CASE REPORT

Pregnancy Unmasking Hereditary Hemorrhagic Telangiectasia: A Case Report

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Abstract

Hereditary hemorrhagic telangiectasia (HHT) is a rare genetic disorder characterized by angiodysplasia which may manifest with telangiectasia and arteriovenous malformations (AVMs) in the central nervous system, lung, liver, and gastrointestinal tract. Women suffering from HHT may have a normal pregnancy course but some pregnancies are at risk of fulminant complications related to hormonal and cardiovascular changes of pregnancy. Hence, it becomes very important to diagnose this condition promptly and monitor women with HHT carefully to reduce morbidity and mortality associated with the disease. We present two women diagnosed with HHT in pregnancy who presented to our institution with different clinical scenarios. Based on the clinical findings, family history and high index of suspicion, we were able to diagnose and manage them accordingly.

Keywords: Case report, Hereditary hemorrhagic telangiectasia, Osler–Weber–Rendu syndrome, Pregnancy.

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Introduction

Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant inherited genetic vascular disorder characterized by recurrent epistaxis, cutaneous telangiectasia, and visceral arteriovenous malformations (AVM).[1] Diagnosis is made either by clinical or genetic criteria. Quiescent disease prior to pregnancy may manifest for the first time during pregnancy. Severe complications like rupture of cerebral AVM and pulmonary hemorrhage due to increased fragility of vasculature can occur in pregnancy resulting in a 1% overall maternal death rate. Though there are guidelines for prepregnancy screening and antenatal care of these women diagnosed with the disorder before pregnancy, diagnosis, and management of women who present with the problem for the first time during pregnancy could be a challenge! We present two patients who were diagnosed with HHT during pregnancy and discuss important clinical implications and how we managed these patients. Our objective is to make clinicians aware of this rare entity, overlapping manifestations of HHT and common symptoms in pregnancy, and hence the need for a high index of suspicion. Our patients presented with pedal edema and epistaxis which are commonly seen in pregnancy but a careful evaluation led to the diagnosis.

Case Description

Case 1

A 25-year-old woman, fourth gravida with two living children was referred at 29 weeks gestational age with complaints of epigastric pain and bleeding from the nose. On her past obstetric history, she had two uncomplicated normal vaginal deliveries. Her third pregnancy however was complicated at 32 weeks of gestation by recurrent epistaxis, hematemesis, intrauterine fetal demise, disseminated intravascular coagulation, and sepsis.

Clinical examination revealed signs of periorbital petechial rash (probably due to telangiectasia) and hyperdynamic circulation which was confirmed on echocardiogram. An ultrasonogram of the abdomen revealed features suggestive of congestive liver and portal cavernoma. Upper gastrointestinal endoscopy revealed normal study and there were no varices. She probably vomited and swallowed a nasal bleed. Investigations revealed a significant drop in hemoglobin and packed cell volume with normal platelet count and coagulation profile. Diagnostic nasal endoscopy revealed diffuse mucosal bleeding points involving the nasopharynx. Chest X-ray revealed well-defined bilateral opacities and a magnetic resonance imaging (MRI) scan of the chest revealed bilateral pulmonary AVMs. Based on the clinical findings and investigations, HHT was diagnosed as she fulfilled three of the diagnostic criteria:

- Presence of portal cavernoma and pulmonary AVMs.
- Recurrent and spontaneous epistaxis.
- Presence of telangiectasia in bilateral periorbital region.

She was readmitted at 31 weeks with shortness of breath and increasing pedal edema due to high-output cardiac failure. She was managed by a multidisciplinary team and an emergency cesarean section was done in view of worsening maternal condition. A 1.66 kg baby girl was delivered with a good Apgar score. Postoperatively, she was transfused 4 units of packed red blood cells and 6 units of fresh frozen plasma. She gradually improved over the next 2 weeks.

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Case 2

A 27-year-old primigravida at 32 weeks gestational age was referred to our hospital with excessive bilateral lower limb swelling without any evidence of preeclampsia (no hypertension or proteinuria). On clinical examination, she had features of hyperdynamic circulation with wide pulse pressure, visible carotid pulsation, bounding pulses, and flow murmur over the pericardium which was confirmed later by echocardiogram. There were no crackles heard on the auscultation of the lungs. She had normal oxygen saturation on pulse oximetry and did not have orthopnea or paroxysmal nocturnal dyspnea. She was evaluated for high-output cardiac failure. She had no anemia or thyrotoxicosis and had no risk factors for thiamine deficiency. She had no improvement with parenteral thiamine supplementation. She had a history of recurrent mild epistaxis which the patient revealed only on further questioning. The patient's sister also had a history of recurrent epistaxis. The possibility of HHT was considered and an MRI chest was ordered to look for lung AVMs. MRI chest did not reveal any pulmonary AVM but revealed multiple telangiectasia of the liver. With this clinical picture, the possibility of HHT was considered because of the following:

- History of recurrent epistaxis.
- Presence of hyperdynamic circulation with multiple telangiectasia on liver imaging.
- Possibility of a family member with HHT (sister with recurrent epistaxis).

