Capillary malformation-arteriovenous malformation

Inherited pial arteriovenous fistula in capillary malformation-arteriovenous malformation (CM-AVM) family [1].

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2 pediatric patients were both found to have pial arteriovenous fistulas (AVFs) with subsequent genetic analysis revealing mutations in the RASA1 gene. Considering their family history of distinct cutaneous lesions, these mutations were likely inherited as opposed to de novo mutations. Patient 1 had large capillary malformations on the left side of the face and neck, associated with macrocephaly, and presented at the age of 32 months with speech delay, right-sided weakness, and focal seizures involving the right side of the body. Patient 2 presented with proptosis at the age of 9 months but was otherwise neurologically intact. Given the chance for definitive single-stage control of vascular shunt (obviating chances for radiation exposure with endovascular treatment) and surgically accessible location of these intracranial lesions, both patients were treated with surgery with excellent clinical and radiological outcomes. In general, given the high mortality secondary to severe congestive heart failure when treated conservatively, the goal of treatment in cortical AVF in young children, even when asymptomatic, is rapid control of the shunt. This was achieved successfully in our cases - both patients experienced significant symptomatic improvement following surgery and remained neurologically stable in the subsequent follow-up visits [2].

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Li J, Yu J, Zhang H, Li G. Inherited pial arteriovenous fistula in capillary malformation-arteriovenous malformation (CM-AVM) family. Ann Neurol. 2022 Feb 13. doi: 10.1002/ana.26316. Epub ahead of print. PMID: 35152468.
2)
Chugh AJ, Shahid A, Manjila S, Gulati D, Bambakidis NC. Pial Arteriovenous Fistula and Capillary Malformation-Arteriovenous Malformation Associated with RASA1 Mutation: 2 Pediatric Cases with Successful Surgical Management. Pediatr Neurosurg. 2017;52(4):261-267. doi: 10.1159/000474942. Epub 2017 May 31. PMID: 28558378.