



Detection of Thalassemia trait amongst persons deferred for blood donation at a tertiary care hospital in Western Rajasthan, India

Sonam Alha¹, Dev raj Arya², Shaina Raheja³, S. P. Bhardwaj⁴, Ravindra Yadav⁵

¹Department of Immunohaematology and Transfusion Medicine, S.P. Medical College and A. G. of Hospitals, Bikaner, Rajasthan, India

^{1,3}Senior Resident, ²Professor, ^{4,5}Resident

Corresponding Author: Dr. Sonam Alha, Senior Resident, Department of Immunohaematology and Transfusion Medicine, S.P. Medical College & A.G. of Hospitals, Bikaner - 334 003, Rajasthan, India

Type of Publication: Original Research Paper

Conflicts of Interest: Nil

Abstract

Background: Thalassemia has been observed as major burden over health services as well as the society, as these patients require regular and repeated blood transfusions to maintain oxygenation of their tissues and keep the erythropoiesis suppressed. Carriers of beta thalassemia trait can have varying degree of anemia. Some of them have no symptoms and therefore can be detected only in a population survey or as a part of family study if other members are symptomatic or have thalassemia major.

Subjects and Methods: The present prospective study was conducted at Department of Immunohaematology and Transfusion Medicine, Sardar Patel Medical College and Associated Group of Hospitals, Bikaner (Rajasthan), among deferred Blood donors at Blood Bank and out-door voluntary blood donation camps during the period of May 2017 to June 2017. The study was based on the Blood donor selection criteria laid down by the Drug and Cosmetic Act and Rules of India. Criteria laid down by director general Health Services, New Delhi and Drug's Controller of India were strictly followed. All the persons which were deferred were further studied. Haemoglobin

estimation was performed on all reported blood donors. Venous blood samples were collected in EDTA tube from the persons who were deferred and tested for complete blood count by using fully automated haematology analyzer. A peripheral blood smear was prepared and stained by Leishman stain and observed microscopically for red cell morphology. Confirmatory test was done on HPLC Shimadzu LC-2010C HT. The data were filled up in a specially designed proforma for the study and transformed into a master chart which was then subjected to analysis.

Results: Total 5227 pre donation screening interviews were conducted. Total number of deferrals due to various reasons were 940 giving an overall incidence of 18%. Most common cause for deferral was low Haemoglobin 526(55.96%), second most cause was low body weight 97(10.32%). Out of 940 deferred donors 71 were positive for β Thalassemia Trait. Distribution of thalassemia trait was 7.55% among deferred blood donors and equally distributed in both gender. High prevalence of β thalassemia trait was detected in Muslim community in comparison to Hindu and other religious group. This is

because of endogamy and consanguineous marriages are still important feature of our population.

Conclusion: Present study proved that prevalence of β thalassemia trait among deferred donor is high, which is more prevalent in microcytic hypochromic donors. All efforts should be made to reduce the burden of genetic disorder which can be preventable through screening process and genetic counselling.

Keywords: Thalassemia trait, Blood donor, Deferral, Anemia.

Introduction

Blood is the elixir of life. It is essential for human life. Blood Transfusion is an integral part of medical practice and it is required in many surgical procedures as well as medical indications. Human blood has no substitute till date. Thus, proper utilization of blood is necessary with minimal wasting. In the developing countries because of limited resources of blood and increasing demands, there is a need to make efficient use of blood. The maximum requirement of blood and its products has been observed for the patients of cancer, obstetric cases, surgeries, trauma, chronic anemia and various pediatric indications; leading pediatric indications for blood transfusion include thalassemias, bleeding disorders, hemophilias, exchange transfusions for ABO-Rh incompatibilities etc. Thalassemia has been observed as major burden over health services as well as the society, as these patients require regular and repeated blood transfusions to maintain oxygenation of their tissues and keep the erythropoiesis suppressed. Beta-thalassemia is the commonest genetic haemoglobin disorders according to WHO report.¹ Approximately 1.5 % of world population carries genes for beta thalassemia. It is common in Greeks, Turks, Cypriots, Italians and to a lesser extent in Indian subcontinent.² Every year, approximately 100,000 children are borne worldwide with thalassemia major, of

