A 13-year-old female with Turner syndrome and achalasia

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SUMMARY: Balcı S, Akcan AB, Şenocak ME, Çiftçi AÖ, Ozaltın F. A 13-year-old female with Turner syndrome and achalasia. Turk J Pediatr 2006; 48: 272-274.

The most common gastrointestinal problems associated with Turner syndrome are intestinal telangiectasia, colon carcinomas, inflammatory bowel, and liver diseases. In this paper we present for the first time a 13-year-old female with 45,X karyotype associated with achalasia. As far as we know, achalasia associated with Turner syndrome has not been reported previously. The aim of this report was to point out the association of Turner syndrome and achalasia. It could be a coincidental or Turner syndrome-associated finding.

Key words: Turner syndrome, achalasia, gastrointestinal malformations.

The most common gastrointestinal problems associated with Turner syndrome[1] are intestinal telangiectasia, gastrointestinal bleeding, iron deficiency anemia[2], colon carcinomas, inflammatory bowel[3,4], elevated liver enzymes, hepatic fibrosis, and hepatic cirrhosis[5]. In this paper we present a 13-year-old female patient with 45,X karyotype associated with achalasia. This association has not been reported previously. The aim of this report was to point out the association of Turner syndrome and achalasia. It could be a coincidental or Turner syndrome-associated finding.

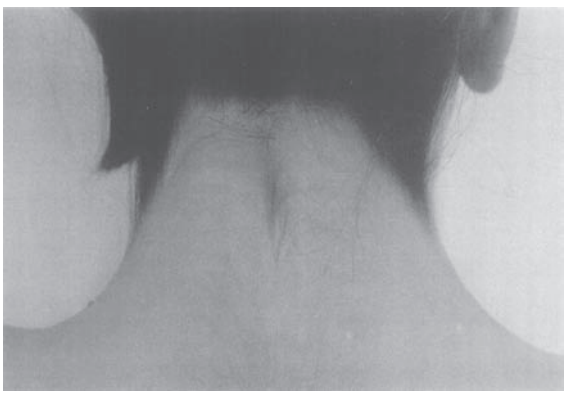
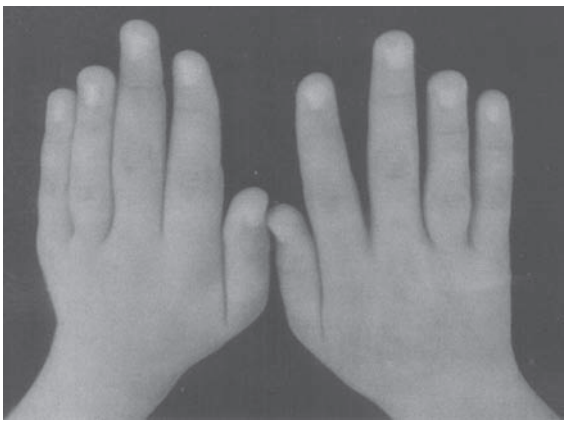


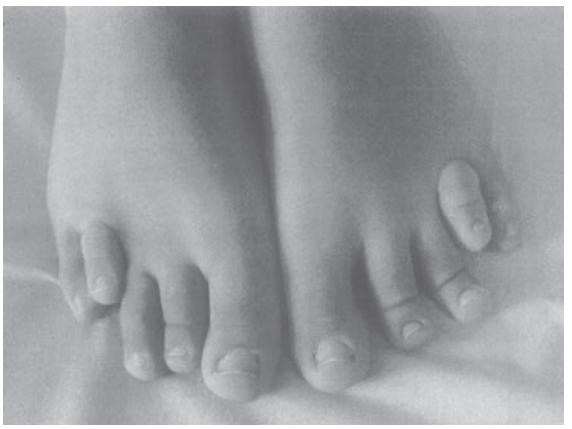
Fig. 1. Turner syndrome characterized by short neck and low hairline.

