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Original Research Paper

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A STUDY ON SPECTRUM OF HEMOGLOBINOPATHIES AMONG COLLEGE GOING STUDENTS, RAJKOT, GUJARAT

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ABSTRACT Hemoglobinopathies are among the most common inherited diseases around the world. They fall into two main groups: Thalassemia syndromes and structural hemoglobin variants. In this study we screened total 1049 college going students, among them 1026 (97.81%) participants belongs to 15-25 years, 20(1.90%) participants belongs to 26-35 years. Hemoglobin estimation was performed by automated cell counter and various hemoglobinopathies were studied by HPLC method. The study showed that out of 1049 (100%) students 126 (12.01%) students were affected with hemoglobinopathies. Total 32 (3.05%) cases of beta thalassemia trait cases were detected. We also found few other hemoglobinopathies like four cases (0.38%) of heterozygous Hb D, two cases (0.19%) of Hb Q India, two cases (0.19%) of Sickle Cell Heterozygous and one case of Hereditary Persistence of Fetal Hemoglobin. By this kind of study we can prevent the double heterozygous and homozygous state in next offspring by mass screening. Some rare hemoglobinopathies can be identified by mass screening doing HPLC method. So, it is necessary to identify heterozygous state in premarital age group by mass screening. By doing such kind of study we can reduce the burden of major diseases like thalassemia major and sickle cell disease in society.

KEYWORDS : Hemoglobinopathies, Mass screening, HPLC

INTRODUCTION

Inherited disorders of hemoglobin, hemoglobinopathies are the most common monogenic diseases. They are a group of autosomal recessive disorders that are classified into two main groups of thalassemia syndromes and structural variants of hemoglobin. World Health Organization figures estimate that 7% world population is carrier for hemoglobin disorders, leading to high degree of morbidity and mortality. [1]The cumulative gene frequency of hemoglobinopathies in India is 4.2%, with a population of over 1.36 billion and over 12,000 infants are born each year with a clinically significant hemoglobinopathies. β-thalassemia is the most common single gene disorder in our country.Hemoglobinopathies are more common in Gujarat compared to other Indian states. Model and Petrou[2] have estimated 12% incidence of major haemoglobinopathy traits in Gujarat.[3]Hemoglobinopathies such as sickle cell anemia, Thalassemias and variant hemoglobins together are responsible for the largest number of genetic diseases.The most common severe hemoglobin opathies are thalassemia major and sickle cell disease. These genetic diseases are controlled by a single gene that is transmitted from parents to offspring from one generation to another. [4]The wide variation in the clinical manifestation of hemoglobin disorders could be attributed to the influence of various genetic modifiers and environmental factors. Heterogeneous distribution of the disease and the presence of high variation in the phenotypic manifestation of a specific mutation are major problems with the development of programs for the control of the hemoglobinopathies. To reduce the birth of homozygotes and double heterozygote of these disorders, it is essential that the carrier stage detection with genetic counseling in young individuals is done by mass screening programs. [5, 6] To add to this, abnormal hemoglobins, nowadays, are genetically discovered during a systematic study performed within programs for prevention of hemoglobinopathies across the country. So, we have done observational study among the college going students in Rajkot, Gujarat. Here we also found some common hemoglobinopathies which are mostly found in Saurashtra region.

MATERIAL& METHODS

A total 1049 college going students enrolled in this study. A total volume of 2 ml of venous blood was drawn from each participant into EDTA (ethylene diamine tetra acetic acid) container for CBC Test and HPLC to diagnose hemoglobin opathies. Blood was drawn by skilled personal. Universal precautions were followed during blood collection, transportation, storage, and disposal to protect the participants as well as researchers. The blood samples were mixed for 1 to 2 minutes and there after analyzed on the Sysmex XP-100, three part differential cell counter to obtain the hemoglobin values and indices as described in the instruction manual. All samples were analyzed on the Bio-Rad Variant-II Hemoglobin system (Bio-Rad Laboratories, Hercules) as described in the instruction manual.

RESULT

In our study we enrolled total 1049 college going students. Among them 1026 (97.81%) students were between 15 to 25

years, while 20(1.90%) students were between 26 to 35 years, and remaining only three(0.29%) students only were above 36 years. Out of 1049 students screened 683 (65.11%) were male and 366 (34.89%) were female. Total 32 (3.05%) cases of beta thalassemia trait cases were detected. We also found few other hemoglobinopathies like four cases (0.38%) of heterozygous Hb D, two cases (0.19%) of Hb Q India, two cases (0.19%) of Sickle Cell Heterozygous and one case of Hereditary Persistence of Fetal Hemoglobin.

Table-1: Age wise distribution (in years):

Age (Years)	15 to 25	26 to 35	36-45	Total
Number of students	1026 (97.81%)	20 (1.90%)	3 (0.29%)	1049 (100%)

Table-2: Sex wise distribution:

Gender	Male	Female	Total
Number of students	683(65.11%)	366(34.89%)	1049(100%)

Table-3: Spectrum of Hemoglobinopathies:

Result	Total no. of Number of cases
Normal	923 (87.99%)
Borderline for β- thalassaemia Minor	85 (8.10%)
β-Thalassemia Minor	32 (3.05%)
Hb D Punjab Trait	4 (0.38%)
Hb Q India	2 (0.19%)
Sickle Cell Trait	2 (0.19%)
Hereditary Persistence of Fetal Hemoglobin	1 (0.10%)
Total	**1049 (100%)**

DISCUSSION:

Haemoglobinopathy is a hematologic disorder due to alteration in the genetically determined molecular structure of hemoglobin. The India has an extreme diverse population of 1.36 billion. It comprises numerous castes, sub castes and tribal groups in the different geographic region, each revealing different genetic traits and reservoir of hemoglobinopathies. Due to large scale migration of population and marriage across ethnic groups, the prevalence of hemoglobinopathy is quite variable.

It is estimated that about 100,000 children with transfusion-dependent thalassemia are born worldwide annually. About 8000-10,000 children with thalassemia major disease are born in India every year.[7] It is seen with highest frequency in North-West and Far East of India. Several studies in the literature have reported that Gujarat has a higher frequency of thalassemia and sickle cell disease.[8-11]

In our present study we screened total 1049 students. 1026 (97.81%) students were of pre-marital age group. So, it will be highly fruitful for him/her to know his/her heterozygous state. It is needful to prevent homozygous state of offspring by marriage of two heterozygotes.

In our present study we found beta thalassemia trait prevalence is of 3.05% which correlate with many previous studies done in Saurashtra region[12, 13, 14].The other small proportion of hemoglobinopathies indicate that asymptomatic hemoglobinopathies also present in young healthy individuals. Hb D heterozygous and Sickle cell trait frequency are same 0.38% and 0.19% as per our previous study done on prevalence of anemia and hemoglobinopathies among girls living in hostel of Rajkot respectively.[15]

CONCLUSION

The present study indicates the spectrum of hemoglobinopathies in college going students. Main aim of our study of mass screening for detection of hemoglobinopathies is to prevent double heterozygous or homozygous state in next offspring. Those found to have asymptomatic hemoglobinopathies should be given counseling regarding marriage so as to reduce the birth of children with major hemoglobinopathies like sickle cell disease and thalassemia major. Some rare hemoglobinopathies can be identified by mass screening. So it is advisable to test by HPLC method to identify such kind of a rare condition before marriage.

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