which 10,000 are born in India.³ Regional incidence varies considerably depending on the gene prevalence and birth rate in the area.^{4,5,6} Due to the high birth rate in India a very large number of infants with genetic disorders are born every year. It is estimated that beta thalassemia has a frequency at birth of 1:2700.⁷ β -thalassaemia has been reported in most of the communities that have been screened so far in India. While the overall prevalence varies from 1.5 to 4 percent in different States, communities like Sindhis, Punjabis, Lohanas, Kutchi Bhanushalis, Jains and Bohris have a higher prevalence (4-17%)⁸⁻¹². The frequency of beta thalassemia trait (β TT) has variously been reported from <1% to 17% and an average of 3.3%.¹³ It is essential to have a more accurate assessment of the gene frequency of β TT in the population for planning control programs for β thalassemia in the country. India is a vast country with considerable regional and ethnic heterogeneity. A study of every region is an impossible proposition due to lack of infrastructural facilities, expertise, and resources.¹³ So, it becomes necessary and important to conduct more such prevalence studies to cover all the regions and ethnic populations of the country. Carriers of beta thalassemia trait can have varying degree of anemia. Some of them have no symptoms and therefore can be detected only in a population survey or as a part of family study if other members are symptomatic or have thalassemia major.¹⁴ The epidemiology of thalassemia is changing during the past few decades in some countries such as Cyprus and Greece. This is mainly due to the successful implementation of prevention programs. A reduction in the birth rate of babies with thalassemia major from 1:250 to 1:4000 over the years has been reported in Sardinia.¹⁵ Prevention is possible by public awareness, carrier screening, genetic counseling, prenatal diagnosis, and selective termination of affected fetuses. Though there is a

definite need for carrier screening in the population. Success of any program depends on the culture, education, and circumstances of the target population.^{16,17,18,19}The positive findings regarding carrier status of thalassemia would be notified to the related persons so that they can get their further genetic counseling done. This would provide an “add on” investigation facility to the persons reporting for the blood donation, which would serve as a motivational tool towards blood donation i.e., recruiting new blood donors and increasing the future donor pool. This would also help reducing the overall burden of blood requirement on the blood banks because of reduced burden of the disease in the society by means of thalassemia trait screening and genetic counseling. The key strategy of this study is carrier screening, public awareness, and planning for further prevention programs in the communities of this region on large scale.

Material and Methods

The present prospective study was conducted at Department of Immunohaematology and Transfusion Medicine, Sardar Patel Medical College and Associated Group of Hospitals, Bikaner (Rajasthan), among deferred volunteers at Blood Bank and out-door voluntary blood donation camps during the period of May 2017 to June 2017. Informed consent was obtained from all the participants before enrolling into the study. The study was based on the donor selection criteria laid down by the Drug and Cosmetic Act of India. Criteria laid down by director general Health Services, New Delhi and Drug’s Controller of India were strictly followed. Each donor was evaluated based on detailed medical history and brief physical examination of donors with regard to hemoglobin, blood pressure, temperature, and pulse rate. All the persons which were deferred were further studied. Haemoglobin estimation was performed on all reported blood donors using the finger-prick method with

Hemocue Hb 301 at the time of donor selection. Venous blood samples were collected in EDTA (ethylene diamine tetra acetic acid) anti-coagulated tube from the persons who were deferred and tested for complete blood count. The complete blood count was carried out by using fully automated haematology analyzer. Peripheral Blood Film were prepared and stained with Leishman stain and were examined under oil immersion lens of microscope for morphological typing of anemia. Test for β thalassemia trait was done on HPLC Shimadzu LC-2010C HT. The data were filled up in a specially designed proforma for the study and transformed into a master chart which was then subjected to analysis.

