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Case Report
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Rendu-Osler-Weber Syndrome in Pregnancy: Case Report

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Abstract

Rendu-Osler-Weber syndrome or Hereditary Hemorrhagic Telangiectasia (HHT) is a rare autosomal-dominant angiodysplasia characterized by the development of mucocutaneous telangiectases and arteriovenous malformations (AVMs) in the central nervous system, lung, liver and gastrointestinal tract. Its most common symptoms are epistaxis and gastrointestinal bleeding. Visceral AVMs cause headache, cerebral ischemia and abscess, stroke, dispnea, cyanosis, hemothorax, embolia, and fatal haemorrhage from the rupture of AVMs. Its diagnosis is mainly clinical, based on the Curacao criteria: recurrent epistaxis, telangiectases, gastrointestinal bleeding and family history of HHT. Treatment is mostly symptomatic.

When HHT is coupled with pregnancy, it may be associated with a higher risk of complications because the typical hormonal and cardiovascular changes of pregnancy carry disease progression. For this reason, it is important to screen and treat women with HHT before pregnancy in order to reduce morbidity and mortality. However, most women with HHT have a normal pregnancy. Most individuals with HHT are unaware of their diagnosis because in youth the only clinical manifestation is often epistaxis, AVMs could be asymptomatic for years or because no direct family members have previously been diagnosed.

We present a case of HHT which had not previously been diagnosed in a young woman at 40 weeks of pregnancy, who came to our attention from the obstetric emergency room due touterine contractile activity.

Introduction

Rendu-Osler-Weber syndrome or Hereditary Hemorrhagic Telangiectasia (HHT) is a rare autosomal-dominant angiodysplasia characterized by heterogeneous clinical manifestations even in families with the same genetic mutation. Its prevalence is estimated to be one case per 5,000–8,000 individuals.

Most cases are due to mutations in the endoglin (*HHT1*) or *ACVRLK1* (*HHT2*) genes involved in the TGF-beta pathway. Initial histological lesions are the increase of submucosal vessels in intact endothelium, dilations of the capillaries, post-capillaries and collector-type venules, with large elongated clusters of erythrocytes with fibrin channels in the connective tissue, red cells spread in the interstice around affected vessels, endothelial discontinuity and degeneration. This causes a focal dilation in the capillary venules followed by a spread to the entire venular structure with formations of arteriovenous anastomoses and telangectases which are poorly resistant to mechanical injury.

The disease is characterized by the development of arteriovenous malformations (AVMs) which may occur anywhere in the body, but that often appear in the skin, lips, face, fingertips, and nasal, oral, gastrointestinal mucous membranes, and internal organs such as lungs, liver, and central nervous system.

Although the disease can affect different organs, the most common clinical manifestations are recurrent epistaxis and gastrointestinal bleeding.

Diagnosis is mainly clinical and is made according to the Curaçao Criteria:

1. Epistaxis, spontaneous and recurrent;
2. Telangectases, multiple, at characteristic sites: lips, oral cavity, fingers and nose;
3. Visceral lesions, such as gastrointestinal telangiectases or pulmonary, hepatic, cerebral or spinal AVMs;
4. Family history, a first-degree relative with HHT according to these criteria.

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HHT diagnosis is:

1. Definitive: if three or more criteria are present
2. Possible or suspected: if two criteria are present
3. Unlikely: if fewer than two criteria are present

Case Presentation

Primipara, 29 years old, at 39+6 weeks of amenorrhea with suspected Rendu Osler Weber syndrome.

She had a diagnosis of thalassemia trait (*HbA2*=5.6%) and thyroid nodules in suppressive treatment with levothyroxine. She had a positive family history of HHT. Her first obstetrician check was performed at 7+3 weeks of amenorrhea, the course of pregnancy was physiological with biometrics and regular growth in subsequent checks. Analytically there was a presence of microcytic hypochromic anemia, compatible with the thalassemia trait and treated with oral iron therapy.

The patient came to our clinical observation at 39+6 weeks of amenorrhea for contractile uterine activity. On admission there were no signs of labor, but based on the clinical and family history, HHT was suspected because there were two of the diagnostic criteria:

1. Frequent epistaxis: the frequency of episodes increased during the course of pregnancy;

2. A positive family history: mother diagnosed with HHT, with oropharyngeal and nasal telangectases and AVM in the gastrointestinal mucosa, lung and liver.

As HHT is characterized by the presence of AVMs that can remain clinically silent, and in order to correctly choose the mode of delivery, otorhinolaryngologist counseling, a full abdomen ultrasound and a brain MRI without contrast were performed to better define the medical case.

The anterior rhinoscopy showed the presence of vascular ectasia of the Locus Valsalvae bilaterally; however, laryngoscopy was negative. The ultrasound of the abdomen showed two small liver hemangiomas and splenomegaly (longitudinal diameter max 16 cm).

According to the documentation of ectasia to the nasal mucosa and liver AVMs, we made a diagnosis of Rendu-Osler-Weber syndrome based on the Curacao criteria (presence of three manifestations).

Since the MRI didn't show the presence of macroscopic brain AVMs, vaginal delivery was considered safe.

The patient began spontaneous labor at 41 weeks of amenorrhea. The second stage of labor progressed quickly, not necessitating the use of epidural analgesia. The delivery was eutocic resulting in a healthy newborn female of 3150g with an Apgar score of 9 at 5 minutes, and 10 at 10 minutes.

The patient reported a second degree laceration of the perineum that was sutured in analgesia. There was no excessive bleeding during the second and third stage of labor. There were no complications in post-partum and the mother and the baby were discharged after 72 hours.

Discussion

Rendu-Osler-Weber disease is a rare haematological pathology that requires a multidisciplinary approach, diagnosed by clinical criteria. Women with HHT who have not been screened for arteriovenous malformations are at risk for serious pregnancy complications: a retrospective descriptive study of women with HHT reported that they have a higher risk of developing serious pregnancy complications, especially if they had not been screened and treated for AVMs before pregnancy. The higher risk of HHT-related complications could be the result of haemodynamic changes due to pregnancy. The most common complication in women with HHT during pregnancy is frequent epistaxis and the report of new telangiectases. However, these manifestations are not associated with significant complications. More serious manifestations are related with visceral AVMs, like the development of hemothorax, deep vein thrombosis, pulmonary embolism and myocardial ischemia. It is estimated that up to 48% of patients with HHT have pulmonary AVMs (PAVMs). Often they are multiple, small and easily missed on routine examination. In addition to the risk of rupture of these PAVMs, there is the risk of significant right to left shunt resulting in hypoxemia, heart failure and the potential passage of emboli across the shunt into the systemic circulation causing complications, such as stroke and cerebral or systemic abscess. For these reasons, women with HHT should be screened and treated for pulmonary and cerebral AVMs before pregnancy in order to reduce maternal risk and to improve fetal outcome. In these cases, an ECHO-Bubble could be recommended. Spinal AVMs have to be excluded in order to allow epidural an aesthesia. However, few bleeds have been reported, therefore if spinal AVMs are detected, pregnancy management could be optimized to minimize risks of obstructed venous drainage.

In our case, collecting a personal and family medical history enabled us to achieve a rare diagnosis. Imaging studies, such as MRI and ultra sonography can help to provide important information about the presence of AVMs in the internal organs to be able to decide the safest modality of delivery. However, if macroscopic AVMs are not detected on imaging studies, vaginal delivery is considered safe.

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