

“Spectrum of Haemoglobinopathies in Anaemic Patients Admitted in Tertiary Care Hospital in Jharkhand”.

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Abstract:

Aim: To identify different types of haemoglobinopathies using high performance liquid chromatography in anaemic patients and correlation of haemoglobinopathies with ethnicity.

Methods: A Prospective cross sectional study done in 42 anaemic patients admitted in Department of Medicine and Department of Paediatrics in RIMS from November 2014 to December 2015. The diagnosis of Haemoglobinopathies was based on High Performance Liquid Chromatography.

Result: From the study it was found that 66.7% of patient were of sickle cell disease, 9.5% of patients were of sickle cell trait and beta thalassemia trait each, 7.4% were of beta thalassemia major, 4.8% of sickle cell beta thalassemia compound heterozygous variety and 2.4% was of haemoglobin E beta thalassemia variety. 18 cases (42.857%) belonged to the tribes of Jharkhand and 24 cases (57.1%) were non tribals

Conclusion: Sickle cell disease was the most common (66.7%) haemoglobinopathies observed in the study. 18 cases (42.857%) belonged to the tribes of Jharkhand and 24 cases (57.1%) were non tribals.

Keywords: Haemoglobinopathies, Sickle cell Disease, Thalassemia

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I. Introduction

Haemoglobinopathies are the disorder affecting the structure, function or production of haemoglobin^[1]. The hereditary disorders of haemoglobin are classified into, the haemoglobinopathies and the thalassemia syndrome^[2]. Haemoglobinopathies and its clinical consequences were noted among African tribes long before they were recognized in the western countries. In West Africa the ethnic tribes gave various names to these conditions; Ga tribe: Chechweechewe, Faute tribe: nwiwii, Ewe tribe: nuidudui, Twi tribe: ahotutuo^[3]. Haemoglobinopathies result from molecular substitution within the Hb chain (e.g HbS or HbC) or from the presence of abnormal chains or abnormal amounts of Hb chains. Clinical disorders include sickling, anaemia, haemolysis, drug-induced haemolysis, and alteration in oxygen affinity (both increased and decreased). They are inherited disorders, but acquired form have also been seen. Haemoglobin consist of a tetramer of globin polypeptide chain. During the first few months of postnatal life, Hba completely replaces HbF, and adult pattern is fully established by 6 months^[4]. Each haemoglobin tetramer are highly soluble and capable of transporting four oxygen molecules. Solubility and reversible oxygen binding are the key properties deranged in haemoglobinopathies^[1]. There are five major classes of haemoglobinopathies and include structural haemoglobinopathies, Thalassemia, Thalassaemic haemoglobin variants, Hereditary persistence of fetal haemoglobin, Acquired haemoglobinopathies. Inherited haemoglobin disorders were originally characteristic of the tropics and subtropics but are now common worldwide due to migration. Hemoglobin disorders were originally endemic in 60% of 229 countries, potentially affecting 75% of birth, but are now sufficiently common in 71% of countries among 89% of birth. At least 20% of the world population carry alfa thalassemia^[5]. Sickle cell disease is a collective term for a number of genetic disorders in which haemoglobin is structurally abnormal, resulting in the episodic formation of sickle shaped red blood cells and a wide range of clinical manifestations. It affects some 12500 people in UK and millions worldwide, particularly^[6] those of black African and Afro-Caribbean descent, and also those from the Mediterranean, Middle East, and parts of India^[7]. The underlying abnormality is a single nucleotide substitution in the gene for beta-globin on chromosome 11, resulting in the replacement of glutamic acid residue with valine on the surface of the protein^[8]. It is now well established that that sickle cell anaemia result from a single change of one amino acid, valine instead of glutamic acid at the sixth position among the 146 amino acids of the haemoglobin beta chain^[9,10]. The thalassaemias are a group of anaemias that result from inherited defects in the production of

haemoglobin. The thalassemias are among the most common genetic disorders worldwide, occurring more frequently in Mediterranean region^[11], the Indian subcontinent, southeast Asia and West Africa^[12]. Ineffective bone marrow erythropoiesis and excessive red blood cells haemolysis together account for the anaemia. Since reticulocytes manufacture equimolar quantities of alpha and beta chains, mature erythrocytes contain essentially equimolar amounts of each chain^[13].

Aims and Objective:

1. To identify different types of haemoglobinopathies using High Performance Liquid Chromatography [HPLC] and to differentiate between different types of haemoglobinopathies.
2. Correlation of haemoglobinopathies with ethnicity.

