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ORIGINAL RESEARCH

An evaluation of clinico-hematological profile of hereditary haemolytic anemias- A prospective study

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ABSTRACT

Introduction: Congenital hemolytic anaemia is mostly caused by hemoglobinopathy and thalassemia. They contribute significantly to morbidity, moderate to severe haemolytic anaemia in young children, and a number of fatalities in India. In India, according to UNICEF estimates from 1996, there are 29.7 million beta thalassemia trait carriers and 10,000 newborns are born each year with homozygous beta thalassemia.

Materials and Methods: All instances of hereditary hemolytic anaemia seen as outpatients and inpatients between the ages of 1 and 20 years were included in this group for the purposes of the study. The patient was disqualified if they had other systemic illnesses such heart failure, liver disease, major infections, etc. With the aid of a pre-designed and pre-tested standard proforma, all the data regarding their socio-demographic profile, pertinent clinical history, and clinical examination were gathered. Data was compiled in an excel spreadsheet and properly statistically assessed.

Results: 50 cases of hereditary hemolytic anaemia were identified. Hereditary hemolytic anaemias are passed to the progeny through a specific pattern of inheritance.A total of 27 patients were born out of consanguineous marriage, in which 1st degree was 2 and 2nd/3rd degree were19 and 6 respectively.

Conclusion: The majority of hereditary hemolytic anaemias are hemoglobinopathies, which place a heavy strain on families and society. Although important, haemoglobin electrophoresis establishes the disease in hemoglobinopathies and the osmotic fragility test in hereditary spherocytosis when differentiating between hereditary hemolytic anaemias. Despite some recent optimistic initiatives in developing nations, hereditary hemolytic anaemia control rarely receives the attention it requires.

Keywords: Haemoglobinopathy and thalassemia, hereditary hemolytic anaemias

INTRODUCTION

In India, hemoglobinopathy and thalassemia account for the majority of cases of congenital hemolytic anaemia. They contribute significantly to morbidity, moderate to severe haemolyticanaemia in young children, and a number of fatalities in India. [1] Hereditary haemolyticanaemias are a category of diseases marked by an intrinsic red blood cell deficiency coupled with hyperbilirubinemia, erythroid hyperplasia, and rapid erythrocyte destruction. For example, inherited spherocytosis and elliptocytosis, aberrant haemoglobin synthesis in sickle cell anaemia, decreased globin synthesis in thalassemia, and erythrocyte

enzyme defects like G6PD and pyruvate kinase deficiency are among these types of illnesses. [2] Hereditary haemolyticanaemias are the third leading cause of mortality and morbidity in developing nations, behind infection and hunger, and they place a significant burden on the patients, their families, and eventually the community. Population screening, genetic counselling, and prenatal diagnosis can all help prevent them. [3] In India, according to UNICEF estimates from 1996, there are 29.[7] million beta thalassemia trait carriers and 10,000 newborns are born each year with homozygous beta thalassemia. 4 In diverse Indian population groups, the carrier frequency of hemoglobinopathies ranges from 3 to 17%, while the carrier frequency of sickle cell hemoglobinopathies in India ranges from 1-44%. [5-8] Congenital hemolytic anaemia with Hereditary Spherocytosis is relatively prevalent. Hereditary spherocytosis was identified as the most frequent kind of hemolytic anaemia in a study conducted in Japan. [9] Hereditary spherocytosis is rarely described in Indian literature. [10,11] Direct Coombs test should be performed in all instances with spherocytes to rule out autoimmune haemolyticanaemia, which is the predominant disease when spherocytes are detected. The most prevalent erythrocyte enzyme deficiency condition is G6PD deficiency. The main clinical symptom is drug-induced hemolytic anaemia. [12]

Due to the spleen's ongoing exposure to red cells containing inclusions made of precipitated globin chain, thalassemia patients experience splenomegaly, which results in the condition known as "work hypertrophy." Both and thalassemia are characterised by progressive splenomegaly, which makes the anaemia worse. [13,14] Secondary thrombocytopenia and leucopoenia typically develop in -thalassemia major along with the potential for severe splenomegaly, increasing the risk of infection and bleeding. [15] Absent secondary hypersplenism, the white cell and platelet counts are slightly higher. Red blood cells in splenectomized subjects in particular show stippling or ragged inclusion bodies when stained with methyl violet. [16]

MATERIAL AND METHODS

The present study was carried out in Department of pathology at government Medical College Jammu & Kashmir. All the hereditary haemolyticanaemia patients reported in the referral hospital between June 2018 to December 2019. Clinical and haematological parameters were correlated.

All hereditary hemolytic anaemia cases diagnosed as outpatients and inpatients between the ages of 1 and 20 years during the study period were included in this category. The patient was disqualified if they had other systemic illnesses such heart failure, liver disease, major infections, etc. With the aid of a pre-designed and pre-tested standard proforma, all the data regarding their socio-demographic profile, pertinent clinical history, and clinical examination were gathered. Data was compiled in an excel spreadsheet and properly statistically assessed.

