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"PREVALENCE OF SICKLE CELL TRAIT AND BETA THALASSEMIA TRAIT IN PREGNANT WOMEN: A STUDY AT TERTIARY CARE HOSPITAL"

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ABSTRACT

Background: Hemoglobinopathies are a group of genetic disorders of hemoglobin. They affect 4.5% of the world population.[6] Approximately 250 million people constituting 4.5% of the world population carry a potentially pathological hemoglobinopathy gene. The sickle cell anemia and thalassemia are the most severe forms of genetic disorders and hence are of great importance to be dealt with from public health point of view in India. The hemoglobinopathies encompasses all genetic diseases of hemoglobin. Early antenatal detection followed by prenatal testing is the key to prevention of the birth of homozygous children. Antenatal screening is the important step to identify women having the risk of producing a child affected with hemoglobinopathy.

Aims and Objectives: The aim of the present study was to estimate the prevalence of sickle cell trait(SCT) and beta thalassemia trait (BTT) amongst the women attending Antenatal Clinics (ANC).

Materials and methods: This cross sectional, retrospective study included all pregnant women attending ANC clinics in a tertiary care hospital from 15 January 2021 to 15 august 2021. After obtaining the ethical approval, the demographic data and HPLC(High Performance Liquid Chromatography) results were recorded. The HPLC was performed on BioRad D10 Analyser. Collected data were compiled in Microsoft office Excel sheet

and analysed using Epi-info statistical software. Descriptive and analytical statistical methods were used for the preparation of results. Data is presented in tabulated as well as graphical format.

Results: Total 6029 patients were screened. Out of this 218 cases were of SCT, 13 cases of SCT+BTT. 160(69%) cases were from age-group of 20-25 years. Total prevalence of SCT was 3.61% and that of cases with SCT+BTT was 0.21%.

Conclusion: We found that sickle cell trait is more prevalent than beta thallasemia trait. Early antenatal screening followed by prenatal testing is the most feasible approach for the prevention of the birth of homozygous childen. Public health education programs plays the vital role to raise the awareness.

Keywords: Sickle cell trait, Thallasemia, Antenatal, Prevalence

INTRODUCTION

The hemoglobinopathies encompasses all genetic diseases of hemoglobin. They fall into three main groups: structural hemoglobin variants (abnormal hemoglobins), thalassemia syndromes characterized by the reduced rate of synthesis of one or more globin chains, and the condition in which fetal hemoglobin synthesis persists beyond the neonatal period, collectively known as hereditary persistence of fetal hemoglobin.[1,12]

Sickle Cell Anemia (also known as Sickle Cell Disorder or Sickle Cell Disease(SCD) is a genetic blood disorder, where the blood cells contain abnormal sickle shaped hemoglobin (HbS) called sickle hemoglobin. With an estimated 5,200 live births each year, SCD is a major public health problem in India. Although SCD has been described in India in numerous ethnic groups, it is most prevalent. Prevalence of Sickle Cell gene is 5 to 34 % in scheduled tribes, who have a high prevalence of socio-economic disadvantage and are frequently medically underserved. India has also a very huge populations of tribal community about 18 crore and expected to have 1.80 crore sickle cell trait(SCT) and 14 lakhs of sickle cell disease. This shows the big burden on the public health of India.[2]

Thalassemia syndromes are a heterogeneous group of single gene disorders, inherited in an autosomal recessive manner, prevalent in certain parts of the world. Worldwide, 15 million people have clinically apparent thalassemic disorders. Reportedly, there are about 240 million carriers of β-thalassemia worldwide, and in India alone, the number is approximately 30 million with a mean prevalence of 3.3%. They are encountered among all ethnic groups and in almost every country around the world.[3]

Every year approximately 100,000 children with Thalassemia Major are born world over, of which 10,000 are born in India. It is estimated that there are about 65,000-67,000 β-thalassemia patients in our country with around 9,000-10,000 cases being added every year. The carrier rate for β-thalassemia gene varies from 1 to 3% in Southern India to 3% to 15% in Northern India. Certain communities in India, such as Sindhis and Punjabis from Northern India, Bhanushali's, Kutchis, Lohana's from Gujarat, Mahar's. have a higher carrier rate. [3-5]

