JCEI /
Journal of Clinical and Experimental Investigations

2012; 3 (2): 199-201
doi: 10.5799/ahinjs.01.2012.02.0144

RESEARCH ARTICLE

Cardiovascular findings of children with Marfan syndrome

Marfan sendromlu çocukların kardiyolojik bulguları

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ABSTRACT

Objectives: The aim of our study is to investigate the frequency of structural heart diseases in patients with Marfan syndrome (MS) and to reveal the importance of clinical follow-up in MS.

Materials and methods: Study population consisted of 17 patients admitted to the Pediatric Cardiology department between January 2005 and March 2010 with the diagnosis of MS according to the Ghent criteria. Patients were evaluated for the eye, genetic and the cardiovascular system abnormalities. Physical examination findings, echocardiographic, and radiological examinations of the patients were evaluated retrospectively.

Results: Of the 17 cases, 9 were girls and 8 were males, ages ranged from 1 month to 17 years (mean 9.7 years). There was a second degree of kinship between mothers and fathers in 5 patients. Respiratory distress, syncope, chest pain and palpitation were the most seen in the presentation complaint of the patients. Skeletal findings observed in 13 patients, 4 patients had subluxation of the lens. Two patients had positive family history. When cardiovascular findings were examined, there were aortic root dilatation with mitral valve prolapse and/or tricuspid valve prolapse in 8 patients, mitral valve prolapse in 3 patients only aortic root dilatation in 3 patients and mitral valve prolapse and tricuspid valve prolapse in 3 patients. Mean follow-up period was 2.6 years, aneurysm and rupture of the aorta wasn't observed during this period.

Conclusion: In patients with Marfan syndrome, regular follow-up and cardiological evaluation should be done because significant structural heart diseases can be seen in these patients. *J Clin Exp Invest 2012; 3(2): 199-201*

Key words: Marfan syndrome, aortic root dilatation, mitral valve prolapse

ÖZET

Amaç: Bu çalışmamızın amacı, Marfan sendromlu (MS) olgularda yapısal kalp hastalıklarının sıklığını araştırmak ve klinik izlemin önemini ortaya koymaktır.

Gereç ve yöntem: Ocak 2005-Mart 2010 tarihleri arasında Çocuk Kardiyolojisi bölümüne başvuran ve Ghent kriterlerine göre MS tanısı konan 17 hasta çalışmaya alındı. Hastalar kardiovasküler sistem muayenelerinin yanı sıra göz ve genetik bakımından da değerlendirildi. Hastaların fizik inceleme bulguları, ekokardiyografik ve radyolojik incelemeleri geriye dönük olarak irdelendi.

Bulgular: Çalışmaya alınan 17 olgunun 9'u kız, 8'i erkek olup, yaşları 1 ay-17 yıl (ort. 9.7 yıl) arasında değişmekte idi. Hastaların 5'inde anne ve baba arasında 2. derecede akrabalık vardı. Hastaların başvuru yakınmaları arasında en fazla solunum sıkıntısı, bayılma, göğüs ağrısı ve çarpıntı görüldü. Hastaların 13'ünde iskelet sistemine ait bulgular görülürken, 4 hastada lens subluksasyonu saptandı. İki hastada aile anamnezi pozitifti. Kardiyovasküler bulgular incelendiğinde, hastaların 8'inde aort kökü dilatasyonu ile birlikte mitral valv prolapsusu ve/veya triküspit valv prolapsusu, 3'ünde mitral valv prolapsusu, 3'ünde sadece aort kökü dilatasyonu, 3'ünde mitral valv prolapsusu ve triküspit valv prolapsusu vardı. İzlem süresi ort. 2.6 yıl olup bu süre içinde aorta anevrizması ve rüptürü gözlenmedi.

Sonuç: Marfan sendromlu hastalarda önemli yapısal kalp hastalıklarının görülmesi nedeniyle kardiyolojik olarak değerlendirilmesi ve düzenli izlemleri yapılmalıdır.

Anahtar kelimeler: Marfan sendromu, aort kökü genişlemesi, mitral kapak prolapsusu

INTRODUCTION

Marfan syndrome (MS) is a hereditary genetic syndrome involving predominantly the skeleton, eyes, heart and aorta, besides various other systems.[1-2] The most serious complications observed in MS affect the heart and blood vessels. Some of these complications are cardiac lesions, aortic root dilata-

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Received: 10.03.2012, Accepted: 23.04.2012

tion (ARD), mitral valve prolapse (MVP), tricuspid valve prolapse (TVP) and the related insufficiencies of these valves.[3] Aortic aneurisms and ruptures are the fatal complications of this syndrome.[4] The aim of our study was to demonstrate the presence of serious structural heart conditions in patients with MS and the importance of the clinical follow-up in order to prevent the related complications.

MATERIALS AND METHODS

Seventeen patients who presented to the pediatric cardiology department of the Dr. Sami Ulus Children Hospital between January 2005 and March 2010 and diagnosed with MS according to the Ghent criteria were enrolled in the study. Besides the cardiovascular examination, the patients were also evaluated by the ophthalmology and genetics departments. All the physical findings and the echocardiographic and radiological examination results of the patients were investigated retrospectively.

