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A study of prevalence of thalassemia in antenatal patients in Deen Dayal Upadhyay Hospital, Delhi – A 1998 scenario

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Abstract

The haemoglobinopathies are autosomal recessive inherited disorders of haemoglobin synthesis (thalassaemias) or structure (sickle cell disorders) that are responsible for significant morbidity and mortality on a worldwide scale. Screening is necessary in pregnancy because of the complications to mother as well as fetus. NESTROFT is a suitable test for screening for beta-thalassaemia and the common haemoglobinopathies seen in India. But HPLC remains the gold standard for detection of thalassaemia. Pregnancy thalassaemias should be considered a high-risk pregnancy and requires proper antenatal and postpartum management with follow up of fetal outcome.

Keywords: Prevalence, thalassemia, antenatal patients

1. Introduction

Haemoglobinopathies are seen mainly in individuals who originate from Africa, the Middle East, the Caribbean, the Mediterranean, Asia and the Far East [4].

Individuals with trait (carriers) are healthy and unaware of their carrier status unless specifically screened. If a couple both carry a clinically significant haemoglobinopathy trait there is a 1 in 4 chance with each pregnancy that their child will inherit a major haemoglobinopathy.

Sickle cell disease (SCD) is one such blood disorder caused by the abnormal hemoglobin that damages and deforms red blood cells.

The abnormal red cells break down, causing anemia, and obstruct blood vessels, leading to recurrent episodes of severe pain and multi-organ ischemic damage.

Hemoglobin is a tetramer composed of two α -globin and two non- α -globin chains working in conjunction with heme to transport oxygen in the blood. Normal adult hemoglobin (HbA) is designated $\alpha_2\beta_2$. 3 Variant hemoglobin is derived from gene abnormalities affecting the α -globin genes (HBA1 or HBA2) or β -globin (HBB) structural genes (exons). More than a thousand hemoglobin variants have been identified relative to changes in the globin chains. Qualitative changes correspond to amino acid substitutions resulting in hemoglobinopathies. Quantitative changes like amino acid insertions, deletions or mutations in the intervening sequences (introns) correspond to thalassemia and result in decreased globin chain production [5, 6, 7].

Thalassemia is another type of blood disorder that is caused by a defect in the gene that helps control the production of the globin chains that make up the hemoglobin molecule. There are two main types of thalassemia:

- Alpha thalassemia occurs when a gene or genes related to the alpha globin protein are missing or changed (mutated).
- Beta thalassemia occurs when a beta globin gene is changed (mutated) so as to affect production of the beta globin protein.
- Beta thalassaemias occur most often in persons of Mediterranean origin. To a lesser extent, Chinese, other Asians and African Americans can be affected [5].

Thalassaemic women have an increased risk for thrombosis, as the disease entity is a chronic hypercoagulable state with high incidence of thromboembolic episodes, especially in TI, with a risk of as high as 29%, especially in splenectomized and non-transfused patients [12-15]. Where both parents are carriers of the same trait (α - α or β - β couple), genetic counseling should be performed so as to achieve prenatal diagnosis. The couple should be informed of the possibility (25%) of a TM fetus.

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The diagnosis is made either by chorionic villus sampling or by amniocentesis. Chorionic villus sampling has some advantages, as it can set the diagnosis earlier during the first trimester (11th week), more DNA is obtained by placental biopsy, and it is perhaps safer to penetrate the placenta than the amniotic cavity. On the contrary, amniocentesis has the drawback of being feasible only after the 16th week. The risk of miscarriage does not differ between these invasive procedures, and is estimated to be less than 1% [16-21].

Screening programs may differ throughout the world, depending on population needs, culture, and/or ethics, and although antenatal diagnosis remains a personal choice, policies are focused on education and counseling. Hemoglobin electrophoresis remains the gold standard for the diagnosis and classification of thalassemia. Quantitative evaluation of HbA₂ can be made by either electrophoresis or by high-performance liquid chromatography. In low resource countries, Nestroft test is used for thalassemia. Nestroft is a suitable test for screening for beta-thalassaemia and the common haemoglobinopathies seen in India. It is easy to perform, simple, inexpensive and does not require sophisticated equipment.

Nevertheless, the latter has the additional advantage of quantifying HbF at the same time. Carriers of the β -thalassaemia trait demonstrate increased values of HbA₂ and HbF [9-11].

2. Objectives

To detect the presence of Thalassemia in antenatal patients, take appropriate measures to deal with complications present or likely to develop, effectively, save the parents from emotional or psychological trauma of having a baby with severe disease by early antenatal diagnosis of carrier states in parents and prevent the birth of second affected child by preconceptional counseling of patients.

3. Study Design: Cross Sectional study

4. Study population

Study population includes pregnant females attending the OPD and IPD department of obstetrics and gynaecology in Deen Dayal Upadhyay hospital, New Delhi in 1997 and 1998.

5. Sample Size

500 antenatal patients were screened for hemoglobinopathies by Nestroft test and if found positive or doubtful for hemoglobinopathies, hemoglobin estimation by sahli's method, PCV by wint robe 's method, TLC, DLC, TRBC COUNT, MCV, MCH, MCHC, peripheral smear for type of anemia and hemoglobin variants quantification was carried out.

