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Study of Heterozygous β Thalassemia in Pregnant Women of Latur Region

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Abstract

The cross sectional study was performed to find out the incidence of â-Thalassemia in pregnant women of Latur ; Maharashtra from October 2011 to September 2013. The study was conducted at MIMSR, Medical College, Latur. In this study we screened 1186 pregnant women from Obstetrics & Gynaecology OPD of MIMSR, Latur. Whole blood Samples were collected in EDTA bulb for on-site Naked Eye Single Tube Red cell Osmotic Fragility Test (NESTROFT). 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 quantization of Hb A2 level, by HPLC machine in special investigation laboratory of GMC & JJ Hospital Mumbai. Out of 1186 women the NESTROFT was positive for 229 subjects, and Hb A2 level was more than 3.5% in 36 subjects, which is a diagnostic of â thalassemia trait. With a 3.03% rate of â thalassemia trait, urges necessity in studying beta thalassemia carrier status in the child bearing group, as a primary step to prevent the birth of beta thalassemia major.

Keywords: β-Thalassemia Trait; NESRTROFT; HB A2.

Introduction

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. Thalassemia is the commonest inherited hemoglobinopathy.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. NESTROF test was employed as a screening method and all the NESTROFT positive subjects were subjected to HbA2 quantitation by HPLC.

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Material and Methods

Study Design

The current cross sectional study was undertaken from Latur; Maharashtra.

Study Period

October 2011 to September 2013.

Ethical Approval

The study was approved by the MIMSR,Latur Institutional Ethical Committee.

Exclusion

The pregnant women with hemoglobin level <10 gm/dl were excluded from the study.

Site of Sample Collection

Obstetrics & Gynaecology OPD of MIMSR,Latur

Site of Sample Study

Pathology Laboratory MIMSR,Latur & Special Investigation Biochemistry laboratory, department of Biochemistry, J.J hospital, Mumbai.

Method

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 HbA2 quantitation by High performance Liquid Chromatography [7]. HbA2 level more than 3.5% was taken as cut off value for beta thalassemia trait.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 in NESTROFT test kit stand having white background with thin black line drawn over it. 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 A2 level >3.5% performed by HPLC.

Results

Blood samples of 1186 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 1186 pregnant women, 229 showed NESTROFT positive. The samples positive for NESTROFT were then followed by HbA2 quantitaion. If the Hb A2 level was >3.5% of the total hemoglobin, then it was considered as positive for beta thalassemia trait, and when the Hb A2 level was <3.5%, then it was considered as negative for beta thalassemia trait. Out of 229 NESTROFT positive women 193 were having the Hb A2 level <3.5%, therefore giving false NESTROFT positive. And 36 women were having Hb A2 level >3.5% which were confirmed as β thalassemia trait.

So out of 1186 pregnant women 36 women having the NESTROFT test positive and Hb A2 level >3.5%, giving 3.03% incidence of β thalassemia trait in pregnant women from Latur.

Discussion

NESTROFT test was carried out as a screening test, to find out the incidence of β thalassemia trait among the pregnant women from Latur. The NESTROFT was given positive in â 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]. 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 Latur, 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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