

# Morgagni Hernia in a Girl With Turner Syndrome

## *Turner Sendromlu Bir Kızda Morgagni Hernisi*

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### Abstract

This report describes the delayed presentation of right-side Morgagni hernia in a 15-year-old girl with Turner syndrome. It is commonly associated with a number of systemic malformations and abnormalities. Morgagni hernia is a rare type of congenital diaphragmatic hernia, which may not be symptomatic until adulthood when the patient presents with acute symptoms or incarceration. As this result, patients with Turner syndrome should be investigated for Morgagni hernia because there may be an association between the two, and Morgagni hernia may be asymptomatic. *Turk Jem 2008; 12: 60-2*

**Key words:** Diyafragma hernisi, Morgagni hernisi, Turner sendromu

### Özet

Bu olgu sunusunda 15 yaşındaki Turner sendromlu bir kızda geç bulgu veren sağ diyafragmada Morgagni hernisi tanımlandı. Turner sendromu sıklıkla çok sayıda sistemik malformasyon ve anormalliklerle ilişkilidir. Morgagni hernisi akut semptomlar ya da inkarasyon olduğu zaman tanınan yetişkin çağa kadar semptomatik olmayabilen nadir bir konjenital diyafragma hernisi tipidir. Turner sendromu ve Morgagni hernisi arasında bir ilişki olabileceğinden Turner sendromlu hastaların Morgagni hernisi açısından da araştırılması gereklidir. *Turk Jem 2008; 12: 60-2*

**Anahtar kelimeler:** Diaphragmatic hernia, Morgagni hernia, Turner syndrome

### Introduction

Turner syndrome (TS) is characterized by the absence of part of or the entire X chromosome in a female (1). Approximately 1:2000 to 1:5000 live-born girls have TS, and it is commonly associated with a number of other systemic malformations (2). The syndrome can cause many problems with lymphatic, skeletal, gonadal, cardiovascular, renal, and cognitive development. Congenital heart diseases are the most common seen anomalies associated with TS (3). However, congenital diaphragmatic hernia is a rare malformation in the patients with TS. We report a girl with Morgagni hernia (MH) and TS.

### Case Report

A 15-year-old female patient was admitted to our clinic for evaluation of growth retardation, primary amenorrhea, weakness, and short stature. There was no family history of short stature. On physical

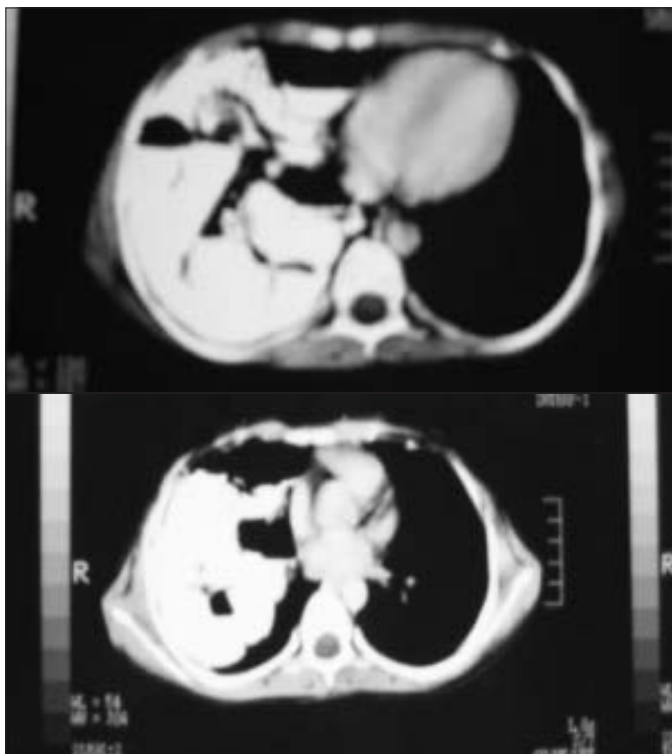
examination, typical signs of Turner syndrome were observed (short stature, underdeveloped breasts, high-arched palate, triangular face, bilateral pedal lymphedema, small retropositioned mandible, and webbed neck) (Figure 1). Body height was 128 cm ( $\leq -3$  SD below the mean), and body weight was 26.2 kg ( $\leq -3$  SD below the mean), BMI was 18.9 kg/m<sup>2</sup>; and bone age and height age were 10 years old. The crown-pubis length was 56 cm, and pubis-heel length was 72 cm (upper segment/lower segment ratio: 0.77). Arm span was 125 cm. According to Tanner classification, genital development was B1, P1. Body temperature 36.8°C; pulse, 78 beats/min; respiratory rate, 22 breaths/min; and blood pressure was 100/60 mm Hg. On cardiac auscultation, a systolic ejection murmur (2/6) was heard at the precordium. Respiratory sounds were found to be diminished at the right basal region. Abdominal examination was normal. Bowel sounds were present with a frequency of 8/min.

