Cherry angiomas associated with familial cerebral cavernous malformations

Case illustration

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A previously healthy 51-year-old man was awakened from sleep by vertigo associated with nausea and vomiting. When attempting to stand, he fell to the left. He was admitted to the hospital after a computed tomography scan demonstrated a pontine hemorrhage. A magnetic resonance (MR) image revealed a large pontine cavernous malformation (Fig. 1 left) as well as several smaller cerebral cavernous malformations. Our review of a cerebral MR image obtained as part of a cerebral ischemia work up in the patient’s mother also revealed multiple cerebral cavernous malformations (Fig. 1 right). The patient’s medical history was remarkable for multiple minor surgical procedures to remove more than 150 skin hemangiomas. Results of the general physical examination were remarkable for thousands of small, pinpoint-sized red macules covering most of his trunk and extremities. He also had a few larger papular lesions consistent with cherry angiomas. Both his mother (Fig. 2) and his sister have similar lesions. We made a diagnosis of familial cerebral cavernous malformations and recommended appropriate follow up for his family.

Skin lesions have been previously reported as part of the constellation of findings seen in familial cerebral cavernous malformations. The majority of these skin lesions are hyperkeratotic cutaneous capillary-venous malformations (HCCVM). On histological investigation, HCCVMs show hyperkeratosis, orthokeratosis, and dilated capillaries in the dermis and hypodermis. Our patient had no skin lesions consistent with HCCVMs. Cherry angiomas have been seen previously in families with cerebral cavernous malformations, but the relevance of the association is unclear because of their common occurrence.2

Although no genetic testing has been performed, the burden of cutaneous lesions in the proband and his mother, both of whom had multiple cerebral cavernous malformations, support a relationship between cherry angiomas and the presence of familial lesions. In addition, the preponderance of angiomas on sun-exposed surfaces of the extremities is consistent with biallelic loss of function in this disease. An endothelial cell containing a germline mutation in one allele will give rise to a cherry angioma if ultraviolet radiation damages the remaining normal allele. Examination of patients with cerebral cavernous malformations must include examination of the skin to ensure recognition of familial disease. Appropriate family screening and counseling regarding the risk of recurrence is a crucial aspect of the care of patients with cerebral cavernous malformations.

References


Fig. 1. Left: Axial T2-weighted MR image obtained in the patient, revealing a large pontine cavernous malformation and several smaller bilateral temporal lobe lesions. Right: Axial T2-weighted MR image obtained in the patient’s mother, demonstrating a cavernous malformation adjacent to the left trigone (white circle).

Fig. 2. Photograph of the patient’s mother’s upper back, demonstrating multiple punctate, red, papular lesions on the skin that are consistent with cherry angiomas. The largest is marked with an arrow.

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