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Case Report

Obstetrics and Gynaecology Section

Hereditary Spherocytosis in Pregnancy- A Case Report

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ABSTRACT

Hereditary Spherocytosis (HS) is the most common inherited red cell membrane disorder included under intracorpuscular defect. The HS has both autosomal dominant and autosomal recessive inheritance. The disease may be mild, moderate or severe and requires a multidisciplinary approach for its management during pregnancy. Hereby, authors present a case of a 30-year-old female G2P1L1 at 34 weeks+4 days presented with anaemia not responding to iron therapy since first trimester. Peripheral blood smear revealed red blood cell's were normocytic and normochromic. Spherocytes were seen with mild polychromatophilia and diagnosed as a case of HS and its management was done with haematological support.

Keywords: Anaemia, Haemolysis, Multidisciplinary approach, Splenectomy

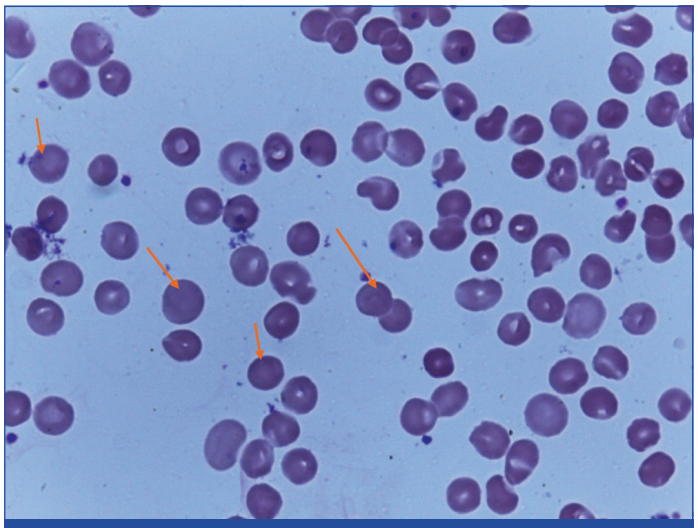
CASE REPORT

A 30-year-old female G2P1L1 at 34 weeks+4 days presented to high-risk antenatal clinic with anaemia not responding to iron therapy since first trimester. She had mild anaemia (haemoglobin was 9.9 gm/dL) in her first trimester. During her late second trimester, she had breathing difficulty, palpitation and fatigue during normal activity and was found to have moderate anaemia (haemoglobin was 8.3 gm/dL). The patient was advised parenteral iron therapy and two months later her Haemoglobin (Hb) was 8.6 gm/dL. Then she was given two units of packed cell transfusion and her Hb was raised to 10.8 gm/dL during the third trimester. All obstetric ultrasounds and doppler done were normal. There was no evidence of foetal anaemia. She had a history of anaemia during the postpartum period of the previous pregnancy despite no history of any postpartum haemorrhage for which she had received two units of packed cell and parenteral iron and it was not evaluated further. The patient's mother had a history of Hereditary Spherocytosis (HS) at 29 years of age and underwent splenectomy.

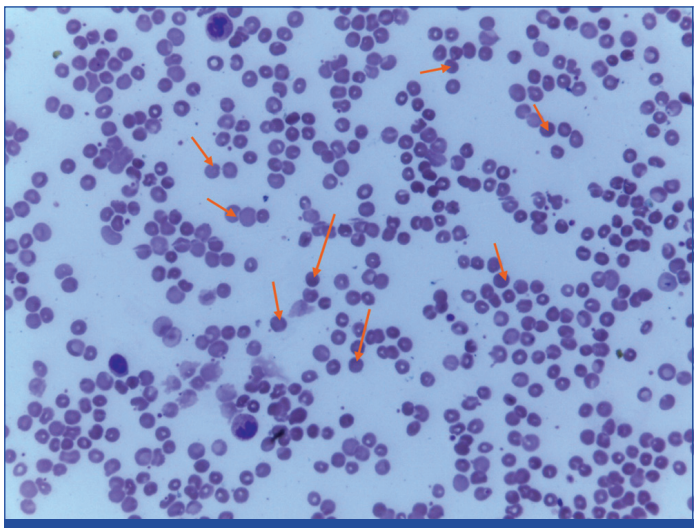
On examination, she had mild pallor and jaundice. Abdominal examination revealed a gravid uterus with fundal height corresponding to 32-34 weeks with a live active baby in a longitudinal lie and cephalic presentation. Investigations are presented in [Table/Fig-1]. Peripheral blood smear revealed Red Blood Cell (RBC) were normocytic and normochromic. Spherocytes were seen with mild polychromatophilia [Table/Fig-2,3].

