ORIGINAL ARTICLE

Prevalence of Thalassemia in Children of Adilabad District

K Ananth Rao
Assistant Professor, Department of Pediatrics, Rajiv Gandhi Institute of Medical Sciences and Hospital [RIMS], Adilabad, Telangana State

Abstract

***Background**: Thalassemias are the common monogenetic diseases belonging to the family of inherited disorders of hemoglobin synthesis characterized by the impaired formation of globin chains of adult hemoglobin. The Region of Adilabad District in Telangana is one of the places where the presence of thalassemia is very common because of geographic location and presence of predominantly tribal population. We in the present study tried to determine the presence of thalassemia among the children admitted in the pediatric ward with severe anemia.*
***Method**: This cross-sectional study was conducted in Rajiv Gandhi Institute of Medical Sciences and Hospital [RIMS] Adilabad. The data was collected from Oct 2017 to Jan 2018. The blood samples were collected from the suspected anemic patients in a vacutainer of 5ml and sent for confirmation of the presence of thalassemia and treatment was done accordingly. The complete records of the patients detected with thalassemia were maintained for future treatments.*
***Results**: Total numbers of inpatients admitted in the pediatric ward during the period from Oct 2017 to Jan 2018 were 2055. Out of that 52 patients were confirmed with Thalassemia. Out the 52 cases, 34 cases were male children having minimum Hb gm% 3.4 and maximum Hb gm% 9.4 with 95% Confidence Interval 5.3 – 6.5. Cases of thalassemia in female were 18 numbers having minimum Hb gm% 3.7 and maximum Hb gm% 9.8 and 95% Confidence Interval of 5.13 - 6.47. The prevalence rates of thalassemia in pediatric inpatient wards were 2.53%. **Conclusion**: The prevalence of Thalassemia is common in Adilabad, there is a need for complete and detailed study collecting baseline data of tribal children with Thalassemia. The data obtained from such a study can be used by the Governmental Authorities to prepare an appropriate action plan for reducing the burden of the disease and providing treatment and support to people already suffering from the disease.*
***Keywords**: Thalassemia, Prevalence, Adilabad*

Address for correspondence: Dr. K. Ananth Rao, Assistant Professor, Department of Pediatrics, Rajiv Gandhi Institute of Medical Sciences [RIMS], Adilabad- 504001. Telangana State. E mail: kachraj@gmail.com Mobile: 9849072897

Introduction

Hemoglobinopathies are a group of inherited hemoglobin disorders in which there is a structural defect in the hemoglobin molecule. Thalassemia was first described by Dr. Thomas Cooley and Lee in 1925 most of them was located in Mediterranean geography and the majority of them suffered from anemia hence the name of these group of anemias are known as Cooley's anemia or Mediterranean anemia. [1] Variants of thalassemias and abnormal hemoglobins interact to produce a wide range of clinical disorders of varying severity. [2-4] In thalassemia there could be either mutation or deletion that results in reduced rate of synthesis or no synthesis of one of the globin chains that make up hemoglobin. [5] This causes the formation of abnormal hemoglobin molecules or fewer hemoglobin molecules thus causing anemia. Severe forms of the thalassemia re usually diagnosed early in childhood and are lifelong conditions and require regular blood transfusions to survive. [4] The clinical manifestations of β-thalassemias are highly variable. They range from asymptomatic cases with mild mutations to those with mild hypochromic anemia or with moderate and server lifelong transfusion-dependent anemia with multi-organ involvement. [2] Nearly all β-thalassemia variants are inherited in a Mendelian recessive manner, but there is a small

subgroup of β-thalassemia alleles that behave in a dominant fashion. [2]

