Pregnancy With Uterine Vascular Malformations Associated With Hemorrhagic Hereditary Telangiectasia: A Case Report

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Abstract

Background: Hereditary hemorrhagic telangiectasia (HHT) is a rare autosomal dominant condition. It is rarely seen in pregnancy and even more rarely has uterine manifestations.

Case: A 29-year-old primigravid woman with HHT was noted to have vascular manifestations of her disease in the lower uterus, distal rectum, pelvis, and bladder before pregnancy. Prior to delivery, a case conference was held, involving representatives of the departments of vascular surgery, hematology, radiology, anaesthesiology, maternal-fetal medicine, neonatology, and laboratory medicine, and other appropriate health professionals. A successful elective Caesarean section was performed at term, with a good outcome for both mother and child.

Conclusion: Pregnancies in women with HHT and associated uterine vascular manifestations have been rarely reported, and published information is minimal. We present a case of a successful operative delivery following careful multidisciplinary antepartum care.

Résumé

Contexte : La télangiectasie hémorragique héréditaire (THH) est une affection autosomique dominante rare. Elle est rarement constatée au cours de la grossesse et il est encore plus rare qu'elle présente des manifestations utérines.

Cas : On a constaté, avant la grossesse, des manifestations vasculaires dans la partie inférieure de l'utérus, la partie distale du rectum, le bassin et la vessie chez une femme primigravide de 29 ans présentant une THH. Avant l'accouchement, une conférence de cas (mettant en jeu des représentants des services de chirurgie vasculaire, d'hématologie, de radiologie, d'anesthésiologie, de médecine fœto-maternelle, de néonatologie et de médecine de laboratoire, ainsi que d'autres professionnels de la santé pertinents) a eu lieu. Une césarienne de convenance a été pratiquée à terme avec succès, de bonnes issues ayant été constatées tant pour la mère que pour l'enfant.

Conclusion : Les grossesses de femmes présentant une THH et des manifestations vasculaires utérines connexes ont rarement fait l'objet de signalements; ainsi, les renseignements publiés sont minimes. Nous présentons un cas d'accouchement opératoire réussi ayant été précédé de soins ante-partum multidisciplinaires méticuleux.

Key Words: Hereditary hemorrhagic telangiectasia, pregnancy, uterus, arteriovenous malformations, Osler-Weber-Rendu disease

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INTRODUCTION

Hereditary hemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu disease, is a rare autosomal dominant disorder caused in most cases by mutations of the genes HHT 1 (ENG) or HHT 2 (ACVRL1).[1] The overall incidence in North America is estimated to be 1 in 10 000.[2] The disorder occurs with wide ethnic and geographic distribution. It is characterized by the presence of multiple arteriovenous malformations (AVMs) that lack intervening capillaries, resulting in direct connections between arteries and veins. The diagnosis is based on family history and the presence of cutaneous or mucocutaneous telangiectases or large visceral AVMs.[3] Common sites for telangiectases are the tongue and mucosal surfaces of the lips, face, conjunctiva, ears, fingers, gastrointestinal tract, and bladder.[4] Affected individuals may have visceral AVMs involving the central nervous, pulmonary, cardiac, renal, and hepatic systems.[4] About 25% of affected individuals have gastrointestinal bleeding. Serious complications include hemorrhage from pulmonary arteriovenous malformations and gastrointestinal telangiectases. Individuals often present in early childhood with recurrent epistaxis. Management includes surveillance for undiagnosed AVMs and treatment for identified complications. The condition is usually benign in pregnancy.

However, there is very little published information regarding pregnancies with HHT and uterine vascular manifestations.[5,6] We present here an unusual case of HHT in pregnancy.

THE CASE

A 29-year-old primigravid woman presented at 20 weeks' gestation for assessment and monitoring of her high-risk pregnancy. At birth she was noted to have extensive cutaneous hemangiomata over the right side of her body. In her early twenties, with no formal investigations or diagnosis having been made, she attempted to have these removed using laser therapy. As part of an evaluation of deep dyspareunia and post coital bleeding, her gynaecologist performed a diagnostic laparoscopy. Telangiectases involving the vagina, cervix, and the entire right side of the parietal peritoneum were noted. There was no mention at that time of uterine involvement.

