CODEN [USA]: IAJPBB ISSN: 2349-7750

INDO AMERICAN JOURNAL OF
PHARMACEUTICAL SCIENCES

http://doi.org/10.5281/zenodo.2229177

Available online at: http://www.iajps.com ***Research Article***

A CROSS-SECTIONAL RESEARCH TO DEVISE PREVENTION STRATEGIES FOR B-THALASSEMIA AMONG NEWBORNS ESPECIALLY THROUGH RELIABLE THALASSEMIA SCREENING

[1]Dr Tayyba Ashraf, [1]Dr Jazab Ali, [2]Dr Naghza Tul Zahra
[1]Govt. General Hospital Ghulam Muhammadabad Faisalabad
[2]Allied Hospital Faisalabad

Abstract

Objectives: *The aim of the research was to design a technique for β-thalassemia prevention in newborns with the help of indexed families reliable screening process.*

Methods: *The researcher completed this cross-sectional research in the timespan of six months at Sir Ganga Ram Hospital, Lahore (February to August 2017). The researcher evaluated various ethnic group children's blood samples and screened them for β-thalassemia at the hospital laboratory. The process of human haemoglobin electrophoretic separation such as (A, F, S & C) was completed. Haemoglobin in the gel was restrained in the solution and a film was made after drying out the gel from the fixative solution. The pattern of the haemoglobin visually appeared after the straining through a specific strain of the protein and this pattern was also enumerated through densitometry.*

Results: *In the total ninety-eight samples 57 samples had β-thalassemia trait (58.2%) with an increased level of haemoglobin alpha-II and 41 had a normal level of haemoglobin alpha-II the (41.8%). Total carriers were fifty-seven with 33 males (57.89%) and 24 females (42.10%). Carried had a mean age of (11.65 ± 6.25) years; whereas, the mean age of the normal patients was (10.93 ± 7.75). Level of mean haemoglobin alpha-II in carriers and normal patients was respectively (5.2% ± 0.56%) and (2.34% ± 0.57%).*

Conclusion: *There is an emerging need for the screening of thalassemia detection programmes throughout the country with the provision of marriage counselling benefits and merits to the newly married couples. This awareness and educational programmes can be beneficial for major thalassemia reduction.*

Keywords: *Newborns, Thalassemia, Straining, Marriage, Carriers and Indexed Families.*

Corresponding author:
Dr. Tayyba Ashraf,
Govt. General Hospital Ghulam Muhammadabad, Faisalabad.

QR code

Please cite this article in press Tayyba Ashraf et al., ***A Cross-Sectional Research To Devise Prevention Strategies For B-Thalassemia Among Newborns Especially Through Reliable Thalassemia Screening.,*** *Indo Am. J. P. Sci, 2018; 05(12).*

INTRODUCTION:

Thalassemia refers to primary single-gene monogenetic disorders all over the globe that is studied with the help of molecular biology techniques for the first time [1, 2]. It is a derivation of two Greek words namely "thalas" and "emia" which respectively means sea and blood [3, 4]. The name was developed because of an anemia type on the Mediterranean Sea bordering countries. Thalassemia was never taken as a clinical entity till 1925 before the description of a syndrome that has an association with bony deformities and splenomegaly [4]. There is an approximation of 240 million β-thalassemia heterozygous affected people with 200,000 annual affected people [1]. These people mainly relate to various regions which are also malaria endemic including the Middle East, Mediterranean and also include some of the parts of Southern China, Africa, Southeast Asia and India [5]. Highest β-thalassemia prevalence is found in Southeast Asia and Indian subcontinent as this belt is also known as thalassemia belt [1, 2].

Pakistan also faces the common genetic disorder of β-thalassemia [6]. In the overall population of Pakistan, the frequency of β-thalassemia is above five percent with an estimation of 9 million β-thalassemia carriers spread all over the country [7]. More than 40,000 children dependent on the transfusion are registered per year and the about five thousand children born with the major thalassemia all over the country every year.

