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CASE REPORT

A unique case of right-sided Poland syndrome with true dextrocardia and total situs inversus

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Abstract Poland syndrome has been reported to be associated with true dextrocardia, but not with true situs inversus. In this report, we describe the first patient with total situs inversus in medical literature and try to highlight the syndrome's probable etiology and pathogenetic mechanisms in utero.

Keywords Poland syndrome · Situs inversus · Dextrocardia · Thrombophilia · Smoking

H. I. Atasoy (✉) · T. Yavuz · E. Yesiller
Department of Pediatrics, Izzet Baysal Faculty of Medicine,
Abant Izzet Baysal University,
Golkoy 14280 Bolu, Turkey
e-mail: atasoy_h@ibu.edu.tr

H. I. Atasoy
e-mail: halilibrahimatasoy@gmail.com

S. Altunrende
Department of Radiology, Izzet Baysal Faculty of Medicine,
Abant Izzet Baysal University,
Golkoy 14280 Bolu, Turkey

M. Guven
Department of Ortopedics, Izzet Baysal Faculty of Medicine,
Abant Izzet Baysal University,
Golkoy 14280 Bolu, Turkey

A. Kılıcgun
Department of Thoracic Surgery, Izzet Baysal Faculty
of Medicine, Abant Izzet Baysal University,
Golkoy 14280 Bolu, Turkey

O. Polat
Department of Neurosurgery, Izzet Baysal Faculty of Medicine,
Abant Izzet Baysal University,
Golkoy 14280 Bolu, Turkey

S. Duzenli
Department of Genetics, Izzet Baysal Faculty of Medicine,
Abant Izzet Baysal University,
Golkoy 14280 Bolu, Turkey

Introduction

Poland syndrome (PS) (OMIM 173800) is an uncommon congenital anomaly characterized by unilateral aplasia/hypoplasia of the m. pectoralis major and ipsilateral deformities in the extremities. Genetics of the syndrome indicates a sporadic mutation without a clear inheritance pattern. The syndrome is believed to be caused by a brief interruption or reduction in the circulation of the subclavian and vertebral arteries or their smaller branches during the first six weeks of embryonic development [6].

A variety of other congenital anomalies including the Klippel–Feil syndrome, Möbius and Sprengel anomalies, abnormalities of the anterior chest wall, breast and craniofacial structures, hemivertebra, and scoliosis may be associated with this syndrome [6]. However, to the best of our knowledge, association of the syndrome with true dextrocardia and total situs inversus (TSI) has never been described [9]. Herein, we report a PS associated with true TSI and try to highlight the underlying pathogenetic mechanisms during fetal development.

Patient

In this 6-year-old boy, a deformity of the chest wall was observed at birth by the parents. History revealed that the boy's father was an active smoker during the patient's intrauterine life. Delivery was uneventful. He had normal motor and intellectual development but had subnormal school performance. There was no history of recurrent respiratory infections, sinusitis, or bronchiectasis. Familial history was unremarkable.

Body weight was 19 kg (p 50–75), height 113 cm (p 25–50), and head circumference 49.6 cm (p 25–50). Physical examination showed depression of the right anterior chest

wall, cranially located right nipple, and the hypoplastic right areola. There was preaxial polydactyly, sternomastoid hypoplasia and hemifacial microsomia on the right side, and torticollis (Fig. 1). He had a short neck; mild nuchal webbing; low posterior hairline; low-set, anterior-facing, and dysmorphic ears; mild scoliosis with concavity to the left; and Sprengel anomaly. Apical cardiac sounds were right sided. He demonstrated a restricted range of cervical and spinal movements due to sternomastoid muscle hypoplasia and atlantoaxial rotatory instability. Neurological examination was normal.

Chest X-ray revealed a right-sided heart (Fig. 2a). Echocardiography showed a mirror image dextrocardia without evidence of cardiac or great vessel malformations. Axial and coronal MR of the chest demonstrated hypoplastic m. pectoralis major and descending aorta located on the right (Fig. 2b), right-sided heart, spleen and left kidney, and left-sided liver and right kidney. Computerized tomography of the neck and upper chest revealed bifid atlas, right sternomastoid hypoplasia, and right-sided main vascular structures.

