Nonsyndromic Congenital Absence of the Pectoralis Muscles

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The congenital absence of the pectoralis muscle is usually a manifestation of Poland syndrome. However, a nonsyndromic congenital absence of this muscle is rare, and such absences are usually partial and unilateral. A complete or bilateral absence is even rarer. Two young men presented to our outpatient clinic with incidentally noted unilateral flat chest walls. By chest computed tomography, they were diagnosed with a congenital unilateral absence of the pectoralis muscles. They did not show any functional disability of the arms. As the congenital absence of the pectoralis muscles is often associated with leukemia and genitourinary anomalies, it is advised that hematological testing and renal ultrasonography be performed, even in nonsyndromic cases. (J Nippon Med Sch 2018; 85: 246–249)

Key words: congenital abnormalities, leukemia, pectoralis muscles, Poland syndrome, thoracic wall

Introduction

Though the partial or complete absence of the pectoralis muscles is the most common muscle defect, their nonsyndromic absence is rare. Its incidence was estimated to be 1 : 11,000. The absence of the pectoralis muscle is usually partial and unilateral. It can be part of a syndrome, such as Poland syndrome, with other associated anomalies. Usually, an absence of the sternocostal portion of the pectoralis major muscle with or without the absence of the pectoralis minor muscle is the most frequent, while the absence of the sternocostal part of the pectoralis major muscle alone is the least frequent. The associated anomalies can be deficiencies of the chest, breast, nipple, ribs, costal cartilage, axillary hair, and sweat glands (on the affected side), and scoliosis can also be present. It is called Poland syndrome in the presence of ipsilateral symbrachydactyly (webbed, short, or missing fingers/toes or hypoplasia of the hand) and called Sprengel’s deformity when resulting from hypoplasia of the serratus anterior muscle. However, nonsyndromic complete or bilateral absence is rare. There have been only 4 cases of nonsyndromic unilateral complete absences of the pectoralis muscle reported in China. The first 3 cases were found in patients, with 1 of them associated with a pterygoid anterior axillary fold, and the fourth case was found during a routine cadaver dissection. In order to document the clinical features of nonsyndromic unilateral complete absence of the pectoralis muscle, a literature review with the cases of 2 young men is presented herein.

Literature Review

The complete or partial absence of the pectoralis major muscle is relatively common as a part of Poland syndrome, with associated ipsilateral upper extremity anomalies, such as syndactyly, but the nonsyndromic complete absence of the pectoralis major muscle (without Poland syndrome) is rare, as seen in the 2 cases reported here. Dermatoglyphic (when present) and limb abnormalities can be diagnostic evidences of Poland syndrome, as the sporadic absence of the pectoralis muscle is unlikely to have such associations. The evident disparity of the associated anomalies thus contributes to the differential diagnosis of Poland syndrome and nonsyndromic congenital absence of the pectoralis muscle; however, in the 2 cases of this report, the pectoralis major muscle was completely absent, with no other deformities (such as syndactyly, hypoplasia of the ipsilateral nipple, areola, and chest wall structures, or scoliosis) leading to a diagnosis of Poland syndrome.

A systematic search of the literature was conducted to...
Absence of the Pectoralis Muscles

Fig. 1 Chest roentgenogram and computed tomography of Patient 1. (A) Chest roentgenogram of the normal right anterioaxillary fold and pectoralis shadows; and (B) Chest computed tomography showed left-sided absence of the pectoralis major and pectoralis minor muscles. Note the normal right-sided pectoralis major (large arrow) and minor muscles (small arrow) on computed tomography.

