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Study of Hemoglobinopathies in Gujarat.

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Abstract: Hemoglobin is a major protein involved in transport of oxygen. Hemoglobinopathies are group of disease characterized by quantitative and/or qualitative abnormality in the production of Hemoglobin, e.g. thalassemia and sickle cell disease respectively. Identification of these disorders is immensely important epidemiologically and they can be prevented by population screening. Approximately 250 million people constituting 4.5% of the world population carry a potentially pathological haemoglobinopathy gene. Beta thalassemias are widely distributed through a belt running from Mediterranean, North West Africa through the Middle East, Pakistan, India and south East Asia. Hbs is prevalent in Africa, Mediterranean countries, and India. The disease is observed in Orissa, Andhra Pradesh, some tribal areas of Madhya Pradesh, Tamilnadu and Maharashtra. In USA and Latin America countries, 6-8% of the black carry sickle cells gene. The aim of the study was study of various Hemoglobinopathies in patients of Gujarat.

Index Terms **– Hemoglobinopathies, Thalassemia, sickle cell disease, HPLC, HbD-Panjab.**

I. INTRODUCTION

Definition: A protein inside the red blood cells that carriers oxygen from the lungs to tissues and organs in the body and carries carbon dioxide back to the lungs.

Haemoglobin is a hemoprotein with two linked pairs of globin chains. Each chain is bound to heme residue in its centre. Haemoglobin molecules are in red blood cells (RBCs) and regulate O_2 and CO_2 concentration in tissues. A structural difference in the globin chains is account for the different haemoglobin types in circulating blood. In normal adults, haemoglobin include HbA ($\alpha_2\beta_2$), HbA2 ($\alpha_2\delta_2$) and HbF ($\boldsymbol{\alpha_2\gamma_2}$), accounting for 95-97%, 1-3.5% and up to 2% of total haemoglobin, respectively. HbA is adult haemoglobin; it begins to form in eighth week of foetal life and reaches the adult levels within a few months after birth. In HbA2 its functional behaviour is probably similar to HbA. HbF is Foetal haemoglobin. It is produced by erythroid precursor cells from 10 to 12 weeks of pregnancy through the first six months of postnatal life.

Hemoglobinopathies or haemoglobin disorders are common genetic diseases that have an autosomal recessive inheritance pattern. Hemoglobinopathies are divided into quantitative e.g. thalassemia and qualitative e.g. sickle-cell disease defects in globin synthesis.

1. Quantitative defect leads to thalassemia syndromes, since no structurally abnormal haemoglobin is synthesized. Quantitative defects lead to haemoglobin variants with point mutations in goblins.

Thalassemia is a group of disorders which result from an inherited abnormality of globin production. This is the commonest form of Hemoglobinopathies. About 250 million people i.e. 4.5% of the world population carry thalassemia gene. Beta thalassemia are widely distributed through a belt running from Mediterranean, North West Africa through the Middle East, Pakistan, India and south East Asia. Thalassemia syndromes results from defects in the rate of synthesis of **α** or β globin chain. Clinical and haematological features of these cases are due to reduced haemoglobin production. And accumulation of α and β globin chains. Clinical syndromes vary from totally asymptomatic carriers to sever anaemia.

They are classified into two types:

β-Thalassemia:

In this type of there is reduced synthesis of β chains of globin. β-Thalassemia is commonest type and is prevalent all over the world with large number of cases in India.

α-Thalassemia:

There is reduced synthesis of α-chains of globin. α-Thalassemia cases are seen mainly in South East Asian countries and Indian subcontinent.

2. Qualitative defect leads to sickle cell syndrome, since no regulatory abnormal haemoglobin is synthesized. Qualitative defects lead to haemoglobin variants with point mutations in goblins.

Hbs is prevalent in Africa, Mediterranean countries, and India. The disease is observed in Orissa, andrapradesh, some tribal areas of Madhya Pradesh, Tamilnadu and Maharashtra. In USA and Latin America countries, 6-8% of the black carry sickle cells gene. It has been observed that there is high prevalence of Hbs in area which are endemic for falciparum malaria, suggesting that HBs provides protection against falciparum malaria.

