Thalassemia Major and Intermedia in Jammu and Kashmir, India: A Regional Transfusion Centre Experience

Rahul Vasudev · Vijay Sawhney

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Abstract 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 96 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. Thalassemia major patients comprised 92 (95.8 %) and intermedia 4 (4.2 %) of the cohort. 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 (76/96, 79.2 %). A positive family history was most often obtained from Muslim patients (8/18, 44.4 %). 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 patients. Data such as ours may help in health management planning for thalassemic patients in this region.

Keywords Thalassemia epidemiology · Transfusion in thalassemia · Thalassemia in India

Introduction

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 -thalassaemias are a major health problem in India but have received little attention because of other health priorities, such as malnutrition and communicable diseases [1]. There are about 60–80 million people in the world who carry beta thalassemia trait alone. The average frequency of β -thalassaemia 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 [2, 3]. The burden of β -thalassaemia is not uniform with some communities having much higher frequencies ranging from 5 to 17 % [4–7]. 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 -thalassaemia is common, not all ethnic groups are at the same risk of possessing the thalassaemia gene. In India, where the average prevalence of the β -thalassaemia trait is about 3.5 %, Sindhis and Punjabis are known to carry the β -thalassaemia gene more commonly than other Indian populations [8].

We investigated the socio-demographic characteristics of symptomatic β -thalassemia including differences in prevalence in different communities in Jammu and Kashmir, India. We also analyzed selected management issues in our resource constrained set-up.

Subjects and Methods

This study was done at Department of Immunohematology and Transfusion medicine at Shri Maharaja Gulab Singh

R. Vasudev (✉) · V. Sawhney
Department of Immunohematology and Blood Transfusion
Medicine, Government Medical College, Jammu, J&K, India
e-mail: vasudev_2285@yahoo.co.in

R. Vasudev
259s 1 Channi Himmat Housing Colony, Jammu, J&K, India

Table 1 District wise distribution of thalassemic patients

District	No. of thalassemics
Jammu	29
Rajouri	24
Reassi	4
Poonch	5
Kathua	6
Akhnoor	3
Kistwar	1
Udhampur	14
Doda	1
Outside state	9

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 6 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 patients previous records at department of transfusion medicine and pediatrics. Patient consent and hospital ethical committee clearance was taken. The diagnosis had been previously established based on blood film, electrophoresis and hemoglobin F quantitation performed at various other centres and departments. Alloimmunization screening was done using DiaMed™ DiaCell-ID I+II+III (Asia) screening cells initially and then identification of screening positive patients was done using 11 cell antibody identification panel. All screening and antibody identification was done on DiaMed™ ID Gel cards (anti IgG +anti c3d). Socio economic status has was measured using Kuppaswamy's Socio-Economic Status Scale and patients were divided into Upper (I), Upper Middle (II) Middle/Lower middle (III), Lower/Upper lower (IV), Lower (V) base on their education, monthly income and occupation [9]. Different ratios were calculated and significance limit was set at <0.05 and chi square test was used to find statistical significance.

Results

A total of 96 patients were registered at Department of Immunohematology and Blood transfusion medicine at SMGS hospital. Out of these 73 % ($n = 70$) were male and 27 % ($n = 26$) were females male: female ratio was 2.7:1.

Table 2 Age group thalassemic patients coming to hospital for treatment

Age group	Male	Female
0–1	2	0
1–5	11	7
5–14	39	12
>14	18	7

Table 3 Distribution of thalassemic patients into major and inter-media types

Type	Male	Female
Major	66	26
Intermediate	3	1
Total	69	27

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.

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). As shown in Table 3.

Most common blood group in thalassemic patients was O ($n = 34$, 35.4 %) followed by B ($n = 32$, 33.3 %), A ($n = 22$, 22.9 %) and AB ($n = 8$, 8.3 %). Most of the children were Rh +ve only five children were Rh –ve.

Two-third of the patient were diagnosed before their 1st birthday ($n = 63$, 65.6 %). 29.1 % ($n = 28$) of children were diagnosed between age of 1–5 and 5.2 % ($n = 5$) children were diagnosed after age of 5 years. The distribution of thalassemia major and intermediate minor in our region is shown in Table 3.

Only 52 % ($n = 50$) of children were on chelation therapy. Table 4 shows detailed description of number of transfusions received by thalassemic patients in relationship to age and sex. Seven out of the 96 thalassemic children showed red cell alloimmunization.

