Case Report

A Unique case of pregnancy with E beta Thalassemia having history of recurrent pregnancy loss

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Abstract :
Recurrent pregnancy loss defined as two or more consecutive failed clinical pregnancies documented byultrasound or histopathology. Patient need careful monitoring and evaluation for better prognosis.In thiswe discuss a case of G3P0+2 with known case of beta thalassemia with history of recurrent pregnancy lossand its management through out the pregnancy.Cause of pregnancy loss if diagnosed early and managedwith strict followup may give better outcome

Key words : Recurrent pregnancy loss,Thalassemia.

Introduction

The Hb E/â-thalassaemia genotype accounts for around half of all severe beta-thalassaemia cases globally, with the highest frequencies being recorded in India, Bangladesh, and throughout Southeast Asia.[1,2]The disorder is characterized by wide clinical diversity, ranging from moderate and asymptomatic anaemia to a life-threatening condition necessitating transfusions from infancy. â-thal/HbE illness has been linked to increased risk of foetalgrowth restriction, premature birth, and low birth weight. The effects are more significant in the â-thal0/HbE subgroup. Pregnancy may be safer for women with â-thal/HbE illness.[3]This case report describes an unusual complication of the disease in women.

Case report

A 26year old G3 P0+2 with known HbE betathalassemia presented at 13 weeks of POG with weakness and yellowish discolouration of sclera and passage of mustardcoloured urine. The present conception was spontaneous. LMP was 14.02.2023 & EDD was 21.11.2023.The past medical history showed multiple blood transfusion since childhood while obstetric history revealed spontaneous miscarriage at 12 weeks and 9weeks in the last 2 years.On general examination moderate pallor and icterus was seen. Abdominally the uterus was just palpable above symphysis pubis and also splenomegaly of grade 3 seen. The per vaginal examination showed uterus to be anteverted, 12 weeks size approximately with bilateral fornices free. The complete antenatal profile done and revealed Hb to be 5.6 g/dl with microcytic hypochromic anemia with serum ferritin levels around 351mcg/dl; iron: 151 mcg/dl; transferrin saturation: 71.85%; TIBC: 210.16mcg/dl; Total bilirubin: 4.2gm/dl; Direct bilirubin: 1gm/dl; TSH: 5.5miu/ml; ferriscan showed hypointensity of liver with diffuse iron deposition. Other investigations, like FBS, PPBS, ECG and ECHO were normal. USG confirmed splenomegaly of 218mm with single live fetus of 13weeks 2 days.

The patient was managed conservatively with strict follow up every 3months for Hb & iron stores and every 2eeks post 28weeks for USG. The 1st& 2nd trimester was uneventful but at 30weeks she presented with cough and distress for which

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she was admitted in HDU considering high risk pregnancy with SpO_2 86% on room air. In HDU patient was diagnosed with pulmonary tuberculosis for the cough and distress after proper evaluation and was started on ATT which improvedher condition dramatically. At 37weeks patient underwent LSCS for fetal distress with MSL in transverse lie. A healthy baby boy of 2.5kgs was delivered. Both mother and baby were discharged in stable condition.

Discussion

HbE is a variant hemoglobin with a mutation in the â-globin gene, causing substitution of glutamic acid for lysine at position 26 of the â-globin chain. HbE is the second commonest abnormal hemoglobin after sickle cell hemoglobin (HbS).[4]HbE is common in Southeast Asia where its prevalence can reach 30-40% in some parts of Thailand, Cambodia, and Laos. HbE is also found in Sri Lanka, Northeast India, Bangladesh, Pakistan, Nepal, Vietnam, and Malaysia.[5]HbE-â-thalassemia is not uncommon in India. Recent studies from West Bengal have shown that the prevalence of â-thalassemia carriers varies from 3.7 to 10.3% and Hb E carriers from 3.02 to 4.0%.[6,7,8]In the North eastern region, there is an extremely high prevalence of HbE being 30 to >50% in some tribal groups while the frequency of â-thalassemia carriers varies from 1.5 to 3.48%.[9] In both these regions around 1% of anemic individuals referred for hemoglobin analysis by HPLC had HbE-â-thalassemia.

Around 15% of HbE-â-thalassemia patients in Southeast Asia are clinically mild and do not require regular blood transfusions.[10]However, it is difficult to predict whether the affected fetus will have a mild or severe clinical presentation as there are no genetic markers for the same. This results in a dilemma while offering genetic counselling to couples at-risk. Chakrabarti et al,[11]showed that in West Bengal, patients who received optimum management through unrestricted funding from corporate houses as a part of corporate social responsibility had a significantly better quality of life than those in routine care. However, this is possible only for around 15% of patients and majority of patients with â-thalassemia major or HbE-â-thalassemia are poorly transfused and not chelated.Pregnancy with HbE thalassemia is associated with potential morbidity for the mother and the fetus, requiring regular antenatal care of obstetricians in collaboration with hematologists and thus needs to be monitored and followed up regularly till safe delivery of mother and baby is ensured.

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