

## Answer and discussion

A diagnosis of hereditary hemorrhagic telangiectasia (*Osler-Rendu-Weber syndrome*) was made, fulfilling all four Curaçao diagnostic criteria<sup>1</sup> (spontaneous and recurrent nose bleeds, telangiectasias in characteristic sites, visceral lesions, and a first-degree relative with the disease).

It is an autosomal dominant genetic disorder with variable penetrance that has a prevalence of more than 1/10,000 inhabitants.<sup>1-3</sup> The manifestations of the disease develop with increasing age and are characterised by epistaxis, telangiectasia, and visceral involvement.<sup>1,2</sup> Among the visceral manifestations, pulmonary arteriovenous malformations (PAVMs) are the most prevalent (30%) and the ones that most commonly result in clinical complications.<sup>2-4</sup>

PAVMs are abnormal communications between pulmonary arteries and veins, bypassing the pulmonary capillary bed. They are usually multiple and predominantly located in the lower lobes. These right-to-left shunts lead to exertional dyspnoea, clubbing, cyanosis, polycythaemia, and thoracic murmurs. They also allow the passage of emboli and bacteria into the systemic circulation, leading to central nervous system infections, transient ischemic attacks (TIAs), and ischaemic strokes. A central nervous system presentation is seen in 20–40% of patients with PAVMs.<sup>2,3,5</sup> Diagnosis of PAVMs can be made with helical computed tomography of the chest.<sup>2</sup>

The treatment of PAVM consists of transcatheter occlusion of the vascular abnormalities, particularly when the feeding vessel is  $\geq 3$  mm in diameter, usually with coils (Figures 3 and 4, showing embolisation coils in the lungs). After treatment, patients should have long-term follow-up, including regular helical chest CT, as recanalisation of occluded vessels or enlargement of non-treated PAVMs is possible.<sup>2,3</sup>

Medications that interfere with platelet aggregation should be used with caution or best avoided as they may increase the risk of bleeding. Patients with evidence of pulmonary shunts should receive antibiotic prophylaxis at the time of dental and surgical procedures to reduce the risk of brain abscess.<sup>3,4</sup>