Indian scenario

The first case of β-thalassaemia/Hb E disease in India was reported by Chatterjee *et al;*[6] and that of β-thalassaemia/sickle cell disease by Naik *et al;*[7] Subsequently other variants haemoglobins were reported – C, D, F, G, H, J, K, L, M, Q in India. [8-12] Haemoglobins S, D and E were observed to be quite common: Hb S has been found mostly in tribal communities, Hb D in Gujaratis and Punjabis and HbE in Bengalis, Assamese, and Nepalese.The average incidence of beta thalassemia trait in India is 3.3 % with 1–2 per 1,000 couple being at risk of having an affected offspring each year. [13] The disease is characterized by its genetic heterogeneity at the molecular level, and more than 300 mutations of the β-globin gene have been characterized all over the world. Most are small nucleotide substitutions within the cluster. All mutations result in either absence of synthesis of β-globin chains (β- thalassemia) or a reduction in synthesis of beta globin (β-thalassemia). [13] Molecular study has been done for different β-thalassemia mutations. The five to six common mutations prevalent are IVS I-5(G - C), - 619 bp deletion, IVS I-1(G -T), Cd 8/9 (+G) also known as FS 8/9 along with Cd 41/42 (-CTTT). The data were provided by Sheth *et al;*[14] and Colah *et al;*[15] for Maharashtra and Gujarat in addition to the information given by Bhukhanvala *et al;*[16] We in the present paper tried to evaluate the prevalence of thalassemia in one of the predominantly Tribal Region, Adilabad of Telangana where there is a higher presence of thalassemia has been reported however the actual extent of the problem till date is not fully identified.

Materials and Methods

This cross-sectional study was conducted in the Department of Pediatrics of Rajiv Gandhi Institute of Medical Sciences and Hospital [RIMS] Adilabad. The data was collected from Oct 2017 to Jan 2018 from the patients admitted to Pediatric Inpatients wards. Institutional Ethical committee permission was obtained for the Study. The blood samples of the suspected anemic patients were collected in vacutainer tubes of 5ml and sent to Medcis laboratory, New Bowenpally, Secunderabad for confirmation of the presence of type of thalassemia as the presence of a fully automated High-Performance Liquid Chromatography (HPLC) system for diagnosis of thalassemia is not present in our Hospital. When HbA_2 is measured as a proportionof the total hemoglobins present, not as an absolute amount HbA_2 measurement is used as a marker for beta thalassemia trait. Carrier detection is important because: β-thalassemia carriers are asymptomatic but homozygous beta thalassemia is a life-threatening disorder. The normal value of HbA_2 in the adult is 1.50% – 3.50%. HbA_2 elevated up to 8% indicates either β- Thalassemia trait or homozygous thalassemia. Once identified as thalassemia the patient's guardians were informed, treatment was done and counseled accordingly. The complete records of the patients detected with thalassemia were maintained for future treatments.

Results

Total numbers of inpatients admitted in the pediatric ward during the period from Oct 2017 to Jan 2018 were 2055. Most of the cases of thalassemia were detected in 2-5 year age group accounting for 46.15% of the patients. Out of the total 52 identified cases 34 were male and 2-5 year age group had 19 male (55.88%) and 5 – 10 years male was 10 (29.41%). The maximum numbers of cases in females were in 5 -10 years of age group a total of 7 cases out of 18 were detected (38.89%) followed by 5 (27.78%) cases in 2 – 5 years age group given in table 1.

Table 1: Age and sex wise distribution of thalassemia

Age in years	Male	Female	Total	%
0 – 1	0	0	0	0.0
1 – 2	1	2	3	11.53
2 – 5	19	5	24	46.15
5 – 10	10	7	17	32.69
10–14	4	4	8	15.38
Total	34	18	52	100

Out the 52 cases, 34 cases were male children having minimum Hb gm% 3.4 and maximum Hb gm% 9.4 with 95% Confidence Interval 5.3 – 6.5. Cases of thalassemia in female were 18 numbers having minimum Hb gm% 3.7 and

maximum Hb gm% 9.8 and 95% Confidence Interval of 5.13 - 6.47. The prevalence rates of thalassemia in pediatric inpatient wards were 2.53%.

Table 2: Hemoglobin levels in Thalassemia patient

Sex	Minimum Hb gm%	Maximum Hb gm%	SD	Average
Male	3.4	9.4	1.79	5.9
Female	3.7	9.8	1.45	5.8

Out of the total 2055 cases, 52 were confirmed to have thalassemia and in that male were 34 accounting to 65.38% female were 34.62%.

