CASE REPORT

Osler-Weber-Rendu syndrome: an anaesthetic challenge?

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SUMMARY

Osler-Weber-Rendu syndrome, or hereditary haemorrhagic telangiectasia (HHT), is an autosomal dominant vascular dysplasia characterised by mucocutaneous telangiectases and arteriovenous malformations (AVMs). Diagnosis is clinical and treatment is supportive. The authors demonstrate a safe anaesthetic approach for a patient with HHT. A 53-year-old woman with a left trochanteric fracture was scheduled for urgent orthopaedic surgery. She was diagnosed as having HHT and presented with recurrent epistaxis, telangiectases and gastrointestinal AVMs. She had undergone a nasal dermoseptoplasty 4 weeks earlier, with total nasal occlusion. Surgery was undertaken with a lumbar and sacral plexus block performed with neurostimulation. She was sedated and spontaneous ventilation was maintained. The procedure was completed without complications. Anaesthetic management of patients with HHT is a challenge. The patients should be carefully studied before surgery, to reduce perioperative morbidity and mortality. Regional anaesthesia may be a good alternative to general anaesthesia, but the use of neuroaxial techniques is still controversial.

BACKGROUND

Osler-Weber-Rendu syndrome, also known as hereditary haemorrhagic telangiectasia (HHT), is an autosomal dominant vascular dysplasia.^{1–3} It is manifested by mucocutaneous telangiectases and arteriovenous malformations (AVMs). Lesions can affect the nasopharynx, central nervous system, lungs, liver, gastrointestinal tract and conjunctiva. Recurrent epistaxis is the most common presentation. Diagnosis of HHT is made clinically on the basis of the Curaçao criteria: epistaxis, telangiectasia, visceral lesions and family history.¹ The prognosis varies, depending on the severity of symptoms. Treatment is focused on prevention of complications and supportive measures.^{1 4 5}

The authors emphasise that this uncommon disease requires careful anaesthetic and surgical planning to guarantee success of the procedure. We believe enhanced regional anaesthesia is a good option in some cases.

CASE PRESENTATION

A 53-year-old Caucasian woman, body mass index 24.7 kg/m² (61 kg; 167 cm), with left trochanteric fracture, was admitted for urgent orthopaedic surgery (DHS—dynamic hip screw). She had been diagnosed with HHT at 40 years of age, and presented with a history of recurrent epistaxis,

telangiectases, severe gastrointestinal bleeding episodes (multiple transfusions requirements) and positive familial history of HHT (father, sister, brother and cousins diagnosed with HHT).

She had been submitted to a nasal septoplasty 4 weeks prior to this surgery, for correction of epistaxis and presented to us with total nasal occlusion.

The patient was taking: aminocaproic acid, pantoprazol and sucralfate.

Physical examination revealed multiple red raised lesions on her tongue, lips and hands, and total nasal occlusion with severe oedema. She was rated at Mallampati classification II, with no predictors for difficult airway management.

Remaining physical examination showed no further exterior lesions. Laboratory data revealed chronic anaemia. Records documented gastrointestinal AVMs (stomach and duodenum). Further examination excluded pulmonary and cerebral vascular malformations.

TREATMENT

The patient was submitted to a lumbar plexus block with 15 mL of mepivacaine 1.5% and 15 mL of ropivacaine 0.56% and a sacral plexus block with 15 mL of mepivacaine 1.5% and 8 mL of ropivacaine 0.56%. The blocks were performed with neurostimulation.

She was sedated with midazolam (total of 3 mg) and propofol (total of 100 mg) intravenous and spontaneous ventilation was maintained.

Standard monitoring was carried out according to the American Society of Anesthesiologists.

The surgical and anaesthetic procedures were undertaken without complications and took 50 min.

Following surgery, the patient was transferred to the post anaesthesia care unit (PACU), where she remained for 9 h. Pain relief in the PACU was performed with: acetaminophen 1 g intravenous and tramadol 100 mg intravenous (numeric pain rating scale—6).

