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Clinical Image

Embolization of Pulmonary Arteriovenous Malformations in a Patient With a Delayed Diagnosis of Hemorrhagic Hereditary Telangiectasia☆

Embolización de malformaciones arteriovenosas pulmonares en una paciente con diagnóstico tardío de telangiectasia hereditaria hemorrágica

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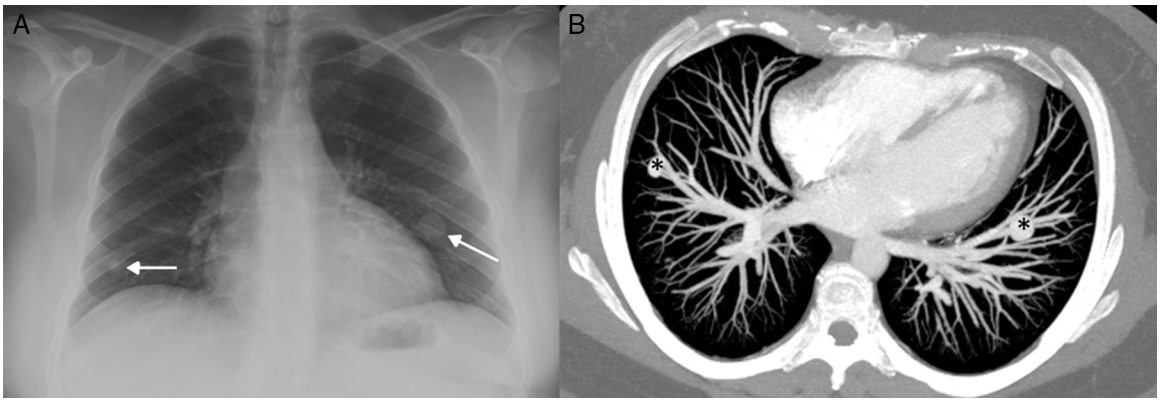

Fig. 1. (A) Chest radiograph showing 2 pulmonary nodules (arrows). (B) Reconstruction, axial computed tomography maximum intensity projection, confirming presence of pulmonary arteriovenous malformations (asterisks).

We report the case of a 42-year-old woman with a history of ischemic stroke in infancy (with resulting sequela of mild psychomotor retardation). A routine pre-operative chest radiograph revealed 2 pulmonary nodules (Fig. 1A). A chest computed tomography confirmed the existence of 2 pulmonary arteriovenous malformations (AVM) (Fig. 1B), suggesting a diagnosis of hereditary hemorrhagic telangiectasia (HHT), also known as Rendu–Osler–Weber syndrome. In view of these findings, we decided to perform percutaneous embolization of the pulmonary AVMs.

HHT is a rare autosomal dominant disease, characterized by epistaxis, mucocutaneous telangiectases, and AVM in the liver, lung, gastrointestinal tract, and central nervous system. Pulmonary AVMs predispose these patients to stroke and brain abscesses. Similarly to many diseases called "rare", HHT patients are often diagnosed late, so this entity must be taken into account in patients with a history of neurological disease who present pulmonary AVMs.[1]

Reference

1. Pierucci P, Lenato GM, Suppressa P, Lastella P, Triggiani V, Valerio R, et al. A long diagnostic delay in patients with Hereditary Haemorrhagic Telangiectasia: a questionnaire-based retrospective study. Orphanet J Rare Dis. 2012;7:53.

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