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Case Report

Rendu-Osler-Weber Syndrome and Pregnancy: Case Report

Uraga-Camacho NC[1], Hernandez-Cordova A[1], Corral-Enriquez C[1], Figueroa-Perez I[1], Inzunza-Castro ME[1], Ochoa MC[2] and Ramirez-Leyva DH[3]*

[1]Department of Gynaecology, Mexican Social Security Institute (IMSS), Mexico

[2]Department of Pediatrics, Mexican Social Security Institute (IMSS), Mexico

[3]Department of Family Medicine, Mexican Social Security Institute (IMSS), Mexico

***Corresponding author:** Ramirez Leyva Diego Hazael, Department of Family Medicine, Family Medicine Unit #1, Mexican Social Security Institute (IMSS), Sonora Delegation, Sonora, México, Colonia Centro, Cd. Obregon, Sonora, Mexico

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Abstract

Hereditary Hemorrhagic Telangiectasia (HHT) is an infrequent disease. There is no predilection for some gender and is produced by genetically inherited mutations of dominant form that intervene in the mechanism of angiogenesis. The typical manifestations of this condition are visceral or mucocutaneous angiodysplasias, which are distributed through the cardiovascular system. The skin, lungs, gastrointestinal tract, and brain are the most commonly affected organs. Most patients have episodes of recurrent epistaxis due to telangiectasia in the nasal mucosa. The prognosis is almost always favourable due to supportive treatment and iron supplementation or transfusion. We report a 20 years old female which presents Rendu-Osler-Weber Syndrome and pregnancy which had to be urgently operated due to respiratory complications of an infectious pulmonary process complicated by pulmonary bleeding in week 32.4 of pregnancy.

Keywords: Rendu-Osler-Weber Syndrome; Hereditary hemorrhagic telangiectasia; Pregnancy

Introduction

First described by Sutton in 1864, it is Rendu [1] in 1896, who recognizes it as a disease with its own entity. Subsequently, Osler [2], in 1901, and Weber [3], in 1907, published the first series of cases. In 1909, Hanes [4] gives it the name Hereditary Hemorrhagic Telangiectasia (HHT). Rendu-Osler-Weber disease is a rare congenital alteration in the structure of the vascular wall, which favours the formation of aneurysms that produce recurrent haemorrhages [5]. It is inherited as an autosomal dominant pattern with a high penetrance. Several genes related to the disease have been identified in several loci associated with different chromosomes: long arm of chromosome 9 (9q33-34), short arm of chromosome 3 (3p22) and long arm of chromosome 12 (12q) [6].

It is divided into two types: hereditary hemorrhagic telangiectasia type I, where the endoglin gene is affected, and type II, where the receptor-like activity kinase 1, vascular endothelial growth factor-β receptor I gene is affected [7,8]. Clinically it is characterized by mucocutaneous telangiectasia, with the presence of multiple microaneurysms disseminated in skin or viscera, which can rupture and cause haemorrhages [9]. The typical lesions of the disease are located mainly in the lips, lobes of the ears, mucous membranes, nose (epistaxis) and gastrointestinal tract, where it causes recurrent digestive haemorrhages with iron deficiency anaemia [10]. About 20% of patients with Rendu-Osler-Weber syndrome have pulmonary telangiectasia [11]; there have been reports of severe liver failure due to arteriovenous malformations [12]. The most serious complications of this syndrome are due to the neurological involvement that can complicate the evolution of the disease in up to 10% of cases, most frequent are cerebral infarction and cerebral arterial embolism [13].

A greater fragility of arteriovenous malformations during pregnancy has been demonstrated [14] and severe complications have been described during gestation, such as rupture of cerebral arteriovenous fistulas [15], shunts, hypoxemia, and even pulmonary haemorrhages [16].

Case Presentation

A 29-year-old female patient, native and resident of Cd. Obregon, Sonora, Mexico, in her second pregnancy, Gyneco-obstetric medical records: a previous caesarean due to pulmonary hemorrhagic complications during the last trimester of the previous pregnancy. Within her medical history, she is a carrier of Rendu-Osler-Weber syndrome in symptomatic treatment with octreotide since 16 years of age; had two surgeries of intestinal resection secondary to gastrointestinal bleeding (eight and ten years ago respectively) and has epilepsy since the age of five years in treatment with phenytoin, last seizure crisis 4 years ago.

In the current pregnancy she maintained a correct prenatal control from the first month of pregnancy in her clinic of first level of care, with monthly assessments by the department of gynaecology and obstetrics in second level of care; currently with 32.4 weeks of amenorrhea by last menstrual period which is reliable. She went to an emergency department for dyspnoea, chest pain, fever and productive cough; vital signs on admission showed the following: heart rate 120 beats per minute, respiratory rate 32 per minute, blood pressure 110/70 mmHg, temperature 38.5 degrees Celsius, oxygen saturation 90%; after initial evaluation, thermal control, oxygen therapy and a complete physical examination was carried out, in which there is presence of a pleuropulmonary syndrome of consolidation type at the basal level of the left lung compatible with community acquired pneumonia.

After assessing the risk-benefit of the patient, a chest X-ray (with all the preventive cares for the fetus) was decided, this confirmed the presence of a consolidation zone at the left basal level compatible with

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Table 1: Diagnostic criteria for HHT (Curaçao criteria).

