Hereditary hemorrhagic telangiectasia: transient ischemic attacks

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See related primer by Grand’Maison, page 833, and clinical images by Nanda and Bhatt, page 838, and by Irani and Kasmani, page 839

A 39-year-old right-handed man presented to his family physician with sudden-onset weakness in his right arm and leg that had lasted 10 minutes and resolved completely. In the year before presentation, he had an episode of weakness on the right side of his face and difficulty comprehending that had lasted for 5 minutes. The patient reported having an unexplained collapse 4 years before presentation that had not been investigated.

The patient did not smoke, and he did not use drugs or alcohol. He had no history of hypertension, diabetes, coronary artery disease or hyperlipidemia. There was no history of hemoptysis, dyspnea or fatigue. The patient reported having recurrent nosebleeds since early childhood, and his mother also had nosebleeds regularly and “low blood oxygen levels.” His maternal grandmother had experienced numerous “mini-strokes.”

On examination, the patient was not in distress. He had a regular pulse rate, his blood pressure was 116/65 mm Hg, and his respiratory rate was 15 breaths per minute. He had finger and toe clubbing and several telangiectasias on his lips, tongue, gums and conjunctiva (Figure 1). The results of the rest of the clinical examination were normal.

Initial investigation showed that the patient had a normal hemoglobin level (151 [normal 120–160] g/L) and reduced oxygen saturation (90% [normal ≥ 95%] on room air). The levels of his electrolytes, glucose, lipids and inflammatory markers were normal, as were the results of a hypercoagulable screen. Because we suspected pulmonary arteriovenous malformations, we measured the patient’s arterial blood gases in the supine and standing positions. The patient had a supine pH of 7.4, a partial pressure oxygen of 60.2 (normal 80–100) mm Hg, a partial pressure carbon dioxide of 34.2 (normal 35–45) mm Hg and an oxygen saturation of 91%. When standing, his pH level was 7.4, his partial pressure oxygen was 56.8 mm Hg and his carbon dioxide level was 32 mm Hg with oxygen saturation of 89%. A shunt fraction of 21% was calculated by use of the 100% inspired oxygen breathing method (upper end of physiological shunt 5%–8%). Pulmonary arteriovenous malformations can lead to unexplained hypoxemia with further desaturation occurring when standing (orthodeoxia), as occurred in our patient. This finding is because of greater gravity-induced blood flow through basally situated pulmonary arteriovenous malformations which thereby increases the right-to-left shunt and hypoxemia.1 The results of cardiography and chest radiography were normal.

Key points

- Hereditary hemorrhagic telangiectasia may present with neurologic manifestations secondary to arteriovenous malformations in the lung, liver, brain or spinal cord.
- Neurologic manifestations may include headache, stroke, seizure, cerebral abscess, encephalopathy and spinal compression syndromes.
- Hereditary hemorrhagic telangiectasia should be considered in younger patients presenting with stroke or transient ischemic attack if the cause is unclear.
- In these patients, a thorough personal and family history of epistaxis and of respiratory and neurologic symptoms should be elicited.
- These patients should be closely examined for the presence of telangiectasias.

Figure 1: A 39-year-old man with clubbing of his fingers (A) and telangiectasia of the gums (B).
Our patient’s diagnosis of hereditary hemorrhagic telangiectasia was made within a few days after presentation. However, he had previously been examined in hospital for neurological and gastrointestinal complaints, and hereditary hemorrhagic telangiectasia had not been recognized. Box 1 presents the neurologic sequelae of hereditary hemorrhagic telangiectasia. If the syndrome had been suspected earlier, appropriate intervention to treat his pulmonary arteriovenous malformations might have prevented his transient ischemic attack and, at the very least, allowed earlier testing and family screening.

We encourage physicians who encounter younger people with stroke or transient ischemic attack to ask questions about epistaxis and their family history of respiratory and neurological symptoms, and to closely examine the patient for telangiectasias as part of usual practice, especially when the cause is unclear.

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REFERENCE