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Case Report

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Poland Syndrome Presenting with Failure to Thrive: A Case Report

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Abstract

Poland Syndrome is a rare congenital anatomic anomaly disorder characterized by ipsilateral breast and nipple hypoplasia and/or aplasia, deficiency of subcutaneous fat and axillary hair, absence of the sternal head of the pectoralis major muscle, hypoplasia of the rib cage, and hypoplasia of the upper extremity. I present the case of a 3-month-old male infant who exhibited symptoms of Poland syndrome, had no family history of the condition, and presented with a right upper chest deformity, including bulging of the lung with breathing, and failure to thrive.

Keywords: Lung protrusion; Poland sequence; Failure to thrive; Hypoplastic rib; Poland's syndrome; Clinical assessment; Diagnosis of PS

Introduction

Poland syndrome (PS), also known as Poland sequence, is a rare congenital chest wall muscle deficiency disorder. Nobody is entirely sure of what causes the condition, but it is likely to be due to a temporary blockage of the blood supply of the area affected during development. In Poland syndrome they usually present with hypoplasia of the subclavian or axillary vessels, which supports the theory of a poorly developing limb bud. This could be caused by many factors such as amniotic bands, tumours, edema, or aberrant muscle during the sixth and seventh weeks of embryogenesis [1]. Diminished supply affects fetal growth at about the 46th day of pregnancy when the fetal fingers and pectoralis muscle are developing. Other etiologies to be considered are teratogenicity (medication-induced, smoking) and genetics [2]. The exact incidence of PS is not known due to the likelihood that it is often underdiagnosed and underreported [1]. Incidence rates are usually estimated. Moreover, many cases are missed and under-represented as mild forms are more common than severe forms. These aspects can lead to an underestimation of the overall incidence rates [1,3]. The incidence of Poland syndrome ranges from 1 to 3 per 100,000 newborns, with a higher prevalence in males more common on the right side than on the left [4].

This report describes the case of a 3-month-old Caucasian infant with failure to gain weight and an upper chest deformity. The patient had no family history of PS and was first brought to the paediatrician by his mother as a newborn of 22 days old with a weight of 7.31 lbs (6^{th} percentile) and height of 1 ft 8 inches (9^{th} percentile) Since then, the patient has been under routine surveillance to help him keep well nourished. This case further highlights the importance of adhering to current surveillance recommendations and prevention strategies in patients with PS to avoid complications.

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Case Presentation

A 3-month-old Caucasian male infant presented at the paediatric office for a routine check-up. At this visit, the patient's measured height was 1 ft 10 inches (less than 1%) and weight 10.63 lbs (less than 1%), with a BMI of 15.4 kg/m. The patient delivered vaginally at full term. The mother's medical and obstetric history was uneventful and there was no maternal drug abuse during gestation; there was no indicative of any physical deformities in the parents upon examination. Since birth, the boy has exhibited a deformity of the thoracic cage while the patient's older sibling has no medical abnormality.

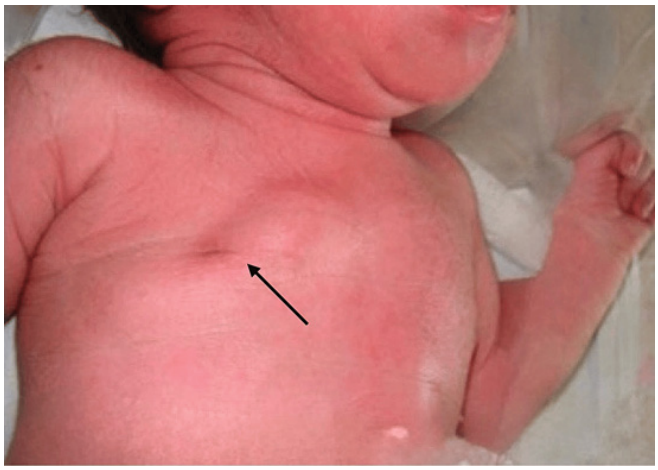

Figure 1: Protrusion of the right lung through the chest wall at the level of 3rd rib cage.

On physical examination the patient exhibited the right lung herniation through the chest wall upon breathing or engaging in forceful exertion, such as crying (Figure 1). However, no other abnormalities were detected during the rest of the physical examination. Thoracic chest radiography was performed at the age of 2 months and 11 days and revealed a hypoplastic right third rib with a normal cardio thymic silhouette, lung impression, and no evidence of pleural effusion. Following the chest X-ray, a renal ultrasound was conducted to ensure that there were no associated abnormalities; fortunately, the results were negative.

During the visit, laboratory results showed that the baby's red blood cell count had significantly improved compared to previous readings (Table 1). Upon further questioning, the parent revealed that the baby drinks around 57 oz of formula per day and occasionally vomits it out but has normal stooling and bladder voiding. The baby appeared active and healthy during the physical examination. At the end of the visit, the parents were advised to thicken the formula with Gerber rice cereal to increase the calorie intake per feeding and to give the baby Vitamin D3 orally daily. Patient's mother was advised to take the patient to emergency care immediately if any severe changes are noted.

Table 1: Comparison of previous and current Lab Blood reports.