Curaçao criteria	
1.	Epistaxis, which should be spontaneous and recurrent
2.	Telangiectasias, multiple and in characteristic sites: lips, oral cavity, fingers, nose.
3.	Characteristic visceral lesions in: - Gastrointestinal telangiectasias (with or without bleeding). - Pulmonary arteriovenous malformations. - Hepatic arteriovenous malformations. - Cerebral arteriovenous malformations. - Spinal arteriovenous malformations.
4.	Family history, with a first-degree descendant diagnosed with HHT according to these criteria.
The diagnosis of HHT is: Definitive, if 3 or more criteria are met. Possible if 2 criteria are met. Unlikely, if less than 2 criteria are met.	

HHT: Hereditary Haemorrhage Telangiectasia.

Table 2: Maternal-fetal complications of HHT.

1.	Maternal complications - Haemorrhages and hematomas during pregnancy (pulmonary, cerebral, hepatic, digestive) - Brain ischemic accidents - Congestive heart failure - Coagulopathy of consumption - Hyperesplenism - Thrombocytopenia - Postpartum metrorrhagia
2.	Fetal complications - Vascular malformations - Intrauterine fetal hemorrhage (pulmonary, cerebral) - Non-immune fetal hydrops - Fetal death

pneumonia. It was decided to hospitalize the patient in department of gynaecology and obstetrics for surveillance and management. Initial management was with cephalosporin of third generation (ceftriaxone), antipyretics (paracetamol) and fetal surveillance.

The laboratory showed: Haemoglobin 10.8 g/dl, Hematocrit 43%, White Blood Cell #18400, Lymphocytes #2010, Neutrophils #12788, Eosinophils #150, Platelets 269.000, Prothrombin time 17.0 sec, Plasma thromboplastin time 24.3 sec. Urinalysis: normal. Electrolytes: Sodium 131 meq, Potassium 3.4 meq, Chlorine 96 meq, Calcium 8.5 meq, Magnesium 1.7 meq, Phosphate 3.2 meq.

Three hours after admission to gynaecology, the patient began to develop significant dyspnea followed by an oxygen saturation to 85%, so that the immediate response team had to be activated; patient with general instability, pregnancy interruption was decided at 32.4 weeks of gestation, which was performed without complications, obtaining a preterm newborn who breathed and cried at birth, with a APGAR scale of 7-8 Points, Silverman-Anderson scale 2-3 points that presented in his first hours of birth pulmonary adaptation syndrome and required hospitalization, with no subsequent complications.

As for the mother, she had self-limiting hemoptysis and syncope, which occurred spontaneously in her first hours of surgical puerperium. A computerized axial tomography was performed, where cardiomegaly, pleural effusion and a left basal consolidation zone were observed without any other alterations. Currently under study for pulmonary arteriovenous malformations. During the next days of her hospitalization she did not present signs or symptoms related the initial illness and was discharged seven days after completing an intravenous antibiotic regimen and without respiratory difficulty. The neonate was discharged at 35 days after birth without complications.

Discussion

A committee of experts met in 1999 on the island of Curaçao and defined the current diagnostic criteria for HHT (known as Curaçao criteria); diagnosis is basically clinical and is performed by the presence of at least 2 criteria (Table 1) [17]. Clinical suspicion is evident with recurrent telangiectasia and bleeding. Biopsy of skin lesions may be necessary for confirmation of diagnosis [9,18]. Treatment consists of symptomatic antihemorrhagic therapy such as administration of thrombin and direct pressure [19]. When these actions cannot control the haemorrhage, it is suggested the resection or embolization of arteriovenous malformations, which could be a complicated surgery [20].

There are few cases reported in the universal literature on HHT and pregnancy, even in Mexico the information on these cases is scarce; Marusov et al. [21] described a case of bleeding at the base of the tongue in a pregnant woman and Barber et al. [22] described a case of postpartum metrorrhagia secondary to this syndrome. Pregnant women with Rendu-Osler-Weber disease have a strict control, particularly during childbirth and immediate puerperium. Perinatal outcomes are generally good, and pregnancy is not contraindicated. In the fetus, complications have been described as the following: vascular malformations, fetal intrauterine (pulmonary or cerebral) hemorrhages, fetal dropsy and increased fetal death [23]. Serious hemorrhagic complications have been reported during childbirth so steps should be taken to avoid these potential complications [24]. Table 2 describes the maternal-fetal complications of Rendu-Osler-Weber disease.

Conclusion

Hereditary hemorrhagic telangiectasia or Rendu-Osler-Weber disease is a rare vascular, congenital, and hereditary disease (autosomal dominant), clinically characterized by the presence of multiple telangiectasia, anemic syndrome, frequent bleeding and different arteriovenous shunt. From the obstetric point of view, the importance of this nosological entity focuses on the severe hemorrhagic complications described during childbirth. Pregnant women with Rendu-Osler-Weber disease should have a strict control throughout pregnancy and in particular during childbirth, in order to prevent or treat possible complications quickly. The use of direct intravenous oxytocin may be indicated, after the exit of the fetal shoulder, with later revision of the vaginal canal and uterine massage in order to diminish possible hemorrhagic complications. It is necessary an extensive interrogation and an adequate physical exploration to discover rare pathologies in the pregnant patient, it is necessary also a familiar screening in order to discover another genetic alterations.

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