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Original Research Article

The Incidence of B Thalassemia Trait in Pregnant Women from South Western Maharashtra

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ABSTRACT

The cross sectional study was performed to find out the incidence of β-Thalassemia trait in pregnant women of south western Maharashtra from October 2007 to August 2011. The study was conducted at Government Medical College, Miraj. In this study we included 1279 pregnant women from Obstetrics & Gynecology OPD, EDTA anti-coagulated whole blood samples were collected for on-site Naked Eye Single Tube Red cell Osmotic Fragility Test (NESTROFT) testing. The screening of β thalassemia trait was done on NESTROFT with 0.36% freshly prepared saline, and β thalassemia trait status of all NESTROFT positive subjects was confirmed by quantitaion of Hb A_2 level, by microcolumn chromatography. Out of 1279 women the NESTROFT was positive for 213 subjects, and Hb A_2 level was more than 3.5% in 38 subjects, which is a determinant of β thalassemia trait. With a 3.1% rate of β thalassemia trait, this indicates need to study beta thalassemia carrier status in the child bearing group, as a primary step to prevent the birth of beta thalassemia major.

KEY WORDS: β-Thalassemia trait, NESRTROFT, Hb A_2

INTRODUCTION

Thalassemia is the commonest inherited hemoglobinopathy. In India β-thalassemia is the most common monogenic disorder. The average incidence of beta thalassemia trait in India is 3.3% with 1-2 per 1,000 couple being at risk of having an affected offspring each year. Prevalence of thalassemia trait varies form 1.0-14.9% in various regions of India. It is estimated that more than 25 million people in India, are carriers of the beta thalassemia gene and 8000 children are born every year with thalassemia major.[1] Only 10 to 15% of these children receive optimal treatment; [2] the cost of such treatment for one thalassemic child amounts to Rs. 90,000 to 1,00,000 annually at around 3 years of age, which increases as the child, grows. [3] The only cure available today is bone marrow transplantation, which is not affordable to almost all patients in India.

The birth of a thalassemic child, thus, places considerable physical, physiological and economic burden, not only on the affected child and its family, but also on the community and the nation at large. With these limitations, along with the treatment, measure for prevention of such births in the future should be undertaken. Prospective prevention, which includes population education, screening of couples in child bearing age group, genetic counseling and prenatal diagnosis, is effective way to cope successfully with such a disease. [4] As a part of prospective prevention, a study was undertaken to measure the incidence of beta thalassemia trait in the pregnant women. A screening method of NESTROF test was employed and all the NESTROF positive subjects were subjected to Hb A_2 quantitation by column chromatography.

SUBJECTS AND METHODS

SUBJECTS

The current cross sectional study was undertaken from Western Maharashtra during the period October 2007 to August 2011. The study was approved by the Institutional Ethical Committee. In this study, a total of 1279 pregnant women were recruited from PVP Civil Hospital, Sangli. The pregnant women with hemoglobin level <10 gm/dl were excluded from the study. 3 ml of blood sample was collected in EDTA bulb, and all samples were screened for beta thalassemia trait by using NESTROFT with 0.36% buffered saline solution. [5,6] The NESTROFT positive samples were subjected to Hb A_2 quantitation by micro-column chromatography. [7] Hb A_2 level more than 3.5% was taken as cut off value for beta thalassemia trait.

METHOD

2 ml of the 0.36% buffered saline solution was taken in one tube (10 cm x 1 cm diameter) and 2 ml distilled water was taken in another tube. A drop of blood was added to each tube and they were left undisturbed for 1/2 an hour at room temperature. Both the tubes were then shaken and held against a white paper on which a thin black line was drawn. The line was clearly visible through the contents of the tube containing distilled water. If the line was similarly visible through the contents of the tube with the buffered saline, the test was considered negative. If the line was not clearly visible, the test was considered positive. A positive test indicates lowered red cell osmotic fragility, suggestive of thalassemia trait, and confirmed by Hb A_2 level >3.5% performed by micro-column chromatography as described by Fairbanks V. F. et al. [7]

RESULTS

Blood samples of 1219 pregnant women were selected for the study. The samples were subjected for NESTROFT as they were available. After analyzing the data it was found that out of 1219 pregnant women, 213 showed NESTROFT positive while 1006 were NESTROFT negative. The samples positive for NESTROFT were then followed by Hb A_2 quantitaion. If the Hb A_2 level was >3.5% of the total hemoglobin, then it was considered as positive for beta thalassemia trait, and when the Hb A_2 level was <3.5%, then it was considered as negative for beta thalassemia trait. Out of 213 NESTROFT positive women 175 were having the Hb A_2 level <3.5%, therefore giving false NESTROFT positive. And 38 women were having Hb A_2 level >3.5% which were confirmed for β thalassemia trait. So out of 1219 pregnant women 38 women having the NESTROFT test positive and Hb A_2 level >3.5%, giving 3.1% incidence of β thalassemia trait in pregnant women from south western Maharashtra.

DISCUSSION

NESTROFT test was carried out as a screening test, to find out the incidence of β thalassemia trait among the pregnant women from south western Maharashtra. The NESTROFT was given positive in β thalassemia trait patients, due to abnormal osmotic fragility of red cell that could occur due to variety of reasons including iron deficiency anemia, giving rise to altered shape and functioning of red cell. The red cells whose shape has been altered due to defective genes or whose functioning has been altered due to production of certain protein in less than normal amount show the positive test. [5, 6]

Worldwide, the highest prevalence of the carrier state in descending order has been found in Sardinia (11% to 34%), [8] the delta region of the river near Ferrara (20%), [9] Sicily (10%) [10, 11] and in Bahrain (3.5%). [12] In Greece, the prevalence varies considerably, ranging from less than 5% to nearly 15% in the southern and central areas, [13, 8] as also in Cyprus. [14]

Out of 1219 pregnant women screened for NESTROFT, 213 (17.4%) reported to be positive and 1006 (82.6%) reported to be negative. After performing HbA_2 quantitaion of 213 pregnant women positive for NESTROFT, 175 women showed HbA_2 less than 3.5% where as 38 women showed HbA_2 more than 3.5%. Therefore, Out of total 1279 pregnant women, 38 were confirmed as β thalassemia trait. So, it is found that prevalence of the beta thalassemia carrier is 3.1% in the women attending ANC OPD at PVP Civil Hospital, Sangli.

The frequency of beta thalassemia carrier varies between 1 to 17 percent in different region in India with mean prevalence of 3.3 percent. [15, 16] In multicentric ICMR study, the carrier frequency was reported to be 5.5 percent in Delhi, 4 percent in Mumbai, and 7 to 8% percent in Kolkata. This calculates to about 29.7million carriers of beta thalassemia in India. Using these carrier frequencies, it can be estimated that almost one out of every 2,700 births has thalassemia major, while almost 9,000 newborn with thalassemia major are born every year. [16] Migration changing marriages pattern among ethnic group, and differences in the relative growth of population can be expected to change the distribution and prevalence of thalassemia.

The incidence of beta thalassemia trait in pregnant women of South Western Maharashtra is comparable to the overall incidence of beta thalassemia trait in India. Hence, it is necessary to consider detection

of beta thalassemia carrier status as a first prospective preventive measure, so as to prevent birth of beta thalassemia major, a disorder with considerable physical, physiological and economic burden on the family of patient and society at large.

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