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Beta-Globin Gene Mutations in India and Their Linkage to β-Haplotypes

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ABSTRACT A total of 124 chromosomes of 64 unrelated Indian β- thalassemia and β-thalassemia patients along with their family members were studied for their haplotype pattern and mutations. These included, 35 with β-thalassemia major, 4 with thalassemia trait, and 25 with β-thalassemia. Fourteen mutations were detected by PCR and Sequencing. The most common mutation IVS1-5 (G-C) was linked with 8 different haplotypes. Nineteen haplotypes were found on β-thalassemia mutations, with haplotype (+ - - - - + -) being the most widespread and was found associated with 39 chromosomes of IVS1-5 (G-C), 2 of HPFH and 1 each of CD41/42(-CTTT) and CD16 (-C).