Isolated giant ascending aortic aneurysm in a child: a novel mutation of the ACTA2 gene

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An asymptomatic 8-year-old boy was referred with a giant isolated ascending aortic aneurysm (Fig. 1). Replacement of the ascending aorta was successfully performed (Fig. 2). DNA analysis revealed no mutation in FBN1, FBN2, TGFBR1, TGFBR2, or SCL2A10. However, analysis of the ACTA2 gene showed a novel missense mutation (p.Arg39Cys (c.115C>T:ex2)).

Fig. 1. Three-dimensional (3-D) computed tomographic scan showing a giant fusiform ascending aortic aneurysm with a maximal diameter of 87 mm. The patient exhibited hypertelorism and thick lips, but did not fulfill the criteria for known genetic connective tissue disorders (A). A right lateral 3-D view of the aneurysm. There was no compression on mediastinal structures such as the superior vena cava, pulmonary arteries or pulmonary veins (pulmonary arteries are not shown) (B).
Fig. 2. Intraoperative picture shows a giant aneurysm originated 10 mm above the sinotubular junction and involved the entire ascending aorta. Replacement of the ascending aorta was performed under cardiopulmonary bypass with selective cerebral perfusion. The aortic valve was structurally normal, and the sinuses and annulus were uninvolved. Histological examination revealed cystic medial necrosis of the aortic wall with presence of type III collagen.