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Case Report

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A Case of a Rare Hemoglobinopathy Causing Symptomatic Anemia

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Case Description

A 35-year-old African-American woman presents with 3 days of nausea, vomiting and abdominal pain, associated with subjective fever, chills, and 3 episodes of syncope with prodromal dizziness. She has a brother with sickle cell disease and father who passed away from syncope of unknown etiology. Her vital signs were normal, and abdominal exam was significant for LLQ and suprapubic tenderness without signs of acute abdomen. Her labs were revealing for severe microcytic hypochromic anemia. Abdominal ultrasound revealed fibroid uterus.

She underwent anemia workup and was found to have no identifiable Hb-A or Hb-B. Hemoglobin electrophoresis interestingly found a rare compound heterozygote with mutation in each beta globin gene, one of which was sickle hemoglobin. It showed Hb S of 37%, Hb D of 56%, Hb F of 1 %. Genomic DNA and nucleotide sequencing revealed she possessed an unusual combination of hemoglobin Korle-Bu, hemoglobin D-Punjab, and sickle cell trait. The patient was given a blood transfusion and then treated for severe iron deficiency with IV iron. Her uterine fibroids were managed with oral contraceptives.

Laboratory Results

CBC

WBC: 7.5 thou/cumm
Hemoglobin: 5.4 g/dl
MCV: 59.8 FL
MCH: 17 PG
MCHC: 28.3 G/dl
RDW: 19.5%
Platelet: 274 thou/cumm
Neutrophil: 83.8%
Lymphocytes: 11.7%
Monocytes: 3.1 %
Eosinophils: 0.1%

Hemoglobin analysis:
Hemoglobin S: 37%
Hemoglobin D: 56%
Hemoglobin F: 1%

Iron Panel

Iron: 19 UG/dl
Iron sat: 4 %
TIBC: 461 MCG/dl
Ferritin: 3.9 NG/ML
Total Bilirubin: 0.2 mg/dl

BMP

Na: 139 mmol/L
K: 4.0 mmol/L
Cl: 104 mmol/L
CO_2: 22 mmol/L
BUN: 5
Creatinine: 0.7 mg/dl
Glucose: 101 mg/dl
Calcium: 9.2 mg/dl
Magnesium: 2.0 mg/dl

Imaging

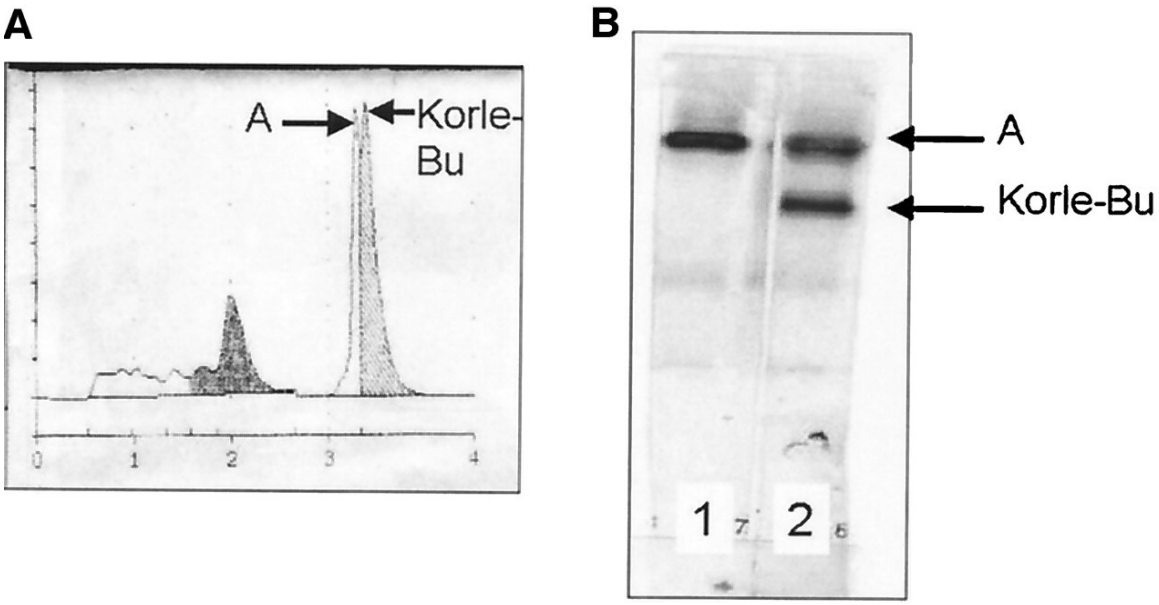

Figure 1: HbA2 vs. Hb Korle-Bu on high-performance liquid chromatography and electrophoresis1.

Treatment

- Patients with hemoglobinopathies should be counseled to maintain adequate hydration, especially during physical activity
- They should be treated with blood transfusions for symptomatic anemia, with emphasis on preventing anemia by treating sources of excess blood loss such as uterine fibroids
- It should be noted that individuals with sickle cell trait are at increased risk of thrombosis with hormonal birth control
- Symptoms often improve by treating the anemia

Discussion

- Hemoglobinopathies are a form of genetic defects that result in the abnormal formation and composition of the globin chains
- Approximately 7 % of the world's population is affected by hemoglobinopathies
- The genetic variations are inherited single-gene disorders frequently found in ethnic populations of Africa, Southeast Asia, and the Mediterranean
- Sickle cell disease is one of the more common forms, while Hemoglobin (Hb) Arab, Lepore, Korle-Bu, Kansas, D. Punjab, and Hasharon are some of the unusual variants
- The combination of Korle-Bu, Hb-D-Punjab and sickle cell trait as seen in this patient is extremely rare. There has been two pediatric cases and no adult cases previously described in literature.
- Hb D is clinically silent in its heterozygous form, but coinheritance of Hb S with Hb D or beta-thalassemia manifests like thalassemia intermedia and may result in hemolytic anemia and splenomegaly
- Hb Korle-Bu is a rare beta-chain variant due to a mutation in codon 73 of Hb gene (GAT à AAT) causing a single amino acid substitution in the Beta chain (Asp à Asn)
- Hb Korle-Bu prevails in West Africa. Hb Korle-Bu along with Hb S defect chain tends to present like sickle cell trait rather than a modified sickle cell disease. Therefore, treatment of Hb Korle-Bu with heterozygous Hb-S is similar to that of sickle cell trait.

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