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CARDIOVASCULAR DISEASE IN CHILDREN WITH MARFAN SYNDROME

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Abstract – To study the cardiovascular changes in children with Marfan syndrome the hospital records of 27 children with this syndrome admitted to our institution between 1975 and 1984 were reviewed. Echocardiographic examination was done in all of them. Heart catheterization and angiocardiography were performed in 7 children. 15 children were without cardiovascular involvement (mean age 7.9 years). Twelve children with cardiovascular abnormalities (mean age 12.3 years) were significantly older than children without cardiovascular disease ($P<0.02$). The most frequent cardiovascular abnormality (9 children) was mitral valve diseas (mitral regurgitation, mitral valve prolapse with and without regurgitation). In one patient artificial mitral valve was inserted. Aortic root aneurysm was found in 7 children, in some of them in combination with mitral valve disease. Fusiform aneurysm of thoracic aorta together with aortic root aneurysm was present in one child. In 2 patients tetralogy of Fallot was diagnosed.
This review shows that the cardiovascular system is more frequently affected in older children and that about half of the children suffer from cardiovascular disease. Mitral valve involvement predominates. Surgery for the correction of the cardiovascular abnormalities in children with Marfan syndrome is seldom required.

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Introduction – The Marfan syndrome is associated with abnormalities of the eye, skeleton, and cardiovascular system (10, 14). The most important cause of death in these patients is cardiovascular disease. Aneurysm of the ascending aorta and aortic regurgitation are the most prominent manifestation of cardiovascular abnormalities in the adult patients (7, 15). In children, mitral regurgitation is the most frequent lesion (13).

The purpose of this report is to review the echocardiographic and angiocardiographic data of children with Marfan syndrome and establish the frequency and nature of cardiovascular lesions diagnosed at University Paediatric Clinic in Ljubljana.

Subjects and Methods – The hospital records of 27 children with the Marfan syndrome admitted to our hospital between 1977 and 1984 were reviewed.

M – mode and cross – sectional echocardiographic examination was done in all the children with this syndrome irrespectively of clinical signs of cardiovascular disease. Increased dimension of the aortic root was considered as a sign of aortic root aneurysm both by echocardiographic and angiocardiographic examination.

The citeria of Perloff and associates were used to diagnose mitral valve prolapse using clinical and cross-sectional echocardiographic examination. (12). Angiocardiographic criteria of mitral valve prolapse consisted of posterior and inferior displacement of mitral valve leaflets beyond the point of their attachment to the fibrous annulus. For the diagnosis of aortic and mitral regurgitation only angiocardiographic criteria were used because Doppler examination was not available. Seven children with clinical and echocardiographic signs of heart involvement which were considered haemodinamically important were invasively investigated.

Results – 15 children were without cardiovascular abnormalities. Their age ranges from 1 to 15 years (mean 7.9 years, SD 4.7 years). Cardiovascular disease were found in 12 children 8 to 12 years old (mean 12.3 years, SD 2.5 years). The children in the first group were significantly youn-

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ger than children in the second group ($P<0.02$). Table lists cardiovascular abnormalities found by echocardiography and angiocardiography. The most frequent cardiovascular abnormality was disease of mitral valve – mitral regurgitation (Fig. 1), mitral valve prolapse with regurgitation (Fig. 2), and mitral valve prolapse without regurgitation (Fig. 3). Aortic root aneurysm was found in 7 cases (fig. 4, 5, 6). Fusiform aneurysm of the thoracic aorta in combination with aortic root aneurysm was present in one child (Fig. 7). In two patients tetralogy of Fallot was diagnosed. In one child artificial mitral valve was inserted.

