ORIGINAL ARTICLE

A PROFILE OF CASES OF HEMOGLOBINOPATHIES AT A MEDICAL COLLEGE

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ABSTRACT

Background: Present study was carried out with objectives of creating a profile for cases of hemoglobinopathies coming at NHL medical college and comparing the results obtained in present study with those of various studies done in India and abroad.
Materials and methods: The study was carried out at the Department of Pathology, N.H.L. Municipal Medical College, Ahmedabad. A total of 35 cases of thalassemias and other hemoglobinopathies were studied. The criteria for case selection were hemoglobin level less than 10 gm%, presence of hepatosplenomegaly, icterus and clinical presumption of hemolytic anemia in general. Details of the cases were recorded in a proforma. Data were then analysed using microsoft excel software.
Results: Out of total 35 cases Beta0 Thalassemia Major was the most frequent (40%) followed by homozygous sickle cell disease (20%). More than one third cases (34.3%) were of 10 or more years of age while 31.4% cases were in 1-3 years of age group. Sex wise distribution showed male preponderance (74.3%). Religion wise majority were hindus (80%). Caste wise majority were of general category (83%). Pallor was found in all cases. Most of the cases showed hypochromia, microcytosis, Anisopoikilocytosis, polychromatophilia.

Key words: hemoglobinopathies, Beta Thalassemia Major, sickle cell disease

INTRODUCTION

Hemoglobinopathies constitute a very important causative factor for anemias of childhood. This is especially so in those regions where abnormal hemoglobin genes are prevalent in a frequency of high order. They may mimic nutritional anaemias and they prove refractory to the usual corrective measures. The two common hemoglobinopathies widely distributed across our country are the beta thalassemic syndromes and the sickling disorders. The former is more common in certain non-tribal ethnic groups and the later among the tribal population[1]. A couple of earlier studies had incriminated them in the causation of anaemias of childhood[2-8].

The thalassemia is not a single disease, but a group of disorders, each of which results from the inherited defects in the rate of synthesis of one or more of the globin chains. They cause imbalance globin chain production, which leads to ineffective erythropoiesis, hemolysis and variable degree of anaemia. The exact nature of the defect is not yet understood, but its result is an unaltered quantity or quality of the m-RNA, which leads to deficient synthesis of haemoglobin polypeptide chains. In contrast to the hemoglobinopathies, no basic chemical abnormality of haemoglobin species lies behind the thalassemias. Different types of thalassemias with different clinical and biochemical manifestations are associated with defects in each kind of polypeptide chains (alpha, beta, gamma, and delta). In India, it has been observed that beta chain production is commonly affected[9].

This study was carried out with objectives of creating a profile for cases of hemoglobinopathies coming at NHL medical college and comparing the results obtained in present study with those of various studies done in India and abroad.

MATERIALS AND METHODS

The study was carried out at the Department of Pathology, N.H.L. Municipal Medical College, Ahmedabad. The study period was of two years from January 2006 to December 2007. A total of 35 cases of thalassemias and other hemoglobinopathies were come

in the attached hospital during that period and were studied. Out of 35 cases, 23 cases were children < 10 years and 12 cases were ≥ 10 years. The criteria for case selection were haemoglobin level less than 10 gm%, presence of hepatosplenomegaly, icterus and clinical presumption of haemolytic anaemia in general. Family study was carried out when needed. Details of the cases were recorded in a proforma which included age, sex, caste, religion, residence and clinical details, blood transfusion requirement and haematological findings. Data were then analysed using Microsoft excel software.

RESULTS

Table 1: Type wise distribution of hemoglobinopathies (N=35)

Nature of Disease	Cases (%)
$Beta^0$ Thalassemia Major	14 (40.0)
$Beta^+$ Thalassemia Major	2 (5.7)
Thalassemia Intermedia	2 (5.7)
Homozygous sickle cell disease	7 (20.0)
Sickle cell trait	4 (11.4)
Sickle cell Beta thalassemia	4 (11.4)
HBE - Beta thalassemia	2 (5.7)

Table 1 shows type wise distribution of hemoglobinopathies. $Beta^0$ Thalassemia Major was the most frequent (40%) followed by Homozygous sickle cell disease (20%).

Table 2: Age distribution of cases of hemoglobinopathies (N=35)

Age Group (years)	Cases (%)
0 - 1	4 (11.4)
1 - 3	11 (31.4)
4 - 6	6 (17.2)
7 - 9	2 (5.7)
≥ 10	12 (34.3)

Table 2 shows age distribution of cases of hemoglobinopathies. Out of 35 cases, more than one third cases (34.3%) were of 10 or more years of age while 31.4% cases were in 1-3 years of age group.

Table 3: Sex distribution of hemoglobinopathies (N=35)

Sex	Cases (%)
Male	26 (74.3)
Female	9 (25.7)

Sex wise distribution showed male preponderance as approximately three fourth cases (74.3%) cases were males, while only one fourth cases were females (Table 3).

Table 4 shows religion and caste wise distribution of cases of hemoglobinopathies. Religion wise majority were hindus (80%), followed by muslim (17.1%) and Christian (2.9%). Caste wise majority were of general category (83%), while rest were schedule tribe (14.3%) and chaudhari (2.9%). Table 5 shows distribution of cases according to blood transfusion requirement.

According to clinical findings, Pallor was found in all cases. While spenomegaly and hepatomegaly were found in 88.6% and 71.4% cases respectively. Icterus was observed only in 22.9% cases (Table 6).

