Intracranial Vessel Stenosis in a Young Patient with an
MYH11 mutation: A Case Report and Review of Two Prior Cases

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Disclosures

• All authors report no disclosures
Introduction

• **MYH11** is a gene that codes for smooth muscle myosin heavy chain which is involved in blood vessel wall integrity\(^1\)

• It is well-understood that patients with **MYH11** mutations have extracranial arterial wall abnormalities including aortic dissections and aneurysms\(^1\)

• Implications of **MYH11** mutations on intracranial vasculature is relatively unknown

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Case Presentation

- A 29-year-old female with a history of aortic dissection presented with diplopia, vertigo and headaches
- Her history was otherwise unremarkable
- Magnetic Resonance Angiogram (MRA) of the brain was obtained
Intracranial Vessel Stenosis in a Young Patient with an MYH11 Mutation

Initial Imaging

Axial MRA demonstrating stenosis of the proximal middle cerebral arteries (MCAs, arrows)

Coronal MRA demonstrating stenosis of the bilateral internal carotid artery (ICA) termini (black arrows), anterior cerebral arteries (ACAs, solid white arrows) and posterior cerebral arteries (PCAs, dashed white arrows)
Intracranial Vessel Stenosis in a Young Patient with an *MYH11* Mutation

**Initial Imaging**

Sagittal MRA showing generalized straightening of the intracranial vessels
Genetic Testing

• Cerebral angiographic phenotype seen on initial imaging has been associated with ACTA2 mutations, which codes for smooth muscle alpha actin

• She was negative for ACTA2 mutations and other connective tissue diseases

• She tested positive for an MYH11 IVS28 c.3858+1G>A (IVS28+1G>A) mutation
Intracranial Vessel Stenosis in a Young Patient with an \textit{MYH11} Mutation

51-month follow-up

- No new neurological symptoms
- No changes of intracranial stenosis
- No evidence of basal collateral vessel development

Axial MRA demonstrating persistent proximal MCA stenosis (arrows).

Coronal MRA demonstrating persistent stenosis of the bilateral ICA termini (black arrows), ACAs (solid white arrows) and PCAs (dashed white arrows). There is no evidence of basal collateral vessel development.
## Prior Reports of MYH11 Mutations with Cerebrovascular Manifestations

<table>
<thead>
<tr>
<th>Study</th>
<th>Age/Sex</th>
<th>Clinical Presentation</th>
<th>MYH11 Mutation</th>
<th>Angiographic features</th>
<th>Treatment</th>
<th>Follow-up time</th>
<th>Clinical/Radiographic outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Keylock et al.¹</td>
<td>2 yr./F</td>
<td>Acute left ACA/MCA infarct</td>
<td>NM_002474:c.4604G&gt;A(p.R1535Q)</td>
<td>Bilateral stenosis of terminal ICA and proximal MCA with Moyamoya-like collateral formation</td>
<td>Bilateral pial synangiosis</td>
<td>48 months</td>
<td>Stable residual asymmetric tetraparesis with pseudobulbar features. Progression of bilateral intracranial stenosis with increased basal collateral formation</td>
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<tr>
<td>Ravindra et al.²</td>
<td>6 mo./F</td>
<td>subarachnoid/subdural hemorrhages</td>
<td>16p13.11 (c.5273G&gt;A)</td>
<td>Some evidence of delayed distal MCA filling, otherwise normal appearance</td>
<td>Microsurgical clipping of original ruptured aneurysm, Onyx embolization of subsequent de novo aneurysm</td>
<td>3 months</td>
<td>Improvement of R-sided hemiparesis. No radiographic follow-up</td>
</tr>
<tr>
<td>Present Study</td>
<td>29 yr./F</td>
<td>Headache</td>
<td>IVS28 c.3858+1G&gt;A (IVS28+1G&gt;A)</td>
<td>Stenosis of terminal ICAs, proximal MCAs, PCAs and ACAs. Generalized straightening of vessels</td>
<td>Continuation of chronic anticoagulation and antiplatelet therapy</td>
<td>51 months</td>
<td>No new neurological events. Stable intracranial stenosis</td>
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Conclusions

• The presence of intracranial vessel involvement of patients with MYH11 mutations, though rare, suggests a role for cerebrovascular screening in these patients

• With similar stenotic phenotypes, the MYH11 gene may play a role in the pathophysiology of Moyamoya disease, though this is uncertain

• Future reporting of similar cases is therefore important