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An empirical investigation of the relationship between consanguineous marriage and prevalence of β -thalassemia in Punjab, Pakistan: A cross-sectional study

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Abstract--It is a striking fact that β -thalassemia has become a common single gene disorder among the children. Among the contributing factors, overwhelmed practice of consanguineous marriage is one of the most prominent reason of the high rate prevalence of β -thalassemia in Pakistan. Therefore, the present research was conducted to investigate the relationship between consanguineous marriage and prevalence of β -thalassemia in Punjab, Pakistan. The nature of study was cross-sectional and quantitative. The study was conducted in the Children's Hospital, Lahore and Multan. Purposive sampling technique was used and sample of 300 parents was drawn. A well-structured research questionnaire was used to collect the data from the parents. The results indicate that rate of β -Thalassemia is significantly higher (76.7%) in first cousin marriage as compared to the second cousin marriage (23.3%). The

trend of thalassemia major among male patients is higher (76.31%) in first cousin marriage as compared to the second cousin marriage (23.69%). Similarly, the prevalence of thalassemia Intermedia is significantly prevalent higher (67.5%) among male patients in first cousin marriage as compared to the second cousin marriage. The above findings can be concluded that there was a strong relationship between consanguineous marriage and prevalence of β -thalassemia in Punjab, Pakistan. In the light of results, it can be recommended that premarital screening and counselling of the couples are highly essential to minimize the incidence of β -thalassemia in Punjab, Pakistan.

Keywords-- β -thalassemia, consanguinity, blood disorder, premarital screening.

Introduction

Among the single gene disorders, β -thalassemia is the most common prevalent disease in Pakistan (Ishfaq & Hashmi, 2015). It is an alarming that rate of β -thalassemia is increasing day by day (Akter, Khatun, & Hossain, 2020). Previous research studies also indicate that there is a close relationship between the consanguineous marriage and the prevalence of β -thalassemia (Aziz, Sadaf & Kanwal, 2012). Speaking statistically, the rate of β -thalassemia in Pakistan is ranged between 5-7% in Pakistan. At present, more than 10 million are carriers and 5000 children are diagnosed with β -thalassemia every year (WHO, 2012).

It is an inherent genetic disorder transmit from one generation to coming generation (Hussain et al., 2021). The bitter fact is that practice of marriage in a similar family with homogenous characteristics is also a contributing factor in β -thalassemia (Baig et al., 2005). There are three common categories of the β -thalassemia have been characterized as under:

- Asymptomatic β -thalassemia (Carrier)
- β -thalassemia intermedia (Moderate anemia requiring intermittent transfusions)
- β -thalassemia major (Severe anemia requiring lifelong transfusions and iron chelation therapy)

Absent production or decreased production of hemoglobin chains triggered by a marriage in a single group referred as thalassemia (Wetherall & Clegg, 2001). Alpha and Beta are characterized depending upon the production of Alpha and Beta Chains.

- Alpha Thalassemia- Decreased/Absent alpha globin chains (due to defective alpha genes)
- β -Thalassemia- Decreased/Absent production of β -globin chains (due to mutations or change in β -genes)

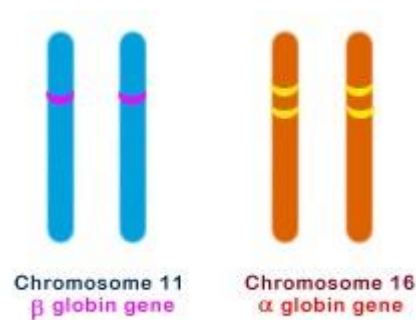


Figure 1

It is an established fact that 2 carriers get marriage, likely to have a chance of 25% of having child with thalassemia major, 25% of a chance of having a normal child and 50% of a chance of having a carrier child in each pregnancy (Shami & Tariq, 1999). Person with thalassemia traits should marry a non-carrier person then there is a chance of 50% of having a normal child, 50% of having a carrier child and zero chance of Thalassemia major during each pregnancy (Asadi & Doroudchi, 2004).

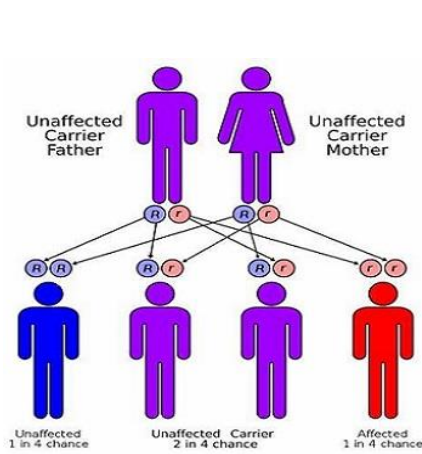


Figure 1

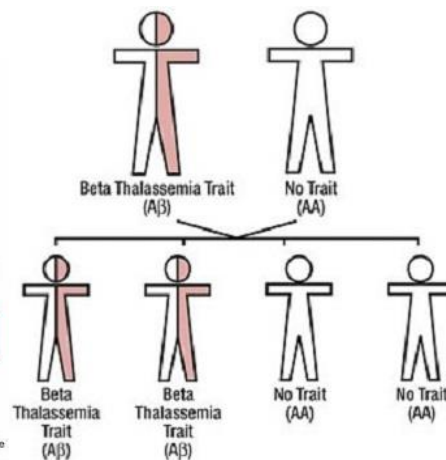


Figure 2

Objective of the research study

The objective of the research study is to investigate the relationship between Consanguineous Marriage and Prevalence of β -Thalassemia in Punjab, Pakistan.

