Molecular Genetics, Genomics, Mechanisms of Diseases

AB128. The first report of a hereditary persistent fetal hemoglobinemia of the Southeast Asian type deletion in the North of Vietnam

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Background: Beta thalassemia is one of the most common single gene disorders worldwide and also in Vietnam. In this study, we reported, for the first time, a hereditary persistent fetal hemoglobinemia (HPFH) of the Southeast Asian (SEA) type deletion in the North of Vietnam.

Methods: Four individuals from a family with Kinh ethnic background were studied. Hematological data were obtained by standard methods. Genomic DNA was extracted from peripheral blood by using Qiagen blood mini kit. β-globin gene amplification, multiplex ARMS PCR, DNA sequencing and duplex Gap PCR system with three primers bridging the 3' breakpoint were performed.

Results: The father had normal hemoglobin A2 and elevated fetal hemoglobin levels. His genotype was heterozygous for HPFH of SEA type. The mother was heterozygous of point mutation IVS2-654C>T. The first child carried no mutations, however the second child was compound heterozygous for the IVS2-654C>T mutation and the HPFH of SEA type deletion. Even though, this child has no severe clinical conditions.

Conclusions: Based on the present case, we conclude that the clinical features of HPFH of SEA type are milder than the β° type of point mutation.

Keywords: Beta thalassemia; deletion; hereditary persistent fetal hemoglobinemia (HPFH)

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