Socio economic status of patients was determined using Kuppaswamy's socio-economic status scale and patients were divided into upper ($n = 1$), upper middle ($n = 9$), middle/lower middle ($n = 46$), lower/upper lower ($n = 31$), Lower ($n = 9$) base on their education, monthly income and occupation shown in Table 5.

Table 4 Number of transfusions received by thalassemic patients in relationship to age and sex

Number of transfusion	Age group							
	0–1 years		1–5 years		5–14 years		>14 years	
	Male	Female	Male	Female	Male	Female	Male	Female
0–10	2	0	0	3	0	0	0	0
10–30	0	0	5	1	1	0	0	0
30–100	0	0	5	3	19	7	4	0
100–200	0	0	1	0	11	4	4	3
>200	0	0	0	0	8	1	10	4

Table 5 Classification of socio economic status of patients according to Kuppuswamy's socio-economic status scale

Socio economic class	Number of patients in this class
Upper (I)	1
Upper middle (II)	9
Middle/lower middle (III)	46
Lower/upper lower (IV)	31
Lower (V)	9

Discussion

Most of the thalassemic patients belonged to Jammu district of the state, this may be due to the fact that Jammu is the single largest city in the region with good medical facilities and the families of thalassemic children have migrated to Jammu for the same reason. Clustering was also seen in Rajouri district of the state. Treatment facilities including transfusion, chelation and specialist should be made available at district hospitals in regions where there is high incidence of thalassemia so that the families don't have to migrate to bigger cities for treatment.

The distribution of thalassemic patients according to sex shows male predominance with sex ratio of 2.7:1 such difference was also noted in other studies. Some of the other studies done in other parts of India have reported 65.5, 62.1 and 56 % of male patients [10–12]. This may be due to the fact that still in rural parts of our country more emphasis is given on the health of male child. Moreover sex ratio of state of Jammu and Kashmir is 883 which is below national average of 940 as per census 2011 [13].

Most of the children visiting the hospital for their transfusion needs were in the age group of 5–14. This can be explained by the fact that if children are not transfused, they die before the age of 6 years and if they are transfused and non-chelated, they die before the age of 20 [14]. It also important for the hospital and health authorities to know the age group of the effected patients while they are creating facilities for these patients.

Family history of thalassemia was more common in Muslims than in any other religion. This may be due to the fact that there is trend of consanguineous marriages in muslims and this may cause increased frequency of genetic diseases like thalassemia in them. The prevalence of β -thalassemia major is especially high in countries where there are close family marriages [15]. Proper genetic counseling can help decrease frequency of disease in these communities.

Most of the children were diagnosed before their first birthday. This may be due to the fact that most of the children were suffering from thalassemia major which tends to cause severe anemia in first year of life [16]. Still a significant number of thalassemia major children are being diagnosed late as the incidence of nutritional anemia is as high as 70 % in this part of world [17] and the primary care physicians misdiagnose thalassemia as nutritional anemia initially. Majority of patients presenting to our centre had thalassemia major (95.8 %). These patients completely dependent on transfusion and more likely to seek medical attention than thalassemia minor or intermediate patients. Talsania et al. [18] in their study done on thalassemic patients in Gujarat also found similar differences in presentation of thalassemia major, minor and intermediate patients.

Blood requirement of patients increases with age and they have to visit blood banks more frequently. Increasing age and transfusion needs puts these patients at risk developing several complications like iron overload, transfusion transmitted diseases and alloimmunization. The incidence of alloimmunization of multi transfused thalassemic patients in our study was 7.2 % this is comparable to that found in other studies [19, 20]. Only 52 % of the thalassemic children were on iron chelation therapy. Kelfer(deferiprone) and desferrioxamine are being given for chelation at our hospital but the effectiveness is not being monitored effectively due to non availability of test for serum ferritin levels at hospital and lack of affordability of most of the patients to get it done from outside hospital. There is a need to study type of mutation in thalassemic patients of these region, a study with 35 patients has been

done by Singh et al. [21] in 2006. Most of these children belong to middle lower/lower socio economic status the patients face the dual burden of poverty and disease. Currently, infrastructure and support for these patients and their family members is inadequate at many levels in India. The state of J&K is no exception. The patients should be managed with proper transfusion regime and given adequate chelation therapy, screening for alloimmunization and transfusion transmitted disease so that they can lead a normal healthy life. Studies such as ours are required to better understand the social, demographic and medical attributes of thalassemics in this region to develop better health management strategies for the future.

Conflict of interest None.

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