Neurovascular Findings in Children and Young Adults with Loeys-Dietz Syndrome

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Introduction

Loeys-Dietz syndrome (LDS) is a recently described autosomal dominant connective tissue disorder resulting from TGF-beta pathway mutations. Manifestations include craniofacial, skeletal, and vascular anomalies (aortic root dilation, neurovascular tortuosity, intracranial aneurysms (IAs), and dissections). Case reports describe unruptured and ruptured IAs in LDS patients. Cerebrovascular screening is thus advised. We present a series of 34 patients with LDS and describe their neurovascular imaging findings.

Methods

Patients with genetically-confirmed LDS were reviewed for neuroradiology findings. At our institution, multidisciplinary care LDS patients is coordinated by a specialized cardiogenetics clinic, which follows screening recommendations for yearly brain/neck imaging. Magnetic resonance imaging (MRI) and radiology reports were reviewed. Findings were recorded with special attention to detect those reported in the literature, including arterial tortuosity, aneurysms, and dissections. Follow-up examinations and reports were analyzed for disease progression.

Results

34 patients were reviewed. Median age was 9.7 years (range 1 week - 34 years); half were male. Mutations included TGBFR1 in 7 (21%), TGBFR2 in 18 (53%), and TGBF2 in 9 (26%). MRI head/neck was available for review in 90%. Average age at first MRI was 12.9 years. 63% showed neurovascular tortuosity. No cerebral aneurysms were visualized. 80% had follow-up MRI for review. 33% of patients with initial and follow-up neurovascular imaging showed new or worsening disease. Patients with worsening or progression (n=6) had either TGBFR1 or TGBFR2 mutations and presented at earlier age (mean 8.3 years). No patients required neurovascular intervention during surveillance.

Conclusions

LDS is a rare genetic disease with multisystem involvement, including the vascular system. Guidelines recommend yearly surveillance for monitoring of cerebrovascular disease in LDS. Our series suggests less frequent imaging in the pediatric LDS population may be reasonable due to lack of clinically actionable vascular findings. Further study and follow-up is warranted, along with correlation with cardiothoracic disease severity.

Learning Objectives

By the conclusion of this session, participants should be able to:

1) Describe the classic presentation and pathology of Loeys-Dietz Syndrome

2) Describe the key neurovascular anomalies in LDS

3) Explain the importance of screening and management of neurovascular issues in LDS

References

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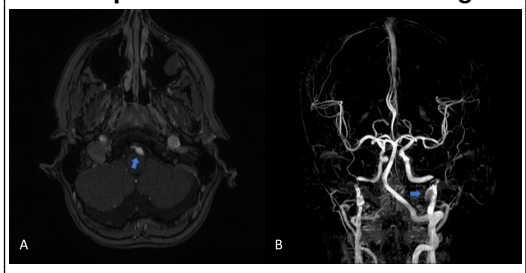
Example of Neurovascular findings



An example of some of the neurovascular pathology found in patients with Loeys-Dietz Syndrome. Axial MRA (a) shows a small 1x2mm aneurysm of the basilar artery near the origin of the right AICA. 3D reconstructed MRA (b) shows a large dissecting aneurysm of the distal left cervical ICA.

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