Brunei International Medical Journal(1999) 1, 449-454

Grand Round

Hereditary haemorrhagic telangiectasia
From coils to genes

Authors

Case presented by: Carlton A W Evans, Registrar in Infectious Diseases and Respiratory Medicine*
Consultant: JMB Hughes, Professor of Thoracic Medicine, Hammersmith Hospital
Radiology: J Jackson, Consultant Radiologist, Hammersmith Hospital
Discussion: J Scott, Professor of Medicine, Hammersmith Hospital
Chairman: Sir Colin Dollery, Dean of Royal Postgraduate Medical School
Series edited by: M Whyte & W Lynn

* Currently: Research Registrar, Department of Medicine, Addenbrooke's Hospital, Cambridge, UK.

Summary

A 36 year old man with hereditary haemorrhagic telangiectasia (HHT) causing pulmonary and cerebral arteriovenous malformations (AVM) was successfully treated with pulmonary steel coil embolisation. Patients with pulmonary AVM are at risk of cerebrovascular accidents or cerebral abscesses if left untreated. This patient suffered such a complication before his pulmonary AVM was discovered and treated. Most pulmonary AVM are associated with HHT and relatives of people with AVM or HHT should be offered screening. By means of a family screening programme, 9 affected relatives of this case have been detected. From 3 large affected families a gene for HHT has been mapped to the long arm of chromosome 9. The implications for diagnósis and therapy are discussed.

Case History

A 36 year old man suddenly developed headache and right arm weakness. CT scan of the brain revealed that this was due to a cerebral haemorrhage and magnetic resonance imaging revealed two small cerebral arteriovenous malformations (AVM), one of which had bled. He made a full spontaneous recovery and the cerebral AVM were treated elsewhere with stereotactically-guided radiotherapy; a technique referred to as the 'gamma knife' (Coffey et al, 1995). He has had no neurological problems since. At that time his chest radiograph was found to be abnormal. He was referred to the Hammersmith Hospital for further investigations, this being a national referral centre for pulmonary AVM.

He was a healthy man, working as a chef and as a soldier in the Territorial Army. His only symptom was epistaxis, occurring 2 to 5 times a week. His brother, sister and father were similarly affected by nosebleeds and his father had died from a cerebral abscess.

Examination was normal except for widespread telangiectasia on his skin and mucous membranes (see Figure 1), suggesting the clinical diagnosis of hereditary haemorrhagic telangiectasia (HHT). There was no residual neurological deficit, no cyanosis and no pulmonary murmur.

Chest x-ray showed a round 5 cm opacity in the lower lobe of the right lung and CT scan of the chest revealed the characteristic feeding and draining vessels of a pulmonary AVM. Radionucleide perfusion scan showed that 8 % of his cardiac output was bypassing the pulmonary capillaries by flowing through the lesion. These investigations confirmed the presence of an asymptomatic pulmonary AVM which put him at significant risk of cerebrovascular accidents or brain abscesses caused by paradoxical emboli.

After careful discussion he elected to have the pulmonary AVM embolised. A 'pig-tail' catheter was passed through the femoral vein, right atrium, pulmonary artery and under direct fluoroscopic vision into the pulmonary AVM. A metallic coil was passed through the catheter into the AVM which was seen to be occluded as a result (Figure 2). This embolisation of the single pulmonary AVM was uncomplicated.

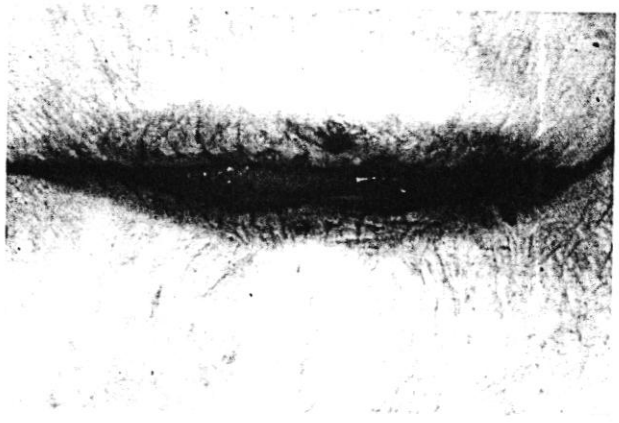

Figure 1
Telangiectasia visible on the lips of a woman with HHT.