Discussion

We in the present study found the prevalence of thalassemia of 2.53% of the total in ward patients. In a similar study by KB Rathod *et al*; for the prevalence of thalassemia in and around Pune district of Maharashtra state found a slightly higher prevalence of thalassemia at around 6.2% of the total number of patients admitted to pediatric wards. [17] The average incidence of β-thalassemia trait in India is about 3.3% with 1-2 per 1000 couples being at risk of having affected offspring. Prevalence of thalassemia trait varies from 1.0-14.9% in various regions in India. [18] It has been estimated that more than 25 million people in India are carriers of β Thalassemia gene and 8000 children are born with β thalassemia major every year. [19] Bhaskar PU *et al*; found that a high frequency of β-thalassemia 9.27% was observed among the Sindhi population of India. [20] R B Balgir evaluating the β-thalassemia trait in tribal lands of middle India found β-thalassemia trait (12.1%) in the general population of Central India. In tribal Odhisa, the highest frequency of β-thalassemia trait (12.7%) was noticed among Paraja Bhuyan tribe of Sundaragarh District, followed by Paraja tribe (8.5%) of Koraput district, Dudh Kharia (8.1%) of Sundargarh district, Santhal (8.0%) of Mayurbhanj district. A low frequency of prevalence of β-thalassemia trait (0.2-3.6 %) was found in Madhya Pradesh. [21] In Adilabad RIMS we found the prevalence of thalassemia of about 2.3% of the patients admitted to pediatric wards. Although it only a very small cross-sectional study, it can only highlight the level of a problem existing here because all the cases of minor thalassemias that do not require treatment may even be unaware of their problem. Adilabad is a small town situated in the North Telangana bordering Maharashtra and the population here is predominantly tribal. Therefore, a large scale study must be done to identify the level of problem in this area. The ultimate aim of the study is to the prevention of thalassemia that is possible only be possible by population screening, genetic counseling, and prenatal diagnosis. One of the effective method advocated by many hematologists in India as a preventive strategy is premarital screening. Countries like Iran have started thalassemia screening in 1995 and experience gained from it is instructive. Their policy is based on couple screening. [22] All couples wanting to marry are required to be checked for carrier status of thalassemia in order to receive a permit for marriage registration. The male is tested first by determining the red blood cell indices. If he is a carrier as suggested by red cell indices the woman is screened. If the results are abnormal, Haemoglobin A_2 (HbA_2) is performed. Couples in which both partners are confirmed to be carriers receive counseling. In the original report by Samavat and Modell [22]over a period of 5 years (1997-2001) 2.7 million couples were tested all over Iran, of whom 10,290 were at risk. Of these, 53 per cent decided to get married, 29 per cent separated, and 18 per cent were uncertain. Even in Saudi Arabia and UAE, more and more couples are choosing prenatal diagnosis. It is only a matter of time when a policy like that is likely to be introduced in India for prevention and reducing the burden of thalassemia in India.

Conclusion

The prevalence of Thalassemia is common in Adilabad, there is a need for complete and detailed study collecting baseline data of tribal children with Thalassemia. The data obtained from such a study can be used by the Governmental Authorities to prepare an appropriate action plan for reducing the burden of the disease and providing treatment and support to people already suffering from the disease.

Conflict of Interest: *None declared*
Source of Support: *Nil*
Ethical permission: *Obtained*

