Neonatal pulmonary arteriovenous malformation in hereditary haemorrhagic telangiectasia

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A 3 week old infant presented with persistent hypoxaemia and was diagnosed with pulmonary arteriovenous malformations. Her family history was positive for hereditary haemorrhagic telangiectasia. She was treated successfully with coil embolotherapy at the age of 4 months. Transcatheter embolisation may be considered the primary treatment for pulmonary arteriovenous malformations in infancy.

Hereditary haemorrhagic telangiectasia (HHT), also known as Rendu-Osler-Weber syndrome, is an autosomal dominant mucocutaneous and visceral vascular dysplasia, characterised by the occurrence of telangiectasia and arteriovenous malformations (AVMs). Patients are usually recognised by the presence of telangiectasia, recurrent epistaxis, and a positive family history. Clinical symptoms usually appear after puberty.

First reports appear to be from around 1864 when Sutton and Babington described patients with recurrent epistaxis and vascular abnormalities. Rendu, Osler, and Weber later recognised the combination of epistaxis in a familial pattern with telangiectasia. Pulmonary AVM is the most dangerous localisation of HHT because of the risk of paradoxical septic embolism. In 1897, the first pulmonary AVM was found at the postmortem examination of a 12 year old boy. Reported series of patients with pulmonary AVM, however, indicate that such findings are uncommon in children. To our knowledge, only 12 cases of pulmonary AVM diagnosed during the first year of life have been described from 1975 onwards, the last one being reported in 2000. Most of the previously described cases were treated primarily with surgery. We report on a 3 week old infant who presented with persistent hypoxaemia as a result of pulmonary AVM.

DISCUSSION

HHT remains a clinical diagnosis despite identification of two genes that cause the disease when mutated (endoglin and ALK-1). The four clinical diagnostic criteria (epistaxis, telangiectasia, visceral lesions, and a positive family history) have been clearly defined. The diagnosis of HHT is definite when three criteria are present, but cannot be established when only two are present. These patients, however, have a high index of clinical suspicion and should be carefully examined for other subtle manifestations of this disease. In children, the diagnosis of HHT is difficult because of age related development of HHT. In our patient, only two criteria were met (visceral lesions and positive family history of HHT).

Abbreviations: HHT, hereditary haemorrhagic telangiectasia; AVM, arteriovenous malformation.
in a first degree relative), and the presence of the mutated gene coding for endoglin on chromosome 9, as has been described in her family,' was recently confirmed. Therefore, we consider her pulmonary AVM to be an expression of the same disease.

We report on one of the youngest patients diagnosed with a pulmonary AVM causing a large right to left shunt. The indication for treatment of the pulmonary AVM was persistent hypoxaemia as well as to prevent neurological sequelae caused by embolic disease. 1 Until the 1980s, the classical treatment for pulmonary AVM was surgery. Since then, embolotherapy using coils or balloons has become the treatment of choice. 1 3 Transcatheter coil embolisation of a large pulmonary AVM has been reported once before in a neonate. 2 This approach is feasible for the treatment of large pulmonary AVMs in infancy, provided that it is performed by a paediatric cardiologist. Arguments favouring surgical intervention are the complexity of the pulmonary AVM and the extent of these lesions, leaving relatively little healthy pulmonary tissue sacrificed in the case of resection of the lobe. In our patient, embolisation was performed because of the selectiveness of the procedure and to avoid the disadvantages of major surgical intervention at this age. Transcatheter embolisation may be considered the primary treatment of pulmonary AVMs in infancy, depending on the angioarchitecture, but long term results of this procedure have to be awaited. Continuing surveillance is necessary to detect short term and long term recurrence, as well as potential development of other pulmonary AVMs, in order to prevent future neurological complications, such as stroke and brain abscess.

References


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References