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Thalassemia: A Genetic Disorder Disease in SMGS Hospital Jammu

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Abstract:
Thalassemia is an inherited blood disorder caused when our body doesn't make enough of a protein called hemoglobin, an important part of red blood cells. Thalassemia can quickly lead to anemia. This condition is marked by a lack of oxygen being transported to tissues and organs. Since red blood cells are responsible for delivering oxygen, a reduced number of these cells means we don't have enough oxygen in the body. Data on status of thalassemia and hemoglobinopathies from the extreme northern part of India is scarce. We investigated socio-demographic characteristics and management issues related to β-thalassemia in Jammu and Kashmir, India. Data from 83 thalassemia major and intermedia patients visiting the department of transfusion medicine for their transfusion needs was collected. Parameters recorded included age group, age at diagnosis, gender, religion, districts of the state they belonged to, family history of thalassemia, blood group, type of thalassemia (major/intermedia), total number of transfusions received and chelation therapy status. Most cases were diagnosed in infancy or early childhood. The districts of Jammu and Rajouri together contributed 53 % of the cases. Most patients were Hindu, A positive family history was most often obtained from Muslim patients. Only 50 % cases were on iron chelation therapy. There is a need to come up with a national/local policy to manage disease in endemic areas and a policy formulated to help families and patient.

Keywords: Patients, Haemoglobin, Anaemia, Genes, Blood Groups, Transfusion.

Introduction

Thalassemia is an inherited (i.e., passed from parents to children through genes) blood disorder caused when the body doesn't make enough of a protein called hemoglobin, an important part of red blood cells. When there isn't enough hemoglobin, the body's red blood cells don't function properly and they last shorter periods of time, so there are fewer healthy red blood cells traveling in the bloodstream. Thalassemia is an autosomal recessive/co dominant disease, the genetic defect results in reduced rate of synthesis of one of the globin chains that make up hemoglobin. The β-thalassemia are a major health problem in India but have received little attention because of other health priorities, such as malnutrition and communicable diseases. There are about 70–90 million people in the world who carry beta thalassemia trait alone. The average frequency of β-thalassemia carriers in the Indian population is 3–4 %, leading to an overall estimate of 30–40 million carriers among a population of over a billion people. The burden of β - thalassemia is not uniform with some communities having much higher frequencies ranging from 5 to 17 %. Among all the inherited disorders of blood, haemoglobinopathies are the major public health problem in the world. Inherited Haemoglobin disorders were originally characteristic of tropics and sub-tropics but are now common worldwide due to migration. World Health Organization estimates that globally at least 5% of adults are carriers for a haemoglobinopathies: approximately 2.9% for thalassemia. There is a tremendous amount of burden of haemoglobinopathies in India. It has been estimated that there would be about 45 million carriers and about 15,000 infants born each year with haemoglobinopathies in India. Major Haemoglobin variant, i.e. HbA ranges from 15 to 45% of the total Haemoglobin in the red cells. More than 100 alpha chain variants have been described in the world. Most of the available

epidemiological data is based on screening that was undertaken in small pockets in individual communities in different states of this vast country. It is well recognized that even in geographic regions where β-thalassemia is common, not all ethnic groups are at the same risk of possessing the thalassemia gene. In India. We investigated the socio-demographic characteristics of symptomatic β-thalassemia including differences in prevalence in different communities in Jammu and Kashmir, India.

Subjects and Methods

This study was done at Department of Immunohematology and Transfusion medicine at Shri Maharaja Gulab Singh Hospital (SMGS), Jammu J&K, India. The data of all the thalassemic patients visiting the department of transfusion medicine for their transfusion needs was collected over a period of 10 months. A Performa was prepared which included the following parameters: age of diagnosis, age group of the presenting patients, sex, religion, districts of the state they belonged to, family history of thalassemia, socio economic status, blood group, type of thalassemia- major/intermediate, total no. of transfusion received so far, alloimmunization status and chelation therapy status. These parameters were obtained from patient's previous records at department of transfusion medicine and pediatrics. The diagnosis had been previously established based on blood film, electrophoresis and hemoglobin F quantitation performed at various other centers and departments. Blood transfusion to thalassemia in SMGS Hospital is regularly, if Blood transfuse to Thalassemia major patient today, then after 10 or 15 days again blood transfuse to these patients.

Fig. **Blood Transfusion to Thalassemia Patients**

Before transfuse the blood to thalassemia patient a Hydrocortisone sodium Succinate injection IP given to only those patient in which blood reaction takes place.

Types of thalassemia

There are two types of thalassemia — alpha thalassemia and beta thalassemia

Alpha thalassemia

We inherit four genes, two from each parent, that make alpha globin protein chains. When one or more genes are defective, we develop alpha thalassemia. The number of defective genes we inherit will determine whether we experience anemia symptoms and how severe they will be.

One defective or missing alpha gene means that we won't experience symptoms. Another name for this condition is alpha thalassemia minima.

