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A Survey of Beta Thalassemia Patients With Various Characteristics Among The Sindhi Families

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Abstract

Thalassemia is one of the major issues in the medical sciences. It is very common genetic disease is globally inherited hemoglobin abnormality. Mutation is only cause of Thalassemia in the alpha globin, hemoglobin A1 and A2 (HBA1/HBA2) and beta globin, Hemoglobin Subunit Beta (HBB) genes are usually inherited in an autosomal recessive manner. Thalassemia has emerged as a major public health concern. Research aim is to identify the prevalence of Beta Thalassemia among Sindhi families along with various connected diseases. This research was governed at the thalassemia health care center, Shaheed Benazirabad. This survey has conducted in the Sindh Province at thalassemia health care center Nawabshah in which 101 families data has been collected from the individuals with different disease are included in the survey. It has been observed that the ratio of beta Thalassemia is more common in male as compared to females. According to the data about 45% male and 65% females, 47% people are affected. During the childhood moderate and severe thalassemia is often diagnosed because symptoms usually appear during the first 2 years of a child's life. It has been concluded that the Thalassemia male patient has the higher frequency of thalassemia-affected as compared to female.

Background History:

Beta thalassemia also called as Cooley's anemia, an American physician Dr. Thomas Cooley first described beta thalassemia in 1925 in Detroit as an inherited blood disease [1]. It is the most common prevalent genetic disease and it affects millions of children throughout the world. Approximate 1.5% (80 to 90 million) people of the world carriers for beta thalassemia, and its increasing with 50 to 60 thousands new cases being born every year [2]. In 2015, 280 million people with thalassemia was reported globally and resulted in 16,800 deaths [3]. It is a genetically hematological disease, caused by over 350 mutations in the beta globin gene [4]. It has estimated about 5-8% are carrier gene frequency and approximately 100,000 people are suffering with beta thalassemia in Pakistan. About 7000-9000, children's were diagnosed every year along with beta thalassemia in Pakistan [5, 6]. Sindh is the one of the province in Pakistan where beta thalassemia cases are prevalent. The aim of our research was to determine the prevalence of Beta thalassemia with various characteristics among the Sindh families.

Introduction

The world's most prevalent hemoglobinopathies is a thalassemia. Thalassemia produced due to cause of gene mutations leading in low level and malfunction respectively alpha and beta globin proteins [7]. Alpha and beta-globin proteins are gene in which form of cluster on chromosome 16, and 11.life comes of different stages in different globin genes. Alpha thalassemia causes of condition hemolytic anemia, or fatal hydrops fetal is depends upon the severity of disease. Beta thalassemia major result of hemolytic anemia growth retardation and skeletal aberration in early childhood [8]. Beta thalassemia is a group of autosomal recessive inheritance disease. They are a heterogeneous grouped of hereditary hemoglobinopathies, it depending on the involvement of globin chain it is classified of beta thalassemia [9]. It is characterized into 3 particular classes: thalassemia major "homozygous disorder resulting in severe transfusion dependent hemolytic anemia "and thalassemia intermediate "severity lies between the minor and major" and thalassemia minor "heterozygous disorders resulting

in mild hypochromic, microcytic hemolytic anemia. Individuals with thalassemia major are typically diagnosed within the two of first years in life and the required frequent blood transfusions on survived [10, 11]. In prevalent regions of gene frequencies for the beta thalassemia ranged in between 2% or 30%. However, the continued or the recent population is migrations also meant that the beta thalassemia could be found by Northern and Western Europe and North America, this disease making by the global health is concerned [12]. The fundamental genetic knowledge of Thalassaemia is included by product hemoglobin structure and location of globin genes, hemoglobin switch, epidemiology, clinical classification of Molecular or cellular pathology and phenotypic or genotypic correlation of genetic modifies [13]. In dominantly inherited, beta-thalassemia of the heterogeneous in molecular level due to mutations of near the (HBB) locus. The resulting variants of beta-chain are very unstable of many cases, or the dominantly inherited on beta-thalassemia is not detectable [14]. The most common genetic disorder in Pakistan is thalassemia, which is a major health care challenge for the country [15]. It is mainly due to a high frequency of hemoglobin in β-subunit of gene mutation, or the high birth rate, and the traditional practices in the consanguineous marriages [16]. Consanguineous marriages are culturally and socially favored and constitute 20–50% of all marriages with first cousins unions accounting for almost one-third of all marriages among populations [17]. It is estimated that currently around one billion of world population has preference for consanguineous marriage [18]. Our research aim is to identify the prevalence of beta Thalassemia in Sindh family with various characteristics is to investigate will focused on recent advanced awareness thalassemia briefly progressive approaches to treatment of this disorder.

