidize and governmentally funded bone marrow transplantation centers and blood banks can be established especially in remote and deprived areas of the country so the poor families can be compensated to bear the financial cost of treatment and medication of the disease. |

## 5.3. Conclusion

Beta thalassemia is a disorder which is categorized in inherited hemoglobin issues, which are linked to a quantitative deficiency of functional beta global chains (Muncie & Campbell, 2009). The chronic and life ending disease-beta thalassemia major is featured as defective production of the beta global chains in human blood (Rehman, Masood, Sheikh, & Mehboob, 2019). An imbalance in the coordination making of the sub units of hemoglobin results in making the bone marrow produce immature red blood cells (Martino et al., 2018). Approximately 50,000 children pass away every year due to thalassemia developing countries (Jyothshna & Kumar, 2016), whereas discernibly 7% of the world's population is hemoglobin disorder carrier (Roth et al., 2018; Weatherall, 2010). Five out of hundred individuals are currently suffering from thalassemia and approximately eight million population of Pakistan is a thalassemia carrier (Azam, 2009) .The approximate life span of thalassemia patient is ten years (Lodhi, 2003).

The present study intended to sociologically analyze the risk factors prominently social, cultural and disease allied of beta thalassemia major and its burden on parents of sick children. For this purpose, one of the populationally dense, geographically scattered and multicultural provinces (Punjab) of Pakistan was selected as a study area. The motive was; to find the cultural, social and practical practices and beliefs of people, for the management and prevention of the disease and the considerations of parents for psychological and social problems as a perceived psychosocial burden for them. The study found that majority of the parents belong to the lower middle class and for the treatment and blood transfusion, they were dependent on thalassemia foundation. Almost all the patients were receiving blood transfusion and being treated with chelation therapies and nobody opted bone marrow transplantation because being an expensive treatment procedure. It has been seen that due to lack of formal education, most of the parents were unaware regarding knowledge of thalassemia and its preventive as well as effective management practices.

Due to lack of knowledge and awareness of beta thalassemia major, this genetic disorder becomes chronic for carriers of thalassemia (Manzoor & Zakar, 2019). It is evident that low level of education in all high rated countries (India,

Bangladesh, Iran, Saudi Arabia, Jordon and Pakistan), triggers thalassemia (Haq et al., 2017; Hossain et al., 2016; Mashayekhi, Jozdani, Chamak, & Mehni, 2016; Olwi, Merdad, & Ramadan, 2018; Sarvestani, Hasanifar, & Bagheri, 2019).

The study found that although parents of sick children are aware of the importance of genetic counselling and screening and they emphasized enough for compulsory premarital, pre/postnatal screening programs but their own knowledge and practical measures were lacked these core preventive practices. The study found that majority of the parents was also influenced by cultural and religious myths for the causes, treatment and management of beta thalassemia major. It is intensively reviewed by the researcher and empirical evidences of study also revealed that regular treatment, blood transfusion and precautionary diet imposed a rigorous economic burden on parents of sick children. They respondents of this study lacked social and financial support from their friends and relatives and their social life was critically affected because of the sickness of their children.

Communities with traditional beliefs and practices for heath management, have greater chances of propagation of thalassemia (Hamamy, 2012) because of angelic emphasize on consanguinity, endogamy, cast and ethnic preferences for marriages. Meanwhile, the falsifications- attached to religion, unwise people go for scientific treatment of beta thalassemia. In all patriarchal and traditional societies generally and in Pakistan particularly, termination of pregnancy is an unlawful act (Moghadam, 1992), the presenters and explainers support such kind of argument with false religious beliefs, because the religious aspects of abortion in Pakistan (through Islam) are very clear, within the particular period of time (Jafri et al., 2012). The study found that majority of the parents of sick children was married with their cousins and especially with 1$^{st}$ cousin. The data also reviled that among respondents of this study, repeated cousin marriages, ethnic and cultural preferences were encouraged along with non-scientific religious practices and beliefs. These factors cause higher propagation of beta thalassemia major and perceived as barricades for the preventive measures of disease.

Due to lack of financial and social support, parents of sick children were facing massive economic and social pressure in their lives. Their social and workplace activities were affected by the sickness of their children and it is a challenge for them

to manage their social participations and maintain their social interaction with other community members. Their pessimism creates many psychological abnormalities e.g. anxiety (Khamoushi et al., 2015), depression (Khoury et al., 2012), mental stress (Messina et al., 2008) and social problems e.g. social isolation (Mashayekhi et al., 2016), stigmatization (Cao & Kan, 2013), weak social interaction (Gharaibeh, Amarneh, & Zamzam, 2009) and denial of acceptance (Ali, Sabih, Jehan, Anwar, & Javed, 2012), for their children and for themselves.

The possibility to overcome this biological disorder is, to create high level of awareness among the families about genetic counseling and screening (Katapodi et al., 2018) and empowering parents to opt the precautionary measures to avoid thalassemia (Bender, 2017). Due to denial and unawareness about genetic counseling and screening, most of the parents blame themselves for becoming the cause of thalassemia in their children.

## 5.4. Hypothetical Conclusion

Based on the findings of the study, the below table explains conclusion of hypothetical statements of this study.

Table 5.2

*Hypotheses Summary*

| Hypotheses | Statements | Results |
|---|---|---|
| H1 | Higher exposure towards the knowledge of disease is likely to decrease the psychosocial burden of disease | Accepted |
| H2 | Higher exposure towards the practices of pre/postnatal diagnosis of disease is likely to decrease the psychosocial burden of disease. | Rejected |
| H3 | Higher exposure towards socio-economic risk factors is likely to increase the psychosocial burden of disease | Accepted |
| H4 | Higher exposure towards cultural risk factors is likely to increase the psychosocial burden of disease | Accepted |
| H5 | Higher exposure towards disease allied risk factors is likely to increase the psychosocial burden of disease | Rejected |

Table 5.2 shows that three hypotheses of the study regarding effects of parental knowledge of the disease, exposure towards socioeconomic risk factors and cultural risk factors on psychosocial burden of beta thalassemia major, on parents of thalassemic children were statistically analyzed and accepted. However, the hypothetical effect of practices of parents regarding pre/postnatal diagnosis and effect of disease allied risk factors on psychosocial burden of beta thalassemia major was rejected in present study.