Once a child is diagnosed to have thalassemia homozygous disorders, he/she has to take lifelong treatment. Management includes regular 3 weekly filtered packed red cell transfusions, chelation therapy for iron overload, management of complications of iron overload and transfusions, including osteoporosis, cardiac dysfunction, endocrine problems, Hepatitis B & C, HIV infection, CMV etc. However, this optimal treatment comes at a prohibitive cost. The cost of treatment of an average weight 4-year-old thalassemic child is around Rs. 90,000-100,000 annually in a private set-up. Therefore, not more than 5-10% of thalassemic children born in India receive optimal treatment. Stem cell transplantation as a curative treatment, which costs between 6 and 16 lac rupees is out of reach for majority of children.[3] So, it is very important to identify the cases with trait to reduce the incidence of the disease.

The present study was done amongst the women attending Antenatal Clinics (ANC) with an aim to estimate the prevalence of sickle cell trait and beta thalassemia trait (BTT) among them.

AIMS AND OBJECTIVES

- To study the age-wise distribution of sickle cell trait and beta thalassemia trait among women attending Antenatal clinics at tertiary care center
- To estimate the prevalence of sickle cell trait among study population.
- To estimate the prevalence of beta thalassemia trait among study population.

MATERIAL AND METHODS

Study Design: Cross sectional, retrospective and observational study.

Study Population: Pregnant women attending ANC clinics in tertiary care hospital.

Sample size: We included all the pregnant women attending ANC clinics in our tertiary care hospital from 15 January 2021 to 15 august 2021 (7 months period). Total 6029 patients were screened in this duration.

Inclusion criteria: All pregnant women attending ANC clinics in our tertiary care hospital from 15 January 2021 to 15 august 2021 (7 months period)

Methods: After obtaining the permission from ethical committee, we have collected the data d from ANC Clinic and Central Laboratory in our tertiary care hospital from 15 January 2021 to 15 august 2021 (7 months period). The data includes demographic details of the patients. E.g Age, Gender, results of HPLC (High Performance Liquid Chromatography) report of patients. The HPLC was performed on BioRad D10 Analyser. All the demographic details and haematological investigation were recorded from the registers.

Statistical Methods

Collected data were compiled in Microsoft office Excel sheet and was analysed using Epi-info statistical software. Descriptive and analytical statistical methods are used for the preparation of results. Data is presented in tabulated as well as graphical format.

RESULTS

We conducted a cross sectional, retrospective and observational study to find out the prevalence of cases of SCT and BTT amongst the women attending ANC in a tertiary care hospital. We included total 6029 patients who were screened for the presence of any hemoglobinopathy by HPLC. In our study there were total 218 cases of SCT and 13 cases of SCT+BTT. Rest of the cases were either not showing any abnormalities on HPLC or having other hemoglobinopathies.e.g. Sickle cell disease(12 cases) etc.

We have studied the age-wise distribution of our cases with SCT and BTT. (Table – 1). This showed highest number of cases in the age-group of 20-25 years female.

Table – 1. Age-wise distribution of cases

Age group (years)	SCT	SCT + BTT	TOTAL
< 20	12	1	13
20-25	151	9	160
26-30	42	3	45
31-35	9	0	9
35-40	4	0	4
> 40	0	0	0
Total	218	13	231

In our study 69% cases were from age-group of 20-25 years.(Figure – 1)