Methodology:

In this research, the data was collected from the thalassemia health care center. This research is conducted at the Department of Molecular Biology and Genetics, Shaheed Benazir Bhutto University, Shaheed Benazirabad. A total 101 families were included in this study survey. We have identified the collection of affiliated family's data to get the proper information and we have noted down the required information regarding the disease. After all, our survey was recognized with sampling questions.

Additionally, we have identified thalassemia disease we have gathered the proper information about a child that is affiliated with the disease. Unfortunately, the diagnosed children are not able to meet the proper treatment because they belong to rural areas they don't have resources. Most probably the mild children die with the disease. This relevant data is collected on this disease, by the help of a CBC machine or blood sample. The thalassemia disease were checked and confirmed by technical staff at center with blood sample from patients.

Fig.1: Map of Sindh Province the targeted area of beta thalassemia patients.

In this map (Fig-1), it estimate the average of population about 5-7% worldwide, to contribute many places in which thalassemia affected patients. From 2007 to the 2022 Year-wise data collection in the area of Nawabshah center at the thalassemia health care center, address: Manawar memorial building housing society Nawabshah District Shaheed Benazirabad, Sindh, Pakistan.
Study location:
This location "Thalassemia health care center Nawabshah, addresses Manawar memorial buildings co-operative housing society Nawabshah" district Shaheed Benazirabad. For this thalassemia disease, we will collect the data in this center.

Questionnaire Survey:
By gathering the relevant data from the target parent whose child is affected by the thalassemia disease. During the data collection process, it was required to get the background information of the target parent because it has been seen that many diseases often genetically transfer from one person to another, it might be the result of predecessors. For more inquiries, it was essential to obtain more information from the parents about the causes of the disease. For the addition of more data, we have used the simple format of a questionnaire paper to approach the main causes of the disease.
The questionnaire paper was filled with some suitable questions that were asked by the target parents about their names and background information.
How many children are affected by the thalassemia disease in the family, their age, and their weight?
How many children are normal in the family? The number of children is survived or not survived?
How many children are expiring with this disease?
Consent form: **Assigned the research documentation, we got permission letter for the chairman of the thalassemia health care center.**

Identification of Thalassemia patients:
Affected families were identified with a disease by visiting the thalassemia center Nawabshah in which variances district of Sindh province is included. The form is contained by the details of disease or/and the number of individuals affected and/or diagnosed, or contact number, or postal address of the family. After receiving the information of families were collected by the number of individuals affected. Contacted the desired families or persuaded them to come to their homes to collect blood samples and information. Affected family members were asked to consent to the study and assist in the sample collection. The selected families were sustained credible of blood disorder pieces in widened of thalassemia patient is frequently pattern of autosomal recessive or/and inheritances. These families are Participants were questions for the confirmation of consanguineous to massage (closely related mortgage). However the rarely of family members many outside and consanguineous were common. We were collected the identification of families data.

Collection of blood samples:
The samples was collected on every member of the family include members who were question of regarded the blood disorders, or the many other diseases such as iron deficiency or bone weakness etc. More information was gathered and files of enrolled families were kept in a locker for future investigation.

Results:
It has been determined that males are more likely than females to have beta-thalassemia. A total of 101 families were observed 45% male or 65% females are parentally affected in thalassemia disorder carry genetically. In the 233 children, both boys/girls are affected on the parentally side. In thalassemia, major patients were no more surviving

for a long term, but bone marrow transplantation and blood transfusion techniques are valid in treatment but maybe a chance genetically affected patients are survive in life. It has also observed but there was no association with age was seen. Because symptoms normally occur during the first two years of a child's life, moderate and severe thalassemia is frequently identified in childhood.

Fig-2: Beta Thalassaemia children patients.

Thalassaemia Children's affected the blood disorder (Fig-2) and the twins children are affected the Thalassaemia and the one family are more than 2 or 3 children's affected in thalassemia.

