Fifteen Cases of Hb J-Meerut: The Rare Association with Hb E and/or HBA1: c.-24C>G (or HBA2) Variants

Khalil MSM, Timbs AT, Henderson SJ, Schuh A, Old JM

Abstract:

ABSTRACT Hb J-Meerut [HBA2: c.362C>A (or HBA1)] is a rare, stable, nonpathogenic α-globin gene variant that peaks in the area between the P3 and A0 windows on high performance liquid chromatography (HPLC). Few cases from different ethnic origins have been published but the majority were Asian Indians. Coinheritance with other hemoglobin (Hb) variants are rarer and can change the Hb J-Meerut phenotype making a diagnostic dilemma. In this study, we have reported 15 cases of Hb J-Meerut, discovered during a wide spectrum study of α-globin chain variants in the UK. The diagnosis was confirmed by forward and reverse DNA sequencing of the α1- and α2-globin genes. The average of the Hb J-Meerut expression was 20.9% of total Hb and characterized by a retention time (RT) of 1.9 min. (on average) on HPLC. The median of isoelectric focusing (IEF) was 5.6mm above Hb A. Among the 15 cases studied, one case coinherited the Hb E (HBB: c.79G>A) mutation in heterozygosity and another case was associated with the Cap þ14 (C>G) [HBA1: c.-24C>G (or HBA2)] variant. We noticed that the coinheritance of the Hb E mutation reduced the Hb J-Meerut expression with the formation of a hybrid peak missed on the HPLC chromatograph. We also noticed an increased expression of Hb J-Meerut in the case showing the coinheritance of the HBA2: c.-24C>G (or HBA1) variant.

Keywords:

α-Globin gene variants; Hemoglobin (Hb); HBA2: c.-24C>G (or HBA1); Hb J-Meerut DOI: 10.1080/03630269.2020.1817755

Published In:

Hemoglobin. 2020 Sep 14:1-4. , 14 , 364-367 (1-4)