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## The Prevalence and Heterogeneity of Beta Thalassemia Mutations in The Western Maharashtra Population: A Hospital Based Study

S.S. Ambekar, M.A. Phadke, D.N. Balpande, G.D. Mokashi, V.A. Khedkar,  
M.P. Bankar\*, P.S. Gambhir, P.M. Bulakh\* and D.G. Basutkar

*Department of Genetics and Pediatrics, \*Department of Biochemistry,  
B.J. Medical College, Pune 411 012, Maharashtra, India*

**KEY WORDS** Beta thalassemia; molecular Heterogeneity; b<sup>+</sup> thalassemia; b<sup>0</sup> thalassemia; Sb<sup>+</sup> and Sb<sup>0</sup> thalassemia in India; Beta thalassemia mutations in India.

**ABSTRACT** The present study was undertaken with the objective to study molecular heterogeneity of b thalassemia, to correlate the phenotype with the genotype in b thalassemia cases, and to know the incidence of different b thalassemia mutations in western Maharashtra population, from the cases referred to the Genetic Clinic, B.J. Medical College, Pune. Total of 114 subjects were studied. These included 85 (74.6%) known b thalassemia carriers, 25 (21.9%) cases of b thalassemia major and 4 (3.5%) cases of sickle thalassemia. Total of 58 (50.9%) males and 56 (49%) females were studied. Amongst the total 114 cases studied 102 (89.5%) showed presence of IVS 1-5 mutation, while 12 (10.5%) cases showed presence of cd 41/42 mutation. Amongst the population studied the types of b thalassemia prevalent are b<sup>+</sup> thalassemia, b<sup>0</sup> thalassemia, b<sup>+</sup>b<sup>0</sup> thalassemia, Sb<sup>+</sup> thalassemia Sb<sup>0</sup> thalassemia. Amongst these b<sup>+</sup> thalassemia is the most prevalent type. Frequency of blood transfusion (B.T.) was different in thalassemia patients with different beta globin genotypes. Hematological findings showed that presence of cd 41/42 mutation in homozygous state produces more severe phenotype than that of IVS 1-5 mutation. IVS 1-5 mutation in combination with sickle mutation produces less severe phenotype than that of cd 41/42 mutation with sickle mutation. The comparative incidence of IVS 1-5 mutation in W.M. population is higher than that reported for Maharashtra and T.N. Incidence of cd 41/42 mutation is similar to that found in Tamil Nadu and North-West Pakistan and higher than that reported for Maharashtra. The communities, which showed higher incidence of cd 41/42 mutation, are Muslim and Navbudha. An exclusive occurrence of IVS 1 5 (G-C) and cd 41/42 mutations in Western Maharashtra population will help us to prevent beta thalassemia by prenatal diagnosis.

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[Home](#)
[Back](#)


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