

Assessment of aortic dilatation and dissection in children with Marfan syndrome

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Introduction

Marfan syndrome (MFS) is a connective tissue disorder characterized by a broad range of clinical manifestations. Cardiovascular involvement is the most serious life-threatening aspect of the syndrome. Although it is well studied in adults, there is paucity of data in children. The aim of the study was to evaluate the rate of aortic dilatation and dissection in children with MFS.

Methods

Study population included 133 children, who were referred with suspicion of MFS. All patients underwent clinical evaluation (physical, ophthalmological, orthopedic, genetic and cardiac examination). Aortic diameter was measured with transthoracic echocardiography at eight different levels using two techniques (leading-edge in end-diastole and inner-edge in mid-systole). The modified Ghent criteria were used to identify patients with MFS.

Results

MFS was recognized in 40 children (5 days - 17 years, mean 9 years). The rest were categorized as Ehlers-Danlos syndrome (n=20), Loeys-Dietz syndrome (n=6), marfanoid (n=57) and other (n=10). Among patients with MFS aortic root dilatation (z-score \geq 2) at the level of sinus of Valsalva was found in 32 (80%), while aortic diameter was at the upper limit of normal range (z-score=1,9-1,99) in 4 (10%). Surprisingly aortic root dilation was similarly frequent in different age groups: 0-7 years (86,67%), 8-13 years (66,67%), 14-17 years (84,6%). Aortic root dilatation ranged from +2,14 to +6,30, mean +3,29 (z-score). The degree of dilatation doesn't depend on patient's sex, age and type of mutation (de novo or familiar). The other segments of ascending aorta were dilated less frequently: valve annulus (10%), sinotubular junction (25%), ascending aorta (22,5%). Only in 1 child (2,5%) entire aorta was dilated. Within 2-year follow-up in 1 patient (2.5%) aortic dissection occurred and 1 patient (2,5%) underwent elective David procedure.

Conclusions

Aortic dilatation at the level of sinus of Valsalva occurs in most children with MFS and is present since early childhood. There is no linear correlation between the dilation of the aortic root and the patient's age. Serious aortic dilatation can be found in infant, however, in children aortic dissection occurs infrequently and aortic surgery is rarely needed.