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Case Report

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A CASE REVIEW OF SICKLE CELL WITH BETA THALASSEMIA

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ABSTRACT

Interaction of Hb S with beta thalassaemia is a very rare case, that has been reported in our institution. Sickle cell disease (SCD) is caused by a mutation in the sixth codon of the -globin gene on chromosome 11. which result in to a single amino acid substitution (β6 glu→val). In India, the Hb S is prevalent in the central part, in the eastern, western and southern tribal belt regions and among the tea tribe communities of Assam. Sickle-thalassemia is a rare variant of sickle cell disease (delta-beta thalassemia occurring in association with sickle hemoglobin, HbS), sparsely reported in literature. We describe a patient who presented with fever and abdominal pain with a negative family history for sickle cell disease. The HPLC report of the patient showed Compound heterozygous for Hb S- β thalassaemia. As this is a rare variant of SCD with potential complications, it is important to establish diagnosis towards planning comprehensive care.

INTRODUCTION

Sickle Cell Disease is a multisystem disease. Which is associated with multiple episodes of acute illness and progressive organ damage. It is the one of the most common severe monogenic disorders worldwide. We report a case of Sicilian $(\delta\beta)^0$-thalassemia, which is a rare variant of Sickle Cell Disease. HbS.β thalassemia is a double heterozygote state of HbS and β thalassemia. Clinical features and hematologic findings are determined by β thalassemia gene. Clinical pictures are resembles that of thalassemia intermedia. Like mild growth retardation, pallor, splenomegaly, vasoocclusive crises, leg ulcers and aseptic necrosis of femoral heads. Peripheral smear shows microcytic hypochromic RBC, basophilic stippling and target cells. MCV and MCH are decreased. There is increase in Hb F and Hb S levels. Prognosis is better than that of thalassemia major or sickle cell anemia.

CASE PRESENTATION

A 4 yrs old male patient, 1st issue of 2nd degree consanguinity, admitted at Dr. DY Patil Medical college, Kolhapur, Maharashtra, India,with following complaints.

Fever since 4 days, high grade with chills, no diurnal variation, temporarily relieved by medication, fever became mild since last night.

Abdominal pain since 3 days. Gradual onset mild vague pain in the left hypochondrial region. Associated with loss of appetite.

On examination

Severe pallor+, mild ictreus+
Frontal bossing+, flat nasal bridge+
Hr 126/min
Rr 28/min
PP Well felt
Bp 100/60 mmhg (in btw 50th and 90th percentile)
Systemic examination

Per Abdomen

Soft, nontender, liver just palpable, spleen palpable 7cm below LCM

Other system are within normal limit.

We sent CBC which show Hb 8.1g/dl,Tlc 8300/mcrl, Tec 3400000/mcrl

PCV 26.5%, MCV fl, MCH 23.6pg, MCHC 30.6gm/dl. RDWCV 22.2%. Peripheral smear shows 97% Nucleated RBC. So the blood parameters are in favor of thalassemia intermedia. So we sent sample for high-performance liquid chromatography (HPLC). The result shows.

Foetal Haemoglobin (HbF) 22.5 %, Haemoglobin A0 (Hb A0) 3.6 % 94%, Haemoglobin A2 (HbA2) 4.0 %, Haemoglobin S (HbS) 69.9%. Surprisingly this reports are suggestive of Sickle Cell Disease (? Homozygous HbS) (? HbS - Beta Thalassaemia). To confirm this we did parental HPLC.

HPLC report of father shows Foetal Haemoglobin (HbF) 0.3 % Haemoglobin A0 (Hb A0) 55.7 % Haemoglobin A2 (HbA2) 3.2 % Haemoglobin S (HbS) 40.8 % which Suggestive of Sickle Cell Trait.
HPLC report of mother shows Foetal Haemoglobin (HbF) 0.4 % Haemoglobin A0 (Hb A0) 95 % Haemoglobin A2 (HbA2) 4.6 % which Suggestive of Beta Thalassaemia Trait.

So the both parentel report suggestive of Father is sickle trait and mother is thal trait so Patient is double heterozygous, that is Sickle thalassemia.

DISCUSSION

δβ-thalassemia is characterized by decreased or absent synthesis of the delta- and beta-globin chains with a compensatory increase in expression of fetal gamma-chain synthesis. The condition is found in many ethnic groups but is most common in Greece, Italy, Middle east and India.

Homozygotes for *δβ*-thalassemia have 100% HbF and, because of the increased synthesis of HbF, may have thalassemia intermedia rather than thalassemia major. The heterozygous form of the condition phenotypically resembles B thalassemia trait but HbA2 is often normal while HbF is elevated varying from 5% to 20%.

Since homozygous -thalassemia presents an identical HPLC finding as homozygotes of hereditary persistence of fetal hemoglobin of 100% HbF, the clinical findings of mild hemolytic anemia rule in favor of -thalassemia rather than HPFH. Family studies also play a role in eliciting the correct diagnosis (thalassemic features). Sicilian $(\delta\beta)^0$-thalassemia presents a deletion of 13,379-bp spanning *δ*-IVS2 to a region located 3′ from the *β*-globin gene within an L1 repeat.

Sickle-$(\delta\beta)^0$-thalassemia is a rare Sickle Cell Disease. variant that has been sparsely reported worldwide . These cases were described to have mild microcytic anemia, as well as Sickle Cell Disease complications which include multiple episodes of VOC (in some cases this occurred prior to diagnosis), osteomyelitis, multifocal avascular necrosis, cholelithiasis, and osteonecrosis.

In our case also the patient had severe anaemia, weakness and complained of abdominal pain. On physical examination hepatosplenomegally was observed in the patient. Only after our diagnosis report, the parents of the patient became aware about their Hb variant carrier state.

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