Poland Syndrome With Renal Stone: A Case Report

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Abstract- Poland syndrome is a rare congenital anomaly with various presentations. The most consistent findings of this syndrome are hypoplasia or aplasia of the sternocostal part of the pectoralis major muscle and upper limb malformations. Here, we are reporting a case of Poland syndrome with renal stone, a new finding that has not been reported in previous literature.

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Keywords: Poland syndrome; Partial rib agenesis; Pectoralis muscle agenesis; Renal stone

Introduction

Poland syndrome is a rare congenital condition with an estimated incidence of 1 in 30000 to 1 in 80000 live births, which was initially described by Alfred Poland in 1841 as a partial or complete absence of the pectoralis major and minor muscles, hypoplasia of the breast as well as ipsilateral upper limb malformations (1,2). This condition has male and right side predominance (3).

There are several other associated findings of this syndrome, including neurologic, cardiovascular, musculoskeletal, genitourinary, dermatologic, and several types of malignancies (Table 1) (4-8).

In this paper, we are reporting renal stone as a new associated finding of Poland syndrome.

Table 1. Poland syndrome associations

Brain	**Microcephaly, cerebral atrophy, myelination disorder, mental retardation**
Ear, eye, and face	External ear malformation, eye anomaly, cleft palate
Heart	Dextrocardia ,atrial septal defect
Lung	Lung cancer, diaphragmatic hernia
Kidney	Ipsilateral renal agenesis duplicated renal system
Reproductive system	Un descendent testis ,inguinal hernia ,hypospadias
Breast	Ipsilateral aplasia, hypoplasia or absence nipple, and areola (athelia)
Thoracic cage	Aplasia or hypoplasia of pectoralis muscles, latisimus dorsi, serratus anterior, external oblique, infraspinatus, supraspinatus Aplasia or hypoplasia of rib cage Agenesis of ipsilateral costal cartilage(2,3,4and 5 ribs) Vertebral defects, hemivertebra
Upper limb	Brachydactyly (aplasia of middle phalanges), syndactyly, absent finger, short arm, underdevelopment of forearm bones Absent shoulder blade or arm bones Sprengel deformity
Lower limb	Knee flexion contracture, club foot, toe syndactyly
Dermatologic	Pectoral and axillary alopecia, absence of sweat gland, anhidrosis
Malignancy	Lymphoma (NHL), leukemia, leiomyosarcoma, cervical cancer, Wilms tumor, neuroblastoma, congenital hemangioma
Vascular malformation	Hypoplasia or aplasia of upper limb vessels

Case Report

An 8-year-old boy came to our center with one week of fever, chills, and cough productive of clear sputum. He was admitted to the hospital with the diagnosis of pneumonia. No history of previous disease or admission was noted. He was the first child of his family, with unremarkable familial history.

Physical examination revealed a boy with normal growth characteristics and good mental status. On inspection,

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asymmetry, and depression of the right side of his chest wall were detected (Figure 1). Heart sounds were normal without a murmur. Lung auscultation revealed rales over the right lung field. Brachydactyly of the ipsilateral upper hand was detected (Figure 2). The remainder of the examination was normal.

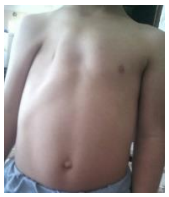

Figure 1. The right side of the chest wall shows asymmetry and depression

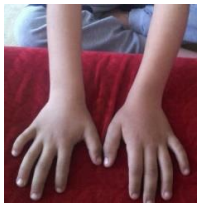

Figure 2. The right hand of the patient demonstrating brachydactyly

Chest x-ray (CXR) revealed deformity of the thoracic cage, absence of the anterior part of the right 3rd, 4th and 5th ribs, as well as consolidation on the right side of the heart border with Silhouette sign. On the chest CT scan, the absence of right side pectoralis major and minor muscles, in addition to previous findings were noted (Figure 3). The diagnosis of Poland syndrome was considered based on these findings.

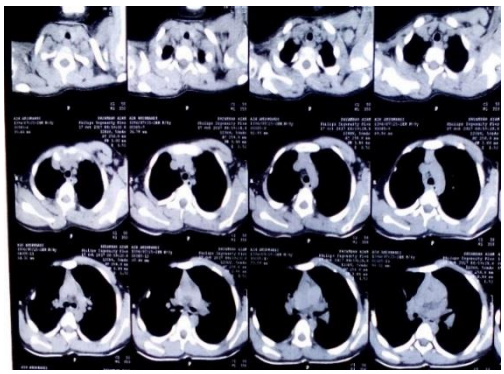

Figure 3. chest CT scan reveals agenesis of the right pectoralis muscles and the anterior portion of the right ribs as well

Furthermore, during the CXR performance, an opacity was observed on the anatomical location of the left kidney. For further investigation, an abdominal and renal CT scan was performed, which detected renal stone (Figure 4). Other investigations, including complete metabolic panel and urinalysis, were unremarkable.

During the hospitalization, the patient received antibiotics for his pneumonia. We recommended our patient to undergo reconstructive surgery after puberty, and annual follow up for hematopoietic malignancies as well.

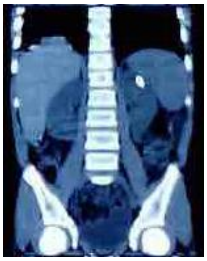

Figure 4. Abdominal CT scan (coronal view), demonstrating the renal stone

Discussion

The etiology of Poland syndrome is not well known, but there is a hypothesis that suggests the interrupted arterial supply of the upper limb (*i.e.*, subclavian, vertebral, or other arterial branches) as a probable cause (9,10). In some studies, tobacco usage in pregnancy has been reported as a risk factor (6). Most cases are sporadic, but in some patients, the autosomal dominant (AD) pattern of inheritance also has reported (11). Our patient had no family history of Poland syndrome, and therefore he was a sporadic case. In addition, his mother did not have a history of exposure to cigarette smoke during her pregnancy.

Patients with Poland syndrome have multiple associated findings that physicians should be aware of them. A complete physical exam, including eye movement, is necessary to rule out Mobius syndrome (4). In order to evaluate associated congenital cardiac malformations, echocardiography should be performed. Further evaluations, such as complete blood count, are essential in the detection of associated hematologic malignancies. All of these studies were normal in our patient.

Based on previous literature, an important association of this syndrome is congenital renal malformations such as ipsilateral renal agenesis and duplicated urinary collecting system (2). Our patient did not have these anomalies but presented renal stone. He had no abnormality in his complete metabolic panel and urinalysis. Hence, the renal stone can be considered as an associated finding of Poland syndrome.

Poland syndrome is a rare congenital disease with a wide variety of associated abnormalities such as renal problems. Considering these associations helps

physicians to perform appropriate interventions and minimize renal damage.

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