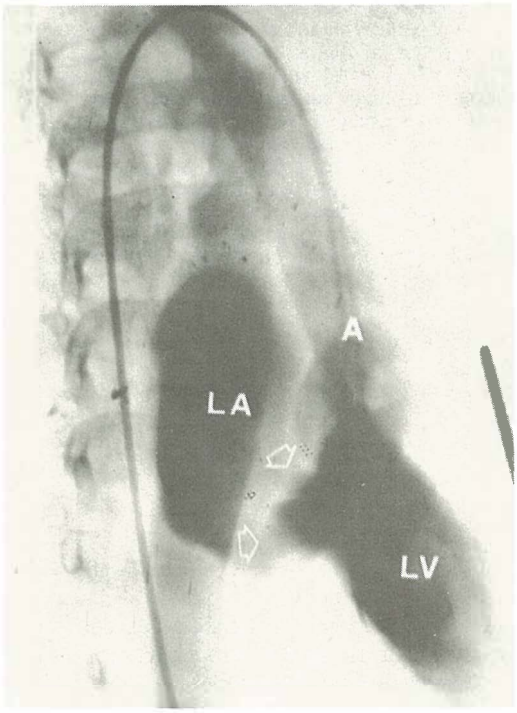

Fig. 1 – Mitral regurgitation with prolapse of the posterior leaflet of mitral valve. LA = left atrium, A = aorta, LV = left ventricle. Arrowheads point to the prolapse

Slika 1 – Mitralna regurgitacija s prolapsom zadajšnjega lista mitralne zaklopke. LA = levi atrij, A = aorta, LV = levi ventrikel. Puščici označujeta mesto prolapsa

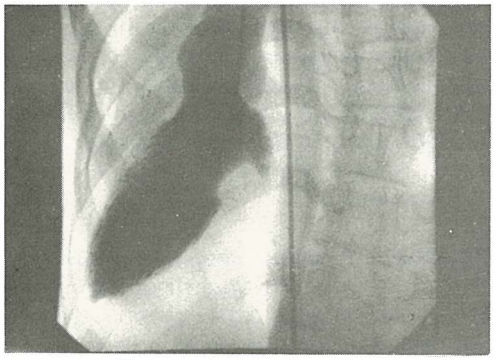

Fig. 2 – Mitral valve prolapse

Slika 2 – Prolaps mitralne zaklopke

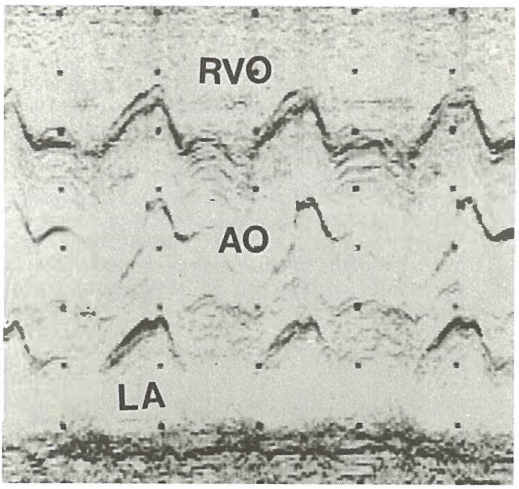

Fig. 3 – M-mode echocardiogram of aortic root aneurysm. RVO = right ventricular outflow tract, AO = aorta, LA = left atrium

Slika 3 – Enodimenzionalni ehokardiogram anevrizme ascendentne aorte. RVO = iztočni del desnega ventrikla, AO = aorta, LA = levi atrij

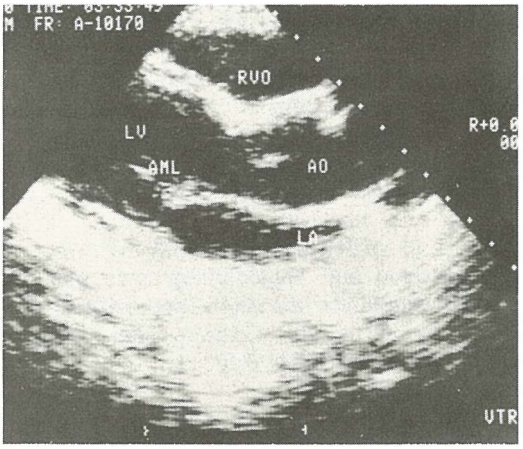

Fig. 4 – Two-dimensional echocardiogram of aortic root aneurysm the same patient as in fig. 3. RVO = right ventricular outflow tract, AO = aorta, LA = left atrium, LV = left ventricle, AML = anterior leaflet of mitral valve

