

Perception of Parents of Children suffering from Thalassemia in Karimnagar District of Telangana state in India

**Dr. Atul Gopal Agrawal¹, Dr. Noori Afifa², Dr. Gantla Akhil³,
Snigdha Mishra⁴, Dr. Ankita Agrawal⁵**

¹Professor and Head, Department of Community Medicine, Prathima Institute of Medical Sciences,
Karimnagar, Telangana

²Final Year Community Medicine Post Graduate Student, Prathima Institute of Medical Sciences,
Karimnagar, Telangana

³Second Year Community Medicine Post Graduate Student, Prathima Institute of Medical Sciences,
Karimnagar, Telangana

⁴MBBS Final Year Part 1 Student, Prathima Institute of Medical Sciences, Karimnagar, Telangana

⁵Physiotherapist, Prathima Institute of Medical Sciences, Karimnagar, Telangana

Abstract

Introduction : Thalassemias are a group of inheritable hemoglobinopathies where abnormal hemoglobin is synthesized leading to decreased hemoglobin levels in the body. Thalassemias are classified according to which chain of the hemoglobin molecule is affected. In α -thalassemias, production of the α -globins chain is affected, while in β -thalassemia, production of the β -globins chain is affected. Thalassemias are a grave menace in the modern world which need to be tackled effectively and quickly to improve the health status of people around the world.

Objectives: To assess knowledge and awareness of parents of thalassemia patients regarding preventive measures and treatment options available for thalassemia.

Methodology: The present study is an observational study design conducted in a district called Karimnagar of Telangana state in India amongst 130 parents of children suffering from thalassemia with the use of a pre-designed direct questionnaire with additional variables.

Results : Out of the 130 parents, 68% knew that thalassemia was a hematological disorder, 57 % parents understood the role of heredity in the transmission of the disease, 54% parents were aware that it could be prevented by pre-natal genetic counseling, 73% parents understood that consanguineous marriage was a risk factor contributing to the disorder, 84% parents were made aware of thalassemia only through their healthcare personnel and nearly 64% parents had an idea about various modalities of treatment of the disorder.

Conclusion: This study provides an important insight into the level of understanding regarding thalassemia disorders amongst parents and the amount of awareness still required to make a difference in improving the statistics surrounding the same.

Introduction

Thalassemias are a group of inherited hematological disorders following autosomal recessive pattern of inheritance. The genetic defect causes a reduction in the rate of globin chain synthesis which causes the formation of abnormal hemoglobin molecules. The resultant microcytic anemia is the characteristic presenting symptom of the thalassemias. These disorders are of two types: alpha (α) thalassemia and beta (β) thalassemia. The various combinations of hemoglobin polypeptide chains present during first trimester of intrauterine life include Hb Gower 1, Hb Gower 2, Hb Portland and HbF. Thalassemias are prominently prevalent in South African and Asian ethnicities. There is a 25% risk for each pregnancy resulting in a diseased child if both parents are carriers of the thalassemia. In 2016, thalassemia was reported in 270 million people worldwide and resulted in 16,700 deaths. Strategies for prevention of thalassemias include providing up-to-date and accurate information to healthcare professionals and screening centre with appropriate counseling. However, since India is a developing country with a large population and almost 4.5 million β -thalassemia carriers - spreading awareness about thalassemias is the need of the hour if there has to be a significant positive change in the statistics for the same.

Material and Methods

It is an observational study design conducted in a district called Karimnagar of Telangana state in India to estimate the knowledge about thalassemia in families whose members are suffering from thalassemia, and to assess the awareness about prevention and treatment in the affected families. The study population consisted of 130 parents of thalassemia patients who visited the thalassemia unit of Prathima Institute of Medical Sciences for their treatment in a span of 2 months (October 1st, 2022 - November 30th, 2022). The samples were selected through non-probability convenient sampling. Thalassemia patients were diagnosed by appropriate diagnostic methods like hemoglobin electrophoresis with quantification of fetal hemoglobin (Hb F). The aim of the study was explained to all volunteer participants and written informed consent was taken from them in their language of understanding. Questionnaires were distributed to parents under proper moderation and guidance of the researcher.

