Abstract

Hemoglobin is a tetrameric protein composed of two pairs of globin chains and four heme groups. Its main function is to realize oxygen transport from lungs to other body tissues.

Hemoglobinopathies are characterized by presence of mutations in the globin genes. These mutations can result in synthesis of hemoglobins with abnormal structure (variants of Hb) or in reduced synthesis of globin chains (thalassemias).

In Unidade de Anemias Congénitas e Hematologia Molecular, Centro Hospitalar e Universitário de Coimbra (CHUC), a reference center for red blood cell disorders, were identified some individuals with following described alpha variants of Hb, Hb J-Paris-I, Hb Hirosaki, Hb G-Pest, Hb Toulon, Hb Setif, Hb Groene Hart, Hb Plasencia and Hb J-Camaguey. Additionally were identified individuals with not described variants of Hb, characterized by following mutations α40(C5)(Lys→Asn)(AAG>AAT (α1) and α104(G11)(Cys→Arg)(TGC>CGC) (α2). These two mutations were designated in laboratory by Hb HUC and Hb Iberia, respectively.

By study of the described variants, *in silico*, was verified the effect of mutations in the structure and function of hemoglobin. This initial study helped to predict the effect of the not described mutations identified in laboratory.

The mutations that characterize Hb HUC and Hb Iberia occur in the contact α1β2 and in the contact α1β1, respectively. At functional level we verified that Hb HUC is a stable variant with slightly high affinity for oxygen (in presence of 2,3-diphosphoglycerate and Hb Iberia is a stable variant.

Key words: Hemoglobin, hemoglobinopathies, variants of Hb and thalassemias