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Case Report

Dextrocardia and Hiatal Hernia in a Patient with Turner Syndrome

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Abstract
Turner syndrome is a sex-chromosome disorder occurring in one out of 2500 female births and characterized by growth retardation, gonadal dysgenesis and cardiovascular anomalies. The 45, XO karyotype is the most frequent type of this disease. Herein, we report on a 6-year-old girl with Turner syndrome and 45, XO karyotype presenting with short stature. She had dextrocardia and hiatal hernia. To the best of our knowledge, the association of Turner syndrome, dextrocardia and hiatal hernia is quite rare.
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Keywords ● Turner syndrome ● dextrocardia ● hiatal hernia

Introduction

Turner syndrome (TS) is the most common chromosomal abnormalities resulting in the loss of all or a part of an X chromosome. The rate of the syndrome reported variably between 1/2000 and 1/5000 live born phenotypically female births.[1] This syndrome is characterized by short stature, gonadal failure, low posterior hairline, thoracic and cardiovascular malformations.[2] The occurrence of cardiac malformations in TS has long been recognized.[3] Coarctation of the aorta and bicuspid aortic valves are the most common heart defects in TS.[4] Other cardiovascular malformations such as aortic stenosis and regurgitation, hypoplastic left heart have also been reported with less frequency.[5]

Case Presentation

A 6-year-old girl referred to the Pediatrics Endocrinology & Metabolism Clinic of Mashhad University of Medical Sciences, Mashhad, Iran, for evaluation of growth retardation. Physical examination at that time of admission disclosed a body weight of 15.5 kg (z-score: -0.51), height of 103 cm (z-score: -2.7), with an upper to lower segment ratio of 1.2/1. She had a short neck, cubitus valgus, micrognathia, low posterior hairline, rotated ears, multiple pigmented nevi and shield chest. There was no webbing of the neck, genu valgum, or edema of hands. Her past medical history was unremarkable except for growth retardation. Bone age was estimated at 6 years and 8 months, using the Greuliche and Pyle radiographic atlas of skeletal development.[6]

Hormonal investigation showed a total T_4 of 96 nmol/l (normal: 58–161 nmol/l), TSH of 7 mU/l (normal: 0.3 to 5 mU/l), FSH of 4 miu/ml (that was high for age–specific reference value). Chromosomal study revealed a karyotype of 45, XO compatible with TS. After 18 months of growth hormone therapy, she had a height of 115 cm (z-score: -1.47) and a weight of 19 kg

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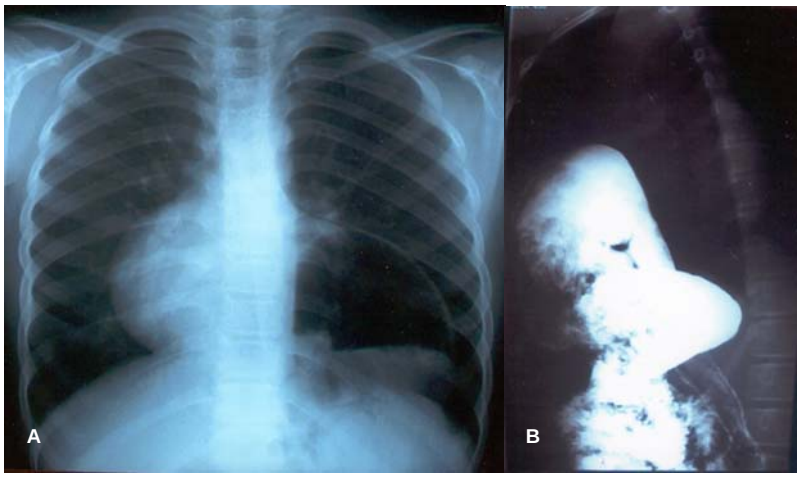

Fig 1: Anteroposterior chest radiograph showing dextrocardia and hiatal hernia (paraesophageal) (A) and Lateral view of barium filled esophagus and stomach in the thorax (B).

(z-score: -1.38). At this time the patient referred to the pediatrics cardiology clinic for routine cardiovascular investigations. In cardiovascular examination, her blood pressure was 100/70 mmHg. The point of the maximum heart impulse was noted in the right. ECG finding revealed a negative P wave in the lead I.

In chest X-ray, dextrocardia with hiatal hernia were noted (Fig 1). Echocardiography with standard right para-sternal and subcostal views, revealed dextrocardia with normal drainage of inferior vena cava to the right atrium in right liver portion of hepatic inferior vena cava—visceral *situs solitous*. There were no significant abnormalities in the heart except for a mild mitral valve prolapse and trivial tricuspid regurgitation with a pressure gradient of 15 mmHg.

Discussion

A wide variation of clinical features seen in females with TS ranging from a severe phenotype with short stature, gonadal dysgenesis, lymphedema and characteristic dysmorphic features to women with only a mild reduction in final height, or premature ovarian failure.[1,7] With a prevalence rate between 23% and 40%, congenital cardiac anomalies are common in females with TS. The structural cardiac anomalies are most prevalent in women with 45, XO monosomy and tend to be less common in those with an isochromosome Xq karyotype.[8]

The most common cardiac anomalies are coarctation of the aorta, bicuspid aortic valve, or both, followed by other aortic valve abnormalities. Whereas, in our case, there was only an isolated dextrocardia with no significant cardiac lesion.

Previous reports revealed a very high incidence of associated cardiac anomalies observed in the form of dextrocardia.[9] Transposition of the great arteries is the most common form of isolated dextrocardia.[9] Other complex cardiac lesions were reported, including ventricular septal defect and pulmonary stenosis, double outlet right ventricle, double outlet left ventricle; complete atrio-ventricular septal defect.[9]

Chest deformity in TS may be observed as shield-like chest with widely separated nipples. Sternal malformation can be detected by lateral chest radiography.[7] In a study reported by Rapaport on 170/343 females with TS in Denmark, 38% of patients with 45, XO chromosome had cardiovascular malformations as compared with 11% of those with mosaic monosomy X.[2]

This finding indicated that Turner syndrome might occur coincidentally in different organs characterized by aforementioned symptoms.

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