Lab Findings	Initial	Follow Up
Complete Blood Count (CBC):		
White Blood Cells (WBC)	10,800 /µL	9,200 /µL
Red Blood Cells (RBC)	3.3 million /µL	4.2 million /µL
Hemoglobin (Hb)	8.5 g/dL	11.0 g/dL
Hematocrit (Hct)	28%	35%
Mean Corpuscular Volume (MCV)	75 fL	83 fL
Platelets	210,000 /µL	250,000/µL
Electrolytes:		
Sodium (Na)	129 mEq/L	138 mEq/L
Potassium(K)	3.3 mEq/L	4.4 mEq/L
Chloride (Cl)	95 mEq/L	103 mEq/L
Bicarbonate (HCO3)	18 mEq/L	24 mEq/L
Iron Studies:		
Serum Iron	36 µg/dL	65 µg/dL
TIBC	324 µg/dL	350 µg/dL
Ferritin	9 ng/mL	46 ng/mL
Thyroid Function:		
TSH	4.9 µIU/mL	5.0 µIU/mL
Free T4	0.9 ng/dL	1.1 ng/dL

Discussion

Most Poland syndrome is found sporadically with some occasional familial genetic descendants. Familial recurrence was observed with different inheritance patterns including autosomal dominant with incomplete penetrance, autosomal recessive, and X-linked.PS has been hypothesized to have a genetic origin secondary to deleterious mutations of genes regulating embryonic development, and particularly affecting pectoral girdle muscles and skeleton structures. A large deletion of chromosome 6q21-q22.1 was also recently reported in a patient with a complex phenotype mainly characterized by mental disability and PS; haploinsufficiency of some of the genes overlapped by the CNV (Copy Number Variation) (in particular WISP3 and COL10A1) may act in the pathogenesis of the musculoskeletal defects as reported in this patient [5].

In a separate case, a child with congenital hyperinsulinism (CHI) and a PS phenotype was found to have a duplication in the region between 10p13 and 10p14. The duplication involves UCMA (upper growth plate area and associated cartilage matrix), which regulates the differentiation of fetal cartilage and cartilage-bone interface. The authors suggested that this could be a contributing factor to the PS phenotype [4].

Poland syndrome is most commonly the only problem affecting the patient, but it can be found together with Dextrocardia, Moebius syndrome, Klippel-Feil, and Pierre-Robin syndromes or the underdevelopment of the kidney or the feet.

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Various variants and associated anomalies have been identified in individuals with Poland syndrome. These include absence scapula, rib hypoplasia, ipsilateral hand brachysyndactyly, and occasionally rare manifestations such as renal agenesis, pneumothorax of the affected hemithorax [4], In 75% of the cases, hypoplasia is of right hemithorax only, elevation of the shoulder blade (Sprengel deformity) and shortening of the arm with underdevelopment of the forearm bones (i.e., ulna and radius) [5] shoulder dislocation, abnormal blood counts, thrombocytopenia, growth hormone deficiency, facial nerve palsy, vertebral abnormalities, and undescended testes, extracorporeal intercostal liver herniation and thoracic myelomeningocele, megacalycosis of the ipsilateral kidney [4]. While associations with malignancies leukemia and carcinoma of the hypoplastic breast and other tumors. and other rare disorders have been identified, these events have pushed the studies toward oncologic awareness [1,4].

Multiple classifications such as the grading system known as Foucras's classification, most frequent abnormalities associated with the pectoral muscle defects, and clinical and radiological classification of thoracic musculoskeletal anomalies have been noted in studies that help to classify the severity of the Poland syndrome. Combining the classifications based on the similarities noted:

Grade 1 of Foucras's classification aligns with type 1 (minimal form) of the proposed Poland syndrome

Grade 2 of Foucras's classification corresponds to a combination of type 2 (partial forms) and type 3 (complete form) of the proposed Poland syndrome classification

Grade 3 of Foucras's classification aligns with type 3 (complete form) of the proposed Poland syndrome classification

The thoracic Musculoskeletal anomalies classification can provide additional details, particularly in the severity of the pectoral muscle and associated skeletal anomalies [1,4,5].

Prenatal sonography can detect unilateral limb defects and chest wall asymmetry, prompting further evaluation for associated anomalies. In adults, physical examination usually diagnoses Poland syndrome, but CT may be used for surgical planning or assessing cardiopulmonary issues like lung herniation. Mammograms reveal unilateral breast and pectoralis major hypoplasia. Chest X-ray shows a hyperlucent unilateral thorax. Ultrasound assesses pectoralis muscle defects [3].

Conclusion

Radiographic imaging plays a crucial role in diagnosing PS as it aids in determining the stringency of the disease as well as comprehending other abnormalities that are related to PS. The diagnosis of PS can also be made through physical examination as it shows classical presentation. The treatment of PS is varied as it is highly dependable on the extent of malformations and also the dire need of surgical and nonsurgical interventions requiring careful evaluation to determine the best course of action. Most of the cases PS require symptomatic treatment such as respiratory rehabilitation which results from chest abnormalities or the use of chest brace to prevent any physical trauma to lung due to herniation as seen in this case. In cases where there is paradoxical chest movement, breast hypoplasia or aplasia in females, or aesthetic concerns due to completed breast development, surgical intervention is indicated.

This case emphasizes the importance of diagnosing PS with the help of clinical evaluation until proven otherwise as well as patient education, routine surveillance, supportive care to avoid complications in individuals diagnosed with PS.

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References

1. Thibodeau Rachel, De Cicco Franco L. Poland Syndrome in Children - PubMed - NCBI. StatPearls Publishing, Treasure Island (FL) (2022).
2. Sharma CM, Kumar S, Meghwani MK, et al. Poland syndrome. Indian journal of human genetics 20 (2014): 82-84.
3. Tafti D, Cecava ND. Poland syndrome - PubMed - NCBI. StatPearls Publishing, Treasure Island (FL); (2023).
4. Chait F, Bahlouli N, Chehrastane R, et al.: Clinical and Radiological Features in Poland Syndrome: Report of 3 Cases and Review of Literature. Global Pediatric Health (2023).
5. Romanini MV, Calevo MG, Puliti A, et al. Poland syndrome: A proposed classification system and perspectives on diagnosis and treatment. Semin Pediatr Surg 27 (2018): 189-199.