In the same way as other countries suffer increasing healthcare burden because of the β-thalassemia especially in the countries of Asia, Pakistan is also no exception to that and it also faces lots of financial burden on the healthcare department. The provision of iron chelation therapy and blood transfusion is next to impossible for affected children with meagre national resources available at hand. Pakistani patients are not capable to bear the financial burden of the bone marrow transplantation as it expensive and scarcely available at few specialized centres. Most effective means are preventive measures taken against β-thalassemia to avoid it for the better and healthy life of the children and families. Proper marriage counselling can bar the marriage between affected couples or even single male of a female affected partner in order to prevent the occurrence of thalassemia. Such restrictions are to made compulsory for the marrying couples and mandatory screening process should be given legal shelter to control the birth of affected children. Which will undoubtedly control the overall disease burden. All positive cases are to be given proper marriage counselling and they are to be convinced for better available options. Few other research studies are also available on the topic of β-thalassemia mutations among different ethnic groups and regions all over Pakistan; whereas, this particular research aims to design a technique for β-thalassemia prevention in newborns with the help of indexed families reliable screening process.

SUBJECTS AND METHODS:

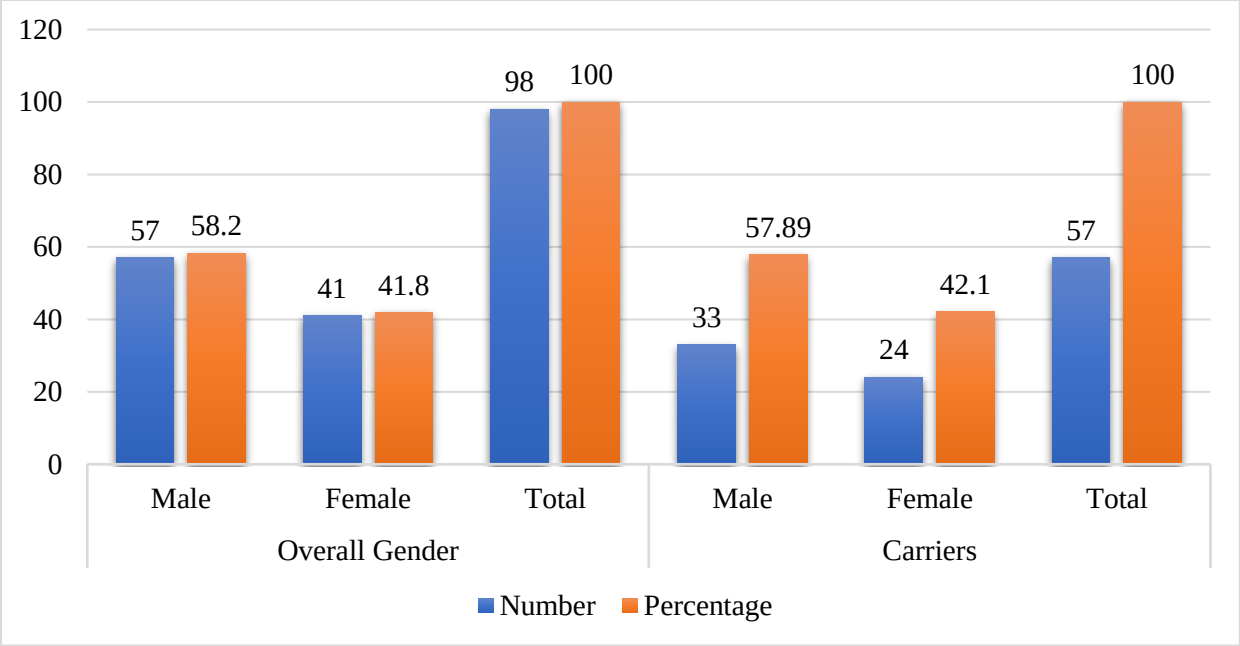
The researcher completed this cross-sectional research in the timespan of six months at Sir Ganga Ram Hospital, Lahore (February to August 2017). The researcher evaluated various ethnic group children's blood samples and screened them for β-thalassemia at the hospital laboratory. The process of human haemoglobin electrophoretic separation such as (A, F, S & C) was completed. Haemoglobin in the gel was restrained in the solution and a film was made after drying out the gel from the fixative solution. The pattern of the haemoglobin visually appeared after the straining through a specific strain of the protein and this pattern was also enumerated through densitometry. Sample collection was made in EDTA tubes from both unmarried males and females. Every patient gave informed consent also completed a questionnaire specially designed for the purpose of this research. The choice specimen for regular Hb investigation is hemolysate (red blood cell) which is the basic reason of using EDTA tube for blood sample collection, heparin anticoagulants or sodium oxalate. Data analysis was done on SPSS software and level of Hb alpha-II with age was showed in Mean and SD values.