Two defective or missing alpha genes means that if we experience symptoms, they will likely be mild. Another name is alpha thalassemia minor.
Three defective or missing alpha genes means that we will experience moderate to severe symptoms. Another name for this condition is Hemoglobin H disease.
Four defective or missing alpha genes usually results in death. In those rare instances when a newborn survives, they'll likely need lifelong blood transfusions. Another name for this condition is hydrops fetalis with Hemoglobin Barts.

Beta thalassemia
We inherit two beta-globin genes, one from each parent. Our anemia symptoms and how severe our condition is depends on how many genes are defective and which part of the beta globin protein chain contains the defect.
One defective or missing beta gene means that we'll experience mild symptoms. Another name for this condition is beta thalassemia minor.
Two defective or missing beta genes means that we'll experience moderate to severe symptoms. The moderate version is called thalassemia intermedia. More severe beta thalassemia involving two gene mutations is called beta thalassemia major or Cooley's anemia.

How do I know if I have thalassemia?
People with moderate and severe forms of thalassemia usually find out about their condition in childhood, since they have symptoms of severe anemia early in life. People with less severe forms of thalassemia may only find out because they are having symptoms of anemia, or maybe because a doctor finds anemia on a routine blood test or a test done for another reason. Because thalassemia are inherited, the condition sometimes runs in families. Some people find out about their thalassemia because they have relatives with a similar condition. People who have family members from certain parts of the world have a higher risk for having thalassemia. Traits for thalassemia are more common in people from Mediterranean countries.

Symptoms of Thalassemia
Beta-thalassemia
Beta thalassemia occurs in two different forms namely thalassemia intermedia and thalassemia major. Thalassemia symptoms appear generally before a child's second year of age and severe Anaemia concerned with this condition can be fatal. Some of the major signs of thalassemia major include: Paleness, Extreme tiredness, shortness of breath, Jaundice, Fussiness, Poor appetite, change in the shape of bones in the face and head etc. This kind of thalassemia is so serious that it needs frequent blood transfusions. Thalassemia intermedia is a less serious kind of beta-thalassemia and do not require the patient to go through blood transfusions.
Beta-Thalassemia trait is found in individuals where there is only one HBB gene mutation in each cell possesses mild Anaemia.
Alpha-thalassemia
It consists of two major forms namely, Hydrops fetalis or Haemoglobin H disease. Haemoglobin H can be responsible for bone complexities. The forehead, cheeks, and jaw may overgrow. Moreover, Haemoglobin H can cause: An intensely enlarged spleen, Malnourishment

Treatment for Thalassemia
The treatment depends on the type and severity of the disease. The doctor provides a course of treatment that suits best for a particular case. Some of the treatments, which are opted for maximum cases include: Bone marrow transplant (BMT), Supplements and Medications, Blood transfusions. Few precautions are prescribed by the doctors that include not taking supplements or vitamins and minerals containing iron. This is true when there is a need for blood transfusions. Patients who go through blood transfusions obtain

extra iron which a body cannot lose. If we are receiving a blood transfusion, we may need chelation therapy. It includes taking a chemical injection that combines with other heavy metals and iron. This helps eliminate extra iron from the body.

Prevention of Thalassemia

Thalassemia cannot be prevented since it is a genetically inherited disorder. However, these disorders can be detected during prenatal tests before birth. Also, genetic counselling helps to detect whether people have altered or missing Haemoglobin genes that cause thalassemia.

How do blood transfusions affect my body?

People who receive a lot of blood transfusions are at risk for iron overload. Red blood cells contain a lot of iron, and over time, the iron from all of the transfusions can build up in the body. When it builds up, the iron collects in places like the heart, liver, and brain, and can make it hard for these organs to work properly. To prevent iron overload, people with thalassemia may need chelation therapy, which is when doctors give a medicine – either a pill or a shot under the skin – to remove excess iron before it builds up in the organs. Every time a person gets a blood transfusion, their risk for a problem called "alloimmunization" goes up. Alloimmunization happens when a person's body reacts to blood from a transfusion because it is seen as harmful by their immune system, and tries to destroy it. Persons with alloimmunization can still receive blood transfusions, but the blood they receive has to be checked and compared to their own blood to make sure that it won't be destroyed by their immune system. This takes time and can mean that persons with alloimmunization have to wait longer for blood, or may have a harder time finding blood that won't be destroyed by their body. Another concern for people who receive a lot of blood transfusions is the safety of the blood they receive. Some infections, like hepatitis, can be carried in blood. In the United States, the blood supply is screened and monitored for safety, and the risk of getting an infection from a blood transfusion is very low. Nevertheless, there is still a very small risk of getting an infection through a blood transfusion.

Results

A total of 83 patients were registered at Department of Immunohematology and Blood transfusion medicine at SMGS hospital. Out of these 73 % (n = 63) were male and 27 % (n = 20) were females male: female ratio was 2.7:1.

