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Compound Heterozygosity of β-Thalassemia Traits of HBB Gene in a Family: A Case Report

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ABSTRACT A report in a Gujarati family in Western India consisting of rare co-inheritance of â-thalassemia ($â^0$/$â^+$) in a proband, son was identified. The trio samples, parents and son, of extracted DNA from blood were subjected to gene sequence analysis, electrophoretic pattern of Hb levels and blood indices. The proband (son) showed altered levels of Hb types with higher levels of HbF (90%) and low values of mean corpuscular volume (MCV) and mean corpuscular hemoglobin (MCH) supporting â-thalassemia major. This case also possessed a compound heretozygotic condition c.92+5 G>C and c.47 G>A ($â^0$/$â^+$). Based on these markers, the proband was suggested blood transfusion by the clinician. Hence, it is suggested that this family must undergo prenatal diagnosis for the next pregnancy to avoid such risky condition.