POLAND’S SYNDROME WITH SPONTANEOUS PNEUMOTHORAX: REPORT OF TWO CASES

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Abstract: Poland’s syndrome is an uncommon congenital anomaly of the chest wall characterized by the absence of the pectoralis major muscle and other nearby musculoskeletal components. Many associated aberrations over the thoracic cage, intrathoracic organs, and upper limbs have been reported. However, spontaneous pneumothorax in these patients has not been reported. Here, we describe two patients with both Poland’s anomaly and spontaneous pneumothorax. One patient was a 16-year-old boy with left chest wall hypoplasia and pneumothorax on the right side. The other was a 27-year-old man with right chest wall hypoplasia, hand brachydactyly, and pneumothorax. Pneumothorax in both patients was treated with bullectomy and mechanical pleurodesis with the aid of videothoracoscopy, and the postoperative courses were smooth. Blood supply disruption has been hypothesized as a pathogenic mechanism of both spontaneous pneumothorax and Poland’s syndrome, suggesting an association between these two diseases.

Case Reports

Case 1

A 16-year-old boy complained of right chest pain, cough, and shortness of breath after exercise on the morning of admission. He had experienced several similar attacks previously with less severe symptoms. At first, he was sent to another hospital for examination and was then transferred to our emergency room for further evaluation. Physical examination revealed a tall, slim boy, with an asymmetric chest wall due to hypoplasia of his left pectoral muscle. He looked uncomfortable, with cold sweating and shallow, rapid breathing. His breathing sounds on auscultation were prominently decreased over the right lung field. No other musculoskeletal or cardiothoracic anomaly was noted. Hemogram and blood chemistry data were normal except for elevated glucose before eating (120 mg/dL). An electrocardiogram revealed sinus tachycardia with right heart strain pattern. Chest roentgenogram revealed hyperlucency over the right lung field. Tube thoracostomy was performed under the impression of right-sided pneumothorax. His symptoms were partially relieved after the intrapleural air was released. However, persistent air-
leakage from the chest tube, a poorly expanded right lung, and repeated attack history made him a candidate for surgical intervention. Thoracoscopy revealed a group of apical blebs at the apex of the right lung surface. Therefore, blebectomy using endo-gastrointestinal anastomosis stapling and mechanical pleurodesis using gauze abrasion were performed with the aid of thoracoscopy. The postoperative course was uneventful. The chest drainage tube was removed 4 days later and the patient was discharged 5 days after surgery. No recurrence of pneumothorax was noted during the 24-month follow-up period.

Case 2
A 27-year-old man had suffered from repeated attacks of right chest pain for 1 year, but he had ignored these pains. Congenital agenesis of the right pectoralis major muscle with right brachysyndactyly had been noted since childhood (Fig. 1). On the occasion of this last attack severe chest pain was noted, and he was admitted to our ward for further evaluation a day later. Besides the thoracic and musculoskeletal anomalies, prominent decreased breathing sounds were noted over the right lung field. Chest roentgenogram and computed tomogram revealed a right-sided hemopneumothorax with agenesis of the right pectoralis major muscle (Fig. 2). His discomfort was not relieved after tube thoracostomy and subcutaneous emphysema appeared over his neck, trunk, and upper limbs. Surgical intervention was indicated due to repeated attacks and resistance to conservative treatment. On thoracoscopic examination, a 2-cm ruptured bulla, based at the apex of the lung, was found adherent to the adjacent chest wall. Pneumolysis, bullectomy, and mechanical pleurodesis were performed with the aid of thoracoscopy. He was dis-

Discussion
Poland’s syndrome manifests as varying degrees of thoracic cage hypoplasia and anomalies of the hand.
This anomaly might be confined, with only partial absence of the pectoralis major muscle, or more severe in configuration, involving agenesis of the ribs, sternum, scoliosis, brachysyndactyly, or mammary aplasia, and absence of the latissmus dorsi, serratus anterior, or other nearby structures [3, 9–11]. Combined anomalies have been reported such as congenital heart disease, pulmonary and renal hypoplasia, diaphragmatic hernia, lower limb anomalies, craniofacial defects, and Möbius’ syndrome. Moreover, certain malignancies such as lung and breast carcinoma, leukemia, Wilm’s tumor, or leiomyosarcoma have also been described in association with Poland’s syndrome [4–6, 12–15]. Bullous pulmonary disease associated with Poland’s syndrome has been reported [16]. However, spontaneous pneumothorax in such patients has not been previously described.

The pathogenesis of Poland’s syndrome is still unclear. However, Bavinck and Weaver proposed that the related Poland, Klippel-Feil, and Möbius anomalies are the result of an interruption of the early embryonic blood supply in the subclavian arteries, the vertebral arteries, or their branches [7]. They hypothesized that the occlusions occur at specific locations in these vessels at around the sixth week of embryologic development. The birth defects represented by these conditions arise through a process called the subclavian artery supply disruption sequence.

The most likely cause of a primary spontaneous pneumothorax is the rupture of small blebs or bullae [8, 17]. This may occur at rest or during exercise, and it is seen most often in young tall males with smoking habits [18]. The pathogenesis of the blebs or bullae is not clear, but might be related to an increased apex-to-base intra-alveolar pressure gradient, small airway partial obstruction, or compromised blood supply. The diagnosis of pneumothorax is best confirmed by erect posteroanterior and lateral chest roentgenogram. Primary spontaneous pneumothorax can be treated by close observation, aspiration and small tube drainage, conventional tube thoracostomy, or ambulatory tube drainage. Indications for surgical intervention include prolonged air-leak, poor re-expansion of the lung, bilateral pneumothorax, or their branches [7]. They hypothesized that the occlusions occur at specific locations in these vessels at around the sixth week of embryologic development. The birth defects represented by these conditions arise through a process called the subclavian artery supply disruption sequence.

The recurrence rate can be remarkably decreased after surgery (2%) as compared with non-surgical treatment (7–50%) [20]. Thoracic anomalies of Poland’s syndrome can also be reconstructed surgically, in cases where these aberrations are severe [2]. The most commonly used reconstruction involves the latissmus dorsi muscle, which is transferred to the anterior chest wall, while preserving the neurovascular pedicle. Breast prosthesis might be included in this reconstruction in women. Chest wall prosthesis is not recommended because of complications associated with its insertion, such as migration, local tissue erosion, and adverse cosmesis. Both of our patients refused surgical reconstruction for their thoracic wall anomaly.

Poland’s anomaly with spontaneous pneumothorax has not been previously reported. Although clinical data on the association of these conditions is limited to this report, they might share some common pathogenic mechanism such as compromised blood supply. Although one of our patients had pneumothorax on the same side as the thoracic wall anomaly, the other had pneumothorax on the opposite side to the anomaly. These patients might have had bilateral pulmonary blebs, and pneumothorax might have occurred incidentally on one side only. Either the blebs (unilateral or bilateral) or chest wall anomaly can be attributed to a common pathogenesis (compromised brachial arch blood supply in embryo). We can also hypothesize that our patient with pneumothorax contralateral to the chest wall anomaly might have had two different brachial arch blood supply defects. However, more such cases need to be reported to clarify the relationship of these two diseases.

References


