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Short Report

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Marfan Syndrome

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Marfan's syndrome is an autosomal dominant disorder of connective tissue that is characterised by skeletal, ocular, and cardiovascular manifestasions. It is estimated to have prevalence of 1:20.000 people, and at least 25 percent of the cases in the absence of a family history. The syndrome shows full penetrance, but there is considerable clinic variability both between and within families (1). The manifestations relating to the skeletion include tall stature, long and thin arms, and legs, aracnhnodactyly (spidery fingers), scoliosis and kyfosis. The ocular manifestations include upward subluxation of the lenses, myopia and retinal detachment. The most life-threatining complications are those of cardiovascular system with dilatation with or without dissecting aneurysm, of the ascending aorta, and less commonly thoracic, abdominal aorta, or pulmonary artery. Mitral valve prolapse is also very common. Without treatment the cardiovascular complications, particularly dissection of the assending aorta is the leading cause of premature deaths (2-4).

In this paper, we report a case of Marfan syndrome with aortic root dilatation, mitral valve prolapsus, and family history.

A thirteen-years-old girl was referred to our outpatient clinic for investigation of tall stature. She had diagnosed as having severe myopia, especially on the left eye, during the eye examination. She had exercise intolerance. Her mental development was normal. She was the 3rd child of the nonconsanguinous marriage. Other members of the family except for the father were normal. It was learned that the father has had similar features.

Physical examination revealed a height of 169 cm (over the 97th percentile for age), a body weight of 37 kg (between the 10-25th percentile for age), an arm span of 176 cm (arm span-height>7 cm) and a ratio of upper segment to lower segment of 0.85. Bone age was determined as 13 years. Her mother's height was measured as 155 cm and father's height as 192 cm. Her target height, estimated from parental height, was calculated as 167 cm. Table 1 shows the clinical characteristics of our patient and her father related to Marfan syndrome Figure 1 despicts the general appearence of the patient, Figure 2 arachnodactyly, and Figure 3 the joint hyperextention. Sexual maturation was at stage II-III, according to Tanner.

Table 1. Clinical features of our patients.

Features	Our patient	Our patient's father
Ectopia lentis	-	-
Myopia	+	+
Archnodactyly	+	+
Pectus escavatum	+	+
Loose joints	+	+
Scoliosis	-	-
Height>95% for age	+	+
Aortic root dilatation	+	-
Mitral valve prolapse	+	+
Aortic regurgitasyon	-	-
Aortic dissection	-	-
Mental retardation	-	-

Figure 1. General apperance of the patient

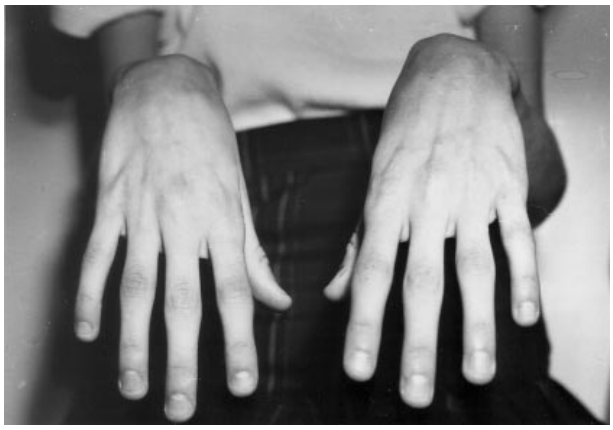

Figure 2. Arachnodacthyly

Biochemical and hormonal evaluation was in normal limits. Blood and urine amino acids were normal. Echocardiograpy revealed aortic root dilatation and mitral valve prolapsus without aortic and mitral regurgitation. Also, mitral valve prolapsus was determined at father's echocardiographic examination. Patient's karyotype was 46-XX.

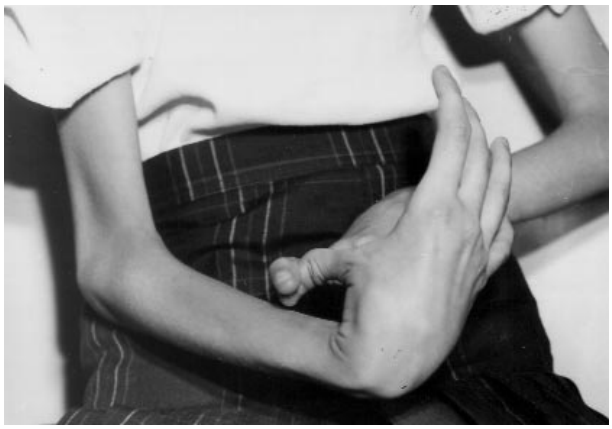

Figure 3. Loose joint

The basic deffect in Marfan syndrome has recently been traced to a deffective fibrillin gene mapped to chromosome 15 (15 q21.1). Fibrillin is a connective tissue protein found in microfibrils, as a constituent of a elastic tissue and abundant in tissues affected in Marfan syndrome, including the aorta, suspensory ligament of the lens and the periosteum (5).

The diagnosis of Marfan syndrome is based on following four major criteria: (a) a positive family history, (b) involvement of skeletal, (c) the ocular and (d) the cardiovascular systems. In the presence of two of these four criteria, the diagnosis of Marfan syndrome can be made (1). Our patient had all four major diagnostic criteria.

Differential diagnosis should be made from Beals syndrome (congenital, contractural arachnodactyly) and homocystinuria, because of the similarities clinical manifestations. Patients with Beal's syndrome have skelatal features, similar to those of Marfan syndrome with long, slender limbs (dolichostenomelia) with arachnodactyly. However, in Beals syndrome, there are joint contractures rather than looseness of the joints, and the eye and heart are not affected. Also in this syndrome, there are camptodacyly of the fingers and folded helixes of the ears. Homocystinuria should be considered in any patient with Marfan like features (dolichostenomelia, arachnodactyly, tall stature) osteoporosis, mental retardation, and history of thromboembolic phenomen. The subluxation of the lens downward, contary to upward dislocation in Marfan syndrome, is the most consistent finding of homocystinuria. Some patient may have no discernible clinical manifestations, except for tall stature. In this situitation, serum and urine amino acid concentrations should be determined for differentiation of homocystinuria (1, 5).

Although several aproaches may reduce the morbidity of Marfan syndrome, there is no specific tretment. Early recognition and correction of a refractive error prevent the amblyopia that often limits vision. Prevention and correction of scliosis ad repair of the pectus deformity may be very helpful. When significant aortic regurgitation is noted, aortic valve prothesis may be utilised. Progressive aortic dilatation may be managed by graft placement. Attempts have been made to limit progression of aortic dilatation by administering propranolol or some other beta adrenergic agents (4, 6).

Life expectancy f patients with Marfan syndrome is abot half that in general population. Half of the affected males are dead by age 40-45, and half of affected females die by age 50-55. In over 95% of patients death is due to cardiovascular complications (6).

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