TABLE 22.2: Important hemoglobinopathies

Hemoglobin	*Point mutation position*	*Amino acid substitution*	*Codon and base substitution*
HbS	Beta 6	Glu → Val	GAG → GUG
HbC	Beta 6	Glu → Lys	GAG → AAG
HbE	Beta 26	Glu → Lys	GAG → AAG
HbD (Punjab)	Beta 121	Glu → Gln	GAG → CAG
HbM	Proximal or distal histidine in α or β chains	His → Tyr	CAC → UAC

Laboratory Diagnosis of Hemoglobinopathies are based on complete blood count results and haemoglobin measurement by cyan-Methemoglobin, electrophoresis, and chromatography. Red blood cell morphology varies from normocytic normochromic to hypochromic microcytic and haemoglobin concentration can reduced or normal.

MATERIAL AND METHODS

The present cross-sectional retrospective study included 70 patients referred for screening of hemoglobin disorders from july 2020 to June 2022 at Unipath laboratory Anand – a reference laboratory which received various samples for testing and diagnosis from many small laboratories and clinicians, from all over Gujarat. A 5 ml intravenous blood sample was collected in EDTA anticoagulant. Laboratory diagnosis of Hemoglobinopathies are based on complete blood count (CBC) analysed on automated blood cell counter.

HbA2, HbF, and other haemoglobin variants were studied by **High-performance liquid chromatography (HPLC)** method used for chromatographic separation of human haemoglobin. These samples are analysed on the Bio-Rad D-10 HPLC system.

We used the Variant Haemoglobin Testing System (Variant II Beta Thalassemia Short Program, Bio-Rad Laboratories Inc., Hercules, CA, USA) under the experimental conditions specified by the manufacturer.

The peripheral smear was stained with Leishman's stain. Grading of hypochromia, anisocytosis, microcytosis, macrocytosis and polychromasia was done according to standard criterion. Inclusion bodies, sickle cells, target cells, nucleated red cells, spherocytes were noted in peripheral smear, when seen.

INCLUSION CRITERIA

1. Hb less than 12 gm%
2. Mentzer's Index: less than 13
3. Pt. of any age

EXCLUSION CRITERIA

1. Hb more than 12 gm%
2. Known case of Hemoglobinopathies.

STATISTICALLY ANALYSIS

Based on t-test, chi-square test.

RESULT

In present study shows that the age distribution highest case in age group 21-30 years. Nearly 50% of the patients were from this age group. (Table 1)

Table No.1 Age Wise distribution

Age (years)	1 to 10	11 to 20	21 to 30	31 to 40	41 to 50	Total
Cases	4	14	35	9	8	70
%	5.71%	20%	50%	12.85%	11.42%	100%

In present study total 70 patient are included. In which 50 patients were female and 20 patients were male.(Table 2)

Table No.1: Gender wise distribution

Gender	Total male/female	%
Male	20	28.57
Female	50	71.42
Total cases	70	100

Present study shows that red cell indices are evaluated in different Hemoglobinopathies and RBC count is highest in thalassemia trait than sickle cell trait.(Table 3)

Table No.3: Mean Hemoglobin and red cell parameters in different Hemoglobin variant.

Disease	Hb (gm %)	RBC (million/cumm)	MCV(fl)
Thalassemia Trait	12.06	7.89	63.77
Sickle cell trait	9.94	5	68.04

DICUSSION

In present study shows that the age distribution highest case in age group 21-30 years. Nearly 50% of the patients were from this age group. Previous study done by Vishal Kadam et al in MAY 2022 shows that Hemoglobinopathies is more common among age group 21-30 years. Nearly 60% were from this age group.
In present study total 70 patient are included. In which 50 patients were female and 20 patients were male. Previous study done by J Patel et al shows that Hemoglobinopathies is more common among female (50.70%) than male(49.29%).
Present study shows that red cell indices are evaluated in different Hemoglobinopathies and RBC count is highest in thalassemia trait than sickle cell trait. Previous study done by Jha R[1] et al also shows that red cell indices are evaluated in different Hemoglobinopathies and RBC count is highest in thalassemia trait than sickle cell trait.

CONCLUSION

Age wise distribution shows the most affected age group is 21-30 years. Gender wise distribution indicates the female are more affected than male. Mean Hemoglobin and red cell parameters in different Hemoglobin variant shows that red cell indices are evaluated in different Hemoglobinopathies and RBC count is highest in thalassemia trait than sickle cell trait.

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