The patients who were positive for hemoglobinopathies, Their husbands were also screened.

If both of them came out to be positive then, the couple was to be referred for chromosomal studies and prenatal diagnosis of fetus was done early in pregnancy. The course of pregnancy and fetal outcome was studied in Thalassemic traits and compared with normal females.

6. Manuscript

In our study, 500 cases were screened by Nestroft Test, out of which, 79 cases were positive or doubtful which accounted population for 15% of the population Under Study. Out of 79 cases, 29 cases were found to be beta thalassemia trait which accounted for 6% of population under study. Thus, it helped in excluding 411 cases from undergoing unnecessary investigations. The prevalence of thalassemia varies from less than 2% to over 7% in Delhi in different ethnic groups. A higher

prevalence has been observed in Punjabis, Sindhis and delhites. A study of Indian council of medical research in school children from different parts of Delhi in 1993, the prevalence was found to be 5.5%. In our study, beta thalassemia trait was present in 6.4% of population of Punjab, 6.1% population from uttar Pradesh, 7.5% of population from Delhi, 6.5% of population from Haryana, 6.2% from Bihar and 4.5% from rajasthan. In our study, the mean hemoglobin was 9.23g %in beta thalassemia and 10.1g%in normal population. It was also seen that cases with beta thalassemia had erythrocytosis and mean RBC count of 5.4X10¹²/L. there was also a decrease in the level of MCH in beta thalassemia trait mean being 62fl. The elevated level of HBA₂ was also seen in beta thalassemia trait, mean being 52%. J.B. Pakes in 1978 [1] studied 43 Greek pregnant females. Out of them, 9 cases had beta thalassemia trait. He found that the hemoglobin in beta thalassemia traits were lower than normal. There were fall in mean hemoglobin level, RBC count and PCV during second and third trimester in patients with beta thalassemia trait as well as normal female. Alger *et al.* [2] studied 42 pregnancies in 33 women with thalassemia between July 1970 and February 1978. He found that females with beta thalassemia traits were more anemic than alpha thalassemia especially during second trimester of pregnancy. A.F. Fleming [3] in 1973 studied 15 Italian cases of pregnancy with beta thalassemia trait. He found that the mean hemoglobin, PCV and MCHC were significantly lower in beta thalassemia traits than the normal control cases. During the course of pregnancy in this study, 93% of beta thalassemia traits developed anemia. Out of them, 59% developed mild anemia, 34% developed moderate anemia. In the normal population, 56% cases developed anaemia. Out of them 49% had mild anemia and 7% had moderate Anaemia. The occurrence of pregnancy complications were similar in two groups under study. Pregnancy induced hypertension was present in 9.6% of beta thalassemia traits and 10% normal population. IUGR was present in 3.2% of normal population where as none of the beta thalassemia traits developed IUGR. 89% of the cases went into labor at term and 11% had preterm labour in beta thalassemia trait whereas 90.1% went into labour at term and 9.9% had preterm labour in normal Population. The delivery was vaginal in 83% of beta thalassemia trait. 17% delivered by caesarean section, out of which 7% was for fetal distress and 10% for non-fetal distress causes. In normal population 81% had vaginal delivery, 2% had forceps assisted vaginal delivery and 17% delivered by caesarean section out of which 7% was for fetal distress. In both the groups the caesarean section was done for obstetric indication.