References

1. Cooley TB, Lee P. A series of cases of splenomegaly in children with anemia and peculiar bone changes. Trans Am Pediatr Soc 1925;37:29-30.
2. Weatherall DJ, Clegg JB. 2001. The thalassemia syndromes. 4th Edition. Oxford: Blackwell Science Ltd 2001.
3. Thein SL. The molecular basis of ß-thalassemia. Cold Spring Harb Perspect Med 2013; 13(5):a011700
4. Cao A, Kan YW. The prevention of thalassemia. Cold Spring Harb Perspect Med 2013; 1:3(2):a011775.
5. Anita J. C.Thalassemias: The Facts and the Controversies. Pediatric Nursing 2003; 29: 26.
6. Chatterjee JB, Saha AK, Ray RN, Ghosh SK. Hemoglobin. E-thalassemia disease. Indian J Med Sci1957; 11: 553-64.
7. Naik SK, Kothari BV, Jhaveri CL, Sukumaran PK, Sanghvi LD. Fatal hemolytic anemia presumably due to the combination of sickle cell and thalassemia gene; case report. Indian J Med Sci 1957; 11: 244-49.
8. Swarup S, Ghosh SK, Chatterjee JB. Haemoglobins E and thalassemia in an Indian family with evidence of interaction between hemoglobin E and thalassemia. J Indian Med Assoc 1966; 46: 587-90.
9. Sukumaran PK, Merchant SM, Desai MP, Wiltshire BG, Lehmann H. Haemoglobin Q India (alpha 64(E13) aspartic acid histidine) associated with beta-thalassemia observed in three Sindhi families. J Med Genet 1972; 9: 436-42.
10. Ajmani M, Sharma A, Talukder G, Bhattacharyya DK. Genetic 19. interaction of beta-thalassemia (Hb βT-) & hemoglobin E (Hb βE) in populations of Eastern India. Indian J Exp Biol 1977; 15: 455-57.
11. Kumar S, Rana M, Handoo A, Saxena R, Verma IC, Bhargava 20. M, et al. Case report of HbC/β0-thalassemia from India. Int J Lab Hematol 2007; 29 : 381-85.
12. Chatterjee JB. Haemoglobinopathies, glucose-6-phosphatedehydrogenase deficiency and allied problems in the Indian subcontinent. Bull World Health Organ 1966; 35 : 837-56.
13. Sobti P. C. and Gautam A. Beta-and-Related Thalassemias: Clinical Aspects. In: Sachdeva A., Lokeshwar M. R., Shah N. and Agarwal B.R. Editors. Hemoglobinopathies. New Delhi: Jaypee Brothers Publishers 2006; 49–59.
14. Sheth J. J., Sheth F. J., Pandya P., Priya R., Davla S., Thakur C. and Flavin V**.** Beta-Thalassemia Mutations in Western India. Indian J Pediatr 2008; 75(6): 567-70.
15. Colah R. B., Goraskshakar A. C., Lu C. Y., Nadkarni A. H., Desai S. N, et al; Application of Covalent Reverse Dot Blot Hybridization for Rapid Prenatal Diagnosis of the Common Indian Thalassemia Syndromes. Indian J Hemat Blood Transf 1997; 15:10-13.
16. Bhukhanvala D. S., Italia K., Sawant P., Colah R., Ghosh K. and Gupte S. C.2013. Molecular Characterization of B-Thalassemia in Four Communities in South Gujarat--Codon 30 (G → A) A Predominant Mutation in the Kachhiya Patel Community. Ann Hematol 2003; 92(11): 1473-76.
17. K B Rathod, DG Kulkarni, ZF Hashmi, SC Puranik. Prevalence of thalassemia in and around Pune District, Maharashtra. International J. of Healthcare and Biomedical Research 2017; 5(02): 28-31.
18. SB Satpute, MP Bankar, AA Momin, et al; The Incidence of B Thalassemia Trait in Pregnant Women from South Western Maharashtra. International Journal of Health Sciences & Research 2012; 103:2(1):103-07.
19. Bobhate SK, Gaikwad ST, Bhaledrao T: NESTROFT as a screening test for detection of β-thalassemia trait. Indian J Pathol Microbiol 2002; 45(3):265-67.
20. Bhaskar PU. The incidence of Sickle Cell Anaemia and Thalassaemia in Central India. Open Journal of Blood Diseases 2012; 2: 71-80.
21. Ranbir S Balgir. Spectrum of hemoglobinopathies and evaluation of beta-thalassemia trait in tribal land of Middle India. Int Public Health J 2013; 5(2):165-77.
22. Samavat A, Modell B. Iranian national thalassemia screening program. BMJ 2004; 329: 1134-37.