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**ANXEURE-A**

## List of Thalassemia Foundations-Punjab Chapter

| | |
|---|---|
| **1. Human welfare Thalassaemia Centre** | **Attock** |
| **Contact Person:** | Prof. Dr. Maqsood |
| **Address:** | A-10/2 Upper story Wapda Customer Service, Near Jamia Masjid, Attock City |
| **Phone:** | 0300-5490600, 0311-1222038 |
| **Email:** | lifeglimp@yahoo.com |
| **Total number of Thalassaemics currently registered:** | NA |
| **Service Provided:**<br>• Blood Transfusion<br>**Any Other Services:**<br>• Iron Chelation & Genetic Counseling | |
| **2. Ali Zeb Foundation** | **Faisalabad** |
| **Contact Person:** | Syed Shahid Ali Zaidi |
| **Address:** | Inside General Hospital, Ghulam Muhammad Abad, Fasilabad |
| **Phone:** | 041-8715997,<br>041-8722090 |
| **Email:** | alizaibfoundation11@gmail.com |
| **Total number of Thalassaemics currently registered:** | NA |
| **Service Provided:**<br>• Free Blood Transfusion<br>• Iron Chelation<br>• Free Medicines<br>• Extended Family Screening<br>• Free Thalassemia Testing<br>• Genetic Counseling<br>• Awarness Lectures and Camps<br>• Publication<br>• Social Media<br>• Conducting Activities and Events For Thalassemia Patients.<br>**Any Other Services:**<br>• Free Laboratory and | |

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|---|---|
| Diagnostic Centre/Blood Bank<br>• Free General OPD | |
| **3. Amina Bashir Memorial Trust** | **Lahore** |
| **Contact Person:** | Arslan Ahmad, Administrator |
| **Address:** | ¾ Aziz Building Aziz Road Misri Shah, Lahore |
| **Phone:** | 04237284913- 03354578728 |
| **Email:** | abmtlhr@yahoo.com |
| **Total number of Thalassaemics currently registered:** | 50+ |
| **Service Provided:**<br>• Free Blood Transfusion<br>• Iron Chelation<br>• Free Medicines<br>• Extended Family Screening<br>• Free Thalassemia Testing<br>• Genetic Counseling<br>• Awarness Lectures and Camps<br>• Publication<br>• Social Media<br>• Conducting Activities and Events For Thalassemia Patients.<br>**Any Other Services:**<br>• Free Laboratory and Diagnostic Centre/Blood Bank<br>• Free General OPD | |
| **4. Fatimid Foundation Lahore Center** | **Lahore** |
| **Contact Person:** | Col. (R) Anwar Iqbal, Administrator |
| **Address:** | 72-A, Blok D-II, Johar Town, Lahore |
| **Phone:** | 042-35210834-6 |
| **Email:** | lc@fatimid.org |
| **Total number of Thalassaemics currently registered:** | 1895 |
| **Service Provided:**<br>• Clinical Services to patients suffering from Thalassaemia and Haemophilia.<br>• Laboratory Services Screening of Blood for HbsAg, HCV, HIV, RPR and M.P.Blood Grouping and cross match.<br>• Blood Products (Fresh Frozen | |

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|---|---|
| Plasma, Platelets, Cryoprecipitate)<br>• Haematology Coagulation Chemistry<br>• Nursing Care.<br>• Physiotherapy.<br>• Creating awareness about Thasalassaemia and blood collection campaign.<br>• Extended Family Screening<br>• Family counseling<br>• Blood and Blood products transfusion.<br>• Iron chelation therapy.<br>• Splenectomy (Through Govt. Hospital)<br><br>**Any other service:**<br><br>• Family Screening Through PTPP | |
| **5. Sundas Foundation** | **Lahore** |
| **Contact Person:** | Mr. Muhammad Yaseen Khan |
| **Address:** | 880, Shadman Colony,1 Lahore |
| **Phone:** | +92-42-7539232-7539233 |
| **Email:** | info@sundas.org |
| **Total number of Thalassaemics currently registered:** | 1596 |
| **Service Provided:**<br><br>• Blood Transfusion<br><br>**Any Other Services:**<br><br>• Iron Chelation & Genetic Counseling | |
| **6. Thalassaemia Patients and Parents Society of Pakistan** | **Lahore** |
| Contact Person | Mr. Muhammad Yaqoob Babar (General Secretary) |
| Address | 2nd Floor, PMA Trade Center, 66 Ferozepur road, Lahore |
| Phone: | +92-42-7524586 |
| Email: | tppsp@thalassaemia.org.pk |
| Total number of Thalassaemics currently registered | |
| **Service Provided:** | |

| | |
|---|---|
| • Blood Transfusion<br><br>**Any Other Services:**<br><br>• Iron Chelation & Genetic Counseling | |
| **7. Thalassaemia Society of Pakistan** | **Lahore** |
| Contact Person | Prof. Dr. Yasmin Raashid (General Secretary) |
| Address | 146/1 Shadman Jail Raod Lahore |
| Phone: | +92-42-7573911-7596589 |
| Email: | info@thalassaemia.org.pk |
| Total number of Thalassaemics currently registered | 1030 |
| **Service Provided:**<br><br>• Physiotherapy<br>• Orthopadic<br>• S-Ferriten tests<br>• Hb Electrophoresis<br>• Echocardiograpy<br>• Endocrine clinic<br>• Food<br>• Free transportation Education<br>• Art and Craft school –etc | |
| **8. Fatimid Foundation Multan Center** | **Multan** |
| Contact Person | Lt Col (R) Syed Abrar Hussain Gillani, Administrator |
| Address | 26- J, T-Chowk, Shah Rukn-e-Alam Colony Multan |
| Phone: | 061-4554520-21, 4555228 |
| Email: | mc@fatimid.org |
| Total number of Thalassaemics currently registered | 1549 |
| **Service Provided:**<br><br>• Blood & Transfusion to Thalassemic Patients.<br>• CP & FFP for Haemophilic patients & carry out transfusion in the centre.<br>• CS for burnt patients<br><br>**Any Other Services:** | |