Observations

Total 5227 pre donation screening interviews were conducted. Among reported donors, most of the donors were males 5038 (96.4%); females accounted was 189(3.6%) only. Total number of deferrals due to various reasons were 940 giving an overall incidence of 18%. Out of 940 deferrals, 802 were males and 138 were females. The deferral rate among male donors was 15.9% and among female donors was 73%. Females were found to have higher deferral rate among female donors than males. Temporary causes of deferral were 856 (91.06%) of the total causes while permanent were only 84 (8.94%). Temporary causes are more predominant in the deferral cases. Most common cause for deferral was low Haemoglobin 526(55.96%), second most cause was low body weight 97(10.32%). The prevalence of anemia is higher among females than males in western part of Rajasthan. The most common type of anemia was microcytic hypochromic 287(54.56%). The next most common type was normocytic normochromic 133 (25.28%). Females in the reproductive age group have higher deferral rate due to anemia. Out of 940 deferred donors 71 were positive for β Thalassemia Trait.

Distribution of thalassemia trait was 7.55% among deferred blood donors and equally distributed in both gender. Most of the persons of thalassemia trait were from young age group. This is because most of donor population comprises of young age group. Mean of hemoglobin was (9.4 ± 2.2). Mean of MCV was (71.6 ± 6.6). HbA2 ranges from 3.5-7. High prevalence of β thalassemia trait was detected in Muslim community (9.36%) in comparison to Hindu (7.16%) and other religious group. This is because of endogamy and consanguineous marriages are still important feature of our population.

Table 1: Prevalence of Thalassemia Trait among deferred donor according to gender.

	Total (n=940)	Thalassemia Trait	Prevalence of Thalassemia trait
Males	802	61	7.61%
Females	138	10	7.25%
Total	940	71	7.55%

In our study, we found that out of 940 deferred donors 71(7.55%) were positive for Thalassemia Trait. In which 61 were male and 10 were female. Percentagewise male and female were 7.61% and 7.25% among their respective gender group so this prevalence of Thalassemia trait was found equal for both gender group.

Table 2: Thalassemia trait according to age group

Age	Total	%
16-20	7	9.86%
21-30	31	43.66%
31-40	23	32.39%
41-50	7	9.86%
51-60	3	4.23%
Total	71	100%

Table 2 shows the age wise distribution of persons who were found positive for thalassemia trait. It is seen that most of the persons were from young age group. This is because most of donor population comprises of young age group.

Table 3: Frequency of Thalassemia trait in different Religious groups.

Religion	Total	βTT	%
Hindu	726	52	7.16%
Muslim	203	19	9.36%
Others	11	0	0%
Total	940	71	7.55%

Table 3 shows the religion wise distribution of persons who were found positive for thalassemia trait. Out of 940 deferred donors, 726 were Hindu, 203 were Muslim and 11 were from other religious group. Prevalence of thalassemia trait were 52 (7.16%), 19 (9.36%), 0, in Hindu, Muslim, and other religious group respectively. The prevalence of thalassemia trait was more in Muslim community as compare to Hindu and Other religious group, as consanguineous marriages are still important feature in Muslim community.

Discussion

Studies	Year	βTT %
Manit Nuinoon (Thailand) ²⁰	2014	0.9%
Alhamdan (Saudi Arabia) ²¹	2007	3.22%
Raiz Ahmed (Pakistan) ²²	2014	4.9%
Rosline H (Malaysia) ²³	2006	5%
Present Study	2017	7.55%

In our study β Thalassemia trait was found 7.55% in the deferred donors which was higher than the previous studies such as Manit Nuinoon (Thailand)²⁰ 0.9%, Alhamdan (Saudi arabia) ²¹ 3.22%, Raiz Ahmed (Pakistan) ²² 4.9%, Rosline H (Malaysia)²³ 5%.

Studies	Year	βTT %
V. K. Meena (BHU, U.P.) ²⁴	2012	1.0%
L.P. Meena (Eastern U.P.) ²⁵	2013	2.8%
Rajesh Kumar (Punjab) ²⁶	2015	3.3%
Rachana Nagar ²⁷ (Eastern Indian States)	2015	3.4%
Malik Mahmood ²⁸ (Azad Kashmir)	2016	5.6%
Sur D (West Bengal) ²⁹	2006	11.25%
Present Study	2017	7.55%