Slika 4 – Dvodimenzionalni ehokardiogram anevrizm ascendentne aorte pri istem bolniku kot na sliki 3. RVO = iztočni del desnega ventrikla, AO = aorta, LA = levi atrij, LV = levi ventrikel, AML = sprednji list mitralne zaklopke

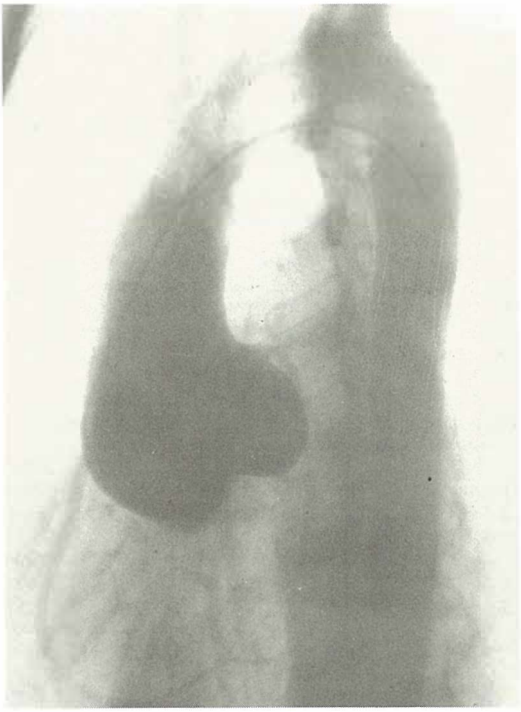

Fig. 5 – Angiogram of aortic root aneurysm
Slika 5 – Angiogram anevrizme ascendentne aorte

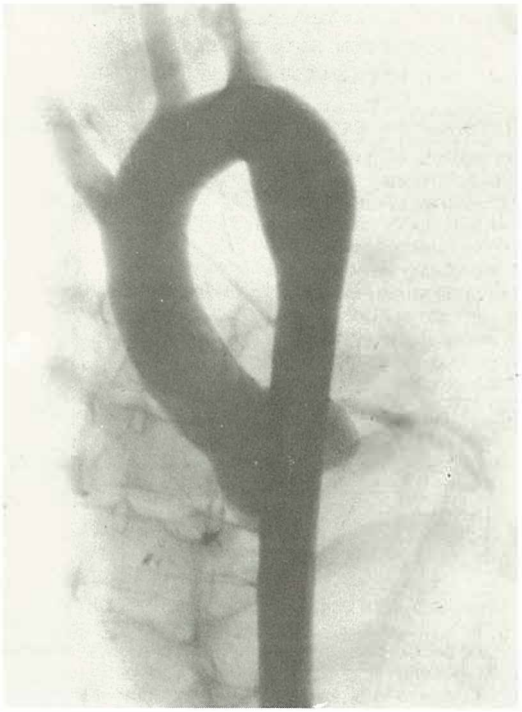

Fig. 6 – Aortic root aneurysm and fusiform aneurysm of the thoracic aorta
Slika 6– Anevrizma ascendetne aorte in vretenasta anevrizma prsne aorte

Discussion – The incidence of cardiovascular complications in patients with Marfan syndrome is estimated to be 30 – 60% (2, 5, 13). In present review the 44% of children had abnormalities of cardiovascular system. In contrast to adults with this syndrome, where aneurysm of the ascending aorta is the most frequent cardiovascular lesion, disease of mitral valve is more common cardiovascular abnormality in children (13). It was encountered in 75% of our patients with cardiovascular involvement. Mitral regurgitation was present in 50% and mitral valve prolapse without regurgitation in 25% of children. Mitral regurgitation was usually mild and in only one patient artificial valve had to be inserted. Aortic regurgitation was found in only one patient but aneurysm of aortic root was present in 58%. This figure differs from the study of Phornphutkul and associates (13) where it was discovered in only 3%.