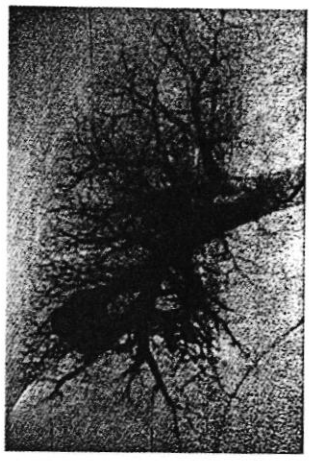

Figure 2(a)

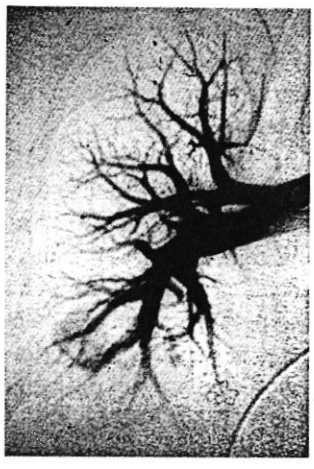

Figure 2(b)

Figure 2: Pulmonary angiogram of the patient described in the text. (a) Diagnostic angiogram with 'pig-tail' catheter in the right pulmonary artery showing the AVM (b) Post-embolisation study showing occlusion of feeding artery and pulmonary AVM.

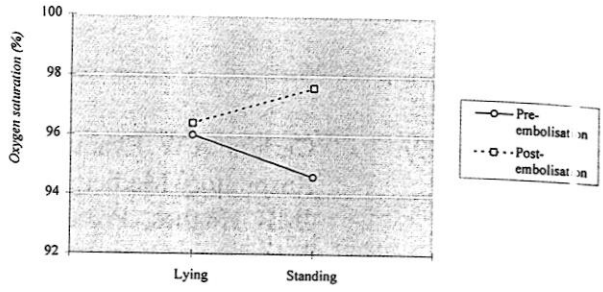

Figure 3(a)

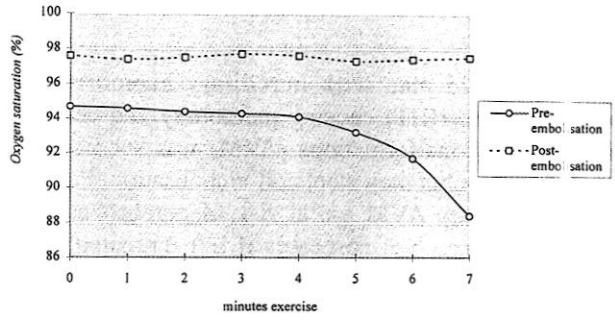

Figure 3(b)

Figure 3: Transcutaneous measurement of blood oxygen saturation (multiple readings over 30 minutes) of the patient described in the text before (solid line) and after (broken line) embolisation therapy. (a) Whilst lying and standing. (b) During exercise (6 minutes exercise was equivalent to 180 Watts)

The response to this therapy was confirmed by pulse oximetry, a simple and non-invasive way of measuring the percentage oxygenation of the blood with a sensor on the finger or ear. Figure 3a shows average oxygen saturation of the blood whilst lying and standing (multiple readings over 30 minutes). Before embolisation there was a fall in oxygenation on standing. This occurs because AVM are concentrated in the lower parts of the lung which are better perfused when standing. After embolisation there was improved baseline oxygenation and no fall in oxygenation on standing. Similarly, Figure 3b shows that before therapy exercise induced significant oxygen desaturation which completely resolved after embolisation.