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|---|---|
| • We also medicate Thalassemic patients for iron chelation & General disease | |
| **9. Amna Thalassaemia & Haemophilia Society** | **Multan** |
| Contact Person | Dr. Muhammad Sohail Iqbal |
| Address | 5- Khan Colony Opp. Shell Petrol Pump, LMQ Road Multan |
| Phone: | 061-4514023 |
| Email: | Sohail_iqbal30@yahoo.com |
| Total number of Thalassaemics currently registered | 308 |
| **Service Provided :**<br>• Blood Transfusion<br>• Iron Chelation<br>• Genetic Counseling | |
| **10. Thalassaemia Center, Safe Blood Bank & Hematological Services** | **Multan** |
| Contact Person | Mr. Hafiz Muhammad Hanif |
| Address | 1967 Aqsa Street HazoorBagh Road, Multan |
| Phone: | 0300-6301473 |
| Email: | safebloodtransfusionservices@gmail.com |
| Total number of Thalassaemics currently registered | |
| **Service Provided :**<br>• Blood Transfusion<br>• Iron Chelation<br>• Genetic Counseling | |
| **11. Jamila Sultana Foundation** | **Rawalpindi** |
| Contact Person | Mr. Muhammad Usman |
| Address | 24,D . 6th Road Rawalpindi |
| Phone: | 051-4842184 |
| Email: | |
| Total number of Thalassaemics currently registered | 230 |
| **Service Provided:**<br>• Blood Transfusion<br>• Iron Chelation<br>• Prenatal Diagnosis<br>**Any Other Services:** | |

| | |
|---|---|
| • Thalassaemia Carrier Screening | |
| **12. Pakistan Thalassaemia Welfare Society** | **Rawalpindi** |
| Contact Person | Lt. Gen. (R) Fahim Ahmed Khan (President) |
| Address | Thalassaemia House, Tipu Road opp. Rawalpindi Medical College, Rawalpindi |
| Phone: | 051-5951269, 5780749 |
| Email: | pakthalassaemia@hotmail.com |
| Total number of Thalassaemics currently registered | 110 |
| **Service Provided:**<br>• Collection of Blood<br>• Screening Before Transfusion<br>• Awareness Campaign, Workshop and Seminars, TV Coverage and in print Medical About Thalassaemia Major being Carried Frequently.<br>• 8th May International Participation of Parents/ Patients | |
| **13. Kalsoom Society** | **Sialkot** |
| Contact Person | Miss Tabsum Munir |
| Address | Airport Road Gohad pur Kalsoom Hospital, Sailkot |
| Phone: | 052-4291717 0323-4293030 |
| Email: | kulsoomsociety@gmail.com |
| Total number of Thalassaemics currently registered | NA |
| **Service Provided:**<br>• Collection of Blood<br>• Screening Before Transfusion<br>• Awareness Campaign, Workshop and Seminars, TV Coverage and in print Medical About Thalassaemia Major being Carried Frequently.<br>• 8th May International Participation of Parents/ Patients | |

| | |
|---|---|
| **14. Safe Life Organization** | **Sargodha** |
| Contact Person | Tanveer Ahmed Sulehry |
| Address | Hemophilia Chwok-st- town Gill wala road, Sargodha cannt |
| Phone: | 0483750638, 0300-6022996 |
| Email: | safelife2002@gmail.com |
| Total number of Thalassaemics currently registered | |
| **Service Provided:**<br><br>• Collection of Blood<br>• Screening Before Transfusion<br>• Awareness Campaign, Workshop and Seminars, TV Coverage and in print Medical About Thalassaemia Major being Carried Frequently.<br>• 8th May International Participation of Parents/ Patients | |
| **15. Thalassaemia Society of Pakistan** | **Sheikhpura** |
| **Contact Person:** | Prof. Dr. Yasmin Raashid (General Secretary) |
| **Address:** | Thalassaemia Daycare Center, DHQ, Sheikhpura |
| **Phone:** | 0300-9479815 |
| **Email:** | info@thalassaemia.org.pk |
| **Total number of Thalassaemics currently registered:** | 67 |
| **Service Provided:**<br><br>• Blood Transfusion<br>• Iron Chelation<br>• Prenatal Diagnosis<br>• Genetic Counseling<br><br>**Any Other Services:**<br><br>• Awareness Advocacy | |

**ANXEURE-B**
**Demographic variables**

**Patients**

1. Gender of the patient i) Male ii) Female
2. Age at the time of thalassemia diagnosed----------
3. Age now ----------
4. Education (class)----------

**Parents**

5. Respondents' (father/mother) marital status i) Widow ii) Divorce iii) Married
6. What is your relation to the child: i) Father ii) Mother
7. Locality i) Rural ii) Urban
8. Father's Ethnicity..... Mother's Ethnicity.....
9. Father's Age ……. Mother's Age……..
10. Father's age when married……..…… Mother's age when married…….
11. Father's Education (years of Education)

| 0 | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 | 11 | 12 | 13 | 14 | 15 | 16+ |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|

12. Mother's Education (years of Education)

| 0 | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 | 11 | 12 | 13 | 14 | 15 | 16+ |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|

13. What is the total number of your children? ----------
14. i) No of Female……ii) No of Male ……

**Socio Economic Variables**

15. What is the type of family you are living in? i) Nuclear ii) Joint iii) Extended
16. Occupation of the family head i) Self employeed ii) Private job iii) Govt job iv) Laborers
17. What is the monthly income of your family in PKRs----------
18. What are the monthly expenditures of your family in PKRs----------
19. What are the monthly expenditures of medication and treatment of thalassemia ----------

**Background Knowledge of the Genetic Diseases**

20. How many of your children are suffering from thalassemia? ----------
21. Is there any other patient in your family suffering from the same disease? i) Yes ii) No
22. When did you come to know that your child is suffering from thalassemia (symptoms)? ----------

