Rare Haemoglobin Variant Hb J Meerut in 27 Years Old Female - A Case Report

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Abstracts: Hemoglobin has plenty of variants and fast moving hemoglobins (FMH’s) are the rare hemoglobin variants. They are having tendency to migrate anodally to hemoglobin A on alkaline gel electrophoresis. Because of the mutation in the globin genes, these hemoglobin variants have the fast moving nature. The basic pathophysiology behind it is the substitution of a negatively charged amino acid residue in either α, β or γ globin chains. Hb J Meerut is an infrequently found α-globin variant. It has previously been reported in various populations around the world. Here, we are reporting a case of Hb J meerut who came to laboratory for thalassemia screening. [Rathod G NJIRM 2014; 5(5):108-110]

Key Words: Hb J Meerut, Alkaline gel electrophoresis, fast moving hemoglobins, Globin genes.

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Introduction: Hemoglobin has plenty of variants and fast moving hemoglobins (FMH’s) are the rare hemoglobin variants. They are having tendency to migrate anodally to hemoglobin A on alkaline gel electrophoresis. Because of the mutation in the globin genes, these hemoglobin variants have the fast moving nature. The basic pathophysiology behind it is the substitution of a negatively charged amino acid residue in either α, β or γ globin chains. Hb J Meerut is an infrequently found α-globin variant. It has previously been reported in various populations around the world. Haemoglobin J family comprising of Hb J Meerut/Hb J Birmingham (α) ¹, Hb J-Bangkok (β) ²Hb J Baltimore (β) ³ and many more. A majority of fast moving hemoglobins (FMH’s) of J family are not showing any clinical significance and therefore are detected accidentally during family or antenatal screening by alkaline gel electrophoresis and/or CE-HPLC. Here, we are reporting a case which came for thalassemia screening and diagnosed as Hb J Meerut.

Case report: A 27 years old Indian female came to laboratory for thalassemia screening. Her native was Delhi – the city situated in north India. She was not having any symptoms of hemolytic process. Her hematological parameters were as follows: Hb – 8.9 g/dl, RBC – 4.91 million/cumm, PCV – 39.7%, MCV – 80.9 fl, MCH – 18.1 pcg, MCHC – 22.4%, RDW – CV – 24.5%. All these hematological parameters were recorded by fully automated KX-21 hematology analyzer, Japan. In hemoglobin electrophoresis of the same blood sample by fully automated HPLC – BIO-RAD D-10, USA, HbJ Meerut was detected. There was peak within the P3 retention time window (P3 – 18.7%) with HbA₂ – 1%, HbA – 62%, HbA1c – 5.5%, HbF <0.8%, LA1c/CHb – 1 – 0.0% and LA1c/CHb-2 – 2.7%. (Figure 1, 2)

Figure 1: Presence of Peak Within The P3 Retention Time Window (P3 – 18.7%).
Discussion: At present, hundreds of different variants of hemoglobin are known. The main etiology for this is structural alteration of alpha, beta or gamma globin chains varying from amino acid replacements, elongated chain, deletions, insertions, or both deletions and insertions. The clinically significant hemoglobinopathies are developed when there is abnormality of beta-chain or alpha chain. The expression and deletion of the disorder fairly depends upon the status of zygosity. In heterozygous variants, the other normal allelic gene produces normal chains which may compensate for the defective gene. In the homozygous state, both allelic genes are affected which results in the production of a large amount of the variants.

Thorup et al. had first described Haemoglobin J (Hb-J) in an African-American patient (1956) and since then more than 50 variants of Hb-J were described. Formation of Hb J-Meerut is due to a C -> A mutation (GCG->GAG) at codon 120 of the α1 or α2 globin gene, changing the alanine to glutamic acid at residue 120 of the α chain. The very first case with Hb J-Meerut was reported in two sisters from Meerut, Uttar Pradesh, India and in two brothers from Bangladesh living in Birmingham, England. Subsequently the same abnormal hemoglobin, was described in one Japanese family and in one Turkish family. Srinivas et al. reported 7 cases of Hb J Meerut from India.

Some of Hb-J variants have abnormal properties and affect respective hematologic indices, whilst majority of them do not result in any abnormal clinical manifestation. The measurement of the oxygen equilibrium curves of Hb J Meerut showed a slightly increased oxygen affinity. The same findings were noted in our patient also.

Nowadays, Electrophoresis and CE HPLC are the important diagnostic tools for hemoglobinopathies. Electrophoresis is the screening test for detecting hemoglobin variants in India and other developing countries. Quantification of these hemoglobins are done by CE HPLC from their characteristic RT’s. Use of alkaline gel electrophoresis is also justifiable because fast moving Nature of Hb J variants is demonstrable only on alkaline gel electrophoresis at pH 8.6. Thus, both these techniques are equally gracious in recognition, quantitation, and categorization of the hemoglobin variant.

One important clinical condition which may lead to a wrong diagnosis of FMH’s is uncontrolled diabetes mellitus. Glycosylated hemoglobin (HbA1C), though not generally considered as true fast hemoglobin variants, should be kept in mind before giving impression of FMH’s. The CE-HPLC can differentiate it from inherited FMH’s such as Hb J-Meerut by their characteristic RT’s.

Conclusion: As Hb J-Meerut is a rare variant of abnormal hemoglobin and not associated with any clinical significance, such cases are detected during family and antenatal screening. Electrophoresis and CE HPLC are the gold standard tool for the validate diagnosis.

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