RESULTS:

In the total ninety-eight samples 57 samples had β-thalassemia trait (58.2%) with an increased level of haemoglobin alpha-II and 41 had a normal level of haemoglobin alpha-II the (41.8%). Total carriers were fifty-seven with 33 males (57.89%) and 24 females (42.10%). Carried had a mean age of (11.65 ± 6.25) years; whereas, the mean age of the normal patients was (10.93 ± 7.75). Level of mean haemoglobin alpha-II in carriers and normal patients was respectively (5.2% ± 0.56%) and (2.34% ± 0.57%). The overall gender distribution included the male, female and total number as 57, 41 and 98 with respective proportions of 58.20%, 41.80% and 100%. Whereas, in the carriers, the males and females were 33 and 24 with respective percentages of 57.89% and 42.10% (Table – I).

Table – I: Gender Distribution in Overall Research Sample and Carriers

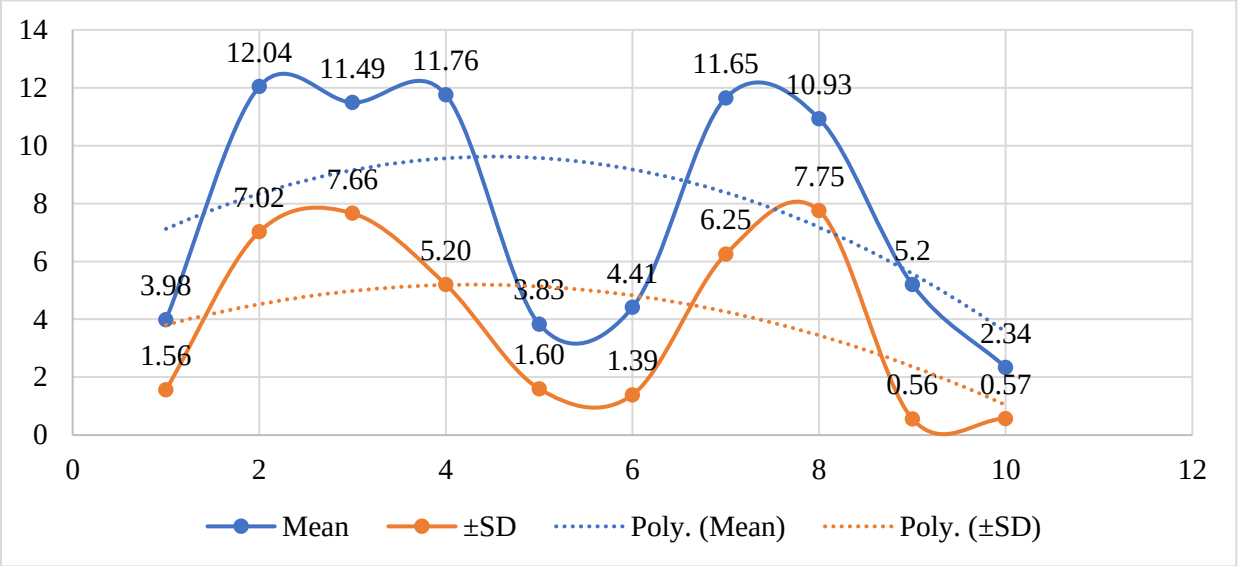
Gender		Number	Percentage
Overall Gender	Male	57	58.20
	Female	41	41.80
	Total	98	100
Carriers	Male	33	57.89
	Female	24	42.10
	Total	57	100



The mean and SD values of mean age (males), mean age (females), patients with consanguineous parents, marriage with distant relatives, consanguineous marriage HbA2, distant relatives HbA2, carriers mean age, normal patients mean age, HbA2% of carriers and HbA2% of normal subjects is given in Table – II.