Fig. 2 Chest roentgenogram and computed tomography of Patient 2. (A) Chest roentgenogram of the normal left anterioaxillary fold and pectoralis shadows; and (B) chest computed tomography showed a flat chest and right-sided absence of the pectoralis major and pectoralis minor muscles. Note the normal left-sided pectoralis major (large arrow) and minor muscles (small arrow) on computed tomography.

look for reports relating to congenital absences of the pectoralis muscles. No limitations were applied to the literature retrieval in terms of publication date, the language of the article, or the type of research. By excluding the syndromic cases, there were 20 related articles retrieved. Only one of these articles referred to a large series (25 patients), while the rest were single cases or small case series. There were, in total, a few more than 50 cases of congenital absence of the pectoralis muscle reported in the literature. Nevertheless, some of the cases featured the partial or bilateral absence of the pectoralis muscles. In contrast, the complete unilateral absence of the pectoralis muscles was even rarer. Moreover, the absence of the lateral or medial pectoral nerve has been noted in cadaver cases that included the unilateral absence of pectoralis major muscle.

Recent Experiences
Two young men aged 19 and 18 years old visited our outpatient clinic on May 26, 2014, and on June 3, 2014, respectively, for the incidentally noted unilateral flat chest walls. Their family histories were investigated to determine the presence of any familial incidence of a similar genetic disorder. Physical, roentgenogram, and computed tomography examinations were performed. Their hematological and renal function tests showed no evidence of associated leukemia or renal disorders. Their past histories and family histories were unremarkable as there were no obvious chest wall or upper limb abnormalities found in their first- and second-degree relatives;
therefore, the possibility that either patient had inherited his condition was judged to be remote. Physical examinations revealed that Patient 1 had a left-sided flat chest wall and that his left anterior axillary fold was absent and that Patient 2 had a right-sided flat chest wall and that his right anterior axillary fold was absent. The nipples and areolas on the affected sides of both patients were smaller than those on the contralateral sides. Their upper limbs were normal, and no syndactyly of the border digits (thumb/index finger or ring/small fingers) was found. Neither showed any significant differences in the lengths and circumferences of their arms, and their muscle strength was normal for flexion, horizontal abduction, adduction, and internal rotation. Roentgenograms and computed tomography scans of the patients’ chests showed the absence of the pectoralis muscles on the affected sides of both men (Fig. 1, 2). Their hematological and renal function tests as well as renal ultrasound scans were normal. The demographic data are shown in Table 1.

Table 1 Demographic data of the two patients

<table>
<thead>
<tr>
<th>Patient data</th>
<th>Normal range of laboratory tests</th>
<th>Patient 1</th>
<th>Patient 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age (years)</td>
<td></td>
<td>19</td>
<td>18</td>
</tr>
<tr>
<td>Sex</td>
<td>Male</td>
<td>Male</td>
<td></td>
</tr>
<tr>
<td>Height (cm)</td>
<td>175</td>
<td>179</td>
<td></td>
</tr>
<tr>
<td>Body weight (kg)</td>
<td>52</td>
<td>56.5</td>
<td></td>
</tr>
<tr>
<td>Body mass index (kg/m²)</td>
<td></td>
<td>17.0</td>
<td>17.6</td>
</tr>
<tr>
<td>Family history</td>
<td>Unremarkable</td>
<td>Unremarkable</td>
<td></td>
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</tbody>
</table>

Hematology

<table>
<thead>
<tr>
<th>Test</th>
<th>Normal range</th>
<th>Patient 1</th>
<th>Patient 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hemoglobin (g/L)</td>
<td>130–175</td>
<td>135</td>
<td>137</td>
</tr>
<tr>
<td>White blood cells (×10⁹/L)</td>
<td>3.5–9.5</td>
<td>7.6</td>
<td>6.8</td>
</tr>
<tr>
<td>Neutrophils (%)</td>
<td>40–75</td>
<td>70.2</td>
<td>68.9</td>
</tr>
<tr>
<td>Platelets (×10⁹/L)</td>
<td>125–350</td>
<td>198</td>
<td>220</td>
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Renal function tests

<table>
<thead>
<tr>
<th>Test</th>
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<th>Patient 1</th>
<th>Patient 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Serum creatinine (μmol/L)</td>
<td>44–133</td>
<td>98</td>
<td>110</td>
</tr>
<tr>
<td>Blood urea (μmol/L)</td>
<td>2.9–8.2</td>
<td>6.8</td>
<td>4.2</td>
</tr>
<tr>
<td>Blood uric acid (μmol/L)</td>
<td>90–420</td>
<td>286</td>
<td>325</td>
</tr>
<tr>
<td>Urinary protein</td>
<td>Negative</td>
<td>Negative</td>
<td>Negative</td>
</tr>
</tbody>
</table>