II. Methodology

This study was performed on 42 anaemic patients admitted in Department of Medicine and Department of Paediatrics in Rajendra Institute of Medical Sciences with special reference to hemoglobinopathy; over a period of one year from December 2014 to November 2015. High performance liquid chromatography was performed to differentiate between various type of haemoglobinopathies.

Inclusion criteria: Both male and female anaemic patients diagnosed to have haemoglobinopathy based on HPLC patients admitted in RIMS.

Exclusion criteria: patients suffering from any other cause of anaemia such as chronic renal failure, liver disease, aplastic anaemia, malabsorption syndrome, anaemia of chronic disease, haemolytic anaemia due to enzyme defect.

Statistical analysis

All data collected and their distribution as per age, gender, ethnicity, religion and diagnosis was tabulated, and their frequency, percentage, valid percent and cumulative percent was calculated.

III. Result

Among the 42 cases of haemoglobinopathies 27 patients were male and 15 were female. 18 cases (42.857%) belonged to the tribes of Jharkhand and 24 cases (57.1%) were non tribals [Table-I]. Distribution on the basis of religion 40.476% cases belonged to Sarna, 35.71% to Hindu, 21.42% to Muslim and 2.4% to Christian. Sickle cell disease was the most common type (66.7%) of haemoglobinopathies followed by sickle cell trait and beta thalassemia trait (9.5% each). Hbe-beta thalassemia was the least common (2.4%). Maximum number of patient (19 cases) belonged to the age group 21-30 years. The minimum and maximum age of the patients in this study is 8 months and 50 years respectively. On ultrasonography examination of abdomen most of the patients had normal ultrasound abdomen, however 19% had splenomegaly, 16.67% had hepatosplenomegaly, 4.76% had hepatomegaly and 2.38% had cholelithiasis demonstrating the various hepatobiliary and abdominal manifestation of haemoglobinopathies.

IV. Discussion

Among the 42 cases, the commonest haemoglobinopathy was sickle cell disease with 28 patients (66.7% of cases), followed by sickle cell trait with 4 patients (9.5%), beta thalassemia trait with 4 patients (9.5%), beta thalassemia major with 3 patient (7.1%), sickle cell disease – beta thalassemia compound heterozygotes with 2 cases (4.8%) and haemoglobin E-beta thalassemia with 1 patient (2.4%) {vide table-}. This result was similarly substantiated by David Weatherall^[14] in his article. The most important forms of thalassaemia are equally divided between beta thalassaemia major and haemoglobin E beta thalassaemia which occurs in high frequency in parts of Indian subcontinent, Bangladesh, Myanmar and throughout southeast Asia^[15-18]. We have included patient in this study belonging to a wide range of age group from 8 months to 50 years, where most of the patients belonged to third decade. In this study the number of non tribals affected (57.1%) appear to be more than tribals (42.9%), probably because we have considered the muslim community (who have high prevalence of the disease probably due to the practice of consanguineous marriage amongst them).

V. Conclusion

Among the different haemoglobinopathies sickle cell disease was the most common (66.7%) followed by sickle cell trait (9.5%) and beta thalassaemia trait (9.5%). Haemoglobinopathy disorder was found more in non tribals (57.1%) than tribals (42.9%).

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Distribution On The Basis Of Ethnicity: Table No-I

	Frequency	percent	Valid percent	Cumulative percent
Valid TRIBAL	18	42.9	42.9	42.9
	24	57.1	57.1	100.0
NONTRIBAL	42	100.0	100.0	
TOTAL				

Disribution On The Basis Of Religion :Table Ii

	Frequency	percent	Valid percent	Cumulative percent
Valid CHRISTAN	1	2.4	2.4	2.4
HINDU	15	35.7	35.7	38.1
MUSLIM	9	21.4	21.4	59.5
SARNA	17	40.5	40.5	100.0
Total	42	100.0	100.0	

Type Of Haemoglobinopathies Table Iii

	Frequency	Percent	Valid Percent	Cumulative Percent
Valid Sickle cell Disease	28	66.7	66.7	66.7
Sickle cell Trait	4	9.5	9.5	76.2
Beta Thalassemia Trait	4	9.5	9.5	85.7
Beta Thalssemia Major	3	7.1	7.1	92.9
Sickle cell Disease/Beta Thalssemia	2	4.8	4.8	97.6
Hbe-Beta Thalssemia	1	2.4	2.4	100
Total	42	100	100	

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