RESULTS

Out of the 17 patients enrolled in the study, 9 were female and 8 were male. Their ages varied between 1 month and 17 years (median age: 9.7 years). The parents of 5 patients were 2nd degree relatives. The most common complaints of the patients included respiratory distress (4 patients), syncope (4 patients), chest pain (3 patients) and palpitations (2 patients). While musculoskeletal findings were detected in 13 patients, lens subluxation was observed in 4 patients (Table I). Two patients had family histories of Marfan syndrome. In terms of the cardiovascular findings, 8 patients had MVP and/or TVP together with ARD; 3 patients had MVP, 3 patients had MVP and TVP and 3 patients had singly ARD. The mean follow-up period was 2.6 years and no aortic aneurism ruptures were observed during this time.

Table 1. Distribution of the cardiac lesions; complaints at presentation according to the cardiac lesions and the involvement of other systems in 17 patients with Marfan syndrome

	MVP	MVP+TVP	ARD	MVP+ARD	TVP+ARD	MVP+TVP+ARD
n=17	3	3	3	4	1	3
Chest pain		2		1		
Tiredness		1	1			
Palpitation				1		1
Syncope			1	1		2
Respiratory distress		1		1		2
Asymptomatic	3	1	3			1
Ophthalmological findings		1	2			1
Skeletal system findings	3	3	3	2		2

n: number of patients, MVP: mitral valve prolapse, TVP: Tricuspid valve prolapse, ARD: Aortic root dilatation

DISCUSSION

Marfan syndrome was first described by Antoine Marfan in 1896.[5] The molecular defect in the patients results from to the defect on the fibrillin gene (FBN1) on the 15the chromosome.[6] As a consequence of this defect, a qualitative and quantitative disruption occurs in the synthesis of fibrillin which is the structural glycoprotein of the microfibrils.[7]. In order to be able to diagnose the Marfan syndrome, the Ghent criteria were set in 1996.[8] Thus, in order to definitely diagnose the MS in a patient with positive family history; at least 2 main criteria (cardiovascular, ophthalmologic or skeletal involvement) should be fulfilled. In patients without a positive family history for MS, at least one of the other 2 criteria should be present in addition to the characteristic cardiovascular involvement.[9] In our study, we have also used the Ghent criteria for the diagnosis of MS.

Mitral valve prolapse is a common feature of MS which is observed in 60-80% of the adult and pediatric patients.[10-11] Extension of the aorta beyond the limits set for the related age and body area is defined as aortic dilatation, whereas an aorta exceeding the normal diameter by 50% or more is classified as an aortic aneurism. Aortic root or ascendant aorta dilatations are present in 75-85% of the patients with Marfan syndrome.[11-13] Among our

patients, 82.3% had an MVP, while 47% had the MVP in combination with an ARD. The ratio of the patients with ARD was 64.7%. The ascending aorta diameters of the patients with ARD were between 19-40.6 mm and mean value was calculated as 27.2 mm.

Aortic root dilatation and MVP are the most commonly observed cardiovascular features of the syndrome.[10,14] Patients with MVP may complain from respiratory distress, chest pain, palpitations or dizziness. On the other hand, ARD is asymptomatic if no apparent aortic insufficiency is present or any ruptures have occurred. Among our patients with the Marfan syndrome,[7] complained from respiratory distress, chest pain, palpitations and dizziness. Six of these patients had MVPs. However, the patients with singly MVP were completely asymptomatic and the complaints arose only when the MVP was combined with a TVP or ARD. Four patients with ARD were also totally asymptomatic.

Patients with MS are followed up closely through echocardiography against the development of cardiovascular complications and to be able to choose the right time point for the surgical intervention.[15] Beta blockers reduce the risk of aortic dissection by preventing or delaying the development of ARD.[16-17] In patients where serious aortic insufficiency (AI) due to the ARD is observed, or the aortic root and/or diameter is growing progressively, indication for surgery is present regardless of the degree of the AI in case the aortic root is detected to exceed the 50 mm limit in the echocardiographic examination. ARD is important due to the aneurismatic dilatation and aortic rupture that may occur.[1,18] Since this is a serious complication that may lead to sudden death, it requires close follow-up and treatment. In patients with Marfan syndrome with ARD and MVR, surgical treatments have been reported in life-threatening conditions.[17-18] No serious valvular insufficiencies were detected in any of our MS patients who had MVP together with ARD. None of our patients had life-threatening aneurisms or large ARDs. Therefore, beta-blockers were prescribed to the 3 patients in which ARD was detected. Since no apparent increase in their aortic diameters were observed in the follow-up period, no surgical intervention was required. We did not lose any patients during the mean follow-up of 2.6 years.

In conclusion, since important structural cardiovascular disorders are observed in patients with MS, these patients should undergo cardiologic evaluations and must be followed up regularly. Patients with Marfan syndrome must be initially diagnosed by the first-step healthcare providers and referred to the cardiology clinic so that they can benefit from an early-stage treatment and from a routine follow-up. Thus, the need for emergency surgeries can be minimized and the survival rates may be improved.

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