23. Do you know that cousin marriages are one of the main causes for genetic disorders? i) Yes ii) No
24. Do you know that apparently normal children of cousin couple may be carrier of genetic diseases? i) Yes ii) No
25. Have you ever heard about genetic counseling? i) Yes ii) No
26. Please indicate blood group of following members

| Individual | Blood group |
|---|---|
| **Child** | |
| **Mother** | |
| **Father** | |

**Treatment of Beta Thalassemia Major**

27. What was the first step you have taken for the treatment of your child?
    i) Govt. hospital ii) Basic health unit iii) Private hospital / clinic iv) Family welfare center v) Hakeem vi) Local wisdom vii) Religious healing
28. If using blood transfusion, then how many times you managed blood transfusion of your child in a month? ----------
29. How you received blood for your child?
    i)Govt. institute ii) Private iii) with the help of relatives iv). Source of family

**Parental knowledge about thalassemia**

| | Statements | Incorrect | Incorrect |
|---|---|---|---|
| 30 | Thalassemia can be identified through blood test | | |
| 31 | Conditions of general illness i.e., fainting, fever and diarrhea worsen thalassemia major | | |
| 32 | A person suffering from beta thalassemia major, lives normal lives with appropriate treatment | | |
| 33 | Thalassemia can only be treated with medications | | |
| 34 | Marriages within the family are significant risk factor for the propagation thalassemia | | |
| 35 | A person suffering from thalassemia minor lives a normal life | | |
| 36 | If one parent (mother/father) is a carrier of thalassemia minor, the chance of having a child with thalassemia disease increases | | |
| 37 | Thalassemia is preventable disease | | |
| 38 | Thalassemia is an inherited disorder | | |
| 39 | Surgery is an effective way to treat thalassemia | | |
| 40 | Thalassemia passes from parents to children through | | |

| | | | |
|---|---|---|---|
| | genes | | |
| 41 | There is a cure for thalassemia major | | |
| 42 | Thalassemia (any kind) transmits sexually | | |
| 43 | Thalassemia minor increases risk of general illnesses | | |
| 44 | Thalassemia can be detected during pregnancy | | |
| 45 | Parents (both mother & father) having thalassemia minor increases the chances of a thalassemic child | | |
| 46 | After identification of thalassemic, child parents should have family planning | | |

**Practices of Prenatal Diagnosis**

| | **Statements** | **SD** | **D** | **UD** | **A** | **SA** |
|---|---|---|---|---|---|---|
| 47 | Screening before marriage is helpful in identification of thalassemia | | | | | |
| 48 | Pre-natal screening is helpful in identification of thalassemia | | | | | |
| 49 | Post-natal screening is helpful in identification of thalassemia | | | | | |
| 50 | Pregnancy with thalassemia major should be terminated | | | | | |
| 51 | Parental education regarding thalassemia prevention is important | | | | | |
| 52 | Counseling and awareness sessions are important for identification and prevention of thalassemia | | | | | |

**Socio-economic Risk Factors**

| | **Statements** | **SD** | **D** | **UD** | **A** | **SA** |
|---|---|---|---|---|---|---|
| 53 | Low financial condition is the main hurdle for treatment of thalassemia | | | | | |
| 54 | Pre-marriage consultation is the only way to prevent the thalassemia incidence | | | | | |
| 55 | Financial management of the expenditure, for treatment of thalassemia is a problem for parents and families | | | | | |
| 56 | Your child loses very important days of her/his education during the treatment of disease | | | | | |
| 57 | Thalassemia foundation/organization support you for treatment of the disease | | | | | |
| 58 | Your friends pay for your child's treatment | | | | | |
| 59 | Your relatives pay for your treatment | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| 60 | Attitude of doctors is sympathetic during treatment | | | | | |
| 61 | Attitude of nurses and other medical staff is sympathetic during treatment | | | | | |
| 62 | It is difficult to participate in family/community gatherings and events due to the illness of your child | | | | | |

**Cultural Risk Factors**

63. Are you married with your cousin?

1. Yes (In case of yes please fill the below table)
2. No

| **Individual** | **1st** | **2nd** | **3rd** | **Distant relative** | **Baradri** | **No info.** |
|---|---|---|---|---|---|---|
| Couple | | | | | | |
| Husband | | | | | | |
| Parents | | | | | | |
| Grandparents | | | | | | |
| Wife | | | | | | |
| Parents | | | | | | |
| Grandparents | | | | | | |

| | **Statements** | **SD** | **D** | **UD** | **A** | **SA** |
|---|---|---|---|---|---|---|
| 64 | Religious believes play an important role regarding prevention and control of thalassemia | | | | | |
| 65 | Your religion encourages cousin marriage | | | | | |
| 66 | Your religion restrains termination of pregnancy | | | | | |
| 67 | Your religion restrains blood screening | | | | | |
| 68 | You believe in unnatural forces, unseen powers causing disease | | | | | |
| 69 | You feel stigmatization in your society due to your thalassemic child | | | | | |
| 70 | Relationship of a married couple break-up after knowing that they have a thalassemic child | | | | | |
| 71 | Patriarchy plays an important role regarding treatment and screening of thalassemia | | | | | |

**Disease Allied Risk Factors**

| | **Statements** | **SD** | **D** | **UD** | **A** | **SA** |
|---|---|---|---|---|---|---|
| 70 | Treatment cost of your thalassemia is affordable | | | | | |
| 73 | Travelling expenses for treatment of your child are affordable | | | | | |
| 74 | Hospital expenses for treatment of your child are affordable | | | | | |
| 75 | Therapeutic facilities are affordable for the prevention of thalassemia | | | | | |
| 76 | Blood supplies are adequate at the center where your child is transfused | | | | | |