India is a vast country with considerable regional and ethnic heterogeneity. Regional incidence varies considerably depending on the gene prevalence and birth rate in the area. Due to the high birth rate in India a very

large number of infants with genetic disorders are born every year. In our study β Thalassemia trait was found 7.55% in the deferred donors which was higher than the previous studies such as V. K. Meena (BHU, U.P.) 24 1%, L.P. Meena (Eastern U.P.) 25 2.8%, Rajesh Kumar (Punjab) 26 3.3%, Rachana Nagar 27 (Eastern Indian States) 3.4%, Malik Mahmood 28 (Azad Kashmir) 5.6% and lower than Sur D (West Bengal) 11.25%. 29 In our study distribution of β thalassemia trait was high as it studied in deferred donor. The most common cause of deferred donor in our study was anemia and microcytic hypochromic persons have high prevalence of β thalassemia trait as studied previously by Aseem k Tiwari. 30 Mean of hemoglobin, MCV was (9.4 ± 2.2) and (71.6 ± 6.6) respectively. HbA2 ranges from 3.5-7.

Conclusion

Present study proved that prevalence of β thalassemia trait among deferred donor is high, which is more prevalent in microcytic hypochromic donors. All efforts should be made to reduce the burden of genetic disorder which can be preventable through screening process. The screening should be done at regular intervals of the same population and record should be maintained by the department implemented by the government. The data of the present study shows that there is a need to carrier screening, public awareness, and planning for further prevention programs in the communities of this region on large scale.

References

1. Patel AP, Parmar PH, Patel RB, Trivedi NM, Bhartiya NA. Factors Influencing Beta-Thalassemia Awareness in Western India. *Ntl J Community Med* 2016;7(3):193-197.
2. Weatherall DJ, Akinyanju O, Fucharoen S, Olivieri N, Musgrove P. Inherited disorders of hemoglobin In: *Disease Control Priorities in Developing Countries* (2nd Edition). Jamison D, Breman J, Measham A et al. (Eds). Oxford University Press and the World Bank, NY, USA, 2006;663–80.
3. Panigrahi I, Ahmed RP, Kannan M, Kabra M, Deka D, Saxena R. Cord blood analysis for prenatal diagnosis of thalassemia major and hemophilia A. *Indian Pediatr* 2005;42:577–81.
4. Alwan A, Modell B, *Community Control of Genetic and Congenital Disorders*, EMRO Technical Publications Series 24. Geneva: WHO Regional Office for the Eastern Mediterranean 1997.
5. Ahmed S, Saleem M, Modell B, Petrou M. Screening extended families for genetic hemoglobin disorders in Pakistan. *N Engl J Med* 2002;347:1162-8.
6. Ansari SH and Shamsi TS. Thalassaemia Prevention Programme. *Hematology updates* 2010:23-8.
7. Verma IC. Burden of genetic disorders in India. *Indian J Pediatr*. 2000 Dec;67(12):893-8.
8. Mehta BC, Dave VB, Joshi SR, Baxi AJ, Bhatia HM, Patel JC. Study of hematological and genetical characteristics of Kutchi Bhanushali Community. *Indian J Med Res* 1972; 60 : 305-11.
9. Sukumaran PK. Abnormal hemoglobins in India. In: Sen NN, Basu AK, editors. *Trends in hematology*. Calcutta: Sree Saraswati Press; 1975. p. 225-61.
10. Chouhan DM, Chouhan V. Epidemiology: Symposium on thalassemia. *Indian J Hematol Blood Transf* 1992; 10 : 1-6
11. Jawahirani A, Mamtani M, Das K, Rughwani V, Kulkarni H. Prevalence of beta thalassemia in sub-castes of Indian Sindhis: Results from a two phase survey. *Public Health* 2007; 121 : 193-8.
12. Mohanty D, Colah R, Gorakshakar A. editors. Report of the Jai Vigyan S & T Mission Project on community control of thalassaemia syndromes- Awareness, screening, genetic counselling and prevention. A National Multicentric Task Force Study