Materials and Methods

The nature of the research study is quantitative and cross section. The present research was conducted in the Children's Hospital Lahore and Multan. The purposive sampling technique was employed to draw the sample from these two settings. The inclusion criteria was as under

- The Married Couples have cousin marriage

- The Married Couples have a child with thalassemia and require blood transfusion support
- The Child with thalassemia should be registered Children's Hospital Lahore and Multan

The target population of the present research was parents of children with thalassemia. A sample of 300 parents was drawn and equitable representation has been given to the Children's Hospital Lahore and Multan. A well-structured research questionnaire was used to collect the data from the parents having child of thalassemia. The consanguinity marriage was operationalized into two categories (i) First Cousin, (ii) Second Cousin. The descriptive analysis i.e. percentage, Z-test and Standard errors of mean was employed to investigate the relationship between consanguinity marriage and prevalence of β -thalassemia in Punjab, Pakistan.

Results and Discussions

Table 1
Percentage of Male and Female with Sex Ratio by β -Thalassemia Major (TM) and Thalassemia Intermedia in Punjab

β -Thalassemia Type	Male Patients (%)	Female Patients (%)	Z-Test	p-Value	Sex Ratio
TM	155 (86.1%)	90 (75%)	7.5	<0.001	172.22
TI	25 (13.9%)	30 (25%)	2.31	0.0061	83.33
Total	180 (60%)	120 (40%)	7.39	<0.001	150

The above table shows that male are highly affected with β -thalassemia major as compared with female children (60% vs 40%). And, there was a strong relationship between the male children and thalassemia major (p-Value <0.001).

Table 2
Percentage Distribution of β -Thalassemia (TM & TI) and Birth Order of Patients

β -Thalassemia Type	Birth Order					
	1	2	3	4	5	6
TM	132 (94.3%)	92 (93.9%)	70 (93.3%)	40 (90.9%)	36 (100%)	16 (84.2%)
TI	8 (5.7%)	6 (6.1%)	5 (6.7%)	4 (9.1%)	-	3 (15.8%)
Total	140 (34%)	98 (23.8%)	75 (18.2%)	44 (10.7%)	36 (8.7%)	19 (4.6%)

The above table presents that rate of β -thalassemia was greater at 1st birth order (34%) and lower at 6th birth order (4.6%). Similarly, the patients of thalassemia major were also highest at 1st birth order (94.3%) and lowest at 6th birth order. On the other hand, patients with thalassemia intermedia were highest at 6th birth order (15.8%) and lowest at 1st birth order (5.7%).

Table 3
Percentage Distribution of β -Thalassemia Patients by Consanguinity Marriage

β -Thalassemia Type	Gender	Consanguinity Marriage		Total
		First Cousin	Second Cousin	
Thalassemia Major	Male	145 (76.31%)	45 (23.69%)	190
	Female	55 (84.6%)	10 (15.4%)	65
Thalassemia Intermedia	Male	25 (67.6%)	12 (32.4%)	37
	Female	5 (62.5%)	3 (37.5%)	8
Grand Total		230 (76.7%)	70 (23.3%)	300

The above table shows the percentage distribution of β -Thalassemia patients by consanguinity marriage. The rate of β -Thalassemia is significantly higher (76.7%) in first cousin marriage as compared to the second cousin marriage (23.3%). The trend of thalassemia major among male patients is higher (76.31%) in first cousin marriage as compared to the second cousin marriage (23.69%). Similarly, the prevalence of thalassemia Intermedia is significantly prevalent higher (67.5%) among male patients in first cousin marriage as compared to the second cousin marriage. The above results indicate that prevalence of β -Thalassemia is highly significant in first cousin marriage.

Conclusion

The above discussions can be concluded that there was a strong relationship between the consanguinity and prevalence of β -Thalassemia in children. The trend of prevailing β -Thalassemia in the first cousin marriage is highly alarming. The above results also show that prevalence of thalassemia major (TM) is significantly higher among male patients. It is the need of the hour to take revolutionary measures to curb the increasing prevalence of β -Thalassemia. It is also recommended that pre-marital screening and counselling can prove very instrumental to minimize the prevalence of β -Thalassemia in Punjab, Pakistan.

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