## ANXEURE-C

**Psychosocial Burden of Beta Thalassemia Major**

| | **Aspects of life** | **SD** | **D** | **UD** | **A** | **SA** |
|---|---|---|---|---|---|---|
| 1 | Thalassemia is affecting education of your child | | | | | |
| 2 | There is a significant effect of thalassemia on school timing of your child | | | | | |
| 3 | Your child can participate in sport | | | | | |
| 4 | You usually feel a certain level of anxiety due to the bad health of your child | | | | | |
| 5 | You have weak family interactions due to your thalassemic child | | | | | |
| 6 | You feel social isolation due to your thalassemic child | | | | | |
| 7 | Thalassemia have a significant effect on your social life | | | | | |
| 8 | Your child can have the feelings of difference due his/her disease | | | | | |
| 9 | Your child can have social stigmatization due to thalassemia | | | | | |
| 10 | Your child has a high level social integration | | | | | |
| 11 | Your child is facing bad expression of self-image | | | | | |
| 12 | You have the feelings of denial | | | | | |
| 13 | You feel confusion | | | | | |
| 14 | You have feelings of guilt | | | | | |
| 15 | You have weak social integration | | | | | |
| 16 | Thalassemia effected/effect your family size | | | | | |

## ANXEURE-D

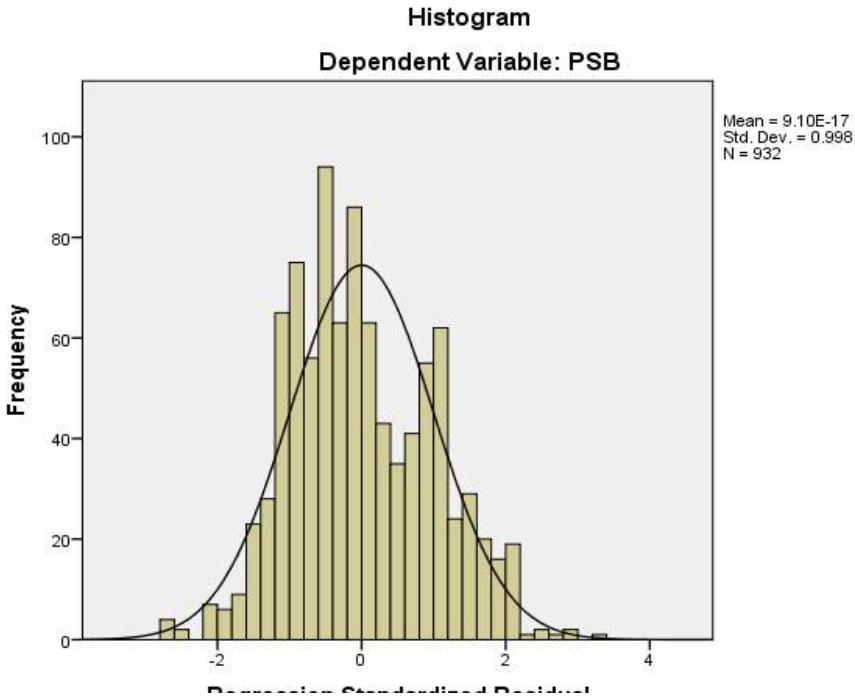
## Reliability Section

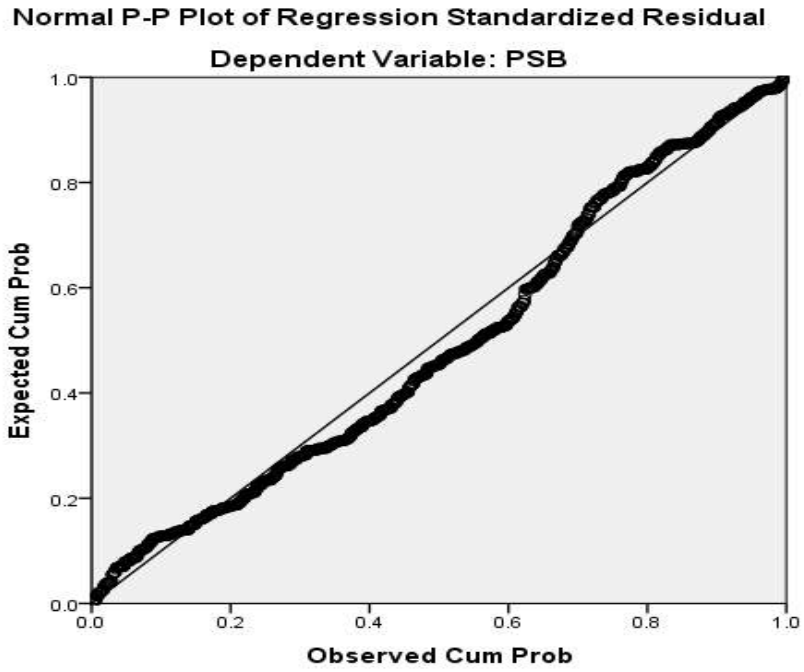
| Variable | Mean | SD | Total Mean | Total SD | Alpha |
|---|---|---|---|---|---|
| KNOWLEDGE1 | 0.8969957 | 0.3041277 | 10.63627 | 2.81924 | 0.6503 |
| KNOWLEDGE2 | 0.7360515 | 0.4410084 | 10.79721 | 2.793016 | 0.6564 |
| KNOWLEDGE3 | 0.4828326 | 0.4999735 | 11.05043 | 2.831204 | 0.6747 |
| KNOWLEDGE4 | 0.7242489 | 0.4471319 | 10.80901 | 2.801336 | 0.6595 |
| KNOWLEDGE5 | 0.6169528 | 0.4863907 | 10.91631 | 2.843099 | 0.6762 |
| KNOWLEDGE6 | 0.7156652 | 0.4513392 | 10.8176 | 2.835821 | 0.6699 |
| KNOWLEDGE7 | 0.6287554 | 0.4833971 | 10.90451 | 2.713395 | 0.6376 |
| KNOWLEDGE8 | 0.6834764 | 0.4653695 | 10.84979 | 2.742624 | 0.6443 |
| KNOWLEDGE9 | 0.6641631 | 0.4725358 | 10.8691 | 2.710133 | 0.6351 |
| KNOWLEDGE10 | 0.2725322 | 0.4455012 | 11.26073 | 2.8567 | 0.6750 |
| KNOWLEDGE11 | 0.7478541 | 0.4344777 | 10.78541 | 2.719046 | 0.6329 |
| KNOWLEDGE12 | 0.2907725 | 0.4543626 | 11.24249 | 2.807692 | 0.6623 |
| KNOWLEDGE13 | 0.8433477 | 0.3636678 | 10.68991 | 2.717526 | 0.6243 |
| KNOWLEDGE14 | 0.8412017 | 0.3656841 | 10.69206 | 2.776807 | 0.6432 |
| KNOWLEDGE15 | 0.8100858 | 0.3924437 | 10.72318 | 2.797225 | 0.6521 |
| KNOWLEDGE16 | 0.8229614 | 0.3819063 | 10.7103 | 2.73651 | 0.6323 |
| KNOWLEDGE17 | 0.7553648 | 0.4301015 | 10.7779 | 2.768362 | 0.6477 |
| Total | | | 11.53326 | 2.924391 | 0.6655 |
| PND1 | 4.22103 | 1.21239 | 20.41309 | 4.31568 | 0.7867 |
| PND2 | 3.92382 | 1.274214 | 20.7103 | 3.730413 | 0.6438 |
| PND3 | 3.92382 | 1.274214 | 20.7103 | 3.730413 | 0.6438 |