Case Report

A 13-year-old female was admitted to Hacettepe İhsan Doğramacı Children's Hospital Department of Clinical Genetics because of short stature and recurrent pulmonary infections. She was the first child of non-consanguineous parents. The second child, a healthy male, was eight years old. The birth weight was 2,250 g, and she was born after 32 weeks of gestation. Swelling on the dorsal side of the feet and hypoplastic nails were noted both at birth and within the first years of life. On admission, height and weight were 112 cm (<5th percentile) and 17.5 kg (<5th percentile). Her facial appearance was characterized by short neck and a low hairline (Fig. 1). The fourth fingers and the fourth toes were short bilaterally (Figs. 2a, 2b). Her chest was broad and her nipples were inverted and widely spaced. Breast development was Tanner stage 2. A chromosomal analysis from peripheral blood revealed 45,X karyotype. The patient had been operated for aortic coarctation which presented with hypertension when she was nine years old. Chest X-ray showed atelectasis of the middle lobe of the right lung. Achalasia was suspected due to typical history of dysphagia for solids and liquids as well as regurgitation of retained esophageal food, in addition to recurrent pulmonary infections. An esophagography performed at eight years of age showed achalasia (Fig. 3). An operation for achalasia (Heller esophagomyotomy and Nissen



(a)



(b)

Fig. 2a, 2b. The short fourth fingers and toes of the patient.

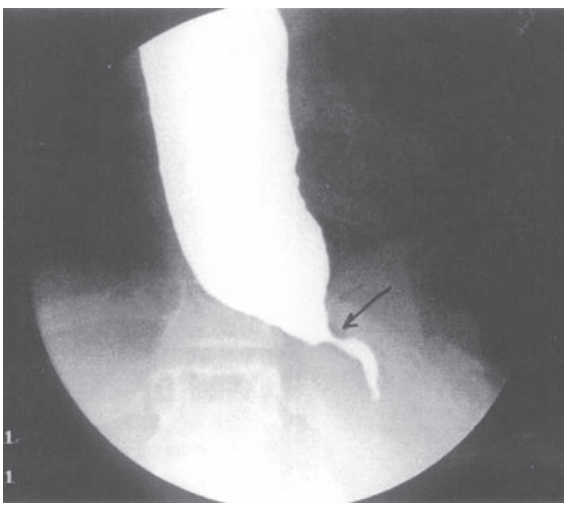


Fig. 3. Barium esophagogram of the patient demonstrating dilated esophagus and narrowing at the lower esophageal sphincter.

fundoplication) was performed at 10 years of age. The postoperative course was uneventful and the patient is well at present.

Discussion

Achalasia is a primary esophageal motor disorder of unknown etiology characterized by loss of lower esophageal sphincter (LES) relaxation and loss of esophageal peristalsis, both contributing to functional obstruction of the distal esophagus. Down syndrome with megacolon[6] is a well-known example among the chromosomal aberrations and gastrointestinal abnormality associations. It rarely occurs in association with some other anomalies or syndromes such as triple A syndrome (achalasia, alacrima, autonomic dysfunction)[7]. Balcı et al.[8] reported familial achalasia with skeletal abnormalities and copper deficiency in two sisters, and in 1994 they reported two further sibs from another family[9]. Recently, Richterich et al.[10] reported a nine-year-old female case with anorexia nervosa mimicking achalasia who was diagnosed by barium swallow. After dilatation and botulinum toxin application, the patient regained weight. Our patient had no similar findings such as anorexia nervosa or other related symptoms. Additionally, our patient had aortic coarctation and short fourth fingers and toes, and cytogenetically proven Turner syndrome (45,X). Achalasia is rarely observed in Steinert's disease (myotonic dystrophy), as was reported by Grazia et al[11]. But our patient had no myotonic dystrophic physical findings. This is the first observation of Turner syndrome and achalasia in our genetic department between 1968 and 2004 from among more than 100 Turner syndrome cases. Recently Akcan et al.[12] studied 28 cases from our department with Turner syndrome. We cannot say whether it is coincidental or a constant finding. However, we recommend investigating achalasia in Turner syndrome when findings indicating LES dysfunction are present. Further observations will clarify the issue.

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