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Prenatal Diagnosis of Thalassaemias

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ABSTRACT In an aim to reduce the birth of children with b-thalassemia and other haemoglobinopathies with immediate effect a study was undertaken to screen the thalassemia carriers from a population of pregnant women attending the antenatal clinic of Ramakrishna Mission Seva Pratisthan hospital over a period of 5 years (1995-1999). From a total of 1962 female pregnant patients 7.04 % thalassemia carriers were identified by electrophoresis and HbA_2 estimation. NESTROFT test was also tried as another screening method as a simple and inexpensive method which showed comparable results. The husbands of these carrier mothers (138 in number) were offered carrier detection and 68.84% responded from which 15.8% carriers (15 in number) were identified. From these high risk couples only 6 agreed for prenatal diagnosis. 19 other couples were also identified as couples "at risk" from 133 referred antenatal cases who came for investigation of anaemia. Thus a total of 25 couples opted for prenatal diagnosis by DNA analysis. 26 pregnancies from 25 couples were investigated (one parent came twice). Prenatal diagnosis by DNA analysis showed 30.77% affected foetus having thalassemia mutations in homozygous or double heterozygous state while 69.23% were unaffected (foetus normal or having mutation in heterozygous state).

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