| PND4 | 3.92382 | 1.274214 | 20.7103 | 3.730413 | 0.6438 |
| PND5 | 4.197425 | 0.9808444 | 20.4367 | 4.577447 | 0.8079 |
| PND6 | 4.444206 | 1.034371 | 20.18991 | 4.516753 | 0.8025 |
| Total | | | 24.63412 | 4.831004 | 0.7695 |
| SERF1 | 3.868026 | 1.326734 | 32.69957 | 7.021058 | 0.7747 |
| SERF2 | 3.166309 | 1.426021 | 33.40129 | 6.911899 | 0.7700 |
| SERF3 | 3.854077 | 1.265146 | 32.71352 | 7.185326 | 0.7870 |
| SERF4 | 3.972103 | 1.146271 | 32.59549 | 7.277269 | 0.7894 |
| SERF5 | 4.47103 | 0.9317285 | 32.09657 | 7.237961 | 0.7762 |
| SERF6 | 2.439914 | 1.404166 | 34.12768 | 7.10031 | 0.7872 |
| SERF7 | 2.319742 | 1.434093 | 34.24785 | 6.912818 | 0.7706 |
| SERF8 | 4.158798 | 1.354666 | 32.4088 | 6.587692 | 0.7290 |
| SERF9 | 4.158798 | 1.354666 | 32.4088 | 6.587692 | 0.7290 |
| SERF10 | 4.158798 | 1.354666 | 32.4088 | 6.587692 | 0.7290 |
| Total | | | 36.5676 | 7.626678 | 0.7843 |
| CRF2 | 3.819742 | 1.296826 | 21.49893 | 5.397119 | 0.6124 |
| CRF3 | 3.343348 | 1.31509 | 21.97532 | 5.2655 | 0.5864 |
| CRF4 | 3.164163 | 1.433032 | 22.15451 | 5.084437 | 0.5589 |
| CRF5 | 2.667382 | 1.340475 | 22.65129 | 5.139688 | 0.5606 |
| CRF6 | 2.607296 | 1.535219 | 22.71137 | 4.986271 | 0.5490 |
| CRF7 | 3.169528 | 1.295789 | 22.14914 | 5.371511 | 0.6070 |
| CRF8 | 2.890558 | 1.514994 | 22.42811 | 5.158516 | 0.5869 |
| CRF9 | 3.656652 | 1.3882 | 21.66202 | 5.117999 | 0.5612 |

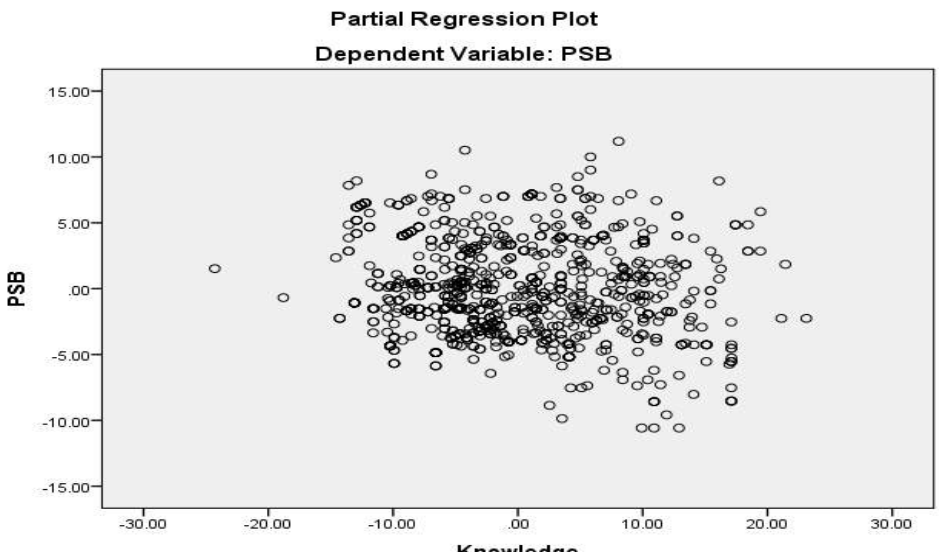
| | | | | | |
|---|---|---|---|---|---|
| Total | | | 25.31867 | 5.775272 | 0.6111 |
| DARF1 | 1.763948 | 1.04366 | 7.290772 | 2.986579 | 0.6211 |
| DARF2 | 1.763948 | 1.04366 | 7.290772 | 2.986579 | 0.6211 |
| DARF3 | 1.763948 | 1.04366 | 7.290772 | 2.986579 | 0.6211 |
| DARF4 | 1.678112 | 0.8352039 | 7.376609 | 3.212959 | 0.6673 |
| Total | | | 26.21247 | 4.663212 | 0.7312 |
| PSBT1 | 3.786481 | 1.253356 | 54.93562 | 9.202703 | 0.8072 |
| PSBT2 | 3.339056 | 1.200992 | 55.38305 | 9.245561 | 0.8080 |
| PSBT3 | 2.084764 | 0.9409548 | 56.63734 | 9.574594 | 0.8193 |
| PSBT4 | 3.991416 | 1.14845 | 54.73069 | 8.771194 | 0.7771 |
| PSBT5 | 3.651288 | 1.356715 | 55.07082 | 8.703329 | 0.7798 |
| PSBT6 | 4.110515 | 0.999257 | 54.61159 | 9.47479 | 0.8153 |
| PSBT7 | 3.651288 | 1.356715 | 55.07082 | 8.703329 | 0.7798 |
| PSBT8 | 3.651288 | 1.356715 | 55.07082 | 8.703329 | 0.7798 |
| PSBT9 | 3.651288 | 1.356715 | 55.07082 | 8.703329 | 0.7798 |
| PSBT10 | 3.061159 | 0.9518561 | 55.66095 | 9.492314 | 0.8152 |
| PSBT11 | 3.991416 | 1.14845 | 54.73069 | 8.771194 | 0.7771 |
| PSBT12 | 3.991416 | 1.14845 | 54.73069 | 8.771194 | 0.7771 |
| PSBT13 | 3.991416 | 1.14845 | 54.73069 | 8.771194 | 0.7771 |
| PSBT14 | 3.991416 | 1.14845 | 54.73069 | 8.771194 | 0.7771 |
| PSBT15 | 3.596566 | 1.382066 | 55.12554 | 9.286021 | 0.8162 |
| PABT16 | 4.181331 | 0.8857114 | 54.54077 | 9.43856 | 0.8108 |
| Total | | | 58.7221 | 9.570195 | 0.8051 |

