Mystery Case: Cutaneous lesions in *KRIT1*-associated cerebral cavernous malformations

Wallace J. Brownlee,
MBCbB, FRACP
Richard Roxburgh,
MBCbB, PhD, FRACP

Correspondence to
Dr. Brownlee:
wj.brownlee@gmail.com

Figure 1 Axial susceptibility-weighted image

The image shows multiple areas of susceptibility in the cerebral hemispheres (A). Similar lesions were seen in the brainstem, cerebellum, and spinal cord. A number of the larger lesions had a typical popcorn appearance on T2-weighted imaging, characteristic of cerebral cavernous malformations (B).

Figure 2 Nodular venous lesions and red macules

Nodular venous lesions (arrows) and red macules (arrowheads). Our patient did not display hyperkeratotic cutaneous capillary-venous malformations. These solitary, plaque-like lesions found on the limbs are present from birth.

An asymptomatic 65-year-old man was seen because of a family history of cerebral cavernous malformations (CCM) and a positive test for the *KRIT1/CCM1* (7q21.2) gene mutation. MRI of the brain

showed multiple CCM (figure 1). The patient was noted to have innumerable skin lesions over the face, trunk, and limbs (figure 2). Cutaneous lesions occur in 9% of patients with familial CCM, particularly

with *KRIT1* mutations.¹ These include red macules, nodular venous malformations, and hyperkeratotic cutaneous capillary-venous malformations.¹ Skin biopsy shows collections of abnormal, dilated thin-walled blood vessels. Familial CCM should be included with the neurocutaneous syndromes.²

AUTHOR CONTRIBUTIONS

Dr. Brownlee: preparation of the manuscript. Dr. Roxburgh: responsibility for the patient's clinical care, critical revision of the manuscript, and supervision.

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DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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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 e-mail sent to the American Academy of Neurology Consortium of Neurology Residents and Fellows and through social media.

All the responses we received came from individuals rather than groups. A total of 25% of respondents accurately identified the CCM demonstrated on MRI, and 13% identified the patient's cutaneous capillary-venous malformations. The most complete response came from Violet M. Aroon, who correctly pointed out that familial CCM is the probable diagnosis and that a mutation in the *KRIT1/CCM1* gene is the most likely underlying etiology.

This case highlights the value of obtaining a family history and performing a systemic examination to reach a rare diagnosis that might otherwise remain elusive.

Andrew Schepmyer, MD

University of British Columbia, Vancouver, Canada

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