Table 4: Religion and Caste wise distribution of cases of hemoglobinopathies (N=35)

Religion and Caste	Cases (%)
Hindu	28 (80.0)
Lohana	5 (14.3)
Scheduled Tribe	5 (14.3)
Sindhi	3 (8.6)
Jain Banias	3 (8.6)
Chaudhari	1 (2.9)
Other (Rajput, Patel, Brahmin, Daraji)	11 (32.9)
Muslim	6 (17.1)
Christian	1 (2.9)

Haematological findings: Hb levels ranging from 2.2 - 10 gm%. MCV and MCH found to be decreased in most of the cases raised from 60.3 - 90.8 fl. and 14 - 28.7 pg. respectively. Peripheral smear study showed many abnormalities regarding the size and shape of RBC. Most of the cases showed hypochromia, microcytosis, Anisopoikilocytosis, polychromatophilia. Target cells and leptocytes were seen in many cases. In some cases there were also presence of howell jolly bodies and basophilic stippling in the RBCs.

DISCUSSION

The results of present study were compared with those of other studies. The maximum numbers of cases were noted between 1 - 3 years of age group. Present study includes 62.5% cases between 1 - 3 years of age group, 25% cases below one (1) year of age and 12.5% cases in 4 - 6 year of age group. Out of 16 cases of Beta thalassemia major, 10 (62.5%) belong to 1 - 3 years of age group, which is expected. Out of 7 cases of sickle cell disease, 6 cases presented after 7 years. Hence, it can reasonably be assumed that thalassemia major disease became manifest in early childhood in most of the cases. Similar age incidence was observed in O.P. Ghai series[3], Manchanda and Khanna series[6], Magotra and Phadke series[5] and Giri, Patra and Patel series[9].

Table 5: Distribution of cases according to blood transfusion requirement (N=35)

Nature of Disease	Total Cases	Transfused cases	Frequency of blood transfusion (in times)		
			2-5	6-10	> 10
Beta^0 Thalassemia Major*	14	14	5	5	4
Beta$^+$ Thalassemia Major*	2	2	2	-	-
Thalassemia Intermedia	2	1	1	-	-
Homozygous Sickle cell disease	7	5	5	-	-
Sickle cell trait	4	0	-	-	-
Sickle cell - B Thalassemia	4	4	1	3	-
HBE - Beta Thalassemia	2	1	1	-	-

* Require regular blood transfusion (At least one unit / 30 - 60 days)

Table 6: Distribution of cases according to chief clinical findings

Clinical Findings	Cases (%)
Pallor	35 (100.0)
Splenomegaly	31 (88.6)
Hepatomegaly	25 (71.4)
Icterus	8 (22.9)

It was observed in present study that compared to Beta thalassemia major, sickle cell anaemia patients presented at later age. Average age for presentation in sickle cell syndromes were HbSS at 13 years, HbAS at 12 years and HbS Beta Thalassemia at 10 years of age. Regarding sex distribution, out of 35 cases, 74.3 % were male with male to female ratio being approximately 3:1. Ghai[3] reported 63.7% males and 36.3% females, while other study[2] found 70.6% males and 29.4% females. In the present study, out of 35 cases, 5 were Lohanas, 6 were muslims, 5 were scheduled tribes, 3 were Sindhis, 3 jain, 1 Chaudhary, 1 Christian and 11 were from other castes i.e. Patel, Brahmin, Rajput, Kachi, Daraji.

Regarding the distribution of thalassemia among various castes of central and Eastern India, it may be mentioned that of the 13 cases reported by Khandelwal and Solanki in 1959[11] from Nagpur, 7 were Sindhis. In the J.J. Group of hospitals, Bombay[8], 85 cases were investigated and their regional distribution was as follows : Saurashtra - 26, Maharashtra - 21, Sindhi - 12, Gujarat excluding Saurashtra - 9, Goa and Adjacent region - 9, Bengal - 7, U.P. - 1. Out of 26 cases from Saurashtra, 16 were Memons and Khojas and 10 were Hindus, mostly Lohanas. Studies[3, 8, 10] show that maximum incidence was noted in Sindhis, Lohanas and Muslims. Present study also shows that there are higher numbers of cases among Muslims, Lohanas, Sindhis along with scheduled tribes in thalassemia cases. Out of 17 other cases in present study, 15 cases are of sickle cell syndrome which includes homozygous sickle cell disease, sickle cell trait, sickle cell beta thalassemia and 2 cases are of HbE Beta thalassemias which were Muslims. In these hemoglobinopathies also there are higher number of cases among Luhanas, Muslims and Scheduled Tribes.

Present study found pallor in all cases, while splenomegaly and hepatomegaly were seen in 88.6% and 71.4% cases respectively. All these three clinical signs were found in all cases by other studies[2-3]. Regarding haematological findings, majority of the cases showed hypochromic microcytic anaemia. This finding is in agreement with the other series of Coelho[2], Pal and Ghai[3]. Raised reticulocyte count was found in 80% cases in the present study, while it was reported in 100% and 76.5% cases by Ghai[3] and Coelho[2] respectively.

CONCLUSION

Beta thalassemias and sickle cell disorders were found to be the commonesthemoglobinopathies in the present study. HbS was found to be the commonest Hb variant followed by HbE. Thalassemia major usually presents in early childhood while sickle cell disordersusually present after five years of age. HbE - beta thalassemia often behaves like Thalassemia intermedia in its clinical presentation. Lohana, Muslim, Sindhi, Scheduled Tribes communities comprised the larger group though other communities are also found to be involved.

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