



DISORDINI EMORRAGICI CONGENITI: ASPETTI
DIAGNOSTICI E CLINICI

A CASE REPORT OF HEREDITARY HEMORRHAGIC TELANGIECTASIA WITH PULMONARY ARTERIOVENOUS FISTULA

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Background. Hereditary hemorrhagic telangiectasia (HHT), is an autosomal dominant vascular disorder characterized by heterogeneous clinical expression (recurrent epistaxis, gastrointestinal bleeding, iron deficiency anemia, mucocutaneous telangiectasia and visceral arteriovenous malformations involving the pulmonary, hepatic, and cerebral circulations). Clinical diagnosis is based on established criteria, including epistaxis, characteristic telangiectasias, visceral arteriovenous malformations (AVMs) and a positive family history. Confirmation can be achieved through molecular testing identifying genetic variants (ACVRL1, ENG, SMAD4). Management includes local and systemic approaches to control bleeding, correction of anemia, embolization of AVMs, and the use of antifibrinolytic or anti-angiogenic therapies.

Case Reports. A 57-year-old woman presented to the Emergency Department with dyspnea, severe hypoxemia (arterial pO₂ 60 mmHg) and anemia. Medical history included recurrent epistaxis and iron deficiency anemia. Contrast en-

hanced chest CT showed two large vascular lesions in the right lower lobe, consistent with arteriovenous malformations. The patient underwent successful embolization of the AVMs. Given the suspicion of HHT, otolaryngologic evaluation revealed small capillary ectasias in the right nasal cavity and mild varicose ectasias at the base of the tongue. Abdominal ultrasound demonstrated hepatic AVMs in the right lobe while brain MRI excluded cerebral involvement. Genetic testing (heterozygous variant in the ACVRL1 gene) confirmed the diagnosis of HHT. After one year of follow-up, the patient is asymptomatic for dyspnea with normal arterial pO₂.

Conclusions. HHT is a diagnostic challenge due to often underrecognized early symptoms, highly variable clinical expression ranging from mild to severe disease and silent vascular malformations. In our case, embolization of pulmonary AVMs has been completely successful. HHT requires multidisciplinary management, appropriate treatment and has a relevant familial impact due to its autosomal dominant inheritance, making family screening essential.