Eight Patients With Multiple Bilateral Thoracic Anomalies: A New Syndrome or Bilateral Poland’s Syndrome?

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Background. Poland’s syndrome is a rare congenital anomaly characterized by an absence or hypoplasia of the greater and smaller pectoral muscles, breast or nipple anomalies, hypoplasia of subcutaneous tissue, chest wall deformities, pectoral and axillary alopecia, and hand anomalies. Poland’s syndrome is usually unilateral. We present 8 patients with multiple bilateral thoracic anomalies.

Methods. Eight patients were admitted to our clinic with an abnormal thoracic appearance and restriction of shoulder mobilization. Bilateral multiple muscles, including the greater pectoral muscle and some other thoracic muscles, could not be palpated during physical examination. All patients were evaluated, with a preliminary diagnosis of bilateral Poland’s syndrome.

Results. All patients exhibited partial or complete absence of bilateral greater pectoral muscles, absence or hypoplasia of bilateral smaller pectoral muscles, bilateral shoulder protrusion to the front, limited abduction of both shoulders, absence or hypoplasia of other bilateral thoracic muscles (serratus anterior, latissimus dorsi, and trapezius muscles), and scapula alata.

Conclusions. All patients with Poland’s syndrome have unilateral hypoplasia or absence of the greater pectoral muscle as the main feature. Poland’s syndrome is routinely described as a unilateral syndrome. Cases of Poland’s syndrome are typically sporadic. Our patients had different additional bilateral anomalies. In particular, the main problems our patients had were the position of the shoulders and limited abduction of both upper extremities. In contrast to patients with Poland’s syndrome, half of our cases were familial. We consider our patients important examples in that they either present a new syndrome or show that Poland’s syndrome can be bilateral.


Poland’s syndrome is very rare and has been defined as the unilateral absence of part or the complete pectoral muscle, with different ipsilateral thoracic and upper limb anomalies [1]. The first description of Poland’s syndrome was a case reported by Alfred Poland in the English literature in 1841 [2, 3]. In this case, there was absence of the greater pectoral and the smaller pectoral muscles with accompanying syndactyly. Breast and nipple anomalies, lack of subcutaneous fat tissue, chest wall anomalies, lack of pectoral and axillary hair, and hand anomalies, in addition to pectoral muscle agenesis, were later identified as the features of the syndrome [2–5]. The absence of many thoracic muscles, Sprengel’s deformity, Klippel-Feil syndrome, and Mobsius syndrome may accompany Poland’s syndrome [6, 7]. The incidence rate of Poland’s syndrome ranges from 1:10,000 to 1:100,000 in different studies [5]. Most cases are sporadic, and cases of familial occurrence are extremely rare [3, 4]. One of the most pronounced features of Poland’s syndrome is that it is unilateral [7]. However, a bilateral case was reported by Karnak and associates [4] in 1998. In light of this information, we present 8 patients with bilateral partial and complete absence of pectoral muscle and associated bilateral multiple anomalies to discuss whether they represent models of a new syndrome or atypical Poland’s syndrome variants.

Patients and Methods

Eight patients were admitted to our clinic with abnormal thoracic appearance and restriction of shoulder mobilization between April 2009 and February 2013 (Figs 1–3). The first of the patients had been described previously in a case report [8]. All patients were men, and their mean age was 22.75 years (range, 20–35 years). Familial occurrence was observed in 4 (50%) patients. In fact, we found 3 additional patients with familial occurrence, but they did not accept the offer to join our case series. One of these patients was the sister of a patient in our case series, and the other 2 were cousins of another of our patients. If these patients had joined our case series, it would have been larger and the familial occurrence rate of the case series would have increased. Bilateral multiple muscles, including the greater pectoral muscle and some other thoracic muscles, could not be palpated...
during physical examination. At first, all patients were thought to have bilateral Poland’s syndrome. After physical examination, laboratory tests and imaging studies—including complete blood count and routine biochemical tests, chest roentgenography, thoracic computed tomography, abdominal ultrasonography, and echocardiography—were performed on all patients, and they were evaluated for Poland’s syndrome bilaterally. No surgical treatment was performed. Our series included only 8 patients, and no statistical analysis was conducted.

