Molecular Genetics, Genomics, Mechanisms of Diseases

AB131. Case report: a couple having a child affected with beta thalassemia major and another child with hydrops fetalis

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Background: Thalassemia is one of the most common single-gene disorders worldwide. There are two major types of thalassemia: alpha and beta thalassemia. In this study, we reported a case of concurrent alpha and beta thalassemia disease.

Methods: This is a case report. A healthy couple had their first child with beta thalassemia major who was treated by blood transfusion once a month. However, the fetus of their second pregnancy was diagnosed of having hydrops fetalis of unclear etiology and the couple decided to have pregnancy terminated at 26 weeks of gestation. The couple then were the referred for genetic counseling at National Children's Hospital. Complete blood counts (CBC), high performance liquid chromatography (HPLC) and molecular testing (ARMS-PCR; Gap-PCR; C-ARMS-PCR) were applied to detect red blood cell profiles, Hb typing, and common gene mutations of alpha and beta thalassemia.

Results: The findings of the husband specimens were as following: RBC 5.25×10^9/L, Hb 13.7 g/dL, MCV 81.1 fL, MCH 26 pg, HbA2 5.7%, HbA 94.3%, and heterozygote for αα/–α3.7 mutation in the *HBA* gene, and heterozygote for Cd71/72(+A) mutation of the *HBB* gene. The wife's results were as follow: RBC 4.34×10^9/L, Hb 108 g/dL, MCV 77.7 fL, MCH 24.8 pg, HbA2 5.8%, HbA 94.2%, heterozygote for αα/--SEA mutation in the *HBA* gene, and heterozygous for the CD41/42 mutation in the *HBB* gene.

Conclusions: Genetic testing is necessary for accurate diagnosis of thalassemia carrier which would lead to more precise genetic counseling.

Keywords: Alpha thalassemia; beta thalassemia; case report; *HBA* gene; *HBB* gene

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