

## MOLECULAR DIAGNOSIS OF HB WOODVILLE [ $\alpha$ 1 6(A4) ASP>TYR].

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**Introduction:** Hemoglobin (Hb) Woodville [ $\alpha$ 2 or  $\alpha$ 1 6(A4) Asp > Tyr] was first reported in 1986 in a Vietnamese female lived in Australia during antenatal and cord blood screening programs for families of South East Asian origin and a Thai female with Chinese ancestry has reported, during premarital screening for thalassemia and haemoglobinopathies due to the detection of an increased HbA2 approximately 10%. This Hb variant is caused by the substitution of an aspartic acid by a tyrosine residue at position 6 of the  $\alpha$ 1- or  $\alpha$ 2-globin.

**Methods:** We recently observed this variant in heterozygous form in a Taiwanese subject. The subject was a 24-years-old health female admitted at the Department of Obstetric and Gynecology of CMUH for premarital screening. CBC data was collected using a full-automated blood cell counter and Hb analysis was done with electrophoresis by automated weak cation-exchange high-performance liquid chromatography (HPLC) compared with alkaline electrophoresis (pH 8.6) on cellulose acetate gel to separate every hemoglobin fraction and to detect Hb variants. Genomic DNA was purified from peripheral blood leukocytes using a standard method.

**Results:** The hemogram showed: Hb 12.2 g/dL, RBC  $4.69 \times 10^{12}/L$ , MCV 78.3 fL, MCH 26.2 pg, MCHC 33.5 g/dL, and ferritin 13.6 ng/mL. Electrophoresis of freshly prepared hemolysates in cellulose acetate at pH 8.6 electrophoresis showed remarkable change, this variant migrated to a position near the position of Hb F. No point mutation was detected in the  $\alpha$ 2-globin gene expect for some know polymorphism. The results showed a G > T substitution at the first base of codon 6 of  $\alpha$ 2-globin gene (GAC>TAC) that resulted in replacement of tyrosine for aspartic acid.

**Discussion:** The rare  $\alpha$ -globin variant can be confirmed by sequencing analysis of  $\alpha$ 2-globin gene product. Several other Hb variant are also at this region include Hb Dunn [ $\alpha$ 2 or  $\alpha$ 1 6(A4) Asp>Asn], Hb Swan River [ $\alpha$ 2 or  $\alpha$ 1 6(A4) Asp > Gly], Hb Ferndown [ $\alpha$ 2 or  $\alpha$ 1 6(A4) Asp > Val], Hb Sawara [ $\alpha$ 2 or  $\alpha$ 1 6(A4) Asp > Gly].

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3] Chang JG, Liu HC, Shih MC, Liu SC, Chan WL, Tsai FJ. Hemoglobin 2002; 26: 91-94.