

## **Beta-Globin Gene Mutations in India and Their Linkage to $\beta$ -Haplotypes**

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**ABSTRACT** A total of 124 chromosomes of 64 unrelated Indian  $\beta$ -thalassemia and  $\beta$ -thalassemia patients along with their family members were studied for their haplotype pattern and mutations. These included, 35 with  $\beta$ -thalassemia major, 4 with thalassemia trait, and 25 with  $\beta$ -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  $\beta$ -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).