Multimodality imaging features of Poland syndrome associated with cervical rib and elongated transverse process of cervical vertebrae
Muammer Akyol1, Onur Gokdemir2, Tulin Ozturk1

Abstract
Background: Poland syndrome (PS) is a rare congenital anomaly characterized by partial or complete absence of the major pectoral muscle which is variably associated with ipsilateral thoracic and upper limb deformities.

Case Presentation: We present multimodality imaging features of PS associated with cervical rib and elongated transverse process of cervical vertebrae in a 25 year-old female patient.

Conclusion: PS is a rare congenital disorder that has a wide spectrum of clinical presentations. The aim of our case report was to show multimodality imaging features of the PS associated with cervical rib and elongated transverse process of cervical vertebrae. We could not find any similar case reported in the literature. (El Med J 2:3; 2014)

Keywords: Cervical Rib, Computed Tomography, Magnetic Resonance Imaging, Poland Syndrome, Ultrasound

Introduction
Poland syndrome (PS) is a rare congenital malformation occurring in approximately one in every 30,000 to 100,000 live births [1]. It was first described by Sir Alfred Poland in 1841 as complete unilateral absence of the pectoralis major-minor, serratus anterior, obliquus externus abdominis muscles with ipsilateral thoracic and upper limb defects. But the term ‘Poland Syndrome’ was first used in an article by Patrick Clarkson in 1962 [2]. Clinical manifestations are extremely variable and are rarely all seen in a single case. Most cases of PS reported are sporadic; however, familial cases have been occasionally described. The right side of the body is affected more often than the left side, and a male to female ratio of 2:1 up to 3:1 can be found. Reports of bilateral agenesis of the muscle have been infrequently reported in literature [2, 3].

Cervical rib is a congenital anomaly which is usually located above the first rib. It is seen bilaterally in 40% of cases and is found twice as frequently in females than in males. Previous studies have reported a wide range of values for the prevalence of cervical rib in various human populations, from 0.05% to 3.0%, based on series using radiographs [4, 5]. Although typically asymptomatic, cervical rib may cause thoracic outlet syndrome (TOS) in adults [4].

We encountered a patient with PS associated with right-sided cervical rib and left-sided elongated transverse process of 7th cervical vertebra. The association of PS with rib anomalies has been described before. But to our knowledge, its association with cervical rib and elongated transverse process of cervical vertebrae has not been reported previously.

Case Report
A 25 year-old female presented to our outpatient clinic of the Department of Plastic and Reconstructive Surgery with the complaint of small left breast. No significant past medical or surgical history was present. Physical examination showed depression of the left anterior chest wall with small left breast volume. The left nipple was sited slightly higher compared to the contralateral side but was otherwise normal (Figure 1). The patient had normal routine laboratory values and was referred to the Radiology outpatient clinic with an initial diagnosis of PS. Radiological examination, including ultrasound, computed tomography (CT) and magnetic resonance imaging (MRI) revealed aplasia of the costosternal portion of the left pectoralis major muscle, left pectoralis minor muscle, left serratus anterior muscle, left anterior intercostal muscles, hypoplasia of the left latissimus dorsi muscle and reduced left breast tissue with right-sided cervical rib and left-sided elongated transverse process of 7th cervical vertebrae (Figure 2). These anomalies were compatible with the diagnosis of PS.

Further systemic evaluation, including examination of upper-lower limbs, did not show other anomalies. Cardiovascular and neurological examination were normal. Renal ultrasound excluded important anomalies which can occur in PS. She was within normal limits in height, weight, and intelligence. The problem was aesthetic. Breast augmentation was performed under general anesthesia with placement of a 100 cc silicone implant (Figure 3). A small suction drain was placed alongside the implant and she obtained a symmetric result. The post-operative period was uneventful and the patient recovered well. Three months following the surgery, she was asymptomatic and was instructed to follow-up on an as-needed basis.

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Discussion

Classical PS is characterized by hypoplasia or aplasia of the pectoral muscles variably associated with ipsilateral thoracic and upper limb anomalies. Cardiovascular, genitourinary, spine and other abnormalities sometimes coexist in this disorder, making it a polymorphous syndrome with differing degrees of severity. The common features of the syndrome are listed in Table 1 [6].

The etiology of this syndrome is still being discussed. Most reported cases are sporadic, but the disease may be inherited as an autosomal-dominant trait. The responsible gene has not yet been mapped.

Different etiologic factors of the PS are taken into account: genetic, vascular disruption during embryogenesis and also teratogenic effect [6, 7].