**Results**

The laboratory test results, including complete blood count and routine biochemical tests, were all within normal limits. Chest roentgenography showed no abnormality other than the presence of scoliosis in 1 patient (Fig 4A). Physical examination findings were confirmed by thoracic computed tomography. Complete absence of bilateral greater pectoral muscles or absence of the sternocostal heads of the greater pectoral muscles, bilateral absence or hypoplasia of the smaller pectoral and serratus anterior muscles, and bilateral scapular winging (scapula alata) were determined in all 8 patients (Table 1). There was a complete bilateral absence of greater pectoral muscles in 3 patients and a bilateral absence of the sternocostal head of the greater pectoral muscle in 5 patients (Figs 1, 4C, 5A, 5B). Bilateral absence of the smaller pectoral muscles was detected in 2 patients, and bilateral hypoplasia of the smaller pectoral muscles was detected in 6 patients (Figs 4C, 5A, 5B). All patients had bilateral scapula alata, and shoulder abduction was up to 90 degrees. In addition, all patients had the same shoulder appearance, ie, both of the shoulders protruded toward the front (Figs 2, 3). The latissimus dorsi, trapezius, and serratus anterior muscles were affected bilaterally in 7 patients (hypoplasia in 4 patients and absence in 3 patients), 1 of whom had bilateral hypoplasia of the rhomboid muscles in addition to absence of the latissimus dorsi muscle and hypoplasia of the trapezius and serratus anterior muscles (Figs 4B, 4D, 5B–5D). Only 1 patient had bilateral hypoplasia of the serratus anterior muscles without affected latissimus dorsi and trapezius muscles, in addition to other anomalies. There was also pectoral alopecia, hypoplasia of the breast and nipple, and hypoplasia of subcutaneous tissue in 7 patients, areolar asymmetry in 2 patients, and paucity of axillary hair in 4 patients. One patient had scoliosis.
Hepatomegaly was detected in 1 patient by abdominal ultrasonography, and mitral valve prolapse was detected in 1 patient by echocardiography.

Comment

Poland’s syndrome was first reported in 1841 by Alfred Poland as the unilateral absence of the greater and smaller pectoral muscles with accompanying syndactyly [9]. Over time, the definition has changed significantly and new features have been added. Today, the commonly accepted features of the syndrome include absence or hypoplasia of the breast or nipple, or both, hypoplasia of the subcutaneous fat tissue, partial or complete absence of the major pectoral muscle, absence or hypoplasia of the smaller pectoral muscle, pectoral and axillary alopecia, aplasia or deformity of the ribs, and hand anomalies [1–5]. The main prominent feature is the unilateral absence of the greater pectoral muscle, especially the absence of its unilateral sternal head. Some authors have considered that only patients who are missing the pectoral muscle have Poland’s syndrome; however, patients with at least 1 of the other associated features are also accepted as having the syndrome [4, 9]. Our patients had bilateral partial or complete absence of the greater pectoral muscle. The patients had additional anomalies on both sides and were considered to have bilateral Poland’s syndrome. Although Poland’s syndrome is a unilateral syndrome, a patient with bilateral Poland’s syndrome was reported by Karnak and colleagues [4] in 1998. Later, some case reports presented Poland’s syndrome with asymmetrical bilateral features. In 2009, Baban and associates [1] reported a patient with bilateral asymmetrical pectoral muscle defects (complete agenesis on the left side and agenesis of the sternocostal head on the right side), nipple hypoplasia, a left rib defect, and right hand symbrachydactyly [1].

If we accept that Poland’s syndrome may be bilateral, our patients are noteworthy for the consolidation of this idea. Conversely, our patients had some features that distinguished them from patients with Poland’s syndrome, which is known as a sporadic syndrome, with an extremely low (<1%) risk of recurrence in the same family [3, 7]. However, the familial occurrence rate was obviously high in our patients. Despite 3 excluded familial cases, half of our patients were familial cases. Gross-Kieselstein and Shalev [10] reported 2 brothers with absence of the trapezius, pectoral, supraspinatus, and serratus anterior muscles in 1987. Their patients had familial occurrence, but they had a 1-sided syndrome, unlike our patients. Nonetheless, these patients had no additional anomalies on the same side to consider them as having Poland’s syndrome, except the absence of the pectoral muscle.

In some cases of Poland’s syndrome, although rare, the latissimus dorsi, external oblique, deltoid, serratus anterior, supraspinatus, and infraspinatus muscles may be affected [11]. To our knowledge, absence of the trapezius muscle has not been reported as a feature of Poland’s syndrome. In our patients, bilateral latissimus dorsi and serratus anterior muscles were absent or hypoplastic. Unusually, bilateral trapezius muscles were affected. Isolated unilateral absence of trapezius muscles has been reported [12]. Horan and Bonafede [13] reported a patient with bilateral absence of the trapezius muscle and sternal head of the greater pectoral muscles in 1977. Similarly, the patient’s shoulders appeared bulbous and prominent, and shoulder abduction was up to 90 degrees. Although there was no information about the scapula and other thoracic muscles in their case report, their patient was similar to ours, as we observed in the picture. In our case series, all patients had a particular shoulder appearance, which was in a position of protrusion. In addition, the bilateral shoulder abduction was limited to 90 degrees. On physical examination, elevation and winging of the scapula, which was

