Large pediatric arteriovenous malformations with associated cutis aplasia: Case report and systematic literature review

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Disclosures

None
Introduction

• Genetic abnormalities leading to arteriovenous malformations (AVMs) are poorly understood outside of inherited syndromes. Evidence suggests that the etiology may differ between pediatric and adult patients.

• Implicated genes include PTEN, VEGF and TGF-β, which can promote the development of these lesions.

• Here we present two pediatric patients with AVMs and cutis aplasia, and review the literature on pediatric AVMs, with a focus what is known about causal genetic abnormalities.
We reviewed two pediatric patients who presented with a yet-undescribed triad including an AVM. We discuss their clinical presentation, demographics, imaging and multi-disciplinary consensus on management.

We review the literature on vascular syndromes in pediatric patients, and the genetics responsible for AVMs in this population.
Results

- Two pediatric patients presented with large, deep AVMs. Both additionally have ipsilateral cutis aplasia, as well as a small path of alopecia on the ipsilateral eyebrow.

- These patients do not meet criteria for known craniofacial vascular anomaly syndromes, and genetic testing is ongoing. They are clinically stable with no AVM rupture at last follow up.
Results

• The second patient was a 5 year old female who underwent imaging due to the cutis aplasia, but was otherwise asymptomatic.

Figure 1. A) Vertex cutis aplasia B) Right eyebrow alopecia C) T2 weighted MRI showing right thalamus AVM
The first patient was a 13 year old male who presented with worsening dystonia, which was initially attributed to cerebral palsy until imaging showed a right thalamic AVM.

Figure 2. A) Coronal T2 weighted B) Axial T2 FFE C) Axial T1 post-contrast MRI showing right thalamic AVM
Discussion

• Cerebrovascular malformations often occur as part of genetic syndromes

• Specific genetic mutations have also been implicated in isolated, non-syndromic vascular lesions

• The patients presented do not fit classic syndromes, and genetic testing is ongoing

• Patients are followed by the vascular anomalies multi-disciplinary team, and the AVMs will be monitored with serial imaging as long as they remain asymptomatic

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Genes</th>
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<tbody>
<tr>
<td>Hereditary Hemorrhagic Telangiectasia (HHT)</td>
<td>TGF-β, SMAD4, Endoglin (ENG)</td>
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<tr>
<td>Sturge-Weber</td>
<td>GNAQ</td>
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<tr>
<td>Familial Cerebral Cavernous Malformation (FCCM)</td>
<td>KRIT1, CCM2, or PDCD10</td>
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<tr>
<td>PHACE</td>
<td>GLUT-1</td>
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<td>Capillary Malformation/Arteriovenous Malformation (CM/AVM)</td>
<td>RASA1</td>
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<tr>
<td>Isolated Lesions</td>
<td>NOTCH, IL-2, TNF-α</td>
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</tbody>
</table>

Table 1. Examples of genetic syndromes associated with cerebrovascular malformations and implicated genes
Summary

• There are innumerable factors which lead to vascular pathology in pediatric patients

• The genetics underlying cerebrovascular development and pathology is still largely uncharacterized

• Pediatric patients with complex vascular lesions should undergo assessment by multi-disciplinary treatment teams