The patient was discharged from hospital 2 days following surgery.

DISCUSSION

HHT is an inherited autosomal dominant disorder with an incidence between 1 in 5000 to 1 in 8000 and high penetration (97%).² Homozygosity appears to be incompatible with life. Distribution is homogeneous for sex and race.

HHT forms include type 1, type 2, type 3 and juvenile polyposis/hereditary telangiectasia

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syndrome. Patients with type 1 usually develop symptoms earlier and are more likely to have pulmonary and cerebral AVMs. Patients with type 2 and 3 are associated with higher risk of hepatic involvement. Juvenile polyposis/hereditary telangiectasia syndrome is a condition in which patients have AVMs and gastrointestinal polyps.^{6 7}

HHT have been linked to mutations in several genes. Type 1 is caused by mutations in ENG 9q33-34 (endoglin gene); type 2 in ACVRL1 gene; and juvenile polyposis/hereditary telangiectasia syndrome in SMAD4 gene. The gene involved in type 3 of the disease is not known but is thought to be located on chromosome 5.⁷

The mutations can be hereditary or a consequence of spontaneous mutation (up to 20% of cases).¹

The first described cases of HHT were published by Sutton (1864). Later, Rendu (1896), Osler (1901) and Weber (1907) described several cases of telangiectasia and epistaxis—but it was Hanes (1909) who named the disease ‘hereditary haemorrhagic telangiectasia’.¹

The initial lesion is due to alterations in angiogenesis (a disruption in balance between proangiogenic and antiangiogenic signals) that lead to friable blood vessels and spontaneous bleeding. Lack of communicating capillaries and veins results in AVM. The mechanism has not been completely elucidated.

Historically, HHT was described as a triad of symptoms: recurrent epistaxis, mucocutaneous telangiectases and positive family history. Currently, the diagnosis is still clinical and based on the Curaçao criteria (1999): recurrent and spontaneous epistaxis, telangiectases (lips, nose, oral cavity, hands), AVMs (lungs, nervous system, gastrointestinal system) and positive family history (first degree family member affected). The diagnosis is made in the presence of three or more of these criteria.

Patients affected with HHT may be asymptomatic, but the majority present with symptoms that began in the fourth decade of life (90%).^{2 8}

The most common manifestations are: epistaxis (90–95%), facial telangiectases (33%) and hand telangiectases (41%). About 15–33% of patients with HHT have pulmonary AVM but the incidence varies according to the gene involved.¹ Pulmonary AVMs are usually located in the lower lobes of the lungs.¹ More than 15% of patients have at least one cerebral AVM (most of them since childhood). Gastrointestinal bleeding from AVMs occur in about 8–12% of patients.¹ AVM can develop in any system, including spinal, coronary and renal.⁸

Nasal telangiectases bleeding varies from mild to severe forms and patients often require multiple blood transfusions due to severe anaemia.

Pulmonary, gastrointestinal, hepatic and cerebral AVM can lead to potentially life-threatening situations.⁸

All patients with HHT should be studied and screened for pulmonary and brain AVM. Hepatic AVMs are uncommon but they can lead to congestive cardiac failure and can pose a great risk to the patient. A liver transplant may highly improve quality of life in patients with hepatic involvement.^{9 10}

Treatment is symptomatic. Chronic bleeding can lead to anaemia, which can be treated with blood transfusions, iron supplementation and tranexamic or aminocaproic acid.^{2 3} Non-steroidal anti-inflammatory drugs, anticoagulants and antiplatelet agents must be avoided. Sometimes, surgery is needed to control nasal bleeding (Saunders or Young procedures) or endoscopic/embolisation techniques to treat nasal, gastrointestinal or pulmonary AVM. Gastrointestinal bleeding can also be treated with hormonal therapy (oestrogen and progesterone). Surgical treatment of a brain AVM depends on its location and

size. There are also some experimental treatments such as use of antiangiogenesis drugs (bevacizumab and thalidomide).

Most people have a normal lifespan but the lesions can affect quality of life.