**Fig 3.** (A, B) The appearance of shoulders protruded toward the front.

**Fig 4.** (A) Posteroanterior chest roentgenogram demonstrating scoliosis. (B) Axial computed tomographic images showing hypoplasia of trapezius and rhomboid muscles, (C) absence of greater and smaller pectoral muscles, and (D) absence of latissimus dorsi muscle and hypoplasia of serratus anterior muscle.
Table 1. Clinical Presentation of Patients

<table>
<thead>
<tr>
<th>Patients</th>
<th>Sex</th>
<th>Age</th>
<th>B/U</th>
<th>Greater Pectoral Muscle</th>
<th>Smaller Pectoral Muscle</th>
<th>Scapula Alata</th>
<th>Shoulders in Protrusion and Limited Abduction</th>
<th>Additional Muscle Anomaly</th>
<th>Additional Anomaly</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>M</td>
<td>20</td>
<td>B</td>
<td>Complete absence</td>
<td>Hypoplasia</td>
<td>Yes</td>
<td>Yes</td>
<td>Absence of latissimus dorsi, trapezius, serratus anterior muscles</td>
<td>Pectoral alopecia, hypoplasia of breast and nipple, hypoplasia of subcutaneous tissue, asymmetrical areola, mitral valve prolapse</td>
</tr>
<tr>
<td>2</td>
<td>M</td>
<td>21</td>
<td>B</td>
<td>Absence of sternocostal head</td>
<td>Hypoplasia</td>
<td>Yes</td>
<td>Yes</td>
<td>Hypoplasia of latissimus dorsi, trapezius, serratus anterior muscles</td>
<td>Hypoplasia of the breast and nipple, hypoplasia of subcutaneous tissue</td>
</tr>
<tr>
<td>3</td>
<td>M</td>
<td>21</td>
<td>B</td>
<td>Complete absence</td>
<td>Absence</td>
<td>Yes</td>
<td>Yes</td>
<td>Hypoplasia of latissimus dorsi, trapezius, serratus anterior muscles</td>
<td>Pectoral alopecia, paucity of axillary hair, hypoplasia of breast and nipple, hypoplasia of subcutaneous tissue</td>
</tr>
<tr>
<td>4</td>
<td>M</td>
<td>35</td>
<td>B</td>
<td>Absence of sternocostal head</td>
<td>Hypoplasia</td>
<td>Yes</td>
<td>Yes</td>
<td>Absence of latissimus dorsi, trapezius, serratus anterior muscles</td>
<td>Pectoral alopecia, paucity of axillary hair, hypoplasia of breast and nipple, hypoplasia of subcutaneous tissue</td>
</tr>
<tr>
<td>5</td>
<td>M</td>
<td>20</td>
<td>B</td>
<td>Absence of sternocostal head</td>
<td>Hypoplasia</td>
<td>Yes</td>
<td>Yes</td>
<td>Hypoplasia of latissimus dorsi, trapezius, serratus anterior muscles</td>
<td>Asymmetrical areola</td>
</tr>
<tr>
<td>6</td>
<td>M</td>
<td>23</td>
<td>B</td>
<td>Absence of sternocostal head</td>
<td>Hypoplasia</td>
<td>Yes</td>
<td>Yes</td>
<td>Hypoplasia of latissimus dorsi, trapezius, serratus anterior muscles</td>
<td>Pectoral alopecia, paucity of axillary hair, hypoplasia of breast and nipple, hypoplasia of subcutaneous tissue, hepatomegaly</td>
</tr>
<tr>
<td>7</td>
<td>M</td>
<td>22</td>
<td>B</td>
<td>Absence of sternocostal head</td>
<td>Hypoplasia</td>
<td>Yes</td>
<td>Yes</td>
<td>Hypoplasia of serratus anterior muscles</td>
<td>Pectoral alopecia, hypoplasia of breast and nipple, hypoplasia of subcutaneous tissue</td>
</tr>
<tr>
<td>8</td>
<td>M</td>
<td>20</td>
<td>B</td>
<td>Complete absence</td>
<td>Absence</td>
<td>Yes</td>
<td>Yes</td>
<td>Absence of latissimus dorsi, hypoplasia of trapezius, serratus anterior, rhomboid muscles</td>
<td>Pectoral alopecia, hypoplasia of breast and nipple, hypoplasia of subcutaneous tissue, scoliosis</td>
</tr>
</tbody>
</table>

B = bilateral; B/U = bilateral/unilateral; M = male.
hypoplastic, was detected in all patients. This appearance of winging scapula (scapula alata) was medial in our patients. Generally, the cause of medial winging is an affected serratus anterior muscle [14]. Another scapular deformity is Sprengel’s deformity, which is the congenital elevation of the scapula [15]. The scapulae of our patients were in an elevated position, and it may not be wrong to consider that there were bilateral Sprengel’s deformities in these patients. We believe that scapula alata was the reason for the abnormal configuration of our patients’ shoulders. The patient reported by Horan and Bonafe [13] had a similar shoulder appearance, but there was no knowledge of the existence of scapula alata in their case report. In our patients, when the scapula was fixed to the thorax by performing external compression, we saw a recovery of the shoulder abduction restriction. Therefore, we determined that the abnormal configuration of the shoulders and scapulae was the reason for limited abduction.