Table – II: Mean and SD Values of Various Variables

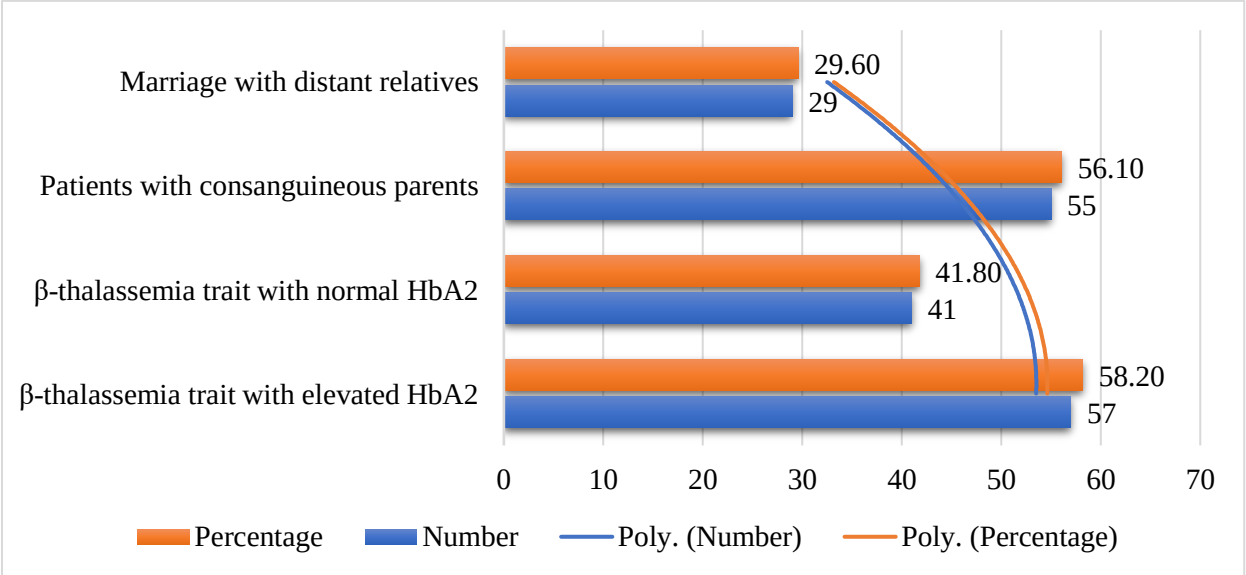
Mean Values	Mean	±SD
Mean Age (Males)	3.98	1.56
Mean Age (Females)	12.04	7.02
Patients with Consanguineous Parents	11.49	7.66
Marriage with Distant Relatives	11.76	5.20
Consanguineous Marriage HbA2	3.83	1.60
Distant Relatives HbA2	4.41	1.39
Carriers Mean Age	11.65	6.25
Normal Patients Mean Age	10.93	7.75
HbA2% of Carriers	5.2	0.56
HbA2% of Normal Subjects	2.34	0.57



The number and percentage of β-thalassemia trait with elevated HbA2, β-thalassemia trait with normal HbA2, patients with consanguineous parents and marriage with distant relatives is given in Table – III.

Table – III: β-thalassemia Trait with Elevated and Normal HbA2

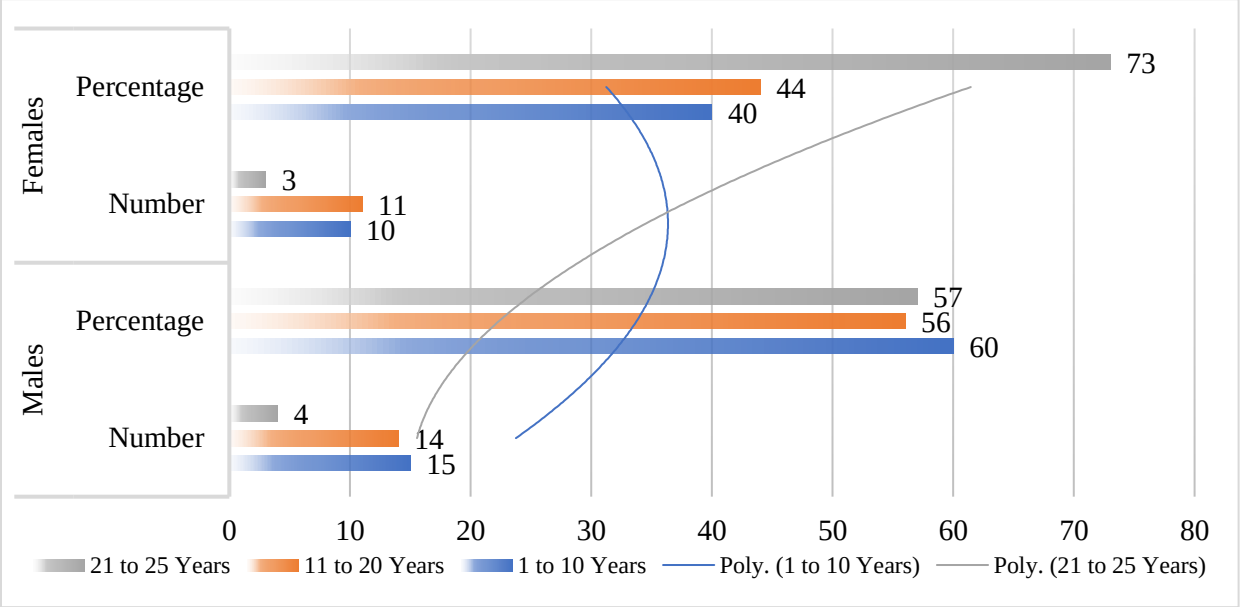
Details	Number	Percentage
β-thalassemia trait with elevated HbA2	57	58.20
β-thalassemia trait with normal HbA2	41	41.80
Patients with consanguineous parents	55	56.10
Marriage with distant relatives	29	29.60



