Mystery Case: Missed diagnosis of hereditary hemorrhagic telangiectasia presenting with recurrent ischemia

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Figure A 35-year-old woman with recurrent strokes

(A, B) T2/fluid-attenuated inversion recovery MRI shows subacute infarct in medial right occipital lobe and bilateral frontoparietal cortices. (C) Photograph of bluish discoloration of left fifth and first toes. (D) Photograph of lingual mucocutaneous telangiectasias. (E) CT angiography demonstrates large left lower lobe pulmonary arterial-venous malformation (AVM). (F) Small left cerebellar AVM on angiography.

A 35-year-old patient presented with hemianopia, decreased consciousness, and bilateral cortical foci of restricted diffusion (figure). Investigations for migraine, vasculitis, and cardiogenic stroke were negative. Four months later, the patient developed left foot claudication and discoloration. She was found to have lingual mucocutaneous telangiectasias and epistaxis. Further workup revealed pulmonary and cerebral arterial-venous malformations (AVMs). The patient met 3/4 Curacao criteria and had a definite hereditary hemorrhagic telangiectasia (HHT) diagnosis. Paradoxical emboli were the cause of her infarcts and lower extremity ischemia.

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HHT is an easily underdiagnosed cause of ischemic stroke in young adults. Delayed diagnosis results in recurrent but preventable peripheral and neurologic ischemic events.

Mystery Case Responses

The Mystery Case series was initiated by the *Neurology*[®] Resident & Fellow Section to develop the clinical reasoning skills of trainees. Residency programs, medical student preceptors, and individuals were invited to use this Mystery Case as an educational tool. Responses were solicited through a group email sent to the American Academy of Neurology Consortium of Neurology Residents and Fellows and through social media.

Fifty-five percent of respondents correctly identified the main radiographic abnormality of subacute medial right occipital lobe infarcts. Sixteen percent of respondents correctly identified the more subtle subacute bilateral frontoparietal lobe infarcts. Sixty-seven percent of respondents correctly concluded that the underlying diagnosis was HHT. The majority (52.6%) of respondents accurately stated that this is an autosomal dominant disorder. The only completely correct answer came from Dr. Reza Bavarsad Shahripour (Neurology Resident, The University of Tennessee Health Science Center).

This Mystery Case illustrates a rare and therefore commonly missed etiology for ischemic stroke in young adults. Further, this case highlights the importance of systemic features that are part of the diagnostic criteria for HHT known as the Curacao criteria.¹ The Curacao criteria include (1) epistaxis, (2) family history, (3) telangiectasias, and (4) visceral

lesions such as pulmonary, hepatic, spinal, or cerebral AVMs. Patients who meet at least 3 of the 4 criteria are classified as definite HHT. This patient had definite HHT because she met 3 out of 4 of the criteria based on epistaxis, lingual mucocutaneous telangiectasias, and both pulmonary and cerebral AVMs.

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Disclosure

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Appendix Authors

Name	Location	Role	Contribution
Waleed Brinjikji, MD	Mayo Clinic, Rochester, MN	Author	Designed and conceptualized study, drafted manuscript
Christopher P. Wood	Mayo Clinic, Rochester, MN	Author	Designed and conceptualized study, reviewed manuscript
Vivek N. Iyer	Mayo Clinic, Rochester, MN	Author	Designed and conceptualized study, reviewed manuscript

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