Clinical Images

Hereditary Hemorrhagic Telangiectasia (Osler–Weber–Rendu syndrome)

Michael A. Santos, MD^{1,2}

¹Department of Medicine, Penn State Milton S Hershey Medical Center, Hershey, PA, USA; ²WellSpan Good Samaritan Hospital, Lebanon, PA, USA.

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A 79-year-old man with hereditary hemorrhagic telangiectasia (HHT, or Osler–Weber–Rendu syndrome) complicated by portal hypertension, hypoxemia from pulmonary shunting, congestive heart failure, and gastrointestinal bleeding was admitted for recurrent epistaxis. Examination revealed extensive telangiectasias on his back (Fig. 1) and classic blanching with pressure applied by a hand (Fig. 2).

HHT is a rare autosomal dominant disorder occurring in 1 in 5000–8000 individuals.¹ Pathologic dilation of post-capillary venules leads to diffuse distribution of arteriovenous malformations in the skin, mucosal sur-

Figure 1 Clinical image of the patient's back showing multiple patches of erythema due to cutaneous telangiectasias.

Figure 2 Demonstration of the blanching over the left scapula using the examiner's right handprint (digits 3, 4, and 5 noted by black arrows).

faces and solid organs that may increase in number with age. Iron deficiency anemia from spontaneous recurrent epistaxis² and gastrointestinal bleeding³ is common. Other complications include hypoxemia from right-to-left pulmonary shunting, high-output heart failure or portal hypertension from liver involvement,^{4,5} and stroke from ruptured arteriovenous malformations in the cerebral vasculature.⁶

The classic clinical triad includes recurrent epistaxis, mucocutaneous telangiectasias, and one affected first-degree relative. Diagnosis is made by the Curaçao criteria,⁷ but up to 20 % of cases report no family history. This patient's skin manifestation is a fascinating correlate of his extensive disease. The initial diagnosis of HHT is often delayed, and therefore, the presence of multiple telangiectasias and recurrent epistaxis should prompt clinicians to consider this syndrome.

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Corresponding Author: Michael A. Santos, MD; WellSpan Good Samaritan Hospital, Lebanon, PA, USA (e-mail: masantosptt@gmail.com).

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