Cytogenetic examination showed normal male karyotype of 46, XY. Protein C, protein S, antithrombin III, and homocysteine levels, as well as prothrombin and partial thromboplastin times, were normal. To evaluate an eventual predisposition to thrombophilia, we carried out molecular detection by real-time PCR: The patient's genotype was homozygous for methylenetetrahydrofolate reductase (MTHFR) A1298C, heterozygous for Factor V H1299R, Factor II G20210A and wild type for Factor V G1691A, plasminogen activator inhibitor-1, and angiotensin-converting enzyme.

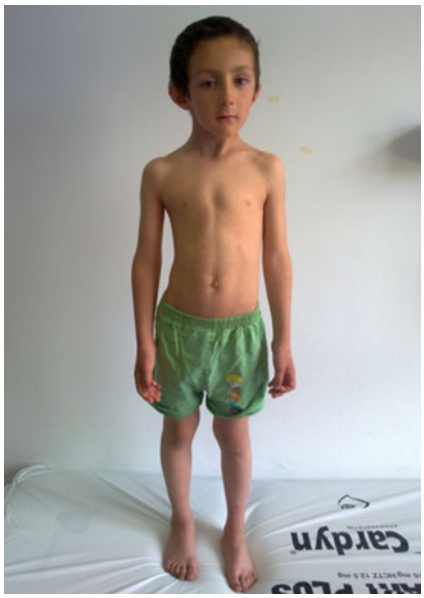

Fig. 1 Depression of the right anterior chest wall indicating pectoral major hypoplasia, cranially located right nipple and hypoplastic areola, hypoplastic sternocleidomastoid, torticollis and mild hemifacial microsomia, and preaxial polydactyly

Discussion

Cardiac malposition observed in PS is usually a dextroposition, not a true dextrocardia. True dextrocardia is very rare in PS, having been reported in only 20 patients. PS with true dextrocardia and TSI has not been reported before in the English medical literature. In contrast to dextrocardia observed in PS, dextrocardia in TSI is always a mirror image dextrocardia, as in our case, not a simple dextroposition. All the vessels originating from the heart had mirror image of the normally situated heart. Interestingly, our patient had right-sided m. pectoralis hypoplasia together with right-sided "true" dextrocardia, in contradiction to the suggestion that dextrocardia results from volume loss of the left hemithorax caused by the syndrome [4]. Additionally, the right hemithorax of our patient was hypoplastic, although the heart was located on the right side. We also did not observe hypoplastic ribs on the right rib cage. In a recent study, it was reported that all patients with left-sided pectoral deformity and partial agenesis of two or more ribs on the left side had dextrocardia. This observation led to a "volume loss hypothesis," which explains the displacement of the heart contralateral to the affected side by shrinkage of one hemithorax [9]. This hypothesis fails to explain the dextrocardia in our patient, who had volume loss of the right hemithorax containing the heart [4].

We have questioned the diagnosis of PS in our case at the first glance since there was preaxial polydactyly but no brachysyndactyly. However, there are cases of PS associated with preaxial polydactyly in the literature [2]. Kartagener syndrome (KS) is unlikely because the patient did not have clinical history or radiologic findings of chronic sinusitis and bronchiectasis [7]. In KS displacement of organs to the opposite site is thought to be caused by abnormal ciliary motility due to congenital defects in the ciliary structure. Unlike the presumed etiology of dextrocardia in KS, we attributed the organ malposition observed in our patient to the impaired ciliary function, which might have resulted from the inhibitory effect of passive smoking which the patient had been exposed in utero.

Sternomastoid hypoplasia has not been reported in patients with PS. Hemifacial microsomia observed in our patient might either be secondary to sternomastoid hypoplasia or an independent component of the syndrome. Hemifacial microsomy and Sprengel deformity

