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FOLLOW-UP OF A PREGNANT WOMAN WITH SPHEROCYTOSIS: CASE REPORT

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Abstract: Hereditary spherocytosis (HE) is a rare genetic condition that can cause hemolytic anemia due to defects in the red blood cell membrane. This case report describes a pregnant woman with HE, highlighting the importance of high-risk prenatal care and the challenges faced during pregnancy. The patient was treated with folic acid and ferrous sulfate supplementation, requiring postpartum transfusion. This clinical case provides valuable insights to improve care for women with HE during pregnancy and the postpartum period, aiming to diagnose complications early and prevent risks for the mother and fetus.
Keywords: Hemolytic anemia; Pregnancy complications; Hereditary Spherocytosis

INTRODUCTION

Although relatively rare, hereditary spherocytosis (HE) is the most common genetic cause of hemolytic anemia due to a defect in the red blood cell membrane.[1]

Its prevalence varies between 1:2000 and 1:5000 people and is due to inherited mutations or mutations that induce quantitative and/or qualitative changes in the membrane of the erythrocyte cytoskeleton.[1] The most frequently altered protein is ankyrin-1 (ANK-1 gene), transmitted by an autosomal dominant pattern, followed by spectrin and band-3.[1–3]

The clinical presentation of HE is variable and can be classified from mild to severe, the latter being characterized by severe anemia (hemoglobin 6 – 8 mg/dL) and hemolytic complications such as jaundice, splenomegaly and thrombosis.[1] Splenectomy is the most effective treatment in severe cases, although it is associated with an increase in thrombotic events.[1] In the literature, rare cases of pregnant women with HE are described.[1] After stating this, this clinical case aims to contribute to current knowledge about HE during pregnancy and the importance of preconception counseling and supervision in high-risk prenatal care, highlighting potential complications associated with the disease during the gestational period.[1,3]

METHODOLOGY

Compile data from the literature available on the UpToDate and Pubmed platforms, using 9 materials dated from 1992 to 2023, with data on hereditary spherocytosis and the gestational period, of which a brief introduction was made on the topic and comparing procedures and outcomes with the case of the patient, E.G.S, 22 years old, pregnant, primigravida and diagnosed with hereditary spherocytosis in childhood.

REPORT OF CASE

Pregnant woman, E.G.S, 22 years old, blood type O+, primigravida, undergoing high-risk prenatal care (PNAR) since 27 weeks and 1 day of gestational age (GA) by early ultrasound (USG), referred due to the diagnosis of hereditary spherocytosis (EH) in childhood. Furthermore, the patient denies a family history of hemolytic anemia.

In this sense, E.G.S states that he has been undergoing monthly monitoring at a blood center for around 5 years and that he receives blood transfusions when necessary. Taking folic acid supplementation of 5 mg/day and ferrous sulfate 40 mg/day. Furthermore, the patient did not have other comorbidities.

After initial assessment by the PNAR team, the prescriptions were maintained, in addition to being admitted for outpatient follow-up at the service with a request for laboratory and imaging tests, as the physical examination revealed 3+/4+ jaundice associated with the presentation of several blood counts. compatible with hemolytic anemia (low levels of hemoglobin (HB), between 7 and 8.5 mg/dL, increased indirect bilirubin (BI), elevated lactic dehydrogenase (LDH) and negative right coombs. During follow-up, the patient

remained if jaundiced and with abnormal laboratory tests. In obstetric ultrasounds and Doppler flowmetry, no fetal abnormalities were detected. In the 30th week of gestation, lung maturation was performed with dexamethasone (6 mg/mL intramuscularly (IM) every 12 hours for 48 hours).

At 31 weeks and 6 days of GA, the patient attended an obstetric emergency consultation at the service, in premature labor associated with broken water, and was admitted for tocolysis with nifedipine (loading dose: 30 mg orally (PO) + 20 mg PO every 8 hours for 24 hours); antibiotic prophylaxis with ampicillin (2,000 mg loading dose + 1,000 mg intravenously (IV) every 4 hours until delivery); and new lung maturation. Upon admission, laboratory tests were collected, which showed: HB: 9.0 g/dL; hematocrit (HT): 28.4%; mean corpuscular volume (MCV): 116.7 fL; mean corpuscular hemoglobin (MCH): 36.9 pg; leukocytes: 16,510/mm^3; platelets: 265,000/mm^3; total bilirubin (BT): 8.6 mg/dL; direct bilirubin (BD): 2.29 mg/dL; BI: 6.3 mg/dL; and LDH 492U/L. Approximately 8 hours after admission, delivery was performed by cesarean section due to prematurity and low fetal weight (estimated fetal weight 1,189g on USG with a GA of 27 weeks and 6 days), with fetal neuroprotection being performed with magnesium sulfate, according to protocols from the service. The newborn (NB) was born weighing 2,160g and with an Apgar score of 3, 5 and 7 in the 1st, 5th and 10th minutes, respectively. The newborn was evaluated by the neonatal team and admitted to a neonatal intensive care unit (ICU).