Results:

For the purpose of reference:

Out of the 130 participants in the study, 69 parents (53%) were illiterate, 82 parents (63%) had consanguineous marriages and 87 parents (67%) belonged to the lower-middle class socioeconomic group (according to Kuppuswamy scale)

Table 1: Participants' perception regarding the system of the body involved in Thalassemia

	Perception of System Affected	Frequency (n)	Percentage (%)
1	Digestive system	21	16
2	Skeletal system	9	7
3	Haematological system	88	68
4	No idea	12	9
	TOTAL	130	100

As shown in Table 1, 88 participants (68%) knew that thalassemia is a hematological disorder

Table 2: Participants' perception regarding the mode of transmission of thalassemia

	Perception of mode of transmission	Frequency (n)	Percentage (%)
1	By sexual contact	20	15
2	By blood	23	18
3	By digestive system	6	5
4	By heredity	74	57
5	No idea	7	5
	TOTAL	130	100

As shown in Table 2, 74 participants (57%) knew that thalassemia is a genetic disease.

Table 3: Participants' awareness regarding the prevention methods that can be adopted to reduce the prevalence of thalassemia

	Perception of Preventive Methods	Frequency (n)	Percentage (%)
1	Only public awareness	31	16
2	Preventing the birth of children amongst two thalassemia carriers	17	13
3	Prenatal Diagnosis	70	54
4	Postnatal Diagnosis	12	9
	TOTAL	130	100

As shown in Table 3, 70 parents (54%) were aware about the need for prenatal diagnosis in people with thalassemia.

Table 4: Participants' awareness regarding the risk factors that may cause thalassemia

	Perceived Risk Factor	Frequency (n)	Percentage (%)
1	Smoking	10	8
2	Alcohol Consumption	10	8
3	Consanguinity	95	73
4	Vitamin Deficiencies	15	12
	TOTAL	130	100

As shown in Table 4, 95 participants (73%) were aware that consanguineous marriages were a risk factor contributing towards the increasing prevalence of thalassemia disorders.

Table 5: The mode of awareness about thalassemia amongst parents of the patients

	Mode of awareness about thalassemia	Frequency (n)	Percentage (%)
1	From health professionals	109	84
2	From peers and colleagues	5	4
3	From media and news outlets	14	11
4	From self interest	2	1.5
	TOTAL	130	100

As shown in Table 5, 109 participants (84%) of the study learnt about thalassemia through their healthcare provider/ other healthcare personnel.

Table 6: Participants awareness about the modalities treatment of thalassemia

	Perceived Modality of Treatment	Frequency (n)	Percentage (%)
1	Iron chelation therapy	22	17
2	Blood transfusion	53	41
3	Stem cell transplant	8	6
4	No idea	47	36
	TOTAL	130	100

As shown in Table 6, nearly 84 participants (64%) were aware of some modality of treatment of thalassemia whereas nearly 36% participants were not aware of any proper modality for the same.

Other findings of the study were as follows:

Nearly 67% participants knew that screening of blood is mandatory before every transfusion. Of them 64% participants understood the reason behind the screening of blood.

Almost 10% participant parents underwent diagnostic testing before marriage and almost 90% parents did not undergo any form of testing.

Unfortunately nearly 8% participants believed that the disorder was a curse upon them by a deity or a supernatural power for their wrong doings.

However, 99% of the participants believed that there should be many thalassemia public awareness programs, and that premarital screening must be performed.

Discussion

The study included 130 parents of patients suffering from β -thalassemia. The main aim of our study was to estimate the knowledge about these disorders among the affected families.

The study found that still 27% of the parents do not know the hazardous aftermath of consanguinity with respect to thalassemia inheritance and nearly 46% of the parents still did not understand the need for prenatal screening to prevent thalassemia disorders.

Nearly 5% of the parents were still unaware of the mode of transmission of thalassemias and nearly 36% of the parents had little to no knowledge about the various modalities of treatment of thalassemia.

It is clearly evident that the need to spread awareness about thalassemias is the need of the hour. India must follow the screening policies which have shown a remarkable decrease in the incidence of thalassemias, to identify carriers of thalassemia.

The life expectancy of thalassemia patients can be increased by national planning and special care but knowledge of the exact rate and age distribution of inherited blood disorders is limited. Many families are still suffering from a menace much bigger than thalassemia - that of misguided beliefs and religious superstitions that are creating stigma around the disorder. It is believed that an increase in maternal education would lead to dispelling such beliefs and while also allowing a significant increase in the utilization of prenatal tests, carrier screening and prenatal diagnosis.