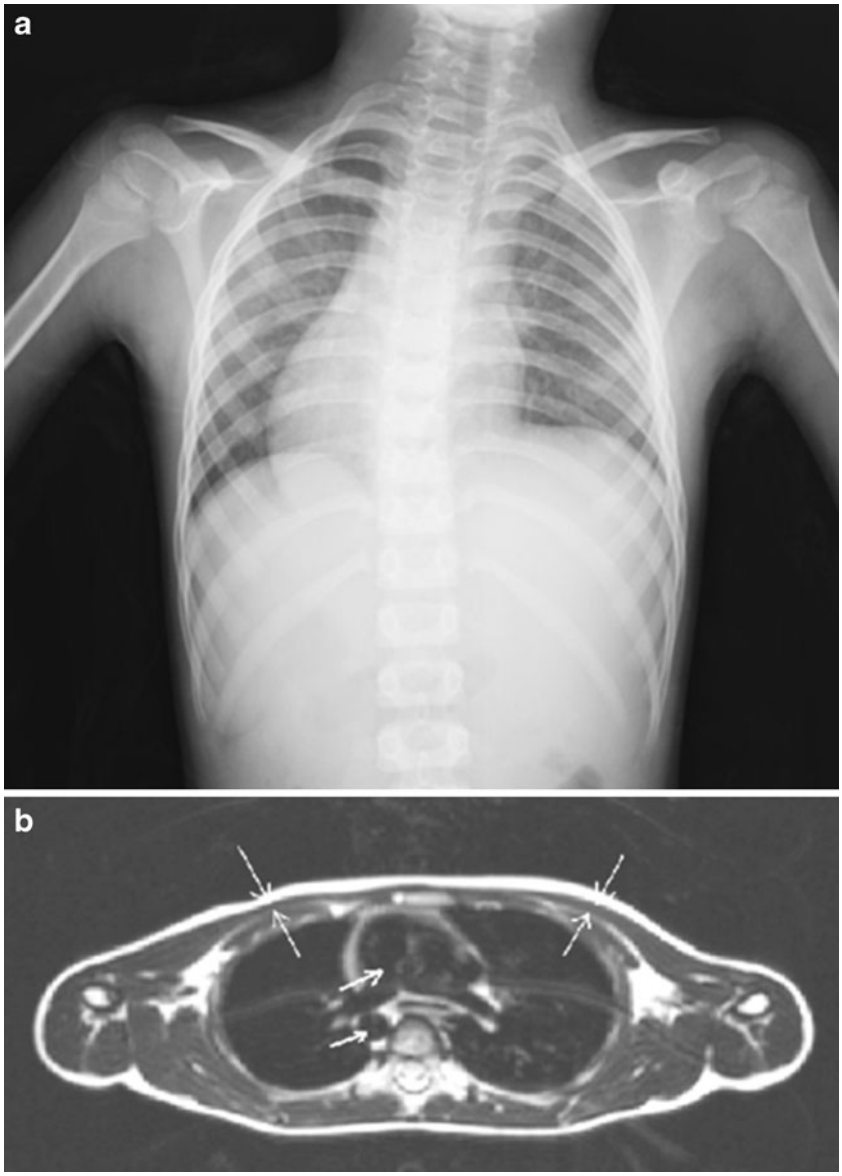

Fig. 2 Conventional radiography of posteroanterior chest shows the cardiac apex is directed towards the right (dextrocardia) (**a**). T2-weighted axial magnetic resonance imaging showing pectoralis major hypoplasia (0.42 cm on the right, 0.63 cm on the left), mirror image dextrocardia both on the right side (*thin arrow*), and right descending aorta (*thick arrow*). Also note the hypoplastic right hemithorax where heart is located (**b**)

might also be regarded as other components of the syndrome since they were reported in the relatives of or patients with PS [1, 3]. Since the torticollis and hemifacial microsomy were both present at the time of birth, we concluded that these features as well as atlas hypoplasia and the associated atlantoaxial rotatory instability were secondary to thrombophilic processes in the subclavian artery branches.

To our knowledge, association of TSI with hypoplasia of the atlas contributing to atlantoaxial rotatory instability has been reported previously in a single case. The authors attributed these abnormalities to a mesodermal malformation affecting the chondrification of the posterior arch of the atlas between the sixth week and fourth month of gestation [8]. As we found homozygote A1298C MTHFR, heterozygote Factor V H1299R, and Factor II G20210A mutations in the thrombophilia genes in our patient, we speculate that the cause of these malformations was in fact a vascular interruption resulting from thrombosed microvasculature in the fetus during the first half of gestation when chondrification of the posterior arch of atlas occurs [8]. Whether this vascular interruption causes a mesodermal malformation is not clear. Additionally, the passive smoking of the mother and fetus in utero may have augmented this thrombotic process. This hypothesis is supported by other studies that showed a twofold increase in PS risk due to maternal smoking [5]. However, literature findings are scarce at the moment and our hypotheses need to be confirmed by further studies.

Conflict of interest The authors declare no conflict of interest related to this work.

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