Clinical vignette

Cardiovascular findings in arterial tortuosity syndrome

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A boy presented at birth with an axial diaphragmatic hernia, hyperelastic skin, hyperextensible joints, and arachnodactyly. During infancy, he developed bilateral inguinal herniae and generalized muscular hypotonia. Echocardiography showed an anomalous tortuous course of the pulmonary arteries with suspected stenosis of the left pulmonary artery. Magnetic resonance (MR) imaging demonstrated elongation and tortuosity of the aortic arch and its branches (Panels A and B) and severe tortuosity of the pulmonary arteries (Panels B and C), leading to a significant obstruction of the left branch pulmonary artery that was subsequently stented. At coronarangiography, both coronary arteries presented with a tortuous course without aneurysm or stenosis.

Connective tissue diseases including Ehlers-Danlos syndrome, Marfan syndrome, cutis laxa, and Menkes disease present typical features consisting of hyperelastic or lax skin, laxity of joints, chest and spine deformities, inguinal, umbilical and/or diaphragmatic herniae, and cardiovascular anomalies. Careful clinical assessment, ultrastructural analysis of the skin, molecular studies of the genes involved, analysis of the collagens produced in cultured fibroblasts, and quantification of the plasma levels of copper and coeuloplasmin may help differentiate among the different forms of the disease. However, severe vascular involvement with tortuosity, elongation, aneurysms, or stenosis of the mid-size and large vessels is suggestive of the rare arterial tortuosity syndrome.

In conclusion, echocardiographic screening is recommended for all patients with connective tissue disease, as early detection and treatment of vascular lesions may have a major influence on prognosis.

Panel A. Contrast-enhanced MR angiography demonstrating an elongation of the aortic arch, with the descending aorta located too far left within the thorax.

Panel B. The 3D reconstruction of MR angiography shows the tortuous course of the aortic branches (asterisk, brachiocephalic trunk; double asterisks, left carotic artery). Ao, aorta; MPA, main pulmonary artery.

Panel C. Tortuosity of both pulmonary arteries. The right pulmonary artery makes a loop (asterisk) before its branching.

Panel D. Kinking of the left pulmonary artery owing to a significant stenosis of the origin of the left pulmonary artery (arrow). This stenosis required catheter-guided stenting.