Table 1: Common features of Poland Syndrome

<table>
<thead>
<tr>
<th>Hypoplasia or aplasia of sternocostal head of pectoralis major muscle</th>
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<tr>
<td>Hypoplasia or aplasia of pectoralis minor muscle</td>
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<tr>
<td>Hypoplasia or aplasia of latissimus dorsi, serratus anterior, external oblique, infraspinatus, supraspinatus, deltid and intercostal muscles</td>
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<td>Hypoplasia or absence of nipple and breast tissue</td>
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<tr>
<td>Bony dysostoses affecting hand (brachymesophalangy with syndactyly, bifid alagony, ectrodactyly), wrist, forearm, upper arm, scapula</td>
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<tr>
<td>Axillary webs and absence of axillary hair; minimal subcutaneous fat</td>
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<tr>
<td>Soft tissue syndactyly</td>
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<tr>
<td>Lung herniation</td>
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<tr>
<td>Scoliosis</td>
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<tr>
<td>Hypoplasia of hemithorax or ribs</td>
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Figure 2(a): Transverse ultrasound images of the chest wall show absence of the pectoral muscles (arrows) and reduced breast tissue (calipers) on the left side compared to the right side.

Figure 2(b): Axial non-enhanced CT scan reveals absence of the costosternal portion of the left pectoralis major muscle, left pectoralis minor muscle, left anterior intercostal muscle and reduced breast tissue compared to the right, arrows: pectoralis major, arrowheads: pectoralis minor.

Figure 2(c): Axial non-enhanced CT illustrates aplasia of the left serratus anterior muscle and hypoplasia of the left latissimus dorsi muscle, arrows: latissimus dorsi muscles, arrowheads: right serratus anterior muscle.

Figure 2(d): Axial fast spin-echo T1-weighted image illustrates absence of the left pectoral major-minor muscles, left serratus anterior muscle, left anterior intercostal muscles, hypoplasia of the left latissimus dorsi muscle and reduced breast tissue.

Figure 2(e): Axial fast spin-echo T1-weighted image demonstrates normal development of clavicular portion of the left pectoralis major muscle and aplasia of the left pectoralis minor muscle.
Depending on the physicians’ specialty and the referral pattern, a variable incidence of the anomalous defects is recognised such as rib defects (usually II-IV or II-V), absence of shoulder girdle muscles, and breast hypoplasia or agenesis athenia, vertebral anomalies and renal malformations [8]. Moreover, dextrocardia, situs inversus, lower limb malformations, Mullerian duct anomalies, malignancies like acute leukemia, non-Hodgkin lymphoma, lung cancer, and breast cancer have been described in association with PS [6, 9].

PS has a wide spectrum of clinical presentations and corrective treatment varies according to the case and surgeon. The operative reconstruction of the chest wall is based on the anatomy, the degree of severity and gender. Several treatment options have been described for chest wall reconstruction in patients with PS, including the use of breast implant or tissue expander combined with a flap, autologous fat injection, a chest wall prosthesis, local or free flap reconstruction such as a transverse musculocutaneous gracilis flap, latissimus dorsi muscle transfer, a partial latissimus dorsi flap, a rectus abdominis muscle transfer, a laparoscopically harvested omental flap, and the use of a free anterolateral thigh perforator flap [9, 10].

Cervical rib is a supernumerary rib that usually arises from the seventh cervical vertebrae. It is asymptomatic in 90% of the cases and is associated with TOS in approximately 10% of the affected population [5].

The criteria for cervical rib have been defined as follows [5, 11]:

1. The cervical rib must articulate with the C7 vertebrae with a well-defined joint; if the rib was fused with the vertebra, it is considered an elongated transverse process.
2. The rib must not originate from the transverse process of the first thoracic vertebra, but rather the 7th cervical vertebral transverse process, which projects horizontally from the spine.

Embryologically, ribs arise from precursor sclerotome cells in the thoracic region and continued growth of these cells in the cervical spine region may lead to the formation of a cervical rib. Mutations in Hox genes (a group of related genes which are instrumental in regulating body formation during development) have been shown to cause the development of cervical ribs from the costal or ventral processes of the primitive vertebral arches [12, 13].

The association of PS with rib anomalies has been reported as hypoplasia or aplasia of the ribs. But to our knowledge, its association with cervical rib and elongated transverse process of cervical vertebrae has not been reported previously in the medical literature. Thus, the
question arises as to whether this condition is a simple coincidence or is an etiological association.

**Conclusion**

PS is a rare congenital disorder that has a wide spectrum of clinical presentations. The aim of our case report was to show multimodality imaging features of the PS associated with cervical rib and elongated transverse process of cervical vertebrae. We could not find any similar case reported in the literature.

**Consent:** Written informed consent was obtained from the patient for publication of this case report.

**Competing interests:** The authors declare that no competing interests exist.

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