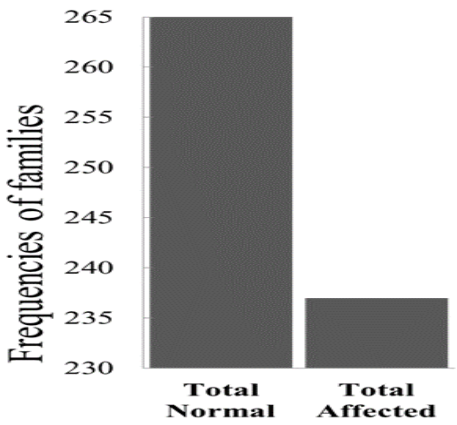

Fig-3. Total number of normal and affected frequencies of families.

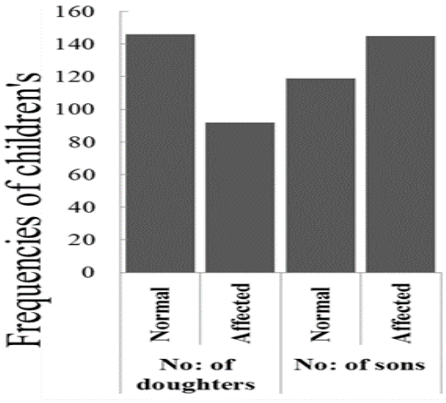

Fig-4. The normal and affected boy or girl frequencies of thalassemia children's.

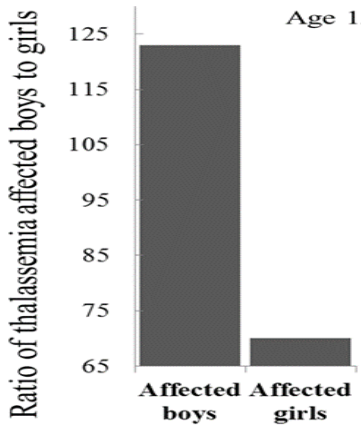

Fig-5. The age wise 1/10 ratio of thalassemia affected boys or girls.

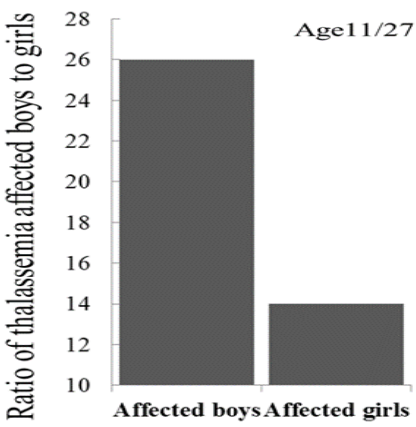

Fig-6. The age wise 11/27 ratio of thalassemia affected boys or girls.