The most useful noninvasive investigation of the cardiovascular system in patients with Marfan syndrome is echocardiography. With this technique aortic root aneurysm, dissection of the ascending aorta, and mitral valve prolapse can be recognized (1–4, 9, 11, 16). Aortic and mitral valve regurgitation can also be evaluated noninvasively by use of Doppler examination (6, 8) but unfortunately this technique, which is the most specific and sensitive nonivasive diagnostic method for the diagnosis of mitral and aortic valve regurgitation, was not used in the present study. Thus the incidence of mitral and aortic valvar regurgitation might be higher, even in younger children. Echocardiography is most suitable way for serial measurement of aortic dimensions in patients with aortic root aneurysm (4). This method is particularly useful in detection of cardiovascular abnormalities because silent heart lesions in children with Marfan syndrome can be easily identified.

Heart catheterization and angiocardiography is indicated only in children with hemodinamically significant lesions and in preoperative patients.

In conclusion, this review shows that in children with Marfan syndrome the cardiovascular system is less frequently affected in younger children and that approximately half of the children suffer from cardiovascular involvement. Mitral valve abnor-

malities predominate. Surgery for the correction of cardiovascular abnormalities in children with Marfan syndrome is seldom required.

Cardiovascular abnormality	Number of children
MR without MV prolapse and aortic root aneurysm	4
MR without MV prolapse, AR with aortic root aneurysm	1
MR without MV prolapse	1
MV prolapse without MR	2
MV prolapse without MR, aortic rood aneurysm	1
Aortic root aneurysm and fusiform aneurysm of the thoracic aorta	1
Tetralogy of Fallot	2

MR = mitral regurgitation, MV = mitral valve, AR = aortic regurgitation

Table: Cardiovascular abnormalities in 12 children with Marfan syndrome

Povzetek

Pregledal sem bolnišnično dokumentacijo 27 otrok z Marfanovim sindromom, ki smo jih zdravili v naši ustanovi med leti 1975 in 1984, da bi ugotovil spremembe srca in ožilja pri tem sindromu. Pri vseh smo napravili ehokardiografsko preiskavo in pri 7 tudi kateterizacijo srca in angiokardiografijo. Pri 15 otrocih srce in ožilje nista bila prizadeta (povprečna starost 7,9 let). 12 otrok je imelo nenormalnost srca in ožilja (povprečna starost 12,3 leta). Ta skupina je bila statistično pomembno starejša kot skupina brez bolezni srca in ožilja ($P<0.02$). Najpogostejša nenormalnost (9 otrok) je bila bolezen mitralne zaklopke – mitralna regurgitacija in proplaps mitralne zaklopke z in brez regurgitacije. Pri 1 bolniku so vstavili umetno mitralno zaklopko. Anevrizmo ascendentne aorte smo našli pri 7 otrocih, pri nekaterih skupaj z boleznijo mitralne zaklopke. Fuziformno anevrizmo prsne aorte skupaj z anevrizmo ascendentne aorte smo odkrili pri 1 bolniku. Dva otroka sta imela tetralogijo Fallot.

Ta pregled kaže, da sta srce in ožilje pogosteje prizadeta pri starejših otrocih, da trpi okrog polovica otrok zaradi sprememb srca in ožilja, da prednjači prizadetost mitralne zaklopke, in da je kirurški poseg pri otrocih redko potreben.

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