Poland’s, Sprengel’s, Klippel-Feil, and Möbius anomalies; isolated absence of greater pectoral and breast hypoplasia; and isolated limb defects are the result of an interruption of early embryonic blood supply in the subclavian arteries, the vertebral arteries, their branches, or a combination [16]. This hypothesis is the most accepted pathogenic mechanism for these anomalies. Although these syndromes have different names and features, they originate from the same pathogenic process. Accordingly, the same pathogenic process may be considered to be the probable cause of our patients’ anomalies.

If the patients in our case series are accepted as models of a new syndrome, partial or complete absence of the greater pectoral muscle, absence or hypoplasia of the smaller pectoral muscle, shoulder protrusion to the front, limited shoulder abduction, and absence or hypoplasia of the serratus anterior, latissimus dorsi, and trapezius muscles may be described as the features of this syndrome. Major features should include affected greater and smaller pectoral muscles, an abnormal configuration of shoulders and scapulae, and limited shoulder abduction. Minor features should include other affected thoracic muscles (serratus anterior, latissimus dorsi, and trapezius muscles), breast and nipple anomalies, lack of subcutaneous fat tissue, and lack of pectoral and axillary hair.

Poland’s syndrome has routinely been described as a unilateral syndrome, resulting from its first description reported by Alfred Poland in 1841. However, a few patients with Poland’s syndrome and bilateral features have recently been reported. Our patients had enough anomalies on each side to be diagnosed with bilateral Poland’s syndrome, and they might confirm the claim that Poland’s syndrome can be bilateral. Conversely, our patients differ from patients with Poland’s syndrome because of additional features, such as an increased rate of familial occurrence, abnormal configuration of the shoulders and scapulae, limited shoulder abduction, and multiple affected thoracic muscles, including the trapezius muscles. For these reasons, these patients should be considered as having a new syndrome, characterized by partial or complete bilateral absence of the greater pectoral muscle, bilateral shoulders protruding to the front, bilateral limited shoulder abduction, and bilateral hypoplasia or absence of multiple thoracic muscles (smaller pectoral, serratus anterior, latissimus dorsi, and trapezius muscles).

References

INVITED COMMENTARY

Alfred Poland, a student demonstrator of anatomy at Guy’s Hospital in London, described a postmortem dissection on a 27-year-old-convict in 1841. The body was found to have complete absence of the sternal and costal portions of the pectoralis major muscle and absence of the pectoralis minor muscle. There was a partial deficiency of the external oblique and serratus magnus (anterior) muscles as well. The patient also had symbrachydactyly of the ipsilateral hand. Since that time, there have been multiple descriptions of isolated patients with absence or hypoplasia of the pectoralis major and various other muscles of the chest wall. Many of them have had associated hand anomalies of varying severity. This anomaly is considered to be ipsilateral, and only rare cases of familial association have been reported. It has become customary to diagnose Poland’s syndrome in any patient with absence of the pectoralis major muscle. Dr Yiyit and colleagues [1] have described the clinical presentation of 8 adult men who came to their surgical clinic over a 4-year interval from 2009 to 2013. All these patients were noted to have anterior protrusion of their shoulders bilaterally with limited abduction of the shoulder. All had bilateral complete or partial absence of the pectoralis major muscles and bilateral absence or hypoplasia of the pectoralis minor and serratus anterior muscles. In half of these cases, there was a familial history of similar anomalies. The authors have also described the relationship of their patients’ anomalies with those seen in Sprengel’s deformity, Klippel-Feil syndrome, and Möbius anomalies. They propose that their 8 patients represent a new syndrome, separate from Poland’s, primarily on the basis of their bilaterality and familial history.

Until our knowledge of the genetic abnormalities involved in these disorders and the embryogenesis resulting in the physical aberrations advances, it is impossible to state whether these patients really do represent a syndrome separate from classic Poland’s. From the standpoint of a thoracic surgeon, it seems that all these disorders could best be included under the rubric of “congenital thoracic musculoskeletal disorders.” The precise anatomic variations are best determined by the use of computed tomographic or magnetic resonance imaging scans. The authors’ contribution will, however, stimulate all of us to evaluate our patients more carefully with regard to these various anatomic findings.

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Reference