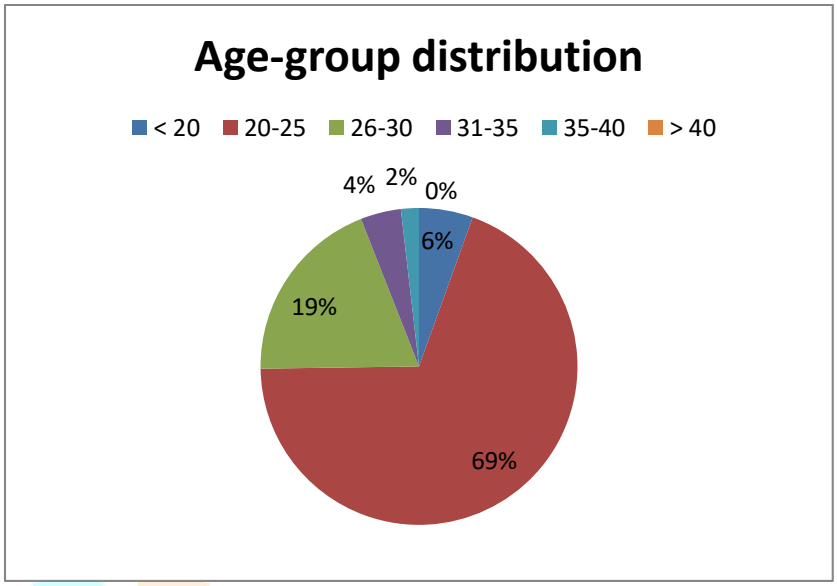

Figure – 1. Age-group wise distribution of cases

We distributed the cases month-wise starting from 15 January 2021 to 15 august 2021 (7 months period).(Figure – 2)

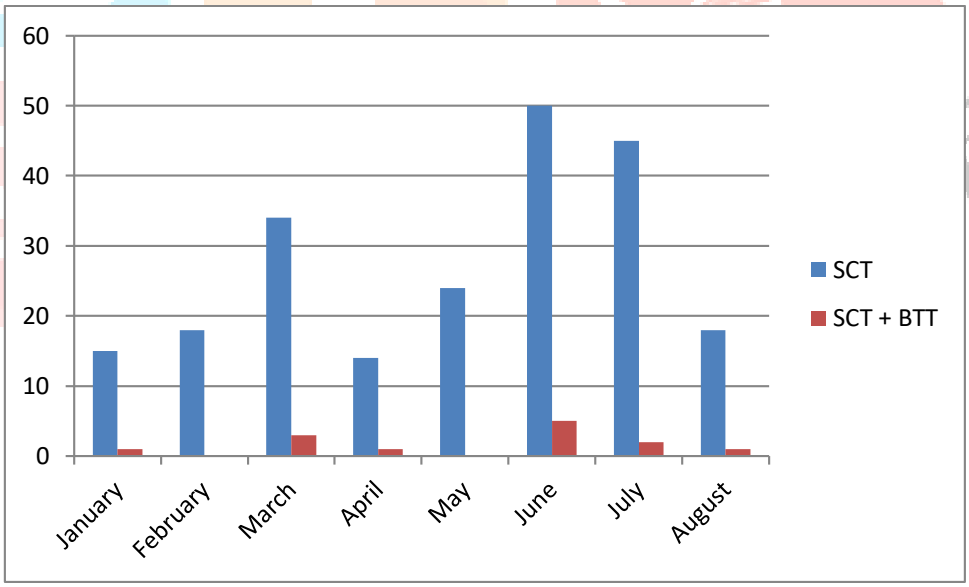

Figure – 2. Month-wise distribution of cases

We found higher number of casesof SCT(218) as compared to cases of SCT+BTT(13). Total 231 cases were detected in this time duration. .(Table – 2)

Table – 2. Month-wise distribution of cases

Month	SCT	SCT + BTT	Total number of cases	Total number of patients screened
January	15	1	16	439
February	18	0	18	849
March	34	3	37	699
April	14	1	15	516
May	24	0	24	692
June	50	5	55	945
July	45	2	47	1038
August	18	1	19	851
Total	218	13	231	6029

In our study, we found the total prevalence of SCT 3.61% and that of cases with both SCT and BTT 0.21%.

