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

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

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.

REFERENCES

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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Wallace J. Brownlee and Richard Roxburgh
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