Hereditary haemorrhagic telangiectasia presenting as cerebral abscess

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Abstract:

A young adult female patient was referred to the neurosurgical unit with a cerebral abscess. She was cyanosed and had clubbing of her fingers. Examination revealed multiple telangiectases and a loud systolic murmur was heard all over the precordium. Investigation showed pulmonary arteriovenous malformations, and a diagnosis of hereditary haemorrhagic telangiectasia was made.

Case history:

A 23 year old woman presented to the neurosurgical unit with a cerebral abscess in the region of the right superior temporal gyrus (Figure 1).

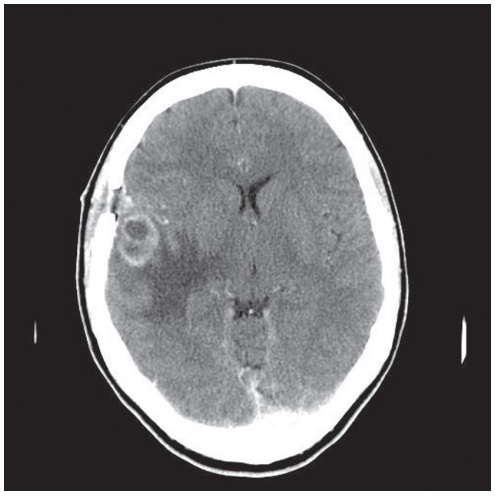

Figure 1: CT head post contrast showing abscess in right tempero-parietal region

She was cyanosed and had finger clubbing. Auscultation revealed a systolic murmur heard all over the precordium. Her partial pressure of oxygen breathing air was 70%.

Following burr hole aspiration of the cerebral abscess she was investigated further. Her trans-oesophageal echocardiogram was normal from a structural point of view; but injection of agitated saline showed delayed appearance in the left heart raising the possibility of a pulmonary arteriovenous malformation (AVM). A subsequent CT angiogram showed a large arteriovenous malformation in the left upper lobe and another malformation in the right middle lobe in addition to several smaller malformations (Figures 2&3).

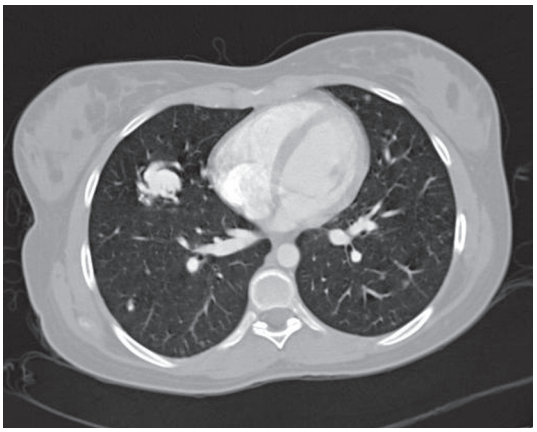

Figure 2: Axial CT showing AVM in right middle lobe

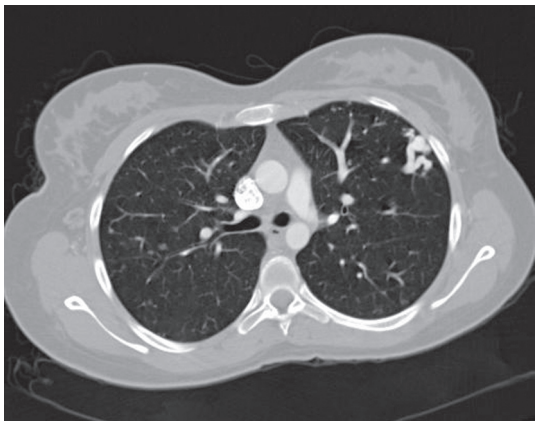

Figure 3: Axial CT showing AVM in left upper lobe

The patient on review showed multiple small telangiectases on her chest, arm and tongue raising the possibility of hereditary haemorrhagic telangiectasia. She gave a history of frequent nose bleeds, haemoptysis and

menorrhagia. Her mother died in her thirties from a haemorrhagic stroke. Her grandfather and aunt both had nose bleeds. In the past she had been investigated for polycythemia. DNA analysis at that time did not detect JAK 2 mutation, excluding polycythemia vera and the cause of her polycythemia was thought to be chronic hypoxia.

She was referred to the vascular radiologist for the coil embolization of the larger pulmonary AVMs.

Discussion:

Hereditary haemorrhagic telangiectasia (HHT) or Rendu-Osler-Weber disease is a rare fibrovascular dysplasia that makes vascular walls vulnerable to trauma and rupture, causing skin and mucosal bleeding. It is of autosomal dominant inheritance. The prevalence varies and may range from 1/3,500 to 1/5,000[1]. Two chromosomal sites have been identified: in HHT1, mutations at chromosome 9 alter the protein endoglin and in HHT2, mutations at chromosome 12 alter the protein activine or ALK-1[1]. Clinical manifestations are characterized by recurrent epistaxis, and telangiectases on the face, hands and oral cavity; visceral arteriovenous malformations and a positive family history. Epistaxis or gastrointestinal bleeding is often the first manifestation. Although rare, it may cause significant morbidity to healthy and young individuals. The cerebral manifestations of this disorder include cerebral abscess, cerebral haemorrhage and embolic stroke[2]. These cerebral manifestations are due to complications associated with cerebral vascular malformation or pulmonary arteriovenous fistulae causing right-to-left shunting, which facilitates the passage of septic emboli into the cerebral circulation. Lung is the most common site for arteriovenous malformations[3]. Transcatheter embolotherapy with detachable balloons or stainless-steel coils has been used in order to occlude such malformations.

The clinical diagnosis is based on four criteria: family history, epistaxis, mucocutaneous telangiectasias and arteriovenous malformations. The diagnosis will be definite if three criteria are present, suspected if two criteria are present, unlikely if fewer than two criteria are present[1].

Conclusion:

- The possibility of hereditary haemorrhagic telangiectasia should be sought in young patients with non-hypertensive cerebral haemorrhage, embolic stroke and iron deficiency anaemia. Search for visceral arteriovenous malformations and their treatment can prevent serious complications.
- A precordial murmur does not only reflect a valvular or structural cardiac abnormality.

References:

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