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SCIENTIFIC LETTER

Compound Heterozygous for Asian Inversion Deletion Gγ $(Aγδβ)^0$ and IVS1-5 (G→C) β Thalassemia Mutation in a Transfusion-Dependent Patient

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To the Editor: An 11-y-old girl along with her parents and sibling was referred to ICMR-RMRC, Bhubaneswar for thalassemia variant characterization. She was transfusion dependent and presented with joint pain, pallor, splenomegaly, thalassemia facies, and anemia (Hb: 8.6 g/dL). A complete blood count (CBC) revealed low MCV and MCH [(father/ mother: 65.5/73.3 fl, brother: 73.1 fl) (father/ mother: 19.2/ 22.32 pg, brother: 12.9 pg), respectively] in parents and sibling.

The VARIANT II System (Bio-Rad Laboratories, USA) uncovered the presence of delta-beta thalassemia (δβ-thal) in the mother (Hb A_2: 2.2%, Hb A_0: 72.5%, and HbF: 16.1%) and brother (Hb A_2: 2.6%, Hb A_0: 72.7%, and HbF: 16.3%) and β thalassemia carrier in father (Hb A_2: 6%, Hb A_0: 83.3% and HbF: 1%). Since the patient was receiving transfusion every 15 d, DNA analysis was carried out by passing HPLC analysis.

DNA analysis performed using ARMS-PCR [1, 2] revealed the patient to be compound heterozygous for Asian Indian inversion deletion Gγ $(Aγδβ)^0$ with β-thalassemia IVS1-5 (G→C) mutation. While the mother and brother were heterozygous for Asian Indian inversion deletion Gγ $(Aγδβ)^0$ and the father was β-thalassemia heterozygote with IVS1-5 (G→C) mutation.

δβ-thal, is a rare cause of increased fetal hemoglobin (HbF) levels in adult life. Thus, this case is being reported because it is the first report from the Yadav caste subsequent to the report of the same variant from the Chasa caste in Odisha [3] suggesting wide prevalence of this variant among different Odia populations and hence needs screening of such rare variants in the state.

Declarations

Conflict of Interest None.

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