Parameters	First trimester	Second trimester	Third trimester
Haemoglobin (gm/dL)	9.9	8.3	10.8
Packed cell volume (%)	27	-	30.4
Platelet (lacs/mm^3)	3.42	-	2.03
Mean corpuscular volume (fL)	-	-	87.1
Mean corpuscular haemoglobin (pg)	-	-	30.9
Mean corpuscular haemoglobin concentration (gm/dL)	-	-	35.5
Total bilirubin (mg/dL)	-	-	1.62
Lactate dehydrogenase (U/L)	-	-	294
Total iron-binding capacity (mcg/dL)	-	-	372.9
Reticulocyte (%)	-	5.03	7.9
Serum ferritin (ng/mL)	-	230	435.8
Vitamin B12 (pg/mL)	-	-	358
Serum folate (ng/mL)	-	20.46	-

[Table/Fig-1]: Blood investigation results.



[Table/Fig-2]: Peripheral blood smear of HS (arrows indicate the spherocytes). (Leishman stain; 40X).



[Table/Fig-3]: Peripheral blood showing HS (Multiple arrows indicate spherocytes). (Leishman stain; 10X).

Osmotic fragility test/Eosin 5 maleimide (EMA) was increased, Antinuclear Antibodies (ANA) was negative and C3 and C4 were

normal. Ultrasound abdomen showed mild splenomegaly and spleen measuring 12.8 cm, mildly enlarged in size and normal echotexture. The liver measures 14.8 cm normal in size shows mildly coarse echotexture. No other pathology was detected. A diagnosis of pregnancy with HS was made in view of peripheral blood smear and other haematological investigations. A multidisciplinary approach was sought including the physician, haematologist, anaesthetist, and neonatologist. As it was a case of previous caesarean section and no other contraindications, the patient was counselled for the trial of labour after caesarean, the risks involved, and the benefits were explained to the patient and relatives and finally, they opted for elective caesarean section.

After proper counselling regarding the need for blood transfusion, risk of postpartum haemorrhage and the chance of the foetus developing HS she underwent elective caesarean section with sterilisation and delivered a term healthy baby. Intrapartum and postpartum periods were uneventful. Her postoperative haemoglobin was 9.8 gm/dL, she was counselled regarding an iron-rich diet and the need for iron supplementation. The initial assessment of the baby was done by the neonatologist and was normal. Both mother and baby were advised to follow-up for the same. During the postnatal visit after six weeks of delivery, haemoglobin of the mother was 10.2 gm/dL.

DISCUSSION

Haemolytic anaemias are defined as the increased destruction of RBC outside the bone marrow resulting in shortened RBC lifespan. It is classified as intracorpuscular defects and extra corpuscular defects. The population incidence among Caucasians is 1:2500-5000 [1]. The sphere-shaped red blood cells present are less resistant to stress and rupture easily leading to chronic haemolytic anaemia [2]. About 75% of cases of HS are inherited as an autosomal dominant pattern. In some cases, an affected person inherits the mutation from one affected parent. In other cases, no history of the disease in the family result from new mutations in the gene. It can also be inherited in an autosomal recessive pattern also [3]. It is caused by a deficiency in red blood cell membrane proteins, ankyrin and spectrin [4].