Limitations

This study included β -thalassemia patients from only one thalassemia centre, which limits the generalisability of the results. A study consisting of patients from different nationalities and centres will give more accurate responses and will help to understand the pattern of behavior common among thalassemia patients/ parents.

This study should also ideally factor in the fact that socio-economic variables like consanguinity, religious apprehensions around abortion and high rates of illiteracy lead to an overwhelming amount of cases going unnoticed and undetected, which could not be accounted for in this study.

Conclusion

The research findings exhibited that awareness about thalassemia, its prevention and treatment is on the correct path but yet not ideal and there is significant room for improvement. There is a long way to go for the healthcare sector to spread awareness about the disorders. Only with awareness of the general public can thalassemias be prevented efficiently, for which the public should be informed and encouraged to

undergo screening to detect carriers. Families that are thalassemia carriers should receive appropriate genetic counseling. An effort to educate parents about prenatal diagnosis must be made. Lastly, the government should organize campaigns to encourage premarital screening.

References

1. Darvishi Khezri, Emami Zeydi A, Sharifi H, Jalali H. Is Vitamin C Supplementation in Patients with beta-Thalassemia Major Beneficial or Detrimental? Hemoglobin. 2016;40(4):293–4. doi: 10.1080/03630269.2016.1190373.
2. Dehshal MH. Addressing adherence to treatment: a longstanding concern. The patients' perspective. Thalassemia Reports. 2014;4(3)
3. Vermeire E, Hearnshaw H, Van Royen P, Denekens J. Patient adherence to treatment: three decades of research. A comprehensive review. J Clin Pharm Ther. 2001;26(5):331–42. doi: 10.1046/j.1365-2710.2001.00363.x.
4. Pedram M, Zandian K, Keikhaie B, Akramipour R, Hashemi A, Ghahfarokhi FK, et al. A report on chelating therapy and patient compliance by determination of serum ferritin levels in 243 thalassemia major patients. Iranian J Pediatr Soc. 2010;2(2):65–9
5. Ragab LA, Hamdy MM, Shaheen IA, Yassin RN. Blood transfusion among thalassemia patients: A single Egyptian center experience. Asian J Transfus Sci. 2013;7(1):33–6. doi: 10.4103/0973-6247.106728.
6. Coifman KG, Ross GS, Kleinert D, Giardina P. Negative affect differentiation and adherence during treatment for thalassemia. Int J Behav Med. 2014;21(1):160–8. doi: 10.1007/s12529-012-9277-7.
7. Pages- Puigdemont N, Mangues MA, Masip M, Gabriele G, Fernandez-Maldonado L, Blancafort S, et al. Patients' Perspective of Medication Adherence in Chronic Conditions: A Qualitative Study. Adv Ther. 2016;33(10):1740–54. doi: 10.1007/s12325-016-0394-6. Lehane E, McCarthy G. Intentional and unintentional.
8. Ward A, Caro JJ, Green TC, Huybrechts K, Arana A, Wait S, et al. An international survey of patients with thalassemia major and their views about sustaining life- long desferrioxamine use. BMC Clin Pharmacol. 2002;2:3. doi: 10.1186/1472- 6904-2-3.
9. Prevalence of β -thalassemic patients associated with consanguinity and anti-HCV- antibody positivity-a cross sectional study. Qurat-ul-Ain LA, Hassan M, Rana SM, Jabeen F. Pak J Zool. 2011;43:29–36.
10. Awareness among parents of children with thalassemia major. Arif F, Fayyaz J, Hamid A.
11. Awareness among parents of β -thalassemia major patients, regarding prenatal diagnosis and premarital screening. Ishaq F, Abid H, Kokab F, Akhtar A, Mahmood S.
12. Ethical, social, and cultural issues related to clinical genetic testing and counseling in low- and middle-income countries: a systematic review. Adrina Zhong et al.
13. Exploring Rare Disease Patient Attitudes and Beliefs regarding Genetic Testing: Implications for Person-Centered Care. Andrew A Dwyer et al.
14. Newborn screening for hemoglobinopathies and red cell enzymopathies in Tripura state: a malaria-endemic state in Northeast India. Upadhye D, Das RS, Ray J, et al.
15. Inherited hemoglobin disorders in Andhra Pradesh, India: a population study. Munshi A, Anandraj MPJS, Joseph J, et al.
16. Distribution and population genetics of the thalassemias. Higgs DR, Thein SL, Wood WG.