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Case Report

Non transfusion dependent thalassemia of pregnancy: a multifaceted approach to maternal and fetal health

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ABSTRACT

Non-transfusion dependent thalassemia (NTDT) patients typically remain asymptomatic until certain triggers, such as pregnancy, necessitate transfusions and increase the risk of pregnancy-related complications if not managed appropriately. We present the case of a 38-year-old female with NTDT, splenectomised at age six, who presented at 17 weeks of gestation with weakness and bipedal edema. She was advised to undergo regular blood transfusions and take aspirin, targeting a haemoglobin level of 9 g/dl. The patient was lost to follow-up and she returned later to underwent an emergency caesarean section at 32 weeks due to absent fetal movements and absence of end-diastolic flow. Postpartum, she experienced thromboembolism and was diagnosed with severe osteoporosis, treated with low molecular weight heparin, calcium, and vitamin D supplements. Pregnancy exacerbates anemia and thrombosis risk in thalassemia patients, leading to complications such as intrauterine fetal death, growth retardation, and spontaneous abortions. Aspirin or low molecular weight heparin, along with blood transfusions, are often required to prevent these complications. Iron chelating agents, contraindicated during pregnancy due to teratogenic risks, are typically recommended post-breastfeeding. Thalassemia also impacts other body systems, including cardiovascular, gastrointestinal, and skeletal systems. Therefore, a multidisciplinary approach with a well-planned treatment regimen and comprehensive family planning counselling is essential for NTDT patients.

Keywords: Thalassemia, Pregnancy, Non-transfusion dependent, Management

INTRODUCTION

Thalassemia is an inherited blood disorder that affects over 19,000 children born worldwide each year having various phenotypes. More than half are sufferers who are highly blood transfusion dependent. These children can have varying severities. Some, diagnosed with β-thalassemia intermedia or mild-moderate haemoglobin E/β-thalassemia, do not require regular blood transfusions (known as NTDT).[1-3] NTDT has three main forms: β-thalassemia intermedia, α-thalassemia intermedia, and mild/moderate haemoglobin E/β-thalassemia.[4] These children either experience no or have mild symptoms throughout their lives. However, pregnancy can act as a stressor, worsening their condition due to physiological changes. This can lead to complications like intrauterine growth restriction, preterm labor, miscarriages, and even fetal death.[5]

CASE REPORT

Patient profile

The patient was a 38-year-old primigravida having a past history of a splenectomy at age 6 without any available health records. She remained asymptomatic for the next 20 years. After age 26, she experienced episodes of generalized weakness and low-grade fever, treated with multivitamins and paracetamol.

Clinical course

In April 2023 (at 17 weeks gestation), she presented with bilateral pedal edema and generalized weakness. Examination revealed hepatomegaly and pallor. Laboratory tests showed low haemoglobin (7.4 gm/dL), elevated platelets, and low albumin. An initial diagnosis of splenectomy-induced thrombocytosis was made, and she was started on aspirin and folic acid.

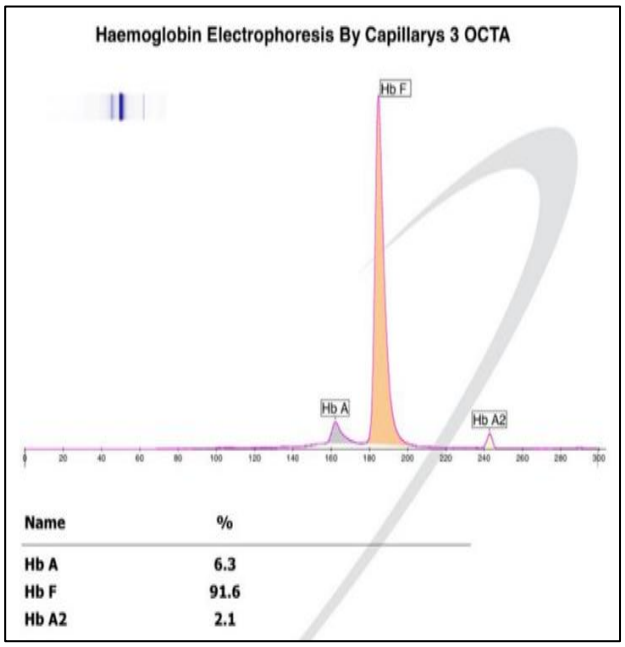

Figure 1: Haemoglobin electrophoresis by capillarys 3 OCTA.