- of Indian Council of Medical Research, New Delhi; 2008. New Delhi: ICMR; 2008.
13. Nishi Madan, Satendra Sharma, S. K. Sood, Roshan Colah, (Late) H. M. Bhatia. Frequency of β thalassemia trait and other hemoglobinopathies in northern and western India. *Indian J Hum Genet*. 2010 JanApr; 16(1): 16–25.
 14. Agarwal M B, Mehta B C. Symptomatic beta thalassemia trait (A study of 143 cases). *JMed*:1982;28:48.
 15. Cao A, Rosatelli MC, Galanello R. Control of beta thalassaemia by carrier screening, genetic counselling and prenatal diagnosis: the Sardinian experience. *Ciba Found Symp*. 1996;197:137–51.
 16. Al Sulaiman A, Saeedi M, Al Suliman A, Owaidah T. Postmarital follow-up survey on high risk patients subjected to premarital screening program in Saudi Arabia. *Prenat Diagn* 2010; 30:478–481.
 17. Al-Odaib AN, Abu-Amro KK, Ozand PT, Al-Hellani AM. A new era for preventive genetic programs in the Arabian Peninsula. *Saudi Med J* 2003; 24:1168–1175.
 18. Mitchell JJ, Capua A, Clow C, Scriver CR. Twenty-year outcome analysis of genetic screening programs for Tay-Sachs and β -Thalassemia disease carriers in high schools. *Am J Med Genet* 1996; 59: 793–798.
 19. Lena-Russo D, Badens C, Aubinaud M, Merono F, Paolasso C, Martini N, et al. Outcome of a school screening program for carriers of hemoglobin disease. *J Med Screen* 2002; 9: 67–69.
 20. Manit Nuinoon, Kwanta Kruachan, Warachaya Sengking, Dararat Horpet, Ubol Sungyuan, Thalassemia and Hemoglobin E in Southern Thai Blood Donors. *Advances in Hematology*, vol. 2014, Article ID 932306, 6 pages, 2014. doi:10.1155/2014/932306.
 21. Alhamdan NA, AlMazrou YY, AlSwaidi FM, et al. Premarital screening for thalassemia and sickle cell disease in Saudi Arabia. *Genetics in Medicine*. 2007;9:372–77.
 22. Raiz Ahmed Qazi, Rabia Shams, Hamid Hassan, Naghmi Asif. Screening for Beta Thalassemia Trait. *Journal of Rawalpindi Medical College (JRMC)*; 2014;18(1):158-60.
 23. Rosline H, Ahmed SA, Al-Joudi FS, Rapiaah M, Naing NN, Adam NA. Thalassemia among blood donors at the Hospital Universiti SainsMalaysia. *Southeast Asian J Trop Med Public Health*. 2006 May;37(3):549-52.
 24. V K Meena, Kailash Kumar, L P Meena, Anju Bharti, A Kumar. Screening for hemoglobinopathies in blood donors from eastern Uttar Pradesh. *National journal of medical research*. 2012 Sept; 2(3):366-67.
 25. L P Meena, K Kumar, V K Singh, Anju Bharti, S K H Rahman, K Tripathi. Study of Mutations in β -Thalassemia Trait among Blood Donors in Eastern Uttar Pradesh. *J Clin Diagn Res*. 2013 Jul;7(7):1394–96.
 26. Kumar R, Gupta S, Jindal A, Kakkar S, Kaur A. Screening of β thalassemia trait and other hemoglobinopathies among blood donors in Punjab. *Int J Med Public Health* 2015;5:106-9.
 27. Rachana Nagar, Sujata Sinha, Rajiva Raman. Haemoglobinopathies in eastern Indian states: a demographic evaluation. *Journal of Community Genetics* January 2015; 6(1) :1–8.
 28. Ahmed MM, Salaria SM, Qamar S, Soaz MA, Bukhari MH, Qureshi AH. Incidence of β -thalassemia carriers in Muzaffarabad, Azad Kashmir. *APMC* 2016;10(1):11-19.

29. Sur D , Mukhopadhyay SP. Prevalence of thalassaemia trait in the state of West Bengal. J Indian Med Assoc. 2006 Jan;104(1):1-15.
30. Aseem K. Tiwari, Iva Chandola1 Comparing prevalence of Iron Deficiency Anemia and Beta Thalassemia Trait in microcytic and non-microcytic blood donors: suggested algorithm for donor Screening. Asian J Transf Sci – July 2009;3(2):99-102.