VASCULAR DISEASE, VASCULITIS

CAPILLARY MALFORMATION-ARTERIOVENOUS MALFORMATION SYNDROME: A REPORT OF 2 CASES

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Background: Capillary malformation-arteriovenous malformation (CM-AVM) syndrome is an underrecognized entity with a wide clinical spectrum. It is an autosomal dominant disorder caused by mutations in the RASA1 gene. CV-AVM is characterized by multifocal cutaneous capillary malformations that are round or oval, red, brown or violaceous, erythematous macules often surrounded by a pale halo and typically distributed on the face, trunk, and extremities. It can cause fast flow vascular anomalies and arteriovenous fistulas which typically arise in the skin, muscle, bone, spine and brain and can be associated with abnormal bleeding, heart failure, and seizures.

Observation: Case 1: An otherwise healthy 2.5-year-old boy presented with multiple café au lait spots and red vascular macules and patches similar to port-wine stains, some with a whitish peripheral halo. Although CV-AVM was not initially considered, upon review of the literature it became a relevant potential diagnosis and appropriate genetic analysis was pursued. The patient was found to be a heterozygote carrier of a nonsense mutation in the RASA1 gene. Follow-up investigations including magnetic resonance imaging (MRI) showed no evidence of arteriovenous malformations in the brain or spine. Case 2: A 7-month-old boy was evaluated for lesions similar to the previous patient that were scattered across his lower extremities, anterior trunk and neck. Genetic analysis revealed a novel heterozygous frameshift variant of the RASA1 gene. His subsequent brain and spine MRI was unremarkable.

Key Message: Clinical diagnosis of CM-AVM is based on characteristic cutaneous lesions. Identification of these lesions warrants farther investigations including a genetic evaluation and appropriate imaging. This allows for definitive diagnosis and early recognition of extracutaneous vascular anomalies. Prompt clinical diagnosis is important for optimal management and may lead to improved outcomes. These cases highlight the importance of including CM-AVM in the differential diagnosis of patients with multifocal capillary malformations.