She was subsequently referred to different specialists for a full assessment. A medical geneticist stated that she had an extensive capillary hemangioma involving the right anterior chest, right flank crossing the midline, right and upper left thigh extending medially, and the right first and second toes on to the ball and arch of her right foot. Involvement of the right labium majus was also noted. The mucosal surfaces appeared normal. The most likely diagnosis was thought to be HHT, given the visceral involvement, although it was also noted that the large cutaneous hemangioma was typical of Klippel-Trenaunay-Weber syndrome. Diagnostic imaging performed included an echocardiogram, a renal scan, and an upper abdominal computerized tomogram (CT) to view the liver. The findings on echocardiography were normal. The CT showed innumerable serpiginous linear densities within the pelvis, the lateral side of the distal rectum, the uterus, and the bladder. No large AVM was noted. The liver, pancreas, spleen, and kidneys were reported to be normal. Magnetic resonance imaging (MRI) of the thoracic spine and head was normal. The findings on ophthalmological examination were normal.

Because the patient wished to conceive, she was referred to an obstetrician for pregnancy counselling. Although the patient was concerned about potential risks associated with pregnancy related to organ expansion and fetal movement, the fact that most of her lesions were cutaneous was reassuring to the patient.

The patient subsequently conceived and had an uneventful early pregnancy. At 20 weeks' gestation, she was referred to the maternal-fetal medicine unit at BC Women's Hospital. She was warned about the increased risk of serious postpartum hemorrhage that could require blood transfusion or hysterectomy. She was hospitalized at 25 weeks' gestation because of weight loss from severe nausea and vomiting, diffuse abdominal pain, and vaginal bleeding. During her hospitalization, a case conference involving members of the departments of vascular surgery, anaesthesia, hematology, and maternal-fetal medicine was held to allow a coordinated multidisciplinary team approach to her care. Autologous blood collection was recommended in view of the potential for severe maternal hemorrhage. Glucocorticoid injections to accelerate fetal lung maturity were administered in anticipation of possible preterm delivery prompted by antepartum hemorrhage. Sonographic colour Doppler flow studies suggested abnormal vascular structures in the vault of the vagina and cervix. Given the extensive telangiectases noted, the management team recommended that the patient have an elective Caesarean section (CS) to avoid the risk of serious hemorrhage during a vaginal delivery.

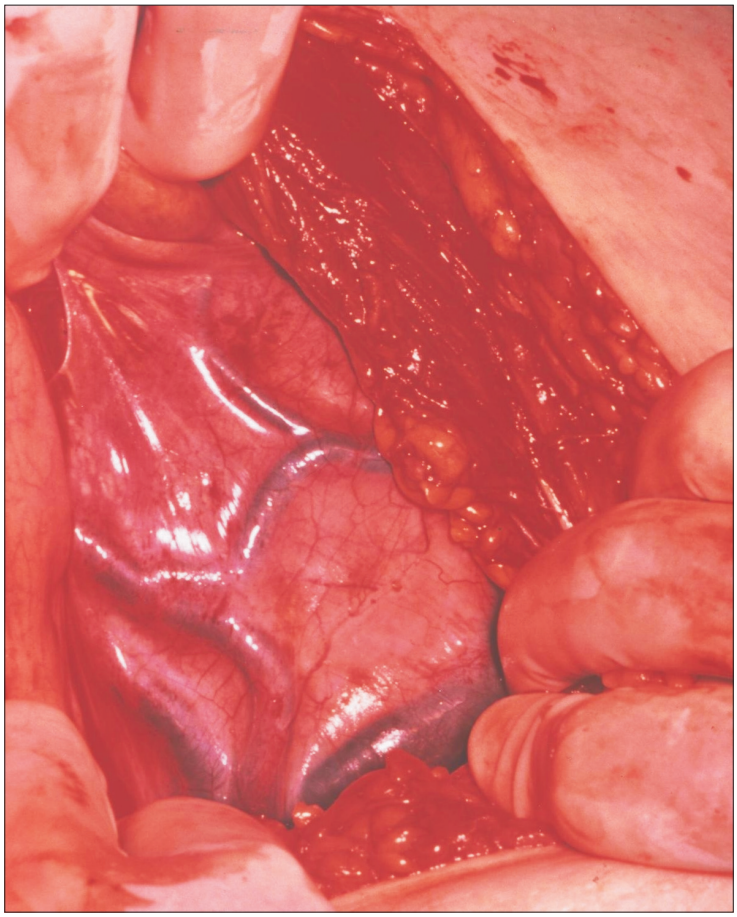
Regular ultrasound examinations for assessment of fetal growth, amniotic fluid, and umbilical artery Doppler flow studies were performed and showed reassuring results. Weekly non-stress testing and fetal biophysical profile were also reassuring.

