

Compound heterozygosity for Hb D-Punjab / β -thalassemia and blood donation: case report

Hb D-Punjab / β -talasemi ve kan bağıışı için bileşik heterozigozite: olgu raporu

Stamatia Theodoridou¹, Michael Alemayechou¹, Parthena Perperidou¹, Clio Sinopoulou¹, Theano Karafoulidou¹, Georgia Kiriakopoulou¹

¹Hemoglobinopathy Prevention Unit, Hippokration Hospital, Thessaloniki, Greece

²National Thalassemia Center, Laikon Hospital, Athens, Greece

To the Editor,

The Hb D gene [β 121 (Glu→Gln)] is encountered mainly in India, Pakistan and Iran. The greatest prevalence is found in the Sikhs of Punjab (~2.5%), and sporadic cases are found in peoples of European and African origin. Hb D-Punjab (or Hb D-Los Angeles) is encountered very rarely in the Greek population. Individuals with compound heterozygosity for Hb D-Punjab / β -thalassemia have mild to moderate disease [1-5]. A subset of patients may manifest moderately severe anemia with splenomegaly and chronic hemolysis, while others may show very mild disease with no apparent clinical signs. Hb levels range between 8 and 12 g/dl. Patients primarily have Hb D and a small percentage of Hb A depending on the type of β -thalassemia mutation (β^+ or β^0). Hb A2 levels may be normal or increased while Hb F levels are usually normal, although slightly increased in a few cases.

We report the case of a 32-year-old Greek male blood donor who was found to have Hb values of 13.9 g/dl on the HemoCue test before his third whole blood donation. Because of the high prevalence of β (beta)- and α (alpha)-thalassemias in the Greek population and the existence of so-called "silent" β -thalassemia mutations, hematological indices were determined using an automated Coulter cell counter (Beckman Coulter;

Fullerton, CA, USA). The sample was further screened for thalassemias and hemoglobinopathies when low red blood cell (RBC) indices were detected. The white blood cell (WBC) and platelet counts were within the normal limits. Screening for thalassemias and hemoglobinopathies was carried out by microscopic examination of a stained peripheral slide, osmotic fragility tests, measurement of serum ferritin, cation exchange high performance liquid chromatography (HPLC) for Hb variants quantitation, alkaline and acid pH electrophoresis, sickle test, and DNA analysis when required. The laboratory results were Hb: 13.9 g/dl, Hct: 43.3%, MCV: 67.3 fL, MCH: 21.7 pg, and RDW: 15.2%. Ferritin level was 35 ng/ml. The HPLC showed hemoglobin in D window of 79%, Hb A of 17.9%, Hb A2 of 2.3%, and Hb F of 0.8%. Electrophoresis showed properties of an Hb D variant. Physical examination did not reveal a palpable spleen.

The results showed that the individual was a compound heterozygote for Hb D / β -thalassemia. The presence of Hb D-Punjab was confirmed by polymerase chain reaction (PCR) amplification followed by digestion with EcoRI DNA analysis. Concomitant analysis of the β -gene showed that the propositus carried the Hb D Punjab + IVSI -110 (G→A) mutation.

The prevalence of β -thalassemia is around 8% in the Greek population, while 2% are carriers of the Hb S

gene. The gene for Hb D is rare. During the 22 years of thalassaemia screening in our Thalassaemia Prevention Unit in northern Greece, 30 cases of heterozygotes for Hb D, 1 case of compound heterozygote for Hb S / Hb D-Punjab and 1 case of compound heterozygote for β -thalassaemia / Hb D-Punjab were detected, among 80,401 subjects screened. Our Thalassaemia Prevention Unit covers the regions of central and western Macedonia, in northern Greece, with a population of around 2.5 million.

Hb D/ β -thalassaemia patients have a disease that has clinical manifestations ranging from mild to moderate disease, resembling either thalassaemia minor or thalassaemia intermedia [4].

Our case can be characterized as exceptionally mild, because the individual was not aware of the disease until it was found on routine pre-donation Hb measurement for his third whole blood donation. He is healthy in appearance and had donated on two previous occasions without untoward effects. He is now exempted from whole blood donation for fear of long-term consequences, although one can argue that, similar to those with the thalassaemia trait, this individual could be kept on a program of long-term blood donation if it is infrequently

done and if Hb values are stringently followed. It is also equally important that the use of such RBCs for transfusion be studied. This report confirms the benign nature of co-inheritance of Hb D-Punjab and β^+ -thalassaemia.

References

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