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SELÇUK TIP DERGİSİ

Marfan Sendromu ile Kombine Situs İnversus Totalis

Marfan's Syndrome Combined with Situs Inversus Totalis

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Özet

Marfan Sendromu bir bağ dokusu hastalığıdır. Başlıca belirtileri iskelet, göz ve kardiyovasküler sistemi tutmakla beraber, nadir durumlarla birlikteliği rapor edilmiştir. Burada 4,5 yaşında bir kız hastayı sunuyoruz. Hastanın uzun boy, artmış kulaç uzunluğu/boy oranı, çift taraflı lens subluksasyonu, atrial septal defekt, aort kapak yetmezliği, aort kökü genişlemesi, araknodaktili, aşırı ekstansiyona gelebilen parmaklar, başparmak işareti ve situs inversus totalisi mevcuttu. Marfan Sendromu'nun Situs İnversus Totalis ile birlikteliği yalnız bir kez tanımlanmıştır. Biz literatüre katkı olarak ikinci vakayı sunuyoruz.

Anahtar kelimeler: Marfan sendromu, situs inversus totalis, tanı kriterleri

Abstract

Marfan Syndrome is a connective tissue disorder. Although cardinal manifestations involve skeletal, ocular, and cardiovascular systems, its association with rare conditions have been reported. We present here 4,5 years-old female patient. She has tall stature, increased arm span/height ratio, bilateral lens subluxation, atrial septal defect, aortic valve regurgitation, aortic root dilatation, arachnodactyly, hyperextensible fingers, thumb sign, and situs inversus totalis. Marfan Syndrome combined with situs inversus totalis has been described only once. We have reported the second case as contribution to the literature.

Key words: Marfan syndrome, situs inversus totalis, diagnostic criteria

Gökçe et al. (1) described the first case of Marfan's Syndrome (MFS) together with situs inversus totalis (SIT) in 2001. Here we describe the second case. MFS is a well-defined autosomal dominant inherited connective tissue disorder. It has an estimated frequency of 1/5000. MFS is caused by mutation in fibrillin gene (FBN1) located on chromosome 15q21. Fibrillin is a component of the extracellular matrix, and particularly rich in the wall of the proximal aorta and the zonule of the ocular lens. So cardinal manifestations involve cardiovascular, ocular and skeletal systems. Cardiovascular involvement is the main cause of major morbidity and mortality in MFS. It is mainly characterized by progressive dilatation of the aorta, a predisposition for aortic dissection, mitral valve prolapse with or without regurgitation. However ectopia lentis is hallmark feature, myopia is the most common sign in ocular manifestations. Skeletal system manifestations are associated with bone overgrowth and joint laxity such as dolichostenomelia, pectus anomalies. MFS may be associated with some rare conditions in addition to these cardinal features. Symptoms can appear at any age and greatly vary between individuals even within the same family. The diagnosis is based on clinical signs and family history according to international diagnostic criteria (Ghent criteria) (2).

Our patient was the fifth child born to a 41-yaers-old G7P5 mother and a 51-years-old father. The mother had two abortions at fourth month of gestation. The patient was born at term by normal spontaneous vaginal delivery. The pregnancy was uncomplicated. However it had been said that her hands, feet and length was tall at follow-up. Birth weight was 4750 gr (> 97 p). Other measurements were not known. There was a history of convulsion at the age of 6-months. Her developmental milestones were normal. Parents were first degree cousin. Neither parents nor their other children were tall. In family history, it has been delineated several individuals who had relevant features such as tall stature, arachnodactyly and/or hyperextensible fingers (Figure 1). However they couldn't be examined due to social causes. During examination, our case was 4,5 years-old. Her weight was 20 kg (75-90 p), height 114 cm (90-97 p) and head circumference 51 cm (25-50 p), arm span 124,5 cm, arm span/height ratio > 1.05. Blood pressure was 90/50 mmHg, pulse rate 115/m. She had deep-set eyes, arachnodactyly, hyperextensible fingers and thumb sign (Figure 2). She doesn't have dolichocephaly, long face, high palate, wrist sign, pectus deformity, hindfoot deformity, flat foot, scoliosis or thoracolumbar kyphosis, skin striae, any hernia. On ophthalmologic examination; right vision is 20/200, left vision is 20/400 by Snellen. After spectacle correction of myopic astigmatism right vision is 20/63, left vision is 20/100. Anterior and post segment examination is performed after dilatation of pupillae by tropicamid %0,5 drop. Both lenses are subluxated to temporal upward which is more obvious in left eye (Figure 3). Lenses are clear. Fundus examination is normal in both eyes. An echocardiogram showed small secundum atrial septal defect, aortic valve regurgitation (minimal), mild dilated aortic root (26 mm). Z-score was calculated as 3.94 on the basis of all these findings. In addition to

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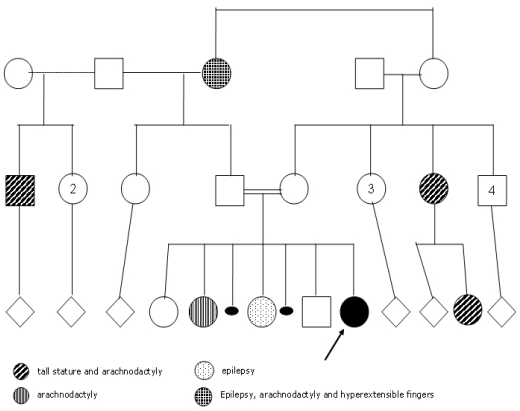

Figure 1. *The pedigree*

MFS, SIT could been diagnosed due to chest X-ray had been revealed dextrocardia and the gas of stomach in right. Subsequently SIT has been confirmed by abdominal ultrasonography. SIT is a rare congenital disorder characterized by transposition of organs to the opposite site of the body (3). In the partial type, it is known as dextrocardia, the heart is located on the right side. However in the complete form, the liver and gall bladder are on the left and the stomach and spleen on the right (4). The incidence is approximately 1/10000 individuals. There is an increased incidence of congenital heart disease (3-9%) compared with 0.6% in situs solitus. It can be associated with Kartegener triad (bronchietasis, sinusitis, and situs inversus) (3). The association of MFS and SIT has been reported in the literature only once (1). Moreover MFS and dextrocardia association has also been reported once (5). We report here the second case who had MFS and SIT. This association may be coincidental, or as a feature of Marfan syndrome. The further cases are required to distinguish these two probabilities.

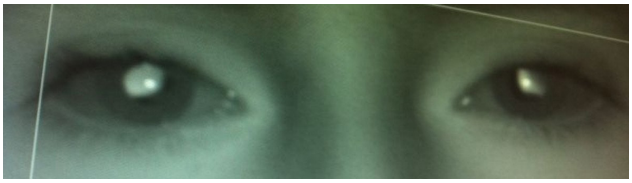

Figure 3. *Bilateral lens subluxation (Photographs are published with consent)*

Figure 2. *A) Arachnodactyly and B) Thumb sign of the proposita*

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