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CASE REPORT

# A rare case of Poland syndrome accompanied by lower extremity anomaly

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## ABSTRACT

Poland syndrome (PS) is a rare congenital anomaly characterized by hypoplasia of the pectoralis major muscle, variable degrees of upper extremity deformities, breast tissue, and anterior chest wall deformities. A 13-year-old male patient had brachydactyly of the fingers of the left hand and symbrachydactyly. On physical examination, the left breast tissue and areola were significantly hypoplasic compared to the right. The patient's right leg was hypoplasic compared to the left. Lower extremity anomalies are rarer compared to upper extremity anomalies in PS. Since hypoplasia of the upper extremities is one of the common findings of PS, we think that hypoplasia of the lower extremities found in our patient may be a rare component of PS.

**Keywords:** Anomalies, lower extremity, Poland syndrome.

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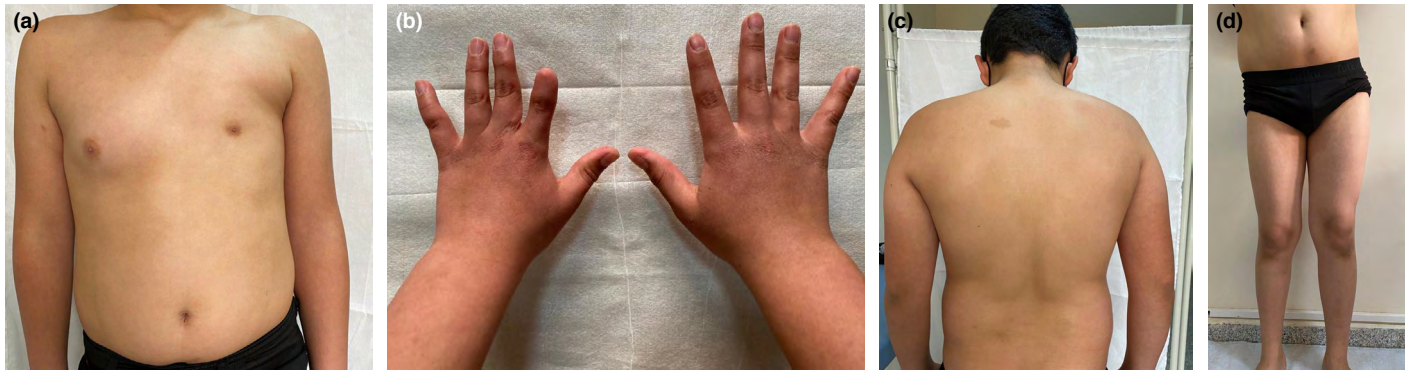


**Figure 1: (a)** Left side hypoplastic areola, breast tissue, and atrophic pectoral muscles. **(b)** Brachydactyly, agenesis, and operation scar on the left hand. **(c)** Cafe au lait spot on the left scapula superomedial. **(d)** Hypoplasic right leg.

## INTRODUCTION

Poland syndrome (PS) is a rare congenital anomaly characterized by (partial or total) hypoplasia of the pectoralis major muscle, variable degrees of upper extremity deformities, breast tissue, and anterior chest wall deformities. It was first described by Alfred Poland in 1841.[1,2] In PS, mild musculoskeletal abnormalities such as breast and subcutaneous tissue under the breast, areola abnormalities, and loss of hair growth in the axilla may be observed, while severe clinical findings such as the absence of cartilage tissues, absence of radius, and ulna may also be encountered.[3] In this syndrome, the right side of the body is affected more frequently than the left side and the syndrome is more common in boys than in girls.[4] The average incidence is one in 10,000–100,000 live births. Although the exact cause is not known, vascular pathologies and teratogenic agents are blamed for the etiology of PS.[5] Decreased blood flow in the vertebral artery, subclavian artery, and/or their branches in the early embryonic period is held responsible for vascular causes.[6]

In this article, we aimed to present a case of lower extremity anomaly which rarely accompanies PS.

