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Case Report

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Pregnancy in patient with hereditary hemorrhagic telangietasia (osler-weber-rendu disease) and pulmonary arteriovenous malformations: case report

Abstract

Background: Hereditary hemorrhagic telangiectasia, also known as Osler-Weber-Rendu disease is autosomal dominant disorder that affects blood vessels throughout the body (causing vascular dysplasia) and results in a tendency for bleeding. It manifested by mucocutaneous telangiectases and arteriovenous malformations in different organs, including the lungs (pulmonary arteriovenous malformations- PAVM). During the pregnancy due to the hormonal and hemodynamic changes bleeding and pulmonary symptoms become more severe, sometimes leading to lifetreating complications.

Case presentation: We report the case of a woman who developed severe dyspnoe, cough, haemoptysis, nose bleedings during 34-week of pregnancy. Diagnosis of hereditary hemorrhagic telangictasia (Osler-Weber-Rendu disease) was established. The lung symptoms were regard to pulmonary arteriovenous malformations. Tests revealed signs of disseminated intravascular coagulation (DIC), acquired von Willebrand syndrome (aVWs). She was successful delivered by cesarean section a then underwent a surgical treatment of pulmonary arteriovenous malformations.

Conclusion: Patients with hereditary hemorrhagic telangiectasia have a great risk of hemorrhage various localizations during pregnancy and delivery and require the multidisciplinary care. Acquired von Willebrand syndrome and DIC syndrome can play an important role in bleeding symptoms during pregnancy and cesarean section in patients with hereditary hemorrhagic telangictasia together with deficiency of vascular wall but in needs to further investigations.

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Background

Hereditary hemorrhagic telangictasia, also known as Osler-Weber-Rendu disease is autosomal dominant disorder that affects blood vessels throughout the body (causing vascular dysplasia) and results in a tendency for bleeding. There are mutations in the endoglin gene in chromosome 9 or the activin receptor like kinase type I gene in chromosome 12, which affect receptors for the transforming growth factor-beta in vascular endothelium. It manifested by mucocutaneous telangiectasia and arteriovenous malformations. Lesions can affect the nasopharynx, central nervous system, lung, liver, and spleen, as well as the urinary tract, gastrointestinal tract, conjunctiva, trunk, arms, and fingers. Arteriovenous malformation in lungs (pulmonary arteriovenous malformations- PAVM) lead to directly shunting the blood from right to left not passing through capillary. Therefore such patients may develop the symptoms of hypoxemia. During the pregnancy due to the hormonal and hemodynamic changes bleeding and pulmonary symptoms become more severe, sometimes leading to lifetreating complications.[1–4]

The term of aVWs is now accepted for patients with predominant underlining diseases that lead to defective von Willebrand factor (VWF).[5] In recent years there has been a progressive increase of publications concerning patients with defective VWF in whom the family history and the late onset of bleeding symptoms speak against an inherited form.[6,7] We report the case of a woman who developed severe dyspnoe, cough, haemoptysis during 34-week of pregnancy.

Case presentation

The patient was a pregnant (34weeks) gravida I, para I, 23-year-old woman who due to severe dyspnoe, cough, haemoptysis and chest pain was refereed from another hospital. She has numerous telangiectases on the face, lips, tongue, oral mucosa, stomach and fingertips. She experienced dyspnoe, cough, haemoptysis and chest pain during the last week. She was healthy, but remarked that spontaneous, recurrent nose bleeds began in her childbearing years and become more frequent during the pregnancy. Her family history was remarkable: her mother and sister suffered from nose bleeds.

Physical examination revealed scattered telangiectases on the face, lips, tongue, oral mucosa, stomach and fingertips. Laboratory tests showed iron deficiency anemia. Due to bleeding symptoms, family history of bleedings we decided to exclude von Willebrand disease and DIC syndrome. Together with routine coagulation tests we evaluate VWF ristocetin cofactor assay, VWF antigen assay, multimer analysis, D-dimer, F1+2, TAT. Tests revealed signs of DIC- syndrome (thrombocytopenia 73×10^9 per liter, prolonged global coagulation assays: APTT- 51, PT-15.2, high levels of FDP- 19mg/mL and DD-2.5μg/mL FEU), acquired von Willebrand syndrome (VWF antigen

-39, VWF ristocetin cofactor assay-48, decreased high molecular weight multimers in multimer analysis).[8,9] Hereditary thrombophylias tests were negative (protein S, C, AT deficiencies, F2, F5 Leiden). There were the signs of hypoxemia (arterial oxygen saturation=92). Thoracic radiography revealed band shaped shadows in the low lobe of right lung.

After multidisciplinary consultation diagnosis of hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu disease) was established according to the Curaçao criteria. Molecular diagnostic revealed HHT2 - *ACVRL1* mutation in the extracellular domain (p.Cys77Phe).[10,11] The lung symptoms were regard to pulmonary arteriovenous malformations.

Due to the great risk of enlargement the pulmonary arteriovenous malformations during the pregnancy leading to fatal hemorrhage it was decide to perform cesarean section followed to surgical treatment of pulmonary arteriovenous malformations. She underwent the cesarean sections because we believe that it can minimize the hemodynamic changes during vaginal delivery.

During the operation the excessive bleeding from small blood vessel was observed. To prevent and treat hemorrhagic complications we used fresh frozen plasma and tranexamic acid. Alive girl with the weight of 2413 and the 7-8 point of Apgar scale was obtained.

During postpartum period because of the signs of intravascular activation of the coagulation (elevated TAT, F1+2, DD) the patient received nadroparin. After delivery she underwent the surgical treatment of pulmonary arteriovenous malformations.

Conclusion

Patients with hereditary hemorrhagic telangiectasia have a great risk of hemorrhage various localizations during pregnancy and delivery and require the multidisciplinary care. Acquired von Willebrand syndrome and DIC syndrome can play an important role in bleeding symptoms during pregnancy and cesarean section in patients with hereditary hemorrhagic telangictasia together with deficiency of vascular wall, thus such patients require testing for acquired von Willebrand syndrome and DIC syndrome.

The prevalence of acquired von Willebrand syndrome and DIC syndrome in patients hereditary hemorrhagic telangictasia is probable underestimated that can lead to unexpected severe bleeding and inadequate therapy but in needs to further investigations.

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Conflict of interest

The authors state that they have no conflict of interest.

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