On an automated haematology analyzer, haematological parameters including cell counts (Total WBC counts, RBC count), red cell indices (Hb, PCV, MCV, MCH, MCHC, and RDW), and others were analysed (sysmex xt1800 and xt2000i). Leishman stain was used to evaluate the morphology of RBCs in peripheral smears. On a glass slide, the sickle cell preparation was carried out by combining 1-2 drops of freshly made 2% sodium metabisulphite solution with a drop of anticoagulated blood (EDTA) and covering the coverslip with wax or nail polish. The preparation was stored in a covered, wet petridish, and the presence of sickle cells was checked at one hour and 24 hours under a 40X microscope. Osmotic fragility test (OFT): Test tubes containing 5 ml of various sodium chloride solution concentrations and 20 l of heparinized blood were combined, centrifuged after mixing, and left at room temperature for 30 minutes. Using a 0.85% tube as a blank, optical density (OD) readings of the supernatant are obtained at 540 nm, and the percentage of lysis is estimated after placing the values on the graph. 2 ml of heparinized blood were incubated with OFT for

24 hours at 37o C. A test tube containing 0.5 ml of anticoagulated blood, 0.025 ml of dextrose sodium nitrate solution, and 0.025 ml of methylene blue was used to conduct the methaemoglobin reduction test. The test was then incubated for three hours at 370 C. To diagnose G6PD deficiency using a standard, positive (blood and dextrose solution) and negative controls were placed on. At a pH of 8.6, 3 l of whole blood-derived haemolysates were applied to strips of cellulose acetate, electrophoresed at 150 V for 30 minutes, and then dyed with Ponceau's satin. The relative percent of each band was calculated after the patterns were scanned on a scanning densitometer. The information was assembled on an excel sheet and examined.

RESULTS

Over a period of two years, 50 cases of hereditary haemolyticanemia were diagnosed during the period study in which 30 were male and 20 were females.

Out of 50, 6 (12%) subjects were in the age group 0-5 years followed by 16(32%) in age group 6-10 years, 20(40%) were age group 11-15 years and 16% were age group 16-20 years. (Table 1) Our results show that HA is more prevalent among younger population (below 15 years) than the elders. Incidences of HA diminishes with increasing age.sex wise distribution of hereditary hemolytic anemiaswas shown in table 2.

age wise distribution of hereditary hemolyticanemiawas shown in table 1. A total of 27 patients were born out of consanguineous marriage, in which 1st degree was 2 and 2nd/3rd degree were19 and 6 respectively (Table 4). Table 3 were shown number of patients born out of consanguineous marriage. Hereditary spherocytosis and Sickle cell anemia were seen in 6(12 %) cases, Sickle cell trait and Sickle / β thalassemia were seen in 2(4 %) cases, β-thalassemia major was shown in 26(52%) cases (Table 7).

Table 1: Age wise distribution

Age group (Years)	Number of cases	Percentage
1-5	6	12
6-10	16	32
11-15	20	40
16-20	8	16

Table 2: sex wise distribution of hereditary hemolytic anemias

Sex	No. of cases	Percentage
Male	30	60
Female	20	40

Table 3: number of patients born out of consanguineous marriage

Consanguinity	Number of cases	Percentage
Present	27	54
Absent	23	46

Table 4: type of consanguinity between parents

Type of consanguinity	Number of cases
First degree	2
Second degree	19
Third degree	6

Table 5: blood group wise distribution of hereditary hemolytic anaemias

Blood group	Number of cases	Percentage
A	10	20
B	16	32
O	20	40
AB	4	8

Table 6: rh wise distribution of hereditary hemolytic anaemias

Rh	Number of cases	Percentage
Positive	48	96
Negative	2	4

Table 7: distribution of various causes of hereditary hemolytic anaemias

Cause	Number of cases	Percentage
Hereditary spherocytosis	6	12
Sickle cell anaemia	6	12
Sickle cell trait	2	4
Sickle / β thalassemia	2	4
β-thalassemia major	26	52
β-thalassemia trait	8	16

DISCUSSION

Although hemoglobinopathies are common everywhere, they are more common in specific regions. 50 patients with congenital hemolytic anaemia between the ages of 1 and 20 years have been examined clinically and by other investigations in the current study. Being a genetic condition, hereditary hemolytic anaemia is frequently observed in specific cultures where inbreeding is common. Being a genetic condition, hereditary hemolytic anaemia is frequently observed in specific cultures where inbreeding is common. Numerous community-based studies carried out across India demonstrate a higher prevalence of hemolytic anaemia in a particular community. In India, the prevalence of hereditary hemolytic anaemia ranges from 0.1% to 0.2%. 5 The prevalence of inherited hemolytic anaemia is 0.2% in the current study as well. Patients in the current study ranged in age from 1 to 20 years. Maximum cases (40%) ranged in age from 11 to 15 years. In order to rule out non-hereditary hemolytic anaemia brought on by immune-mediated mechanisms like infant Rh-mediated hemolytic illness, patients of less than two months were not included. Males were more frequently affected by hereditary hemolytic anaemia in the current study, which is consistent with research by Juwah et al.[17,]Shivashankara et al.[18] and RS Balgir. [19]

As screening tests for detecting enzyme deficiencies were not conducted, there were no instances of the enzyme defects G6PD and pyruvate kinase, which are mentioned in other research. The prevalence of these diseases has not changed significantly, hence the current study, despite being outdated, is still relevant. It provides a thorough, descriptive explanation of the many hereditary hemolytic anaemias that are common in the area and helps to paint a picture of them in this part of India.

CONCLUSION

Hereditary haemolyticanaemias have a particular pattern of inheritance and thus transmitted to the offspring. Haemoglobinopathies which constituted the major group of hereditary haemolyticanaemia has a great burden on the families and the society. Differentiation of hereditary hemolytic anaemias using hematological investigations although useful,

hemoglobin electrophoresis establishes the disease in hemoglobinopathies and osmotic fragility test in hereditary spherocytosis. Despite some recent encouraging initiative in the underdeveloped countries, the control of hereditary hemolytic anaemias is generally not given the importance it deserves.

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