

Comparison of cardiovascular system status between children and adults with Marfan syndrome

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Introduction

Marfan syndrome (MFS) is connective tissue genetic disorder caused by mutations in FBN1 gene. MFS affects mainly the cardiovascular, skeletal and ocular system. Cardiovascular complications are the most serious life-threatening aspect of the syndrome. It is still unclear whether cardiovascular manifestation of MFS differs significantly between children and adults.

Methods

The study population included 230 patients (133 children and 97 adults), who were referred with suspicion of MFS. All patients underwent complete clinical evaluation including cardiac examination (ECG, Holter ECG, and transthoracic echocardiography), ophthalmological, orthopedic and genetic consultations. The modified Ghent criteria were used to identify patients with MFS.

Results

MFS was recognized in 105 patients (40 children and 65 adults). The rest were categorized as Ehlers-Danlos syndrome (n=25), Loeys-Dietz syndrome (n=7), marfanoid (n=79) and other (n=14). Among MFS patients the most common cardiovascular abnormality was aortic root dilation, which was similarly frequent in adults and children (86,05% vs 82,14%, p=0,742). It was found that dilation of the aortic root progresses with age (mean z-score in children: +3,18±0,97, in adults: +4,56±1,92), however without linear correlation. In contrast to aortic root dilation, aneurysms of aortic arch and descending aorta (both thoracic and abdominal) were much more common in adults than in children (20% vs 2,5%). Similarly, aortic dissections occur more often in adults than in children (12,5% vs 2,5%). There was no significant difference between children and adults with regard to aortic regurgitation (AR; 37,04% vs 37,93%, p=1,000), mitral regurgitation (MR; 57,14% vs 78,05%, p=0,109) and mitral valve prolapse (MVP; 71,43% vs 60%, p=0,441). Surprisingly, pulmonary artery dilatation (PAd) was much more common in adults than in children (44,19% vs 25%, p=0,035).

Conclusions

Both adults and children have similar high rates of aortic root dilatation, but the degree of dilatation is much higher in adults. Dilation of aortic arch and descending aorta, the same as aortic dissection occur more common in adults. The other cardiac abnormalities (AR, MR, MVP) don't differ between the groups. The only exception is PAd which is much more frequent in adults.