## ANXEURE-E

## Histogram, P-P Plot and Residual Scattered Plots

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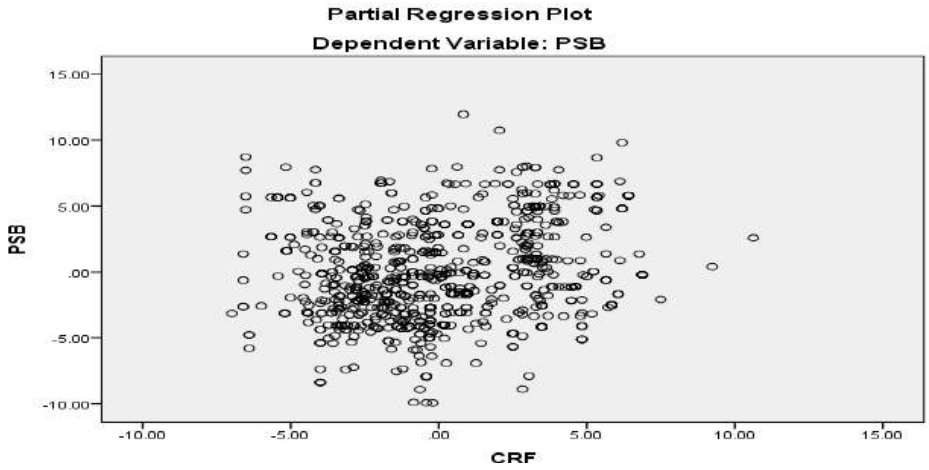
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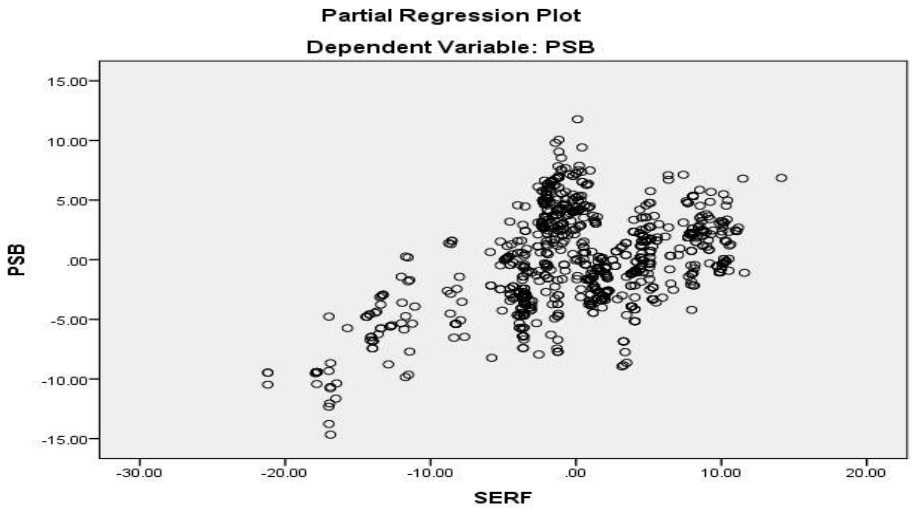
CRF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SERF

## ANXEURE-F

## Thalassemia Prevention Act

**(AS PASSED BY THE NATIONAL ASSEMBLY)**

**A**

**BILL**

*to provide compulsory blood test for relatives of thalassaemia patients.*

**WHEREAS** it is expedient to take concrete steps for controlling the hazardous disease of thalassaemia and to make a law for testing of blood relatives of those persons who have thalassaemia and manifest the disease;

It is hereby enacted as follows:-

**Short title and commencement.-** (1) This Act may be called the Compulsory Blood Test of the Relatives of Thalassaemia Patient Act, 2017.

(2) It extends to the whole of Islamabad Capital Territory.

(3) It shall come into force at once.

