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STUDIUM PRZYPADKU / *CLINICAL VIGNETTE*

Huge ascending aortic aneurysm in a 7-year-old patient with Marfan syndrome

Olbrzymi tętniak aorty wstępującej u 7-letniego chorego z zespołem Marfana

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Aneurysm of the ascending aorta represents a rather rare disease condition. It may remain silent over the course of several years, without symptoms or physical examination signs. While the principal causes in adults of what is generally an isolated aneurysm are degenerative processes and atherosclerosis, in children the presence of systemic disease must be carefully investigated. A 7-year-old male patient was consulted for a swelling in the right chest. While asymptomatic, he had a 1/6 early diastolic murmur at the left midsternal border and a right parasternal swelling on physical examination. Chest X-ray showed cardiomegaly and an enlarged mediastinum (Fig. 1). Echocardiography found an aneurysmal dilatation of the ascending aorta immediately past the aortic valve, measuring 85 mm (z-score: +13.9) at its widest (Fig. 2) and mild aortic insufficiency was noted. Computerised tomographic angiography of the chest confirmed the presence of an aneurysmal dilatation measuring 88 mm at its widest point, extending from the aortic root to the left subclavian artery ostium (Fig. 3). Genetic studies performed by cDNA array analysis in polymerase chain reaction showed the presence of an exon 10 deletion. This result was diagnostic for Marfan syndrome. Aortic valve was preserved during the operation and graft replacements of the ascending and transverse aorta were performed. No postoperative complications were seen. Even in the absence of other findings suggestive of Marfan syndrome, in particular with large aneurysms, this should be kept in mind in investigating a patient. Dissection and rupture of the aortic root is the most frequent cause of death in these cases, with a peak incidence in the third and fourth decades of life. The substantial mortality and morbidity of acute aortic events contrast with the important increase in life expectancy achieved by preventative interventions. Composite valve graft and valve-sparing aortic root replacement procedures are performed under elective conditions with low morbidity and mortality.

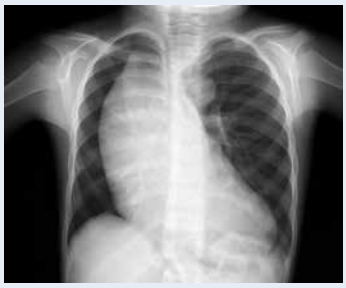

Figure 1. Chest X-ray showing cardiomegaly and an enlarged mediastinum

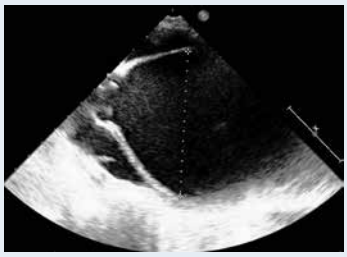

Figure 2. Echocardiography study showing an aneurysmal dilatation of the ascending aorta, immediately past the aortic valve

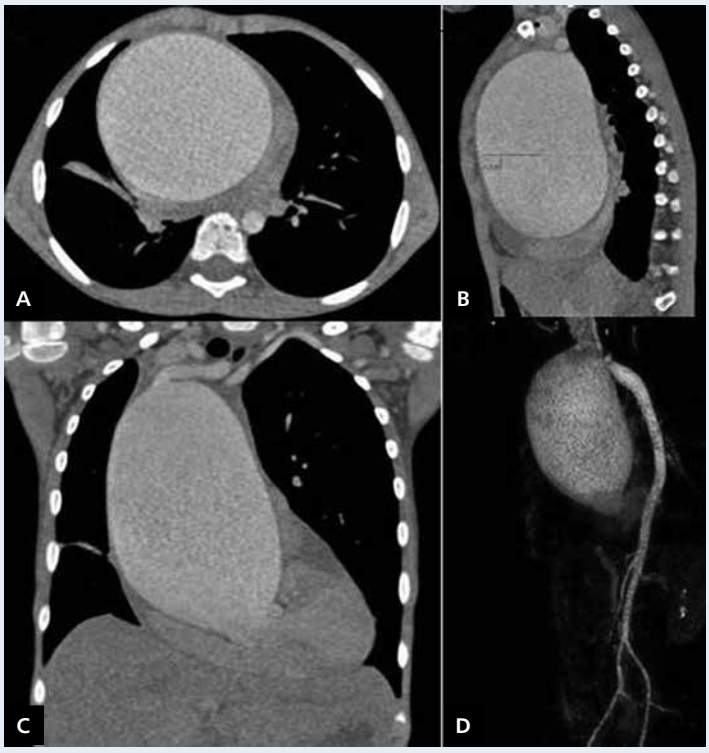

Figure 3. Axial (**A**), sagittal (**B**), coronal (**C**) and three-dimensional (**D**) computerised tomography angiography images showed aneurysmal dilatation of the ascending aorta

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Conflict of interest: *none declared*