Discussion

A variety of theories have been proposed to explain the pathogenesis of the absence of the pectoralis muscle; however, Stein did not support the presence of a genetic background as being one of them. At present, the absence of the pectoralis muscle might be interpreted as a fetal insult of pectoralis mesenchyme separation into the primordium of the pectoralis major and minor muscles at the 6th to 7th gestational weeks. It is probably due to a vascular injury during limb bud formation, one that compromises the circulation in the region. David and Winter reported that a familial case of the absence of the serratus anterior and partial absence of the latissimus dorsi muscles in association with absence of the pectoralis major muscle might be either an autosomal-dominant or a Y-linked genetic disorder. During a routine cadaver dissection, Dayal et al. found a unilateral complete absence of the pectoralis major and serratus anterior muscles. As the family history of the deceased individual revealed no abnormalities specific to the upper limbs, Dayal and his team concluded that the anomaly either had been caused by a sporadic variant of Poland syndrome or was an isolated instance of the congenital unilateral absence of the pectoralis major and serratus anterior muscles. It should be noted (for application in other cases) that while our patients were free from renal difficulties, the combination of an acral anomaly and renal malformation has been identified as a polytopic developmental field defect.

Patients with an absence of the pectoralis muscle on one side of the body usually present with a flattened chest wall, hypoplastic ribs, an elevated nipple, and possible unilateral hyperradiolucency of the lung on chest roentgenogram. Such an absence often causes little or no functional disability and often remains unnoticed. Enhanced muscle strength that compensates for the missing pectoralis muscle on the affected side has been observed. As was observed in a cadaver without pectoral muscles, the enhanced clavicle portion of the deltoid...
muscle on the affected side might serve as a compensatory mechanism in terms of the abduction and internal rotation of the arm. Furthermore, greater potential activities were noted on the affected side with a surgically removed pectoralis muscle than on the contralateral side. While patients may tend to experience a reduction in shoulder strength, no significant muscle dysfunction is present. Lee and Chun reported on a 22-year-old soldier with unilateral congenital absence of his pectoralis major muscle. The patient had difficulties with upper extremity movement, including throwing and climbing, but no visible or radiological signs of associated anomalies were found in the ipsilateral arm.

Patients with Poland syndrome are prone to developing hematological neoplasms, namely acute lymphoblastic and myeloblastic leukemias and non-Hodgkin's lymphomas, as well as chronic myelogenous leukemia. Parikh et al. summarized a series of 8 patients of Poland syndrome with acute leukemia, with 6 of them being acute lymphoblastic leukemia and 2 being acute myeloid leukemia. Their own patient (with acute lymphoblastic leukemia) had a complete remission 16 weeks after chemotherapy. It was supposed that there might be a chance association between Poland syndrome and leukemia, owing to the doubtful link between them. Moreover, associations between the absence of the pectoralis muscles and renal disorders, such as megacalycosis of the ipsilateral kidney and absence of the ipsilateral kidney, have been noted. The renal disorders in Poland syndrome have been proposed to be due to the maldevelopment of the metanephrons during the developmental processes that results in the formation of the ureteric bud and in that of the mesonephric duct, early in gestation. It is therefore advised that hematological testing and renal ultrasonography should be performed, even in nonsyndromic patients.

Nonsyndromic congenital absence of the pectoralis muscle is rare. Isolated congenital absence of the pectoralis muscle with no associated anomalies should be differentiated from Poland syndrome. The pathogenesis of the nonsyndromic cases warrants further investigations. As the congenital absence of the pectoralis muscles is often associated with leukemia and genitourinary anomalies, hematological testing and renal ultrasonography should be performed in these nonsyndromic cases.

**Conflict of Interest:** The author declares no conflict of interest.

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**References**