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Case Report

Rare association of Hemoglobin variant Hb J (α mutation) with haemophilia A: case report

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ABSTRACT

Hb J α mutation is a rare haemoglobin variant. No case of Hb J α mutation with hemophilia A has been reported. Here we are reporting a rare variant of Hb J α mutation found accidentally in our patient in which electrophoresis done to find out the cause of severe anemia with hepato-splenomegaly and association of this hemoglobin variant with Hemophilia A.

Keywords: Electrophoresis, Haemoglobin variant

INTRODUCTION

Hemoglobin J α mutation is an alpha chain variant found in heterozygous state and presents normal haematological blood picture. Mostly the patients are silent carrier and asymptomatic with this variant of hemoglobin. Hemophilia A is an inherited, X linked recessive disorder caused by deficiency of functional plasma clotting factor VIII. There is a rare association of these two different entities, found in our patient.[1]

CASE REPORT

We are reporting a case of 30 months old male child 1st issue of non-consanguineous marriage of healthy parents had complaints of fever and cough for 1 month. Child also had a history of delayed clotting of blood even after trivial trauma. No prior history of hospitalization and any blood products transfusion.

Family history of similar bleeding tendency in maternal uncle (documents not available). Birth history was non-significant. History of pica was present. On examination patient had severe pallor, hepato-splenomegaly liver size of 2 cm and spleen size of 2 cm palpable below costal margin. weight 9 kg, OFC 46 cm, Height 83 cm.

On Investigation, complete blood count revealed microcytic hypochromic anemia with mild anisopoikilocytosis (Hb 4.5 gm/dl, Hematocrit 17.6, mild thrombocytopenia platelet counts 1.36 lacs) and malaria parasite not detected by PBF. His coagulation profile was deranged (PT 17.5 seconds/14.6 seconds, APTT 95.0 seconds/34.8seconds) and iron studies showed (raised Ferritin 724.8 ng/ml, low serum iron 37.08 µg/dl and normal TIBC 320.58 µg/dl).

HB electrophoresis done to rule out thalassemia showed normal HbA 74.6%, Hb A2 1.5%, HbF 1.6% while raised HBP3 levels 22.3% suggestive of Hbj alpha mutation. On the basis of history suggestive of bleeding disorder factor VIII levels were done showed very low levels of factor VIII <3%.

Other biochemical parameters were within normal range.

Patient was treated as clinical malaria with oral chloroquine and given blood transfusion in form of PCV

to correct anemia. Also factor VIII was given for the prophylaxis. Patient improved with treatment.

DISCUSSION

Fast moving hemoglobin's (FMH) are the rare haemoglobin variants. FMH of J family revealed about 48 fast moving Hb J variants. Fast moving haemoglobin predominantly are α globin derived.[1] Hemoglobin J-Rajapppen (alpha) 90 Lys to Thr is an alpha chain variant found in heterozygous state and present normal haematological picture. Hb J is a structural hemoglobinopathy. These is clinically silent and mostly diagnosed accidentally.[2]

These variants can be discovered by high performance liquid chromatography (HPLC), electrophoresis and by mass spectrometry.[3,4]

Hemophilia A is X linked recessive hereditary bleeding disorder caused by lack of clotting factor VIII. Males are the affected one and females are the carrier. Patients presents with bleeding into joints, bruising, nasal bleed, prolonged bleeding from tooth extracts, cuts and surgery. Tests used to perform to diagnosed hemophilia A includes prothrombin time, partial thromboplastin time, bleeding time, fibrinogen levels and Factor VIII levels.[5]

Usually the patients are asymptomatic and not require blood transfusion by this hemoglobin variant. So, no specific treatment required for this. While in case of hemophilia A Factor VIII requirement is there to maintain the normal levels so that spontaneous bleeding can be prevented.

Hemoglobin J α variant is itself a rare and accidental finding when electrophoresis is done to find out the cause of anemia. So, it is the rare association found in our patient having Hb J (α mutation) with haemophilia A.

CONCLUSION

RBC indices, electrophoresis, family history is sufficient to detect and manage the most of the haemoglobin variant and X- linked recessive disorder in our country. Genetic studies are indicated to confirm borderline cases and to detect the silent carrier.

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