## CASE REPORT

A 13 years and 6-month-old male patient was admitted to the pediatrics outpatient clinic because his left breast was smaller than the right one. He was born at 39 weeks of gestation with a birth weight of 3500 g from a gravida 2, parity 1, 28-year-old mother by normal spontaneous vaginal delivery in the hospital. There was no kinship between the parents. At birth, the distal phalanx of the left hand's 2nd finger was agenetic. He had brachydactyly of the fingers of the left hand and symbrachydactyly of the second and third fingers. The patient's developmental milestones progressed in accordance with his age. His brother did not have any health problems. The patient was operated on for hypospadias and symbrachydactyly at the age of 4 years. The patient does not have any chronic diseases. On physical examination, the left breast tissue and areola were significantly hypoplasic compared to the right, the left areola was located at an upward level compared to the right, and the pectoral muscles were atrophic on palpation (Fig. 1a). He had brachydactyly on his left hand, agenesis in the 2nd distal phalanx, and an operation scar between the 2nd and 3rd finger (Fig. 1b). Other examinations of the left upper extremity were normal and there was a 2×1 cm cafe au lait stain on the superomedial aspect of the left scapula (Fig. 1c). During the measurements made between the spina iliac anterior superior and medial malleolus in the lower extremity examination, the actual length of the right leg was 81 cm and the actual length of the left leg was 84.5 cm, while there was 3.5 cm shortness in the right lower extremity. The right leg was hypoplastic compared to the left (Fig. 1d). Other lower extremity examination results were normal. Echocardiography and abdominal ultrasound did not reveal any pathology. Thoracic CT revealed no musculoskeletal abnormalities other than pectoralis major sternocostal fibers and pectoralis minor aplasia (Fig. 2). Pulmonary parenchyma and vascular structures were normal. No pathology was found in other systemic examinations and laboratory tests. Considering the present findings, the patient was diagnosed with PS due to pectoralis major muscle hypoplasia, pectoralis minor muscle agenesis, and left upper extremity deformities.

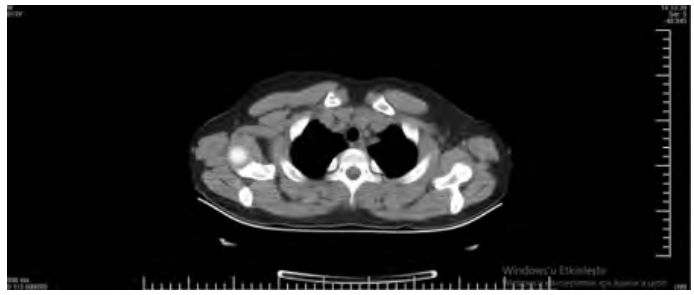

**Figure 2:** Pectoralis major sternocostal fibers and pectoralis minor aplasia on thorax CT.

Written informed consent was obtained from the patient's parents.

## DISCUSSION

The major finding of PS is aplasia-hypoplasia of the unilateral pectoralis major muscle, especially of the sternocostal fibers; and the clinical presentation of PS varies according to the anomalies accompanying the syndrome. PS may be accompanied by upper extremity anomalies such as adactyly, adactyly, arm hypoplasia, arm hypoplasia, absence of radius bone in severe cases, phocomelia, often with ipsilateral symbrachydactyly, syndactyly, brachydactyly; breast and nipple pathologies such as hypomastia, hypotelia, amastia, ateli, polytomyelia; cardiac anomalies such as dextrocardia, atrial septal defect; and genitourinary system anomalies such as renal agenesis, undescended testis, or vesicoureteral reflux.[7–9]

