Dental Management of ELMO2-Associated Arteriovenous Malformations

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INTRODUCTION

Engulfment and Cell Motility 2 (ELMO2) gene is a protein that facilitates cell movement and maintains vascular integrity in intramembranous bones. Mutations in the ELMO2 gene are associated with development of arteriovenous (AV) malformation and subsequently intraosseous vascular malformations (VMOS).[1] The most affected bones in the skull are in the maxilla and mandible.[1]

ETIOLOGY AND EPIDEMIOLOGY

- Pathogenic variants in ELMO2 mutations associated with vascular malformations have been reported in literature in only six families.[2]
- VMOS is an autosomal-recessive disorder caused by the loss-of-function in the ELMO2 gene, a protein that is essential for translating extracellular signals into cytoskeletal movements to maintain vascular integrity within bone via interactions between endothelial and vascular smooth muscle cells.[1]

DIAGNOSIS AND MANAGEMENT

VMOS associated with ELMO2 mutation is diagnosed based on clinical features, radiographic findings and genetic testing.[3]

Clinical diagnosis involves history and physical examination to identify symptoms such as gingival bleeding, swelling, or bony expansion. MRI, CT or angiography imaging is essential to determine the flow dynamics, extent and involvement of neighboring structures. Genetic testing is utilized to confirm diagnosis, prognosis and initiate management with a Vascular Anomalies Care Team (VACT).[4] The goal is to reduce the risk of complications, alleviate symptoms and maintain function as there is no current cure. The VACT may include an interventional radiologist, hematologist, maxillofacial surgeon, anesthesiologist and pediatric dentist, working collaboratively to coordinate comprehensive, individualized care for patients with complex vascular malformations.

CASE REPORT

This presentation outlines a 5-year-old male who was referred to Children's Hospital Los Angeles with a chief complaint of spontaneous gingival bleeding. Patient's medical history is significant for vascular malformation (ELMO2 mutation), autism, anemia, and history of a stroke. Current medications include iron supplements. The patient has a significant history of preemptive embolizations and multiple visits to the emergency department due to severe, life-threatening spontaneous bleeding exacerbated by exfoliation of primary teeth.

Clinical evaluation shows displacement of primary dentition associated with mandibular expansion. Radiographic evaluation reveals irregular opacities consistent with previous interventional radiology (IR) embolizations. The patient subsequently underwent dental extractions with an oral maxillofacial surgeon.

Figure 1. Mandibular expansion with displacement of multiple teeth.

Figure 2. Mandibular region more than one year post-extractions.

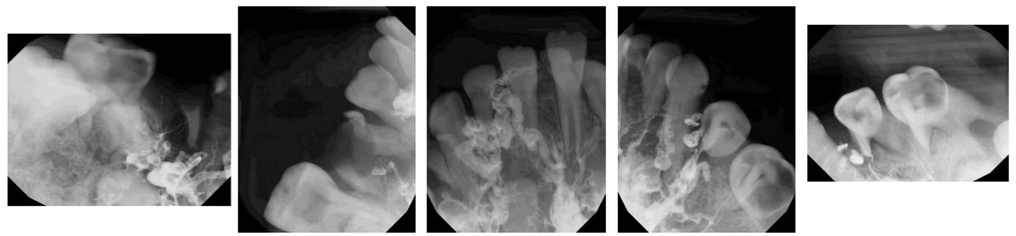

Figure 3. Multiple irregular radiopacities in the mandibular anterior region consistent with embolization materials placed to occlude abnormal blood vessels as well as bony expansion.

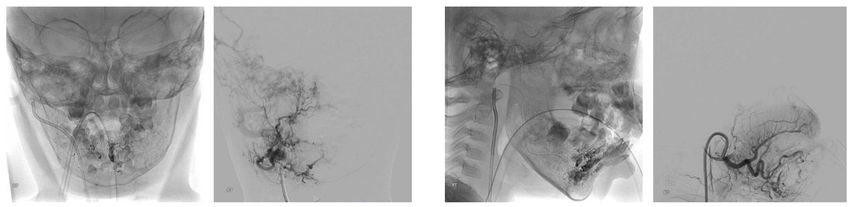

Figure 4 and 5. The frontal and sagittal view of the angiogram.
Vascular malformation highlighted by contrast with delineated abnormal vessels.

DENTAL IMPLICATIONS or CLINICAL FEATURES

- Patients with VMOS associated with ELMO2 mutation present significant dental considerations due to the high-flow vascular nature of the lesions and increased risk of life-threatening hemorrhage.
- Involvement of the maxilla and mandible may result in progressive osseous expansion, facial asymmetry, displacement of primary and permanent dentition, malocclusion, and gingival swelling with spontaneous bleeding.
- Exfoliation of primary teeth may precipitate severe hemorrhage as seen in the case report.
- Emphasis should be placed on preventive dental care, minimally invasive treatment approaches, and avoidance of unnecessary trauma to reduce the risk of bleeding complications.

CONCLUSION

VMOS associated with ELMO2 mutation is an extremely rare autosomal-recessive vascular disorder characterized by intraosseous high-flow malformations, progressive craniofacial expansion, and significant hemorrhagic risk.

This case highlights the importance of recognizing abnormal gingival bleeding and mandibular expansion in pediatric patients as potential indicators of an underlying vascular malformation. Accurate diagnosis through clinical evaluation, imaging, and genetic testing, combined with multidisciplinary collaboration, is essential for safe and effective management. Pediatric dentists serve a vital role in early detection, timely referral, and collaboration with specialized teams to ensure safe dental management in patients affected by complex vascular malformations.

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