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VIEWPOINT

Intraoral Excision and Preoperative Embolization of a Von Hippel-Lindau Disease–associated Facial Arteriovenous Malformation

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Sir,

We have recently managed the case of a 38-year-old woman with genetically proven Von Hippel-Lindau (VHL) disease, who developed a growing, pulsatile lesion on her left upper lip, confirmed as an arteriovenous malformation (AVM). Although she exhibited many of the classical manifestations associated with the disease, including recurrent pheochromocytomas, a retinal angioma, and cerebellar hemangioblastoma, AVM was not a noted feature.

The AVM was confirmed angiographically and managed with Onyx (Medtronic, Irvine, CA) embolization,[1] administered preoperatively by interventional radiology. Subsequent surgical resection, through an intraoral route, was performed 4 days later (Fig. 1). AVMs of the face can be extensive and can cause significant deformity. The novel strategy used here resulted in a reduction in the size of the AVM, a clearer delineation of the lesion, and an excellent scar placement. With a firm lesion, blunt dissection could be performed with protection of facial nerve branches. The result was minimal disruption of facial soft tissue volume and a favorable cosmetic outcome (Fig. 2).

The VHL disease is an autosomal-dominant inherited syndrome seen in approximately 1 in 36,000 individuals.[2] It results from germline mutations in the VHL tumor suppressor gene and is characterized by highly vascularized tumors,[2] including hemangioblastomas of the cerebellum and spine, retinal angiomas, renal cell carcinomas, pheochromocytomas, endolymphatic sac tumors, and neuroendocrine tumors of the pancreas.[2] To date, there is no literature describing AVMs as part of the VHL syndrome; however, given that the genetic defect promotes angiogenesis via hypoxia-inducible

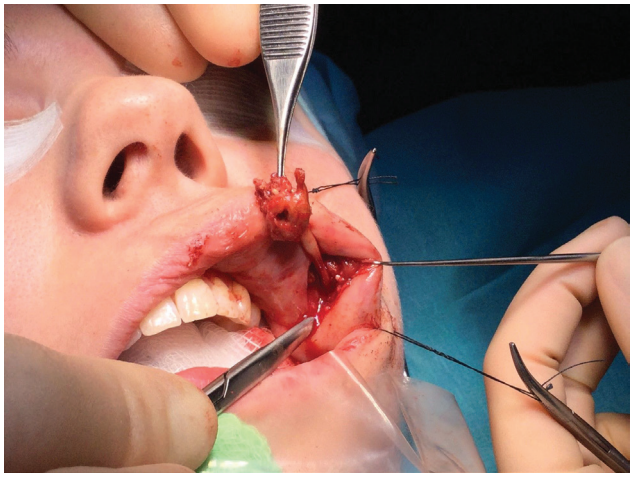

Fig. 1. An intraoperative photograph demonstrating excision of the embolized AVM as a solid mass through an intraoral route.

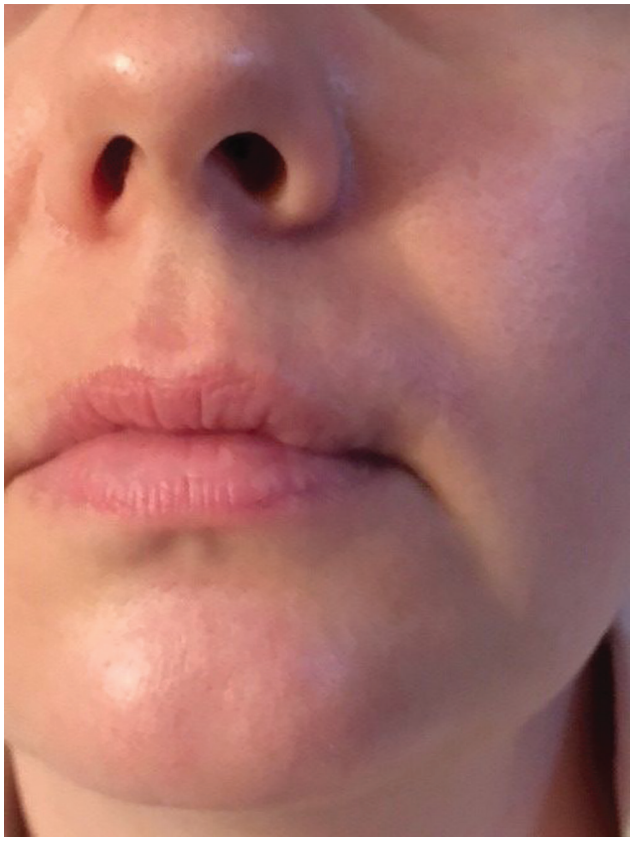

Fig. 2. A satisfactory postoperative result taken at 6-month follow-up with minimal soft tissue disruption and asymmetry.

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factor, we suggest that the 2 processes are likely to be linked.[3]

AVMs are often presumed to be congenital, but there is no direct evidence that they form in utero. Although a small number of AVMs manifest themselves at or shortly after birth, most of them present later in life with the mean age of detection being the mid-30s.[4] In this case, the tissue sample is disrupted by embolization; thus, it is not suitable for analysis to detect loss of heterozygosity of the VHL gene to confirm pathogenicity.

The potential association between AVMs and VHL disease has significant implications. The pheochromocytoma-associated catecholamine excess may be associated with hypertensive crises and intraoperative hemodynamic instability. Although we do not advocate genetic testing for a VHL mutation in every case of AVM, it would be prudent to ensure that the patient is normotensive before any surgical intervention to avoid missing catecholamine-associated hypertension from an occult pheochromocytoma, however rare.

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DISCLOSURE:

The authors have no financial interest to declare in relation to the content of this article.

REFERENCES

1. Giurazza F, Corvino F, Cangiano G, et al. Transarterial embolization of peripheral high-flow arteriovenous malformation with ethylene vinyl alcohol copolymer (Onyx): single-center 10-year experience. *Radiol Med*. 2019;124:154–162.
2. Champion KJ, Guinea M, Dammai V, et al. Endothelial function of von Hippel-Lindau tumor suppressor gene: control of fibroblast growth factor receptor signaling. *Cancer Res*. 2008;68:4649–4657.
3. Komiyama M. Pathogenesis of brain arteriovenous malformations. *Neurol Med Chir (Tokyo)*. 2016;56:317–325.
4. Haase VH. The VHL tumor suppressor: master regulator of HIF. *Curr Pharm Des*. 2009;15:3895–3903.