Because MRI of the maternal spine showed extensive subcutaneous vascular lesions and an increased prevertebral vascular space with distortion of the thecal sac from L1 to L3, the management team elected to use general anaesthesia rather than spinal anaesthesia for the CS to avoid an epispinal hematoma. Amniocentesis to confirm fetal lung maturity was performed at 36 weeks; early delivery was planned to decrease the risk of spontaneous labour with possible antepartum hemorrhage requiring emergency CS. The test confirmed maturity, and an elective CS was performed.

A Pfannenstiel skin incision was used. On opening the vesical peritoneum, large veins were seen coursing along the anterior lower uterine segment. Four or five large veins (Figure) traversed from the fundus to the lower segment. Each vein, measuring 5 to10 mm in diameter, was ligated, and a transverse lower uterine segment incision was made. Lateral extension of the uterine incision was performed by stretching the margins.

A healthy 2725 g female infant was delivered, with Apgar scores of 4 at one minute, 8 at five minutes, and 9 at 10 minutes. Estimated blood loss was 1200 mL. The fallopian tubes and ovaries appeared normal. There were no intraoperative complications, but as a precaution the patient was given two units of autologous blood. Her postoperative hemoglobin level was unchanged from the preoperative level (97 g/L). She was discharged on the fourth postoperative day in stable condition.

Large veins on the lower uterine segment



DISCUSSION

The successful outcome of this pregnancy in a woman with HHT was in large measure due to the multidisciplinary nature of the team providing antenatal care. Diagnostic imaging, including MRI, provided valuable information that allowed the management team to take the necessary precautions to minimize the risk of serious maternal and neonatal morbidity. It would appear reasonable to recommend that all such cases have the benefit of assessment with MRI as well as ultrasound with colour Doppler studies.

Some women with HHT may present for prenatal care without having a formal diagnosis. The diagnosis of HHT should be considered in a patient who presents for prenatal care and exhibits cutaneous hemangiomas. If no prior investigations or diagnosis have been made, referral to other specialists, such as a medical geneticist, may ensure that the appropriate evaluation occurs to formalize the diagnosis.

In 1995, Shovlin et al.[7] reviewed a cohort of 161 pregnancies in 47 women with HHT. None of these women were identified as having uterine involvement. Unfortunately, the authors do not describe the mode of delivery for their cohort. In the 131 pregnancies without pulmonary AVMs, one serious maternal complication (a maternal cerebrovascular accident) occurred. In contrast, in the 23 pregnancies with untreated pulmonary AVMs, there were 10 serious maternal complications that included two fatal pulmonary hemorrhages. The authors concluded that women with HHT who are planning pregnancy should be screened for pulmonary AVMs. They also suggest that a pregnant woman with HHT who has not had a recent pulmonary evaluation should be evaluated as soon as pregnancy is confirmed. Women who have pulmonary AVMs identified when they are already pregnant should be evaluated for treatment during the second trimester because an untreated pulmonary AVM puts such women at high risk for lung hemorrhage.[7]

CONCLUSION

Women with hereditary hemorrhagic telangiectasia and uterine vascular manifestations can have a successful pregnancy and uneventful operative delivery if a multidisciplinary approach is used. The use of MRI and ultrasound to identify areas of significant vascularity contributes to safe management.

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The woman whose story is told in this case report has provided signed permission for its publication.

REFERENCES

1. Berg JN, Gallione CJ, Stenzel TT, Johnson DW, Allen WP, Schwartz CE, et al. The activin receptor-like kinase 1 gene: genomic structure and mutations in hereditary hemorrhagic telangiectasia type Am J Hum Genet 1997;61:60–7.
2. Marchuk DA, Guttmacher AE, Penenr JA, Ganguly P. Report on the workshop on Hereditary Hemorrhagic Telangiectasia. Am J Med Genet 1998;76:269–73.
3. Shovlin CL, Guttmacher AE, Buscarini E, Faughnan ME, Hyland RH, Westermann CJ, et al. Diagnostic criteria for hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). Am J Med Genet 2000;91:66–7.
4. Guttmacher, AE, Marchuk, DA, White, RI Jr. Hereditary hemorrhagic telangiectasia. NEJM 1995;333:918.
5. Shanberge JN. Hemangioma of the uterus associated with hereditary hemorrhagic telangiectasia. Obstet Gynecol 1994; 84(4) Pt. 2:708–9.
6. Sütterlin MW, Müller T, Rehn N, Rempen A, Dietl J. Successful abdominal delivery in a woman with sonographic diagnosis of diffuse cavernous hemangioma of the uterus. Am J Perinatol 1998;15(7):423–5.
7. Shovlin CL, Winstock AR, Peters AM, Jackson JE, Hughes JM. Medical complications of pregnancy in hereditary haemorrhagic telangiectasia. QJM 1995;88:879–87.