Haemoglobin electrophoresis confirmed NTDT (β-thalassemia intermedia). She received one unit of packed red blood cells (PRBCs) and was advised for regular transfusions to reach a target haemoglobin level (9 gm/dL). Aspirin and folic acid were continued, and she was advised for regular prenatal check-ups.

She received a total of 3 PRBC units (2+1) at 3–4-week intervals. Her symptoms improved, leading to non-compliance with further transfusions and she lost to follow up. At 32 weeks she reappeared in OPD, an ultrasound revealed severe compromise of the fetoplacental circulation with absent end-diastolic flow. An emergency caesarean section delivered a very low birth weight baby (1.5 kg). She took an early discharge and went 300 km away to her in native place due to social issues along with poor compliance of medications.

Seven days later, the patient presented with stormy appearance. She had right-sided neck swelling, abdominal distension, pedal edema, and weakness. Doppler studies confirmed a jugular vein thrombus. Further investigations revealed severe pulmonary artery hypertension and right-sided heart failure. A DEXA scan showed severe osteoporosis (Z-score of -2.7).

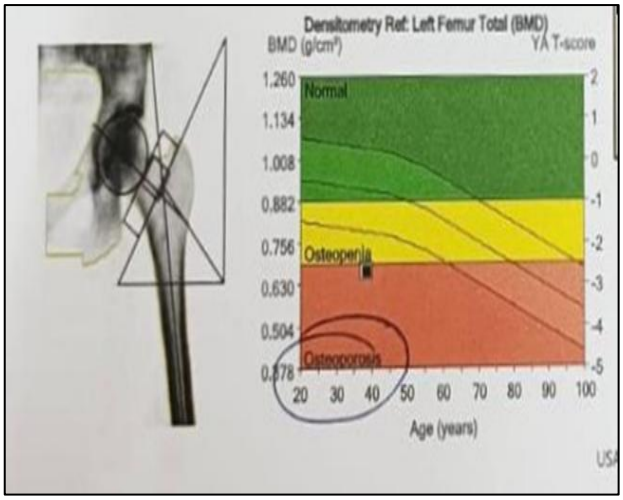

Figure 2: DEXA scan.

Management

The patient received PRBCs (3 units), deferasirox (iron chelation), hydroxyurea (to reduce ineffective erythropoiesis), and apixaban (anticoagulant) for 7 days. Vitamin D and calcium supplements were initiated for osteoporosis. Due to breastfeeding, apixaban was replaced with low-molecular-weight heparin, and hydroxyurea and deferasirox were discontinued due to potential effects on the baby. At 6 weeks and 3 month follow up patient was doing well.

DISCUSSION

NTDT patients typically don't require lifelong transfusions. They may be asymptomatic or mildly anaemic with haemoglobin levels between 7-10 gm/dL.[5] However, stressors like pregnancy can lead to occasional transfusion dependence. In rare cases, poor growth and development may necessitate more frequent transfusions.[6]

During pregnancy, physiological anaemia due to plasma volume expansion and increased demand for fetal growth can worsen pre-existing anaemia in NTDT patients. This can necessitate blood transfusions.[7,8] Chronic anaemia leads to hypoxia which can adversely affect the uteroplacental circulation, potentially leading to complications like IUGR, preterm labor, and intrauterine fetal death. In such case to avoid hypoxia blood transfusion remains the key.[4]

Though blood transfusion is game changer in feto maternal outcome; the main concerns with blood transfusions in NTDT pregnancies are not limited to alloimmunization but iron overload and quality of life matters. The risk of developing antibodies against transfused red blood cells increases with subsequent transfusions, potentially complicating future pregnancies and leading to miscarriage.[9] While frequent transfusions and increased gastrointestinal iron absorption in NTDT can lead to iron

overload. Iron chelation agents like deferasirox have potential teratogenic effects and are usually started after breastfeeding stops.

One more risk is Thromboembolic phenomena due to deadly combination of pregnancy and NTDT. Hormonal and physiological changes during pregnancy enhance the risk of blood clots. This risk is further increased in NTDT patients, especially those who are splenectomised.[10,11] Blood transfusion may play preventive role by reducing pathological RBCs and platelets having hyper thrombogenic properties. Hence the decision to transfuse should be made after careful patient evaluation.[12]

CONCLUSION

Close monitoring of mother and fetus with the help of team approach including haematologist, cardiologist, fetal medicine expert, and neonatologist. Judicious use of blood transfusion, preventive treatment path against infection, stress and hypoxia; along with medical, family and social support remains cornerstone of successful management. Apart from crucial genetic counselling, throughout pregnancy holistic counselling also play major supportive role.

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