CASE REPORT

Osler-Weber-Rendu syndrome during pregnancy

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SUMMARY

Osler-Weber-Rendu syndrome is a very rare systemic fibrovascular dysplasia. Rupture of angiomas can cause haemorrhages, which sometimes can be severe with difficult bleeding control. The main manifestation is recurrent epistaxis. Treatment of this disorder is symptomatic. During pregnancy, there may be an increased risk of complications. We describe a case of a pregnant woman with Osler-Weber-Rendu syndrome. Besides frequent epistaxis and microcytic hypochromic anaemia that resolved with oral iron treatment, she had a normal pregnancy, vaginal delivery and puerperium without complications.

BACKGROUND

Osler-Weber-Rendu disease or Osler-Weber-Rendu syndrome, also known as hereditary haemorrhagic telangiectasia (HHT), is a rare genetic disorder that leads to abnormal blood vessel formation in the skin, mucous membranes, and often in organs such as the lungs, liver and brain.^{1 2} There is a defect in activation and migration of various types of cells in the process of angiogenesis, causing abnormal formation of the elastic and muscular layers of the walls of blood vessels, making them more vulnerable to trauma and spontaneous ruptures.^{3 4}

HHT is an autosomal dominant genetic disorder. Five genetic types of HHT are recognised. Although in more than 80% of affected patients, these are due to mutations in genes *ENG* and *ACVRL1*,⁵ genetic diagnosis is difficult, mainly because mutations occur in numerous different locations in the linked genes, without particular mutations being highly frequent.

In most cases, the diagnosis is clinical and/or by the 'Curação criteria':⁶

- ▶ spontaneous recurrent epistaxis
- ▶ multiple telangiectasias in typical locations
- ▶ proven visceral arteriovenous malformation (AVM): lung, liver, brain, spine
- ▶ first-degree family member with HHT

The presence of three or four criteria define the patient as having Osler-Weber-Rendu syndrome, while the presence of only two criteria define the patient as having possible HHT.

Although the disease can affect various organs, epistaxis and gastrointestinal bleeding are the most predominant locations. Treatment is symptomatic and palliative, since the defect in angiogenesis is incurable.

CASE PRESENTATION

Primipara, 23 years old, with Osler-Weber-Rendu syndrome known since childhood. The diagnosis

was made by clinical criteria, without requiring genetic study.

First obstetric consultation was performed at 9 weeks of gestation. At that moment, the pregnant woman had presence of lip telangiectasias, but no severe haemorrhagic symptoms (only frequent epistaxis). Analytically: non-anaemic, but with microcytosis and hypochromia (transferrin saturation of 6% and ferritin 8 ng/mL). She was treated with 90 mg oral iron twice a day, with normalisation of haemoglobin, mean corpuscular volume and mean corpuscular haemoglobin concentration. The remaining pregnancy was uneventful.

Assuming that HHT is inherited in an autosomal dominant manner, and carrying two abnormal copies of the gene is not compatible with life (homozygotes have never been described),¹ we informed the pregnant woman that she would be a carrier of the abnormal gene and, therefore, she would have a 50% transmission risk of passing the genetic disorder to her offspring.

As pregnant women with HHT can present clinically silent but potentially life-threatening AVM, MRI was performed at 36 gestational weeks, in order to exclude, particularly, cerebral and or spinal AVM (relative contraindication to regional techniques). Since the MRI showed no AVM, vaginal delivery, as well as epidural analgesia, were considered to be safe.

She was admitted at 39 weeks for spontaneous labour. Epidural analgesia was offered and accepted. The delivery was dystocic (vacuum extraction with no complications), resulting in a healthy male newborn, weighing 3430 gr, with Apgar score 09 and 10, on the first and fifth minutes of life, respectively. The newborn does not present any signs of Osler-Weber-Rendu syndrome. During labour, delivery and postpartum, there was never any abnormal bleeding. She was discharged after 48 h.

DISCUSSION

This haematological pathology, due to its specificity, implies a multidisciplinary surveillance.

In a study of Shovlin *et al*⁷ that included 199 women and 484 pregnancies, 13 women experienced life-threatening events during pregnancy: 1.4% (95% CI 0.2 to 2.5) of pregnancies resulted in a major AVM bleed; 1.2% (95% CI 0.3 to 2.2%) in stroke; and 0.2% (95% CI 0.2 to 0.6%) myocardial infarction. There were no cases of spinal haemorrhage. Five maternal deaths have been reported—1% (95% CI 0.1 to 1.9%).

Although most HHT pregnancies proceed normally, women are at risk of major complications, including death (high-risk pregnancies). Awareness

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of HHT or AVM diagnosis was associated with improved survival⁷.

Both general and regional anaesthetic techniques can be performed in pregnant women with Osler-Weber-Rendu syndrome.⁸ MRI seems to be of great value particularly excluding cerebral or spinal AVM.

In our case, there were no complications during pregnancy, vaginal delivery and puerperium.

Learning points

- ▶ Osler-Weber-Rendu Syndrome or hereditary haemorrhagic telangiectasia (HHT) is a rare autosomal dominant inherited disease, commonly diagnosed by clinical criteria.
- ▶ Assuming that homozygosity for HHT does not exist, when one of the parents has the affected gene, we should counsel the couple of a 50% transmission risk of this disorder to their offspring.
- ▶ HHT is characterised by systemic fibrovascular dysplasia.
- ▶ MRI can provide vital information about the presence or the potential development of life-threatening arteriovenous malformation during pregnancy, particularly to obstetricians and anaesthesiologists, in order to choose the safest delivery method and anaesthetic technique, respectively.
- ▶ Women with the Osler-Weber-Rendu syndrome can have an uneventful pregnancy with normal vaginal delivery.

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REFERENCES

- 1 Govani FS, Shovlin CL. Hereditary haemorrhagic telangiectasia: a clinical and scientific review. *Eur J Hum Genet* 2009;17:860–71.
- 2 Dupuis-Girod S, Bailly S, Plauchu H. Hereditary hemorrhagic telangiectasia (HHT): from molecular biology to patient care. *J Thromb Haemost* 2010;8:1447–56.
- 3 Rapoport PG, Uvo IP, Costa KS, *et al.* Síndrome de Rendu-Osler-Weber: tratamento clínico e cirúrgico. *Rev Bras Otorrinolaringol* 2003;69:577–80.
- 4 Maudonnet EN, Gomes CC, Sakano E. Telangiectasia Hemorrágica Hereditária (Doença de Rendu-Osler-Weber): um diagnóstico otorrinolaringológico. *Rev Bras Otorrinolaringol* 2000;66:172–80.
- 5 Abdalla SA, Letarte M. Hereditary haemorrhagic telangiectasia: current views on genetics and mechanisms of disease. *J Med Genet* 2006;43:97–110.
- 6 Shovlin CL, Guttmacher AE, Buscarini E, *et al.* Diagnostic criteria for hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). *Am J Med Genet* 2000;91:66–7.
- 7 Shovlin CL, Sodhi V, McCarthy A, *et al.* Estimates of maternal risks of pregnancy for women with hereditary haemorrhagic telangiectasia (Osler-Rendu syndrome): suggested approach for obstetric services. *BJOG* 2008;115:1108–15.
- 8 Lomax S, Edgcombe H. Anesthetic implications for the parturient with hereditary hemorrhagic telangiectasia. *Can J Anaesth* 2009;56:374–84.

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