Bilateral PS has been reported very rarely in the literature.[10] In general, the right side of the body is more affected by this syndrome. In our case, there was agenesis of the left pectoralis major sternocostal fibers and pectoralis minor muscles.[4] Al-Qattan, in 2001, classified the hand abnormalities observed in PS into seven subtypes. According to this classification, in type 1, there is no hand involvement; in type 2, the hand on the affected side is functionally normal but more hypoplasic than the intact side. Symbrachydactyly is present in type 3, mild hypoplasia is specified as type 3a, and severe hypoplasia as type 3b depending on the severity of the accompanying hypoplasia. Type 4 is characterized by the absence of some digits, type 5 by the absence of functional digits, type 6 by accompanying rudimentary metacarpals, and type 7 by phocomelia.[9] The hand findings of our patient were compatible with the operable type 3a. In a case series prepared by Güzel Nur et al.[11] hyperpigmented spots were reported in two of eleven patients. In our case, there was a 2×1 cm hyperpigmented lesion on the superomedial aspect of the left scapula. Our patient was operable hypospadias, and we did not find any case report of PS with hypospadias in the literature. PS is less frequently accompanied by lower extremity anomalies compared to upper extremity anomalies. Four cases with lower extremity anomalies were reported in the literature review together with the case report prepared by Baban et al.[12] In 1932, Glos reported a case of bilateral breast tissue aplasia with an absence of fingers on the right hand and congenital deformity of the right foot. The details of lower extremity deformity are not mentioned.[13] In 1994, Kabra et al.[14] detected multiple anomalies in a patient with left-sided chest and upper extremity findings, bilateral talipes equinovarus, syndactyly, and nail hypoplasia in the 4th and 5th toes, medial cleft in the right foot, and large and wide hallux in the left foot.[12] In a case published in 2003, contralateral lower extremity hypoplasia and talipes valgus accompanying left upper extremity anomalies were found.[12] In a study reported from our country in 2005, symbrachydactyly and absence of the 5th finger were found in the right foot accompanying bilateral upper extremity anomalies.[12] We found contralateral lower extremity hypoplasia in our patient with PS. Our patient had a 3.5 cm shortening of the right lower extremity. The patient was consulted by orthopedics; the surgical operation was not considered, reinforced insoles were prescribed and the patient was followed up.

In PS, there are different treatment options according to the functional loss and esthetic problems caused by the existing deformities. The age and gender of the patient should be taken into consideration before surgical operations. For operations planned due to esthetic problems, the completion of the patient's growth and development should have waited. Autologous fat injections and silicone implants are frequently preferred interventional methods. Autologous muscle transpositions, muscle transfers, and costal grafts are used in the treatment of anomalies of the anterior chest wall. In cases with syndactyly, interventions for adhesions should be performed early to prevent the progression of loss of function. Toe transfers can be applied in finger agenesis. Prostheses may be preferred in limb absences.[7] In the approach to lower extremity inequalities, for shortness below 5 cm preferred conservative treatments such as reinforcement insoles. For differences above 5 cm, epiphysiodesis, shortening osteotomies and limb lengthening operations are surgical options that can be preferred in appropriate patients.[15] Our patient underwent operations for syndactyly and hypospadias at the age of four. No surgical operation was considered for lower extremity shortness, and the patient was followed up with conservative treatment. Since there was no loss of function and growth and development continued, surgical operation for the anterior chest wall was not considered, and the family was informed about esthetic operations. The patient was followed up.

Lower extremity anomalies are rarer compared to upper extremity anomalies in PS. Since hypoplasia of the upper extremities is one of the common findings of PS, we think that hypoplasia of the lower extremities found in our patient may be a rare component of PS. Therefore, length measurements should also be evaluated in the examination of the lower extremities in patients diagnosed with PS. We aimed to present this case to contribute to the literature.

## Statement

**Informed Consent:** Written, informed consent was obtained from the patient's family for the publication of this case report and the accompanying images.

**Peer-review:** Externally peer-reviewed.

**Author Contributions:** Concept – CYG, EU; Design – CYG, EU; Supervision – CYG, EU; Resource – CYG, EU; Materials – CYG, EU; Data Collection and/or Processing – CYG, EU; Analysis and/or Interpretation – CYG, EU; Literature Search – CYG, EU; Writing – CYG, EU; Critical Reviews – CYG, EU.

**Conflict of Interest:** The authors have no conflict of interest to declare.

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