During the postpartum period, the patient progressed well. On the 3rd day postpartum, the patient, without complaints, presented the following laboratory tests: HB: 7.2 g/dL; HT: 22.5%; VCM: 118.8 fL; HCM: 37.9 pg; RDW (erythrocyte distribution width): 13.4%; platelets: 237,000 /mm^3; BT: 4.59 mg/dL; BD: 2.5 mg/dL; BI: 2.10 mg/dL; and LDH: 510 U/L. Therefore, a transfusion of a bag of packed red blood cells was indicated by the hematology team.

He was discharged the following day, with supplementation of ferrous sulfate 40mg/day and folic acid 5mg/day, in good general condition and with hemodynamic stability, presenting laboratory tests with continued improvement in hematimetric levels. Therefore, she was referred for follow-up at a postpartum outpatient clinic. Finally, the patient appeared on the 56th post-operative day at the puerperium outpatient clinic accompanied by the infant, with no anemic symptoms or jaundice, and was discharged for follow-up at a blood center.

DISCUSSION

Hereditary Spherocytosis (HE) is a spectrum of hematological diseases related to an autosomal dominant inheritance pattern (recessive inheritance of variable expression can also occur) and, although it is a rare manifestation, it is the most common etiology of hemolytic anemia, as well as can cause intermittent jaundice, splenomegaly and cholelithiasis.[4] This pathology is a consequence of mutations in one of the five genes encoding red blood cell membrane proteins, which are associated with links between the cytoskeleton and the lipid bilayer.[1]

HE during pregnancy is extremely rare, with a prevalence of 0.02% to 0.05%.[1]

Therefore, publications about pregnancies complicated by HE are few, and recommendations for postpartum women are not documented. Therefore, there is uncertainty about maternal and fetal outcomes, so this clinical case provides support to improve pre- and postnatal care for women with HE. Some cases reported in the literature demonstrate a high risk of pregnancy loss, while others present favorable

fetal prognoses.[4,5] A clinical case refers to the diagnosis of fetal growth restriction in a pregnancy complicated by HE, similar to the case presented in the present report.[6]

The severe clinical manifestation of this pathology occurs in the form of fatigue, chest pain, pallor and dyspnea, accompanied by intermittent jaundice, gallstone disease and splenomegaly.[7] The diagnosis is confirmed, if there is hemolysis, through a negative direct Coombs test, increased mean corpuscular hemoglobin concentration levels, positive family history and/or peripheral blood smear with spherocytes.[1] In cases of diagnostic uncertainty, additional confirmation is necessary using osmotic fragility tests, Eosin 5-Maleimide binding (EMA) or glycerol lysis.[1]

Mild cases of the pathology do not require treatment, while moderate to severe cases can be treated with folic acid, blood transfusions and/or splenectomy, which helps resolve anemia and is extremely efficient.[8] Patients of any severity who plan to become pregnant and during pregnancy must be treated with folic acid 2 to 4mg/day.[1]. Furthermore, splenectomy is associated with improved obstetric outcomes, however, its performance during pregnancy increases the risks of premature birth, cesarean section, pneumonia, anesthesia complications, severe neonatal thrombocytopenia and the need for transfusions.9 Thus, despite differing from the cases present in the literature, the patient was treated with folic acid 5 mg/day and ferrous sulfate 40 mg/day prenatally, as well as, in the postpartum period, she received a transfusion.

Finally, given the low rate of pregnancies complicated by HE described in the literature, this clinical case contributes to the improvement of care during pregnancy and the postpartum period, thus enabling the prior diagnosis of changes in fetal growth and prevention of complications.

POTENTIAL CONFLICT OF INTEREST

I declare that there is no relevant conflict of interest.

FINANCING SOURCE

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