A 3-year-old boy being followed up for bilateral club foot underwent a routine thorax radiography that revealed aortic arch enlargement. Echocardiography showed sinus of Valsalva dilatation. Because of clinical features such as hypertelorism, bifid uvula, and prominent forehead, a genetic investigation was conducted that confirmed Loeys-Dietz syndrome (LDS) by identifying a heterozygous mutation in the gene encoding transforming growth factor-β receptor type 2 (TGFBR2). Whole-body magnetic resonance angiography was performed on a 1.5T machine. The magnetic resonance protocol included 3-dimensional contrast-enhanced angiographic sequences of the body and time-of-flight sequences of the brain. Whole-body magnetic resonance imaging demonstrated typical cardiovascular findings: dilatation of the aorta at the level of sinus of Valsalva; tortuosity of the aortic arch, the 2 internal carotid arteries, the 2 vertebral arteries, and left subclavian artery; and a superior mesenteric artery aneurysm (Figures 1 and 2). Brain and cervical spine computed tomography examinations were also performed and showed characteristic features of this genetic disorder, such as dolichocephaly, hypertelorism with bony interorbital distance of 2.8 cm (>95th percentile), and retrognathia (Figure 3).

LDS is a recently identified connective tissue disorder. It is caused by heterozygous mutations in the genes that encode
transforming growth factor-β receptor types 1 or 2. LDS is typically characterized by the triad of hypertelorism, cleft palate or bifid uvula, and arterial aneurysm/tortuosity. It is clinically classified into types 1 and 2. LDS type 1 can be recognized by craniofacial characteristics (hypertelorism, bifid uvula, or cleft palate). LDS 2 patients have an isolated bifid uvula but no other facial features, although subtle features such as broad forehead or frontal bossing may exist. Arterial involvement is widespread in patients with LDS; all vessels can be involved by vascular malformations such as tortuosity, aneurysm, dissection, and stenosis. Because approximately half of all individuals with LDS have an aneurysm distant from the aortic root that would be missed by echocardiography, magnetic resonance angiography or computed tomography with 3-dimensional reconstructions from head to pelvis must be performed. Advantages of magnetic resonance imaging over computed tomography include the fact that the patient does not receive ionizing radiation during the scan. The skeletal findings are characterized by Marfan syndrome–like features: Joint hyperlaxity, arachnodactyly, pectus deformity, and scoliosis. Other skeletal findings are observed, such as spondylolisthesis and acetabular protrusion.

Moreover, spine anomalies, including congenital cervical vertebrae malformations and cervical spine instability, are common. All these abnormalities should be excluded by orthopedic evaluation, flexion-extension cervical radiographs, and thoracolumbar spinal imaging. Skin findings are similar to those seen in vascular Ehlers-Danlos syndrome and include velvety, translucent skin with visible veins and easy bruising.

The differential diagnosis of LDS includes atypical Marfan syndrome, vascular Ehlers-Danlos syndrome, Shprintzen-Goldberg craniosynostosis, and familial aortic aneurysm and dissection syndrome. Arterial tortuosity syndrome is a closely related syndrome that is also characterized by severe tortuositities, stenosis, and aneurysms of large and mid-sized arteries. It is caused by a genetic mutation in SLC2A10, which encodes the glucose transporter GLUT-10, a mutation that likely leads to upregulation of transforming growth factor-β.

As in other connective tissue disorders, the risk of aneurysms rupture and dissection in LDS justifies a close follow-up by noninvasive imaging methods such as whole-body magnetic resonance angiography.

Disclosures

None.

References

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