Loeys-Dietz syndrome

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A 3-month-old boy presented with respiratory tract infection and was found to have a posterior mediastinal mass on chest radiography. CT of the chest (Fig. 1) showed an aortic dissection in the descending thoracic aorta causing a pseudoaneurysm measuring approximately 4×4 cm. The child had a family history of intracranial aneurysms. MR angiography of the head (Fig. 2) showed elongated, tortuous intracranial vessels. The child also had a bifid uvula. The thoracic pseudoaneurysm was repaired with an end-to-end anastomosis.

Fig. 1 Chest CT image

Fig. 2 MR angiogram of the head

Loeys-Dietz syndrome (LDS) is an autosomal dominant syndrome caused by mutations in genes encoding transforming growth factor beta receptor (TGFB β 1 or TGFB β 2) [1]. Marfan and Ehlers-Danlos syndromes share many features, including aortic and arterial aneurysms/dissections with pronounced tortuosity (corkscrew structure) of arteries [2]. It is important to distinguish among them because vascular surgery has a better prognosis in children with LDS. Cleft palate or bifid uvula and orbital hypertelorism are helpful distinguishing features seen in LDS.

References

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