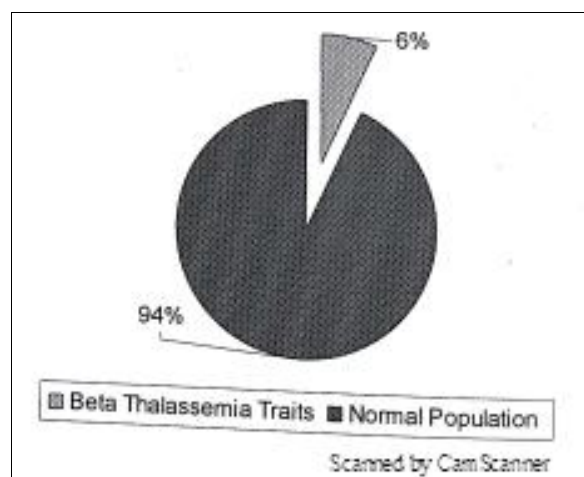


Fig 1: Distribution of beta thalassemia in population under study

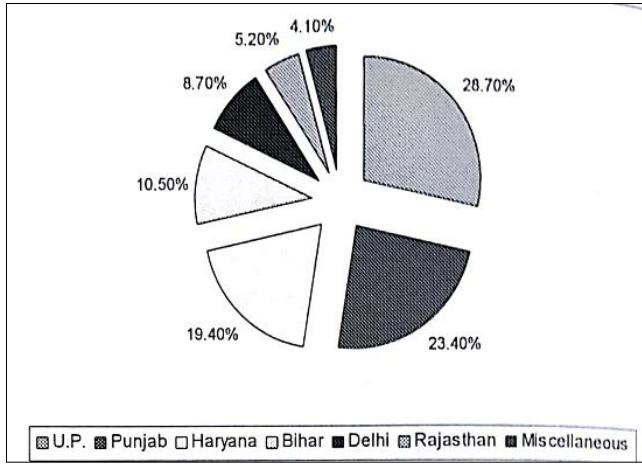


Fig 2: distribution of population according to origin

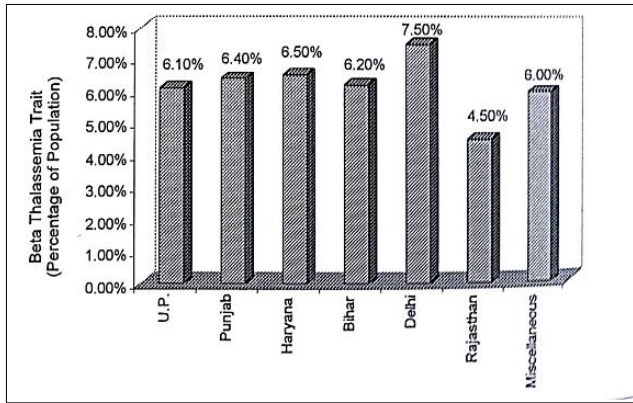


Fig 3: proportion of population with beta thalassemia trait

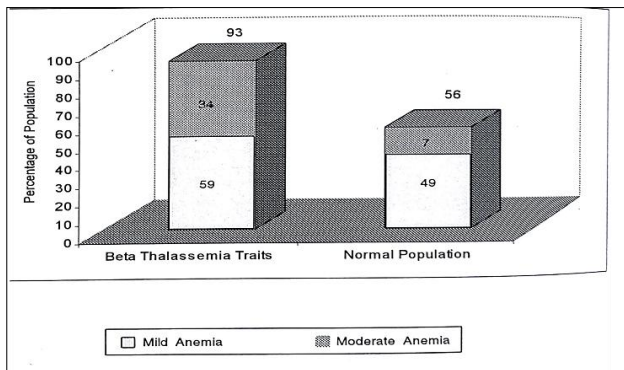


Fig 4: distribution of anemia in normal population beta thalassemia traits

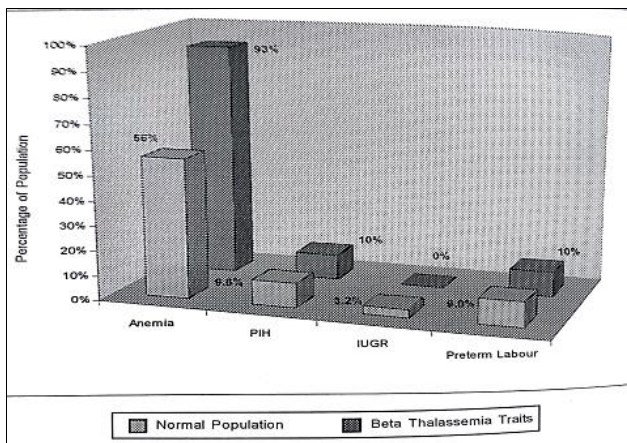


Fig 5: comparison of pregnancy Complications

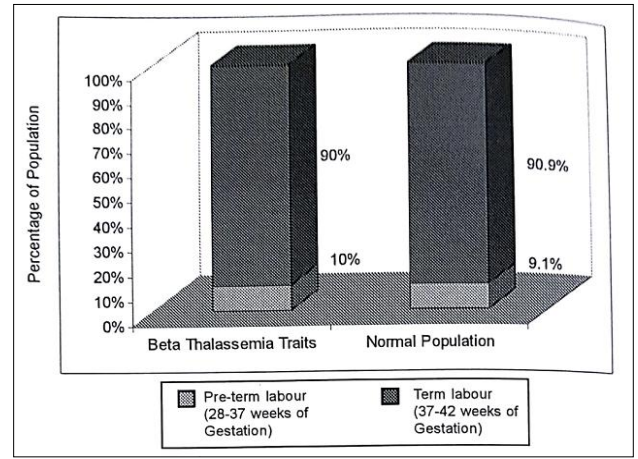


Fig 6: comparison of labour

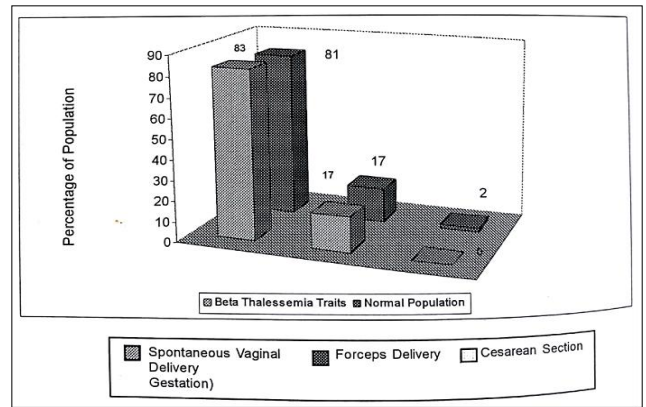


Fig 7: comparison of delivery

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