Table – 3. Prevalence of SCT and BTT

Cases	SCT	SCT + BTT	Total number of cases	Total number of patients screened
Number of cases	218	13	231	6029
Percentage (%)	3.61	0.21	3.83	100

REVIEW OF LITERATURE AND DISCUSSION

Hemoglobinopathies are a group of genetic disorders of hemoglobin. They affect 4.5% of the world population.[6] Approximately 250 million people constituting 4.5% of the world population carry a potentially pathological hemoglobinopathy gene. Each year about 0.3 million infants are born with a major hemoglobinopathy. The prevalence of β-thalassemia trait (BTT) and sickle cell trait (SCT) in India varies between 3-17% and 1-44% respectively because of consanguinity and caste and area endogamy. Hemoglobinopathies are prevalent worldwide, but it is more prevalent in some geographical areas.[6]

In India, sickle hemoglobin was first discovered by Lehman and Cutbush about 50 years ago among the tribles of Nilgiri hills of Southern India.[13] The gene is not only prevalent in tribal peoples, but is prevalent in scheduled castes and some Hindu castes.[14] According to one hospital based study, average frequency of sickle cell gene is around 5%. The highest frequency of sickle cell gene in India is reported in Orrissa(9%), followed

by Assam(8.3%), Madhya Pradesh(7.4%), Uttar Pradesh(7.1%), Tamil Nadu(7.1%) and Gujarat(6.4%). The prevalence of sickle cell trait (3.61%) in our study is in concordance with the other reported studies.[6]

Every year in India 6000 to 8000 children are born with thalassaemia major. The birth of such a child produces considerable physical and economic strain on the affected child, its family and the community at large. Thus, the emphasis must shift from the treatment to the prevention of such births in the future. The approaches to prevent the birth of a thalassemia major child include carrier screening, premarital counseling, and prenatal diagnosis.

Antenatal screening is the important step to identify women having the risk of producing a child affected with hemoglobinopathy. In India, many studies have reported the success of antenatal screening followed by prenatal diagnosis. Gujarat has many high-risk communities, which need antenatal screening and prenatal diagnosis for hemoglobinopathies.[7]

A large scale study conducted by Patel AP et al reported the prevalence of β-thalassemia trait and sickle cell trait in Gujarat to be 1.95% and 6.54% respectively.[6] This findings were similar to our study findings.

Bhatia and Rao have reported the prevalence of SCT in tribal populations in South Gujarat, which ranges from 0 to 31.4 %. The overall prevalence of BTT was found to be 3.38 % among antenatal women.[7] Out of the 210 pregnant women who were studied by Kulkarni P et al, 8.5% were thalassaemia carriers.[9]

A descriptive study conducted by Ishaq F et al concluded that prenatal knowledge about thalassemia and its preventive measures were inadequate; this requires interventions in the form of public health education progress concentrating on high/targeted population.[8]

The sickle cell anemia and thalassemia are the most severe forms of genetic disorders and hence are of great importance to be dealt with from public health point of view in India. These two forms of hemoglobin variants prevalent at higher magnitude pose a great threat to population imbalance. Therefore, these inherited abnormalities of hemoglobin synthesis are the most serious public health problem in central India in particular and in India in general reflecting the genetic heterogeneity of the population. A blood test often employed called HPLC will tell about the carrier or disease state of hemoglobinopathies.[10]

Reduction of the birth rate of thalassemia major from 1:250 live births to 1:4,000 in Turkey was reported after execution of comprehensive genetic preventive program based on voluntary screening and non-directive counseling.[11]

Proper identification of various hemoglobin variants including β-thalassemia trait can prevent occurrence of more serious disorders like thalassemia major in newborns. Thus preventing the birth of affected children is the best option for India. A prerequisite for this is the knowledge of the prevalence of β-thalassemia and other hemoglobinopathies in different regions of the country.[11]

The data regarding prevalence and distribution from our study can be useful in prevention and management of various hemoglobinopathies.

CONCLUSION

Our study concluded that sickle cell trait is more prevalent than beta thallasemia trait. Early antenatal detection followed by prenatal testing is the key to prevention of the birth of homozygous children. The various approaches for prevention include mass screening for carriers, premarital counseling, antenatal screening, and prenatal diagnosis. In India, many studies have shown that antenatal screening followed by prenatal diagnosis is the most feasible approach for the prevention of the birth of homozygous childen. Late antenatal registration, non – cooperation of patient and relatives, and refusal for prenatal diagnosis are the main hurdles in hemoglobinopathy prevention program. Public awareness regarding usefulness of antenatal diagnosis plays a very important role in prevention of birth of diseased children and ultimately to the well beingness of society. Public health education programs should be increased to raise the awareness.

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