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# Clinico-Haematological Profile of Thalassemia Intermedia Patients

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**ABSTRACT** Clinico-haematological profile of 93 patients (60 males, 33 females) of thalassemia intermedia with age between 6 months to 46 years (mean 13.5) is being presented. They comprised of 76 homozygous and 17 heterozygous for b thalassemia. The mean haemoglobin at presentation was 7.4 g/dl. Anthropometric measurements in 62 children below 19 years of age revealed height less than 3rd percentile in 63.4% male and 57.1% female children while weight was less than 3rd percentile in 76% males and 81% female children. Fifty-three of 93 patients were followed up for a mean period of 3.7 years (range 1-14). Twenty-five of 53 (47%) patients had progression of thalassemic facies. Pathological fractures, osteoporotic changes, compression fracture, and gall stones were observed in 8, 3, 1 and 2 patients respectively. Fifty-four of 93 patients required infrequent blood transfusions ranging from 1-2 unit per year, while 34 patients never required blood transfusions. Serum ferritin levels were done in 16 cases which varied between 132-5600 ng/ml. Sixteen of 27 patients who were above 15 years of age or had clinical suspicion of endocrinopathies were found to have endocrine abnormalities like hypocalcemia (5 cases), hypothyroidism (3 cases), growth hormone deficiency (2 cases) and primary gonadal failure (2 cases). Twelve of 33 (36.4%) women had 19 pregnancies during the study. Ten of these had full term normal deliveries, while 5 were premature neonates. Two neonates died because of prematurity and/or infection. Four women had abortions. Nineteen patients were investigated for 5 common genetic mutations. Eleven of these patients showed compound heterozygous pattern while 7 cases were true homozygous with highest frequency of IVS1-nt-1 (G-T) mutations (44.7%). Blood transfusion policy, endocrine problems, pregnancy outcome, other therapeutic options and inheritance pattern for these patients have been reviewed.

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