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Prevention of b-Thalassemia Major and Eb-Thalassemia by Prenatal Diagnosis in Eastern India

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ABSTRACT Prenatal diagnosis is one of the most effe ctive and direct approaches for prevention of b-thalassemia and other Hemoglobinopathies. In this paper prenatal diagnosis was offered to 21 couples 'at risk' from populations of Eastern India with a view to assess the possibility of establishing a comprehensive prenatal diagnosis service. The basis of this consideration was the high prevalence and heterogeneity of b-thalassemia and Hemoglobin E disease (HbE) in Eastern India with predominance of 7 common mutations for b-thalassemia and codon 26 for Hemoglobin E disease. It was also observed that the most common mutation for b-thalassemia was IVS1 nt5 (G->C) followed by codon 26 (G->A) for HbE disease and both these mutations were found to interact very often to produce Eb-thalassemia. DNA analysis for parents and prenatal samples was carried out using *in vitro* amplification by polymerase chain reaction (PCR) based amplification refractory mutation system (ARMS) and restriction fragment length polymorphism (RFLP). Prenatal diagnosis was offered in the second trimester of pregnancy following amniocentesis. In this study 100% diagnosis could be done for prenatal samples using these above techniques. Analysis of prenatal samples showed 36.4% affected and 63.6% unaffected pregnancies. Only 13.6% unaffected pregnancies were found to be normal while 50% were identified as carriers. The couples having affected pregnancies were counselled to terminate the pregnancies. Postnatal follow up confirmed the unaffected pregnancies. Poor awareness about the prevalence and adverse effects of this disease in the population of Eastern India was also observed in this study as 20 (95.2%) out of 21 couples 'at risk' came for prenatal diagnosis only after they had one or more affected child.

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