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Distribution of b Thalassemia Mutations and Its Correlation with a Thalassemia in Gujarati Families

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ABSTRACT We have characterized b-thalassemia mutations in 36 unrelated Gujarat (72 chromosome) families with at least one index case of severe anemia. The predominant mutations found were IVSI-5 (G-C) 61.1%, IVS I-1 (2.7%), –619bp (13.8%), CD8/9 (5.5%) and CD41/42 (8.3%). Among the less common mutations, only CD 15 (C-T) was found with a frequency of (1.3%). However, one allele of each was found for bE (CD 26 G-A) and bD (CD121 G-C) mutations. Using the amplification refractory mutation system (ARMS) technique we were able to identify mutations successfully in all the cases. We have also characterized common a-thalassemia mutations among the b-thalassemia subjects by using Gap PCR. Out of 35 thalassemia major patients 5 showed $-a^{3.7}/aa$ genotype (13.8%) and 3 had $-a^{3.7}/-a^{3.7}$ (8.3%). These findings should prove useful for suggesting the first trimester prenatal diagnosis program based on direct mutation detection.

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