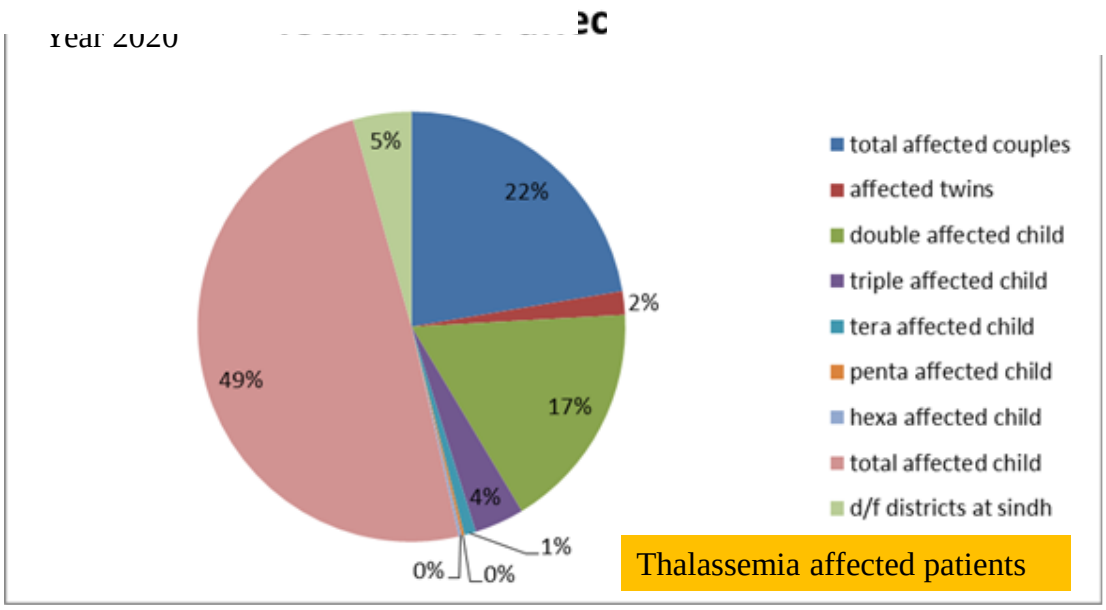

Fig-7: The affected children's including twins children's and also including the families data from different areas at district Shaheed Benazirabad, Sindh, Pakistan.
Data of 101 families was collected (Fig-7), In this graph ratio of affected boys to girls through the thalassemia disease in which variance affected traits of the age wise 11/27 ratio of thalassemia affected boys or girls.

Discussions:
Our study of thalassemia is a genetic blood disorder transferred from parent to offspring. Collected the data of thalassemia-affected child's through the treatment of blood transfusion and also with a daily route in CBC (complete blood count) machine test. Surveyed at the thalassemia health care center in Nawabshah, Sindh, and also will discuss the thalassemia-affected parents with their children. All participants asked the questions about their family members with hematological indices were questions about their medical history In total, 101 families are selectively affected by a genetic blood disorder, in case patients are inherited affected in parental. Approximately 233 children both boys/and girls are affected by the blood disorder thalassemia, major cause of thoroughly parents into offspring. People with thalassemia have fewer healthy hemoglobin proteins made by their bone marrow, as well as fewer healthy red blood cells. Because thalassemia is inherited, the condition sometimes runs in families. Some people are finding out about their thalassemia because they have relatives with a similar

condition. People who have family members have certain parts of the worldwide range have a higher risk of thalassemia.

The majority of the information included in this research is to identify the prevalence of beta Thalassemia in the Sindh family with various characteristics will focus on recently advanced awareness and thalassemia about briefly progressive approaches to the treatment of this disorder. In the results, we were observed that thalassemia carried 65% of females are maternally affected by thalassemia disorder carry genetically. Figure no, 3 and 4 show the frequency of families including affected and normal children it clearly defines thalassemia's impact on the generation sometimes affected traits transferred from parents to their offspring, and fig no, 4 shows the frequencies of children in which the number of daughters affected the frequency and the number of normal frequency are compared it also included the sons and daughters normal to affected. It has been estimated total of 233 affected children and the different ages grouped from 1/10 to 11/27, graph those shown such as figures no, 5 and 6 the ratio of thalassemia affected sons is 63% and affected daughters is 36%. We will also include the current data mentioned in the pie chart as described below.

Our findings were Similar of majority studies is thalassemia by affected children and all of that variance characteristics transferred from parents to their offspring, but other studies are reported variable results in variations disease and the prevalence of various regions in the world or the non-standardized methods of transfusion for some centers due to lack in facilities [19, 20, 21 and 22].

Limitation: we were conduct the information and data collection in thalassemia disease affected patients; this is a genetics blood disorder in which disease transferred parents to their offspring.

Delimitation: Three students were used in the sample. This study's main purpose is to identify and survey at the thalassemia center Nawabshah, Sindh. All participants were asked the questions and their family members with hematological indices were questioned about their medical histories such as how many family members, how many children, in which children are affected in thalassemia disease, family postal address, and contact number. Each of the students is given the same questionnaire or answer.

Conclusion:

The genetic inheritance condition is affecting the blood disorder of Thalassaemia. Thalassemia is different in types Depending on the type you have, Thalassaemia in caused no illness at all and maybe any seriously lifelong condition required by treatments. The estimates are 6,000 to 8,000 thalassemia major children by born in Pakistan every year. The estimated carries are 5-6%, with 9.8 million carriers in the total pollution. As a rough estimate, at any given time, more than 150,000 thalassemia major children may be found in our society and country are getting incomplete treatment by infrequently blood transfusion and inadequate iron chelation therapy. It has been concluded that in 101 families/couples there is a total of 233 children are affected included 45% males or 56% females are affected. The frequency of thalassemia-affected children is observed among every boy/girl who is genetically affected.

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