She was admitted in view of shortness of breath on exertion. After 2 days of admission, she became drowsy, disoriented, and developed jaundice. With possible differential diagnoses of intracerebral bleed, acute fatty liver of pregnancy or hepatic encephalopathy due to portosystemic shunting in HHT, she was transferred to a multispecialty hospital for multidisciplinary care. Her neuroimaging was normal and a final diagnosis of hepatic encephalopathy due to liver involvement by HHT was made. Pregnancy was terminated by cesarean delivery at 34 weeks gestational age owing to her medical condition. However, she rapidly deteriorated in the postoperative period and despite supportive treatment, she succumbed after 2 days.

Discussion

Hereditary hemorrhagic telangiectasia (HHT), also known as Osler–Weber–Rendu disease is a rare vascular genetic disorder that requires a multidisciplinary approach, diagnosed by clinical criteria and diagnosis can be supported by genetic testing. Clinical diagnosis is made by Curacao criteria defined in 2000 by the scientific advisory board of the HHT foundation (Table 1).[2]

The HHT diagnosis is definitive if three or more criteria are present, possible or suspected if two criteria are present, and unlikely if fewer than two criteria are present.[2] A retrospective descriptive study of women with HHT reported that they have a higher risk of developing serious pregnancy complications especially if they are not screened and treated for AVMs before pregnancy.[3] The most common complication in women with HHT during pregnancy is frequent epistaxis and the appearance of new telangiectasia.[4] More serious manifestations are associated with visceral AVMs. Serious complications of AVMs include the development of hemothorax, deep vein thrombosis, pulmonary embolism and myocardial ischemia.[5] It is estimated that up to 48% of patients with HHT have pulmonary AVMs (PAVMs). Besides the risk of rupture of these PAVMs, there is a risk of significant right to left shunt resulting in hypoxemia, heart failure, and the potential passage of emboli causing complications like stroke and cerebral or systemic abscesses.[6] Hence these women should be screened and treated for pulmonary and cerebral AVMs before planning pregnancy in order to reduce maternal risk and improve fetal outcome.[7]

Table 1: Curacao criteria

1. Epistaxis occurring spontaneously on more than one occasion
2. Telangiectasia involving the nose, fingers and oral cavity
3. Visceral involvement such as pulmonary, hepatic, or cerebral arteriovenous malformations
4. A family history of HHT (first-degree relative diagnosed with HHT with the same criteria)

Most of the literature in the area of HHT in pregnancy is focused on women known to have HHT and their pregnancy outcomes. In this case report, our patients were not known to have HHT and were diagnosed for the first time during pregnancy. One of our patients presented with excess pedal edema which is a common manifestation of normal pregnancy or indicative of preeclampsia. She had clinical features suggestive of hyperdynamic circulation and high-output heart failure. She had no anemia or thyrotoxicosis which could explain high output cardiac failure. High output heart failure may be mistaken as exaggerated hyperdynamic circulation which is a physiological phenomenon in normal pregnancy! In view of the presentation, we had to consider multiple differential diagnoses in her and exclude them. We were able to diagnose HHT in her because of a high index of suspicion and a systematic approach. The commonality of the above-mentioned conditions and rarity of HHT may delay diagnosis when the presentation occurs during pregnancy. A common manifestation like excess pedal edema in pregnancy could be a presenting feature of HHT.

The other patient also presented with features of cardiac failure but had a rapidly progressive course because of hepatic telangiectasia and encephalopathy resulting in mortality despite prompt diagnosis and care. Mothers with HHT need multidisciplinary team care and timely intervention in order to reduce maternal morbidity and mortality. There are no clear-cut guidelines regarding timing or mode of delivery for women with HHT. These women require detailed counseling and close monitoring throughout pregnancy and may need preterm termination of pregnancy.

Conclusion

We need to consider the possibility of HHT in pregnant women with recurrent unexplained epistaxis, unexplained excess pedal edema or heart failure symptoms with normal ejection fraction, and normal blood pressure. Clinicians need to have a high index of suspicion to diagnose HHT in pregnancy. Literature and international consensus guidelines provide information mostly on screening for complications and management of women known to have HHT who become pregnant. Documentation of cases that manifest for the first time during pregnancy, their presentation and management further add to this existing information.

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