The carriers respective age of 1 to 10 years, 11 to 20 years and 21 to 25 years stratification in males and females with respect to number and percentage is given as under (Table – IV).

Table – IV: Carriers Age Distribution with Respect to Gender

Carriers Age	Males		Females		Total
	Number	Percentage	Number	Percentage	
1 to 10 Years	15	60	10	40	25
11 to 20 Years	14	56	11	44	25
21 to 25 Years	4	57	3	73	7



DISCUSSION:

An overall beta-thalassemia trait occurrence in the indexed families was about (58.2%) with an increased level of HbA2. Moreover, it is important to note that the frequency of carrier among males was more than females with respective proportions of 57.89% and 42.10%. The conclusion clearly quotes that thalassemia carrier status is higher among the families with major thalassemia.

There is a proper and necessary implementation of the screening of the unmarried and young women for carrier detection in order to control and restrict the increasing major thalassemia incidence among infant children all over the world [8]. For instance, in Italy, Cyprus, UK and USA and also among a few other regions of Africa and Europe, proper measures implementation about thalassemia has significantly reduced the disease prevalence markedly. The example of Cyprus is very much notable as there is a marked decrease in disease incidence by 96%. In Iran, the carrier identification through premarital screening in unmarried males and females has significantly reduced the disease incidence seventy percent of the annual birth rates with a significant decrease in the medical expenses and healthcare burden [9, 10].

In Pakistan, the practice of following these precautionary measures are still not so far which needs a real work with a research strategy, persuasion, educational programmes for family members and parents for proper thalassemia screening before the change of marital status. There is a need to make these screening facilities easily available with an affordable price for the people of all walks of life.

CONCLUSION:

The counselling of guided marriage is important it is also supported by an authentic process of screening with a molecular genetic diagnostic for the thalassemia preventive measures. There is an

emerging need for the screening of thalassemia detection programmes throughout the country with the provision of marriage counselling benefits and merits to the newly married couples. This awareness and educational programmes can be beneficial for major thalassemia reduction.

REFERENCES:

1. Hafeez M, Aslam M, Ali A, Rashid Y, Jafri H. Regional and ethnic distribution of Beta thalassemia mutations and effect of consanguinity in patients referred for prenatal diagnosis. J Coll Physicians Surg Pak 2007; 17: 144-7.
2. Zlotogora J. Population programs for the detection of couples at risk for severe monogenic genetic diseases. Hum Genet 2009; 126:247-53.
3. Christianson A, Streetly A, Darr A. Lessons from thalassemia screening in Iran. BMJ 2004; 329: 1115-7.
4. Karimi M, Jamalian N, Yarmohammadi H, Askarnejad A, Afrasiabi A, Hashemi A. Premarital screening for beta-thalassemia in Southern Iran: options for improving the programme. J Med Screen 2007; 14: 62-6.
5. Weatherall DJ, Clegg JB. The biology of the thalassemia. The thalassemia syndromes. 4th ed. Oxford: Blackwell Scientific Publication; 2001, 121-284.
6. Weatherall DJ. Molecular biology at the bedside. Br Med J 1986;292: 1505-8.
7. Weatherall DJ. Phenotype-genotype relationships in monogenic disease. Nat Rev Genet 2001; 2: 245-55.
8. Colah R, Nadkarni A, Gorakshakar A, Phanasgaonkar S, Surve R, Subramaniam PG, et al. Impact of beta-globin gene mutations on the clinical phenotype of beta-thalassemia in India. Blood Cells Mol 2004; 33: 153-7.
9. Cao A, Saba L, Galanello R, Rosatelli M. Molecular diagnosis and carrier screening for B-thalassemia. J Am Med Assoc 2007; 278:1273-7.
10. Weatherall DJ, Clegg JB. The β-thalassemia syndromes. 4th ed. Oxford: Blackwell Scientific Publication; 2001, 635-87.