Clinical presentation varies from asymptomatic mild anaemia to fulminant haemolytic anaemia with pallor, jaundice and splenomegaly requiring aggressive treatment. Family history includes the history of splenectomy or cholecystectomy. Triggers such as fatigue, stress, emotional distress, and pregnancy can unmask the previously compensated disease. It is diagnosed by anaemia, increased reticulocyte count, Mean Corpuscular Haemoglobin Concentration (MCHC), spherocytes in the peripheral smear, elevated bilirubin and Lactate Dehydrogenase (LDH) levels, and increased osmotic fragility test. The confirmative test is the Sodium dodecyl-sulfate polyacrylamide gel electrophoresis (SDS-PAGE) [1]. The disease may be mild, moderate, or severe based on various haematological parameters and the need for splenectomy as shown in the [Table/Fig-4] [2].

Classification	Trait	Mild	Moderate	Severe
Haemoglobin (gm/dL)	Normal	11-15	8-12	6-8
Reticulocyte count (%)	Normal (<3)	3-6	>6	>10
Bilirubin (µmol/L)	>17	17-34	>34	>51
Spectrin/erythrocyte (% of normal)	100	80-100	50-80	40-60
Splenectomy	Not required needed in childhood and adolescence	Usually not needed in childhood and adolescence	Necessary in school age before puberty	Necessary delay until 6 years if possible

[Table/Fig-4]: Classification of anaemia.

The mild disease usually requires no treatment. The moderate disease is treated with folic acid at a daily dosage of 5 mg. Severe disease needs close haematological supervision and may require splenectomy after the age of six years with proper counselling on the subsequent risk of infection post splenectomy. When spherocytosis is diagnosed during childhood splenectomy is usually carried out [5]. In such cases, there will be no significant maternal effects during pregnancy. In cases where splenectomy is not done life-threatening aplastic and haemolytic crises can occur during pregnancy. Elmezughi K and Ekpebegh C, proved splenectomy done in the second trimester had an uneventful pregnancy [1]. Splenectomy during pregnancy is associated with complications such as sepsis, perioperative morbidity to the mother and the foetus and thrombosis [3]. Prior to surgery, immunisation against pneumococcus, meningococcus and haemophilus influenza should be done. Antibiotic prophylaxis must be given for the first two years following splenectomy and lifelong in immunocompromised patients. Indication for splenectomy depends on the severity of the disease. Maberry MC et al., identified out of 50 cases of HS, 23 cases did not require splenectomy and had uncomplicated pregnancy [6]. Pager A et al., reported eight cases of hereditary spherocytosis required only haematological support during pregnancy [7] same as in the present case. Khadke B et al., also proved that haemolytic crisis can be overcome without the need for splenectomy [8]. If any of the parents are affected, then there is a chance that 65% of the neonates will have hereditary spherocytosis [9].

In the present case report, the patient did not have autosomal dominant mode of inheritance as her mother was a diagnosed case of hereditary spherocytosis for which she underwent splenectomy and her first child was unaffected by the disease. Khadke B et al., [8] also described the same mode of inheritance as in the present case. Haemolytic crises mentioned in case report by Pajor et al., required splenectomy [7] whereas the case discussed by Khadke B et al., needed only blood transfusion [8], depending on the signs and symptoms. In the present case, the patient had only mild splenectomy and symptoms of moderate anaemia such as fatiguability and breathing difficulty on normal activity for which she was managed with haematological support and did not require splenectomy. Not all cases of hereditary spherocytosis with pregnancy requires splenectomy. Splenectomy is advised only in pregnant women who have haemolysis or any symptoms, signs and complications. Early diagnosis and management can lead to better outcome. Genetic counselling should be offered, as it has an autosomal dominant mode of inheritance and the future offspring can be affected.

CONCLUSION(S)

The most common congenital haemolytic anaemia that may get decompensated during pregnancy is hereditary spherocytosis. Usually, pregnancy is well-tolerated without the need for splenectomy. Present case was managed conservatively without the need for splenectomy by a multidisciplinary team and close monitoring during pregnancy.

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