2. **Definitions.**- In this Act, unless the context requires otherwise,-

(a) "thalassaemia" means a disease in which a child or an adult becomes anemic because of a genetic defect of hemoglobin;

(b) "thalassaemia major'' means a stage of thalassaemia which clinically manifests itself as severe anemia requiring treatment with repeated blood transfusion and medicines;

(c) "thalassaemia minor or trait'' means thalassaemia which results in mild anemia and is often misdiagnosed as an iron deficiency anemia if appropriate blood tests are not carried out;

(d) "genetic defect'' means abnormalities of genes;

(e) "hemoglobin'' is the protein in red blood cells which is responsible for carrying oxygen;

(f) "Prenatal diagnosis" means test carried out during pregnancy;

(g) "blood relatives'' means directly related aunts and uncles including sisters and brothers of mother and father of the patient, the children of these uncles and aunts and the siblings of the patient;

(h) "chromosomes'' means double helix structures present in the nucleus of the cells, these carry the genes;

(i) "electrophoresis'' means a test used to identify different types of hemoglobin; and

(j) "hemolytic anemia'' means an anemia caused as a result of shorter life span of red blood cells.

3. **Test of thalassaemia mandatory.-** (1) All clinics, hospitals and centers notified by the Government for handling and treating thalassaemia patients shall ensure that blood relatives of children having thalassaemia are all screened for thalassaemia minor or trait or referred to centers having requisite facility for screening. This test is to be made compulsory for siblings, first cousins

0

and uncles and aunts who are blood relatives. In this regard it is particularly compulsory for those blood relatives of thalassaemia patients who are getting married to get a pre-marital blood screening to ensure that they are not carrying the trait. It will be ensured by the designated Hospital to provide counseling to those blood relatives of the Thalassaemia patients who are getting married and it will not be a public document.

(2) Antenatal tests are to be carried out on pregnant women who are known carriers and whose partner is also a carrier for the trait.

(3) All non-governmental organizations (NGOs) running centers dealing with thalassaemia shall ensure that they spend at least 10% of their budget on developing facilities for prenatal diagnosis of thalassaemia.

(4) All clinics, hospitals and centers should counsel relatives of patients on risks of consanguineous marriages and on their chances of having thalassaemic children.

(5) For pre-marital testing both partners are to have their blood indices done if both the partners have blood reports showing anemia their hemoglobin electrophoreses should be undertaken to ensure that they are not carrying the trait.

(6) Diagnosis of the disease is to be carried out through such facilities so as to carry out tests and procedure for diagnosis during pregnancy.

(7) In case a non-governmental organization, clinic or hospital fails to carry out the necessary action they shall face administrative action from the concerned Ministry. An amount of rupees fifty thousand shall be imposed on the non-governmental organization, clinic or hospital who fails to carry out the test.

4. **Reporting of test result.-** (1) The test results are to be reported to those who are tested and if they are carriers (have thalassaemia minor or trait) they are to be given counseling regarding their marrying someone with the same trait and the risk of passing on the disease to their offspring. The test results are to be entered into a data bank for registration of carriers of the trait.

(2) Antenatal test result are to be reported to the women tested and her partner and if the test is positive the parents are to be advised about the condition of the fetus and offered an option of terminating the pregnancy.

## STATEMENT OF OBJECTS AND REASONS

Thalassaemia is a hereditary genetic disorder causing impaired production of hemoglobin. The two types of thalassaemia are-

a) Thalassaemia major usually becomes symptomatic as a severe, progressive hemolytic anemia during the 2nd and 6th month of life. Normally red blood cells spend 100-120 days in circulation and about 1% are removed from the blood each day and are replaced. In thalassaemia the rate of red blood cell destruction increases. As a response there is an increase in bone marrow activity in order to replace the destroyed red blood cells. However production of red blood cells cannot keep pace with their rate of destruction so that repeated blood transfusion is required. Growth is impaired in these children, puberty rarely occurs, cardiac

complication occurs and are the cause of death. In transfusion dependent thalassaemia death usually occurs during the 2nd decade only a few patients survive to their 30's; and

b) Thalassaemia minor is thalassaemia associated with mild anemia. They do not have the symptoms. So that the condition is not diagnosed and the individuals are misdiagnosed as having iron deficiency anemia.

2. Patients with thalassaemia minor usually do not require treatment. Thalassaemia major patients however require treatment which includes chronic repeated blood transfusion to combat anemia caused by increase red cell destruction, medication to reduce the burden of iron accumulated in the patient as result of repeated transfusion, operation to remove spleen which is the site where red blood cells destruction occurs.

3. The prevalence in Pakistan is 3-8%. Pakistan is seeing a large increase in thalassaemic patients due to lack of screening and genetic counseling. Foci of prevalent groups are present in Pakistan where the disease runs in families. Thalassaemia major (the clinically more serious disease) is more prevalent where there are higher rates of inter marriages within relatives. There is growing concern that thalassaemia may become a very serious problem in the next 50 years, one that will burden the blood bank supplies and the health system in general. Each year 5000 children are born with transfusion dependent thalassaemia major. A recent study in thalassaemia (Ahmed et al, 2002) strongly suggests that due to consanguineous marriages thalassaemia genes are trapped within the family. It was discovered that families with a history of thalassaemia have a high carrier rate (30%). In the study thalassaemia was almost nonexistent in families that did not have any known history of thalassaemia in Pakistan. Thalassaemia in fact is confined to 5-10% of families in Pakistan while 90% are free from thalassaemia. As thalassaemia in fact is confined to 5% of families and since most of these families are already within the loop of blood transfusion centers either private or public, the best way therefore is to legislate for thalassaemia. To screen families of indexed patients registered with them, these centers should also provide facilities such as diagnosis during pregnancy for the prevention of thalassaemia. Screening to identify carriers, genetic counseling and diagnosis during pregnancy of beta thalassaemia, this can greatly reduce the rate of birth of affected infants and save a lot of families from the anguish and trauma of dealing with a child with a disease.

4. The child of two thalassaemia minor carrier parents have 25% (that is 1 chance in 2) of inheriting the abnormal gene from each parent and of being (Thalassaemia Major) which is a more serious type of disease and is fatal.

5. Thalassaemia is a dangerous disease which kills a large number of children in Pakistan every year. It is very much needed to take appropriate steps to step further spread of this disease and arrange for proper care of those patients who have fallen victim of this disease.

6. The Bill seeks to achieve the aforesaid objectives.

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