Laboratory findings included normal findings for age and sex for growth hormone (GH: 0.71 ng/mL [normal range 0.06-5]); insulin-

like growth factor-1 (IGF-1; 145 ng/mL [normal range 220-972]), and IGF binding protein-3 (IGFBP-3: 5.53 µg/mL [normal range 3.3-10]) Growth hormone stimulation tests using insulin and L-dopa were performed. Increase in GH was demonstrated during both stimula-



**Figure 1.** Typical appearance of Turner syndrome in our patient



**Figure 2.** Thorax CT demonstrated right-sided Morgagni hernia in our patient with Turner syndrome

tion tests (L-dopa: GH at 0 min = 0.71 ng/mL, at 30 min GH = 6.38 ng/mL, at 60 min GH = 3 ng/mL, and at 90 min GH = 8.81 ng/mL; insulin tolerance test: 0 min GH = 0.75 ng/mL, and at 60 min GH = 12.2 ng/mL). Other laboratory values included the following: FSH: 178.3 mIU/mL, LH: 44.7 mIU/mL, E2: 8.15 pg/mL; progesterone: 0.515 ng/mL, prolactin: 20.91 ng/mL (3.4-24.1), cortisol: 7.47 µg/dL (6.2-10.4), ACTH: 16.4 pg/mL (5-46), DHEA-S: 174.1 µg/dL (70-300), testosterone: 0.520 ng/mL (0.22-0.80), and 17-OH progesterone: 0.09 nmol/L; thyroid function tests, including free T3, free T4, and TSH levels were normal. These laboratory findings were consistent with hypergonadotropic hypogonadism. In the chromosomal analysis, 45,X0 chromosomal structure was determined. Barr body (X chromatin) was negative.

In the chest radiography, a heterogeneous increase in opacity in the right paracardiac area was observed. There was no history of trauma. Transthoracic echocardiography revealed a bicuspid aortic valve. An ultrasonographic (US) investigation showed a defect in the anterior region of the diaphragm, with intestine protruding into the right hemithorax. Ovaries were not demonstrated, but rudimentary uterus could be detected (38X20X10 mm) in pelvic US imaging. Thoracic and abdominal computed tomography (CT) were performed for further investigation. The right part of diaphragm was not seen on CT scan, and a heterogeneous mass lesion was observed anteromedially, which starting parasternally and extended to the carina; the mass was noted to be pushing cardiac structures into the left hemithorax (Figure 2). CT scan did not show any gas or contrast material in thoracic cavity. These findings suggested that a congenital diaphragmatic hernia of Morgagni type was present in the right thoracic cavity. Ovaries were not seen on CT scan. Hormone replacement treatment (conjugated estrogen 0.625 mg) was given by mouth daily for 6 months in the medical treatment.

## Discussion

Turner syndrome is the most frequent sex abnormality in females. It is generally associated with a 45,X0 karyotype, even if more than half patients have a mosaic chromosomal component (e.g., 45,X0/46,XX). TS is characterized by typical stigmata, including short stature, primary amenorrhea, estrogen insufficiency, pterygium colli (webbed neck), lymphedema, and cardiovascular malformations.

Turner syndrome associated with Morgagni hernia is extremely rare condition. It is well known that there is a relationship between MH and various genetic abnormalities, such as Down syndrome and Noonan syndrome (4). We searched Medline (PubMed), using keywords of Turner syndrome and Morgagni hernia. We found only one report (published in 1989) about this association (5). TS was also found in our previous reported series of MH associated with chromosomal anomaly (6). However, MH is not listed among with stigmata of TS. However, some cases may remain undetected and present only later in life. Therefore, future investigation is necessary to clarify this association.

Various karyotypes and phenotypes exist in TS. It is commonly associated with a number of other systemic malformations and abnormalities. TS predominantly involves the vessels of the left side

of the heart, although venous malformations have also been documented. Aortic dissection occurs early and with increased frequency in TS and is often accompanied by hypertension. The increased risk of coronary disease may well be explained by the very frequent occurrence of hypertension (~50%), increased carotid intimal thickness, aortic augmentation index, and pulse wave velocity. Epidemiological data indicate that metabolic syndrome, involving hypertension, obesity, dyslipidemia, and type 2 diabetes mellitus, is more frequent among adults with TS. We did not find any metabolic or cardiovascular abnormality or other specific systemic disturbance except MH in our patient. In 2001 Swerdlow et al (7) extended the British data and described how mortality in TS is elevated, with an increased risk of death from diseases of the nervous, cardiovascular, respiratory, digestive, and genitourinary systems. Therefore, appropriate management of females with TS requires multidisciplinary efforts from a geneticist, an endocrinologist, and other specialists.

Morgagni hernia may be asymptomatic for years, as in our case. We think the presence of MH in Turner syndrome should be considered because of its tendency to remain asymptomatic. Most patients who have reached adulthood are asymptomatic, and the diagnosis is made incidentally from a chest radiograph taken for another reason (8). Some cases may be discovered incidentally by visualizing air-fluid levels or solid masses in the retrosternal region or on

the right side of the chest radiograph. It is confirmed with barium enema or CT scan. In our case, CT of the thorax gave the most useful knowledge for diagnosis of MH.

In conclusion, we report a case of Turner syndrome and Morgagni hernia. Morgagni hernia may be asymptomatic for years in patients with TS. A relationship may be present between TS and MH; and screening chest radiograph for patients with TS is recommended.

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