The anaesthesiologist must be prepared to manage a patient with HHT. These patients are frequently proposed for embolisation procedures or other procedures not related to the disease.

A patient with HHT proposed for surgery requires a careful anaesthetic plan of care.^{4 5 8 11} The main anaesthetic objectives are: maintenance of normal haemodynamic parameters, and prevention of bleeding and formation of emboli.

A thorough preoperative assessment is essential. The first priority is to avoid bleeding from telangiectases or AVMs. Since telangiectases are frequent in the oral cavity, the airway management must be gentle and, if possible, avoided. Airway materials should be lubricated to minimise tissue trauma and nasal instrumentation should be avoided (nasotracheal intubation and gastric nasal probes).⁵

The patient can present with hypoxaemia due to right-to-left shunt in the presence of a pulmonary AVM. Positive airway ventilation can exacerbate shunting, so spontaneous ventilation should be maintained whenever possible.⁵

Patients with cerebral AVM require careful haemodynamic management and increases in intracranial/systolic blood pressures must be avoided (especially during intubation/extubation).⁵

Patients with systemic AVM can have left-to-right shunting and a decreased systemic vascular resistance, so anaesthetic induction should be carefully performed and fluid status optimised before intervention. Response to hypotension may be unpredictable and sometimes patients do not respond to vasoconstrictor drugs.⁵

These patients are at a higher risk for endocarditis due to pulmonary AVM so a prophylactic antibiotic is recommended before surgical or dental procedures.⁵

Patients should be in a state of deep anaesthesia at the extubation, to avoid an increase in intracranial/systolic blood pressures.

In the case reported, all airway manipulation was avoided to prevent bleeding from oral telangiectases. Since the patient had had a very recent nasal septoplasty and occlusion with visible signs of pronounced oedema, the anaesthesiologist at all cost avoided assisted ventilation with a facial mask. Regional anaesthesia was performed with clinical success. Neuraxial anaesthesia is not contraindicated in patients with HHT (unless coagulation studies are abnormal), but is controversial especially if the patient was not studied for spinal AVM.^{8 12}

Conclusions

Anaesthetic management of patients with HHT is a challenge. There is no standard approach. Patients should be carefully studied before surgery and anaesthesiologists should have a meticulous plan to reduce morbidity and mortality in patients with HHT.^{4 5 8 11} Clinical presentation is characterised by mucocutaneous telangiectases (especially on the nose and in the oral cavity) and AVM. AVMs can affect the gastrointestinal system, lungs, brain, spine and liver.¹ Symptoms vary from mild to severe, and acute complications can be life-threatening. The disease has no cure and treatment is symptomatic.

Airway assessment must be gentle to avoid bleeding from tissue trauma. Non-steroidal anti-inflammatory drugs, anticoagulants and antiplatelet agents must be avoided.

Contraindications to neuraxial anaesthesia are the same for paravertebral blocks.

Paravertebral blocks are not contraindicated in patients with HHT, although their use still remains controversial.

It is important to exclude spinal AVM in patients with HHT before giving neuraxial/paravertebral anaesthesia.

The authors opted for a paravertebral block to avoid general anaesthesia and airway manipulation.

Peripheral regional anaesthesia is a safer option than general anaesthesia.

Taking into account the patient's disease, the surgery proposed and the absence of contraindications to neuraxial anaesthesia, the authors considered a combined lumbar and sacral blockade a good anaesthetic option.

Learning points

- ▶ Anaesthetic management of patients with hereditary haemorrhagic telangiectasia (HHT) is a challenge and there is no standard approach.
- ▶ The anaesthetic management of patients with HHT should be meticulous planned to reduce morbidity and mortality.
- ▶ Whenever possible, regional anaesthesia is a valid option for these patients.
- ▶ Although controversial, neuraxial and paravertebral anaesthesia after exclusion of spinal arteriovenous malformation is a safe option.

Contributors All the authors contributed to the anaesthetic planning, provided the anaesthesia and wrote this clinical report.

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