Tables 1 and 2 shows district-wise distribution of thalassemic patients in our study. Clustering is seen in Jammu and Rajouri region. This difference was not found to be statistically significant.

Table 1:- District wise distribution of thalassemic patients

District	No. of thalassemics
Jammu	25
Rajouri	20
Reasi	04
Poonch	04
Kathua	06
Akhnoor	02
Kishtwar	01
Udhampur	11
Doda	01
Outside state	09

Table 2:- Age group thalassemic patients coming to hospital for treatment

Age group	**Male**	**Female**

0–1	2	0
1–5	11	05
5–14	33	10
>14	14	05

Only 20.8 % (n = 20) of the patients overall had a family history of thalassemia. Significantly higher numbers of Muslim patients (8/18, 44.4 %) gave a positive family history as compared to Hindu (12/72, 16.6 %) or Sikh patients (0/6, 0 %). This difference was statistically highly significant (p value .017). Most of the patients visiting the department were in the age group of 5–14 (range 8 months to 27 years).

REFERENCES:

1. Colah R, Gorakshakar A, Phanasgaonkar S, D'Souza E, Nadkarni A, Surve R, Sawant P, Master D, Patel R, Ghosh K, Mohanty D. Epidemiology of β-thalassaemia in Western India: mapping the frequencies and mutations in sub-regions of Maharashtra and Gujarat. Br J Haematol. 2010;149:739–747. doi: 10.1111/j.1365-2141.2010.08131.x.
2. Sood SK, Madan N, Colah R, Sharma S, Apte SV, editors. Collaborative study on thalassaemia, report of an ICMR task force study. New Delhi: Indian Council of Medical Research; 1993.
3. Verma IC, Bijarnia S. The burden of genetic disorders in India and a framework for community control. Community Genet. 2002;5:192–196. doi: 10.1159/000066335.
4. Mehta BC, Dave VB, Joshi SR, Baxi AJ, Bhatia HM, Patel JC. Study of hematological and genetic characteristics of Kutchi Bhanushalis. Indian J Med Res. 1972;60:305–311.
5. Sukumaran PK. Abnormal hemoglobins in India. In: Sen NN, Basu AK, editors. Trends in hematology. Calcutta: Saraswati Press; 1975. pp. 225–236.
6. Jawahirani A, Mamtani M, Das K, Rughwani V, Kulkarni H. Prevalence of beta thalassemia in subcastes of Indian Sindhis: results from a two phase survey. Public Health. 2007;121:193–198. doi: 10.1016/j.puhe.2006.10.017.
7. Mulchandani DV, Fulare MB, Zodpey SP, Vasudeo ND. Prevalence and some epidemiological factors of β-thalassaemia in sindhi community of Nagpur city, India. Indian J Public Health. 2008;52:11–15.
8. Madan N, Sharma S, Sood SK, Colah R, Bhatia HM. Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India. Indian J Hum Genet. 2010;16:16–25. doi: 10.4103/0971-6866.64941.
9.https://www.nsgc.org/page/find-a-genetic-counselor.
10. Harsha Y. Post counseling follow-up of Thalassemia in high risk communities. Indian Paediatrics. 1997;34(12):1115–1118.
11. Chandramouli C. Census of India 2011—a story of innovations. Government of India: Press Information Bureau; 2011.
12. Agouzal M, Arfaoui A, Quyou A, Khattab M. Beta thalassemia major: the Moroccan experience. J Public Health Epidemiol. 2010;2(2):25–28.
13. Ghosh S, Bandyopadhyay SK, Bandyopadhyay R, Roy D, Maisnam I, Ghosh MK. A study on endocrine dysfunction in thalassaemia. J Indian Med Assoc. 2008;106(10):655–656, 658-659.
14. Schettini F. Diagnosis of thalassemia major in the first months of life. Ann N Y Acad Sci. 1969;165:387–393. doi: 10.1111/j.1749-6632.1969.tb27808.x.
15. Prakash K. Nutritional anemia in young children with focus on Asia and India. Indian J Community Med. 2011;36(1):8–16. doi: 10.4103/0970-0218.80786.
16. Talsania S, Talsania N, Nayak AH. Cross sectional study of thalassemia In Ahmedabad city, Gujarat. Healthline. 2011;2(1):48.

17. Bilwani F, Kakepoto GN, Adil SN, Usman M, Hassan F, Khurshid M. Frequency of irregular red cell alloantibodies in patients with thalassemia major: a bicenter study. J Pak Med Assoc. 2005;55(12):563–565.
18. Shamsian BS, Arzanian MT, Shamshiri AR, Alavi S, Khojasteh O. Frequency of red cell alloimmunization in patients with β-major thalassemia in an Iranian referral hospital . Iran J Pediatrics. 2008;18
19. Singh SP, Gupta S. Molecular pathogenesis and clinical variability of homozygous beta0-thalassemia in populations of Jammu region of J&K state (India) Hematology. 2006;11:271–275. doi: 10.1080/10245330600921956.