

Type B Aortic Dissection in childhood and MYBPC3 mutation: a Case Report

*Aries E, El Louali F., Aldebert P., Pico H., Bal L., Chabrol B., Ovaert C.
Timone enfants, AP-HM, Marseille, France*

Introduction: Non-traumatic aortic dissection is a rare disease in children. Management is standardized in adults. In children the diagnosis, etiology and treatment is less clear. We report an uncommon clinical observation with review of the literature.

Clinical Observation: This young boy presented at the age of 11 years with an ischemic stroke. Hypertrophic cardiomyopathy with non-compacted left ventricle but preserved systolic function was discovered. Genetic work-up revealed mutation of MYBPC3. Anti-coagulant therapy was introduced. At the age of 13 years he underwent surgery for significant scoliosis and pectus carinatus. Three months later he presented with thoracic pain concomitant with mild fever and inflammatory signs. The history was compatible with dry pericarditis and anti-inflammatory therapy was introduced. The pain improved but reappeared a few weeks later. A hyperechogenic retrocardiac mass was seen on echocardiography. CT scan revealed type B aortic dissection with 2 active entry points into the false lumen. There was no visceral impact. After medical-surgical multidisciplinary discussion, conservative approach was decided with medical control of arterial hypertension, discontinuation of anticoagulant therapy but introduction of anti-platelet therapy. Progressive thrombosis of the false lumen and closure of the 2 entry points was seen during the following 3 months, confirmed by CT Scan.

Discussion: The average age of patients with type B aortic dissection is 65 years. Acute mortality is high, approaching 40%. The incidence among those under 21 years is extremely low (about 0.37% of cases), and exceptional before 15 years. It is most often associated with connective tissue diseases such as Marfan syndrome. Spontaneous aortic dissection together with MYBPC3 mutation and non-compaction cardiomyopathy has so far not been described.