Presentation and Treatment of Poland Anomaly

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Abstract

Background: Poland anomaly is a sporadic, phenotypically variable congenital condition usually characterized by unilateral pectoral muscle agenesis and ipsilateral hand deformity. **Methods:** A comprehensive review of the medical literature on Poland anomaly was performed using a Medline search. **Results:** Poland anomaly is a sporadic, phenotypically variable congenital condition usually characterized by unilateral, simple syndactyly with ipsilateral limb hypoplasia and pectoralis muscle agenesis. Operative management of syndactyly in Poland anomaly is determined by the severity of hand involvement and the resulting anatomical dysfunction. Syndactyly reconstruction is recommended in all but the mildest cases because most patients with Poland anomaly have notable brachydactyly, and digital separation can improve functional length. **Conclusions:** Improved understanding the etiology and presentation of Poland anomaly can improve clinician recognition and management of this rare congenital condition.

Keywords: Poland anomaly, Poland syndrome, congenital hand, pectoral agenesis, symbrachydactyly

Introduction

Poland anomaly is a sporadic, phenotypically variable congenital condition usually characterized by unilateral, simple syndactyly with ipsilateral limb hypoplasia and pectoralis muscle agenesis.^{9,23,25,46,47} Several clinical descriptions of the limb deficiency in Poland anomaly exist,^{1,19} and the hand and chest wall deformities can vary in severity, making accurate diagnosis difficult. A hand surgeon may focus on the presentation of simple syndactyly and may miss the possibly subtle associated findings: hypoplasia of the forearm, arm, ribs, and pectoralis musculature. Identifying all clinical manifestations of Poland anomaly is an important aspect of diagnosis and may ultimately lead to better treatment. This review will focus on the most common physical findings and associated surgical treatment options.

The first description of Poland anomaly is attributed to Alfred Poland of Guy's Hospital, London, based on the autopsy of a 27-year-old male convict, in which he described syndactyly of the hand with ipsilateral absence of the left sternocostal head of the pectoralis major.³³ However, the condition was not named eponymously until 1962, when Dr. Clarkson, also of Guy's Hospital, described a series of 3 cases of syndactyly with accompanying ipsilateral breast hypoplasia.⁹ Much controversy surrounds the naming of this complex condition as the first case description appears to have occurred prior to Poland's report by Froriep,¹⁸ and Poland may not have been aware of a possible association between the two conditions.^{27,36} In addition, several physicians described the association of ipsilateral

syndactyly and breast hypoplasia prior to Clarkson's naming of "Poland syndrome." Nonetheless, the association of hand deformity and ipsilateral pectoral hypoplasia has been generally referred to as Poland syndrome or Poland anomaly and serves a valuable purpose by identifying associated findings of a condition under one common name.³⁶ The non-eponymous terms to describe the condition are "pectoral aplasia-dysdactylia syndrome"³⁰ or "hand and ipsilateral thorax syndrome."¹⁷

The true incidence of Poland anomaly is unknown. The reported incidence ranges from 1 in 17000 to 1 in 100000^{17,25,28} births. The rate of pectoral hypoplasia among patients presenting with syndactyly is reported from 2.5% to 13.5%,²³ although hand deformities are not present in all presentations of Poland anomaly. Poland anomaly is reported to be more common in males, ranging from a ratio of 2:1²⁸ to as high as 5:1 and more common on the right side than on the left side (3:2).⁴³ Analysis of the gender and sidedness indicates that males are more likely to have a right-sided anomaly, whereas females have an approximately equal distribution of left- and right-sided anomalies.^{5,28}

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Although the etiology of Poland anomaly remains unknown, rare reports of familial transmission exist.^{10-12,17,28,37,42} Most cases of familial transmission present with chest wall abnormalities only, without substantial hand deformities, and do not demonstrate the gender and sidedness present in the sporadic presentation of Poland anomaly.⁵

Etiology/Embryology

Despite isolated reports of familial cases, Poland anomaly is usually a sporadic presentation. The prevailing theory or etiology is that there is some insult during the early stages of development that leads to upper extremity and chest wall deformity.²⁰

It has been suggested that disruption of embryonic development of the subclavian artery leads to Poland anomaly.⁶ At the end of the sixth week of gestation, when the upper limb bud is still adjacent to the developing chest wall, an interruption in embryonic blood supply may cause hypoplasia of the developing subclavian artery or its branches. Disruption of the developing internal thoracic artery may result in hypoplasia or aplasia of the sternocostal pectoral muscle and chest wall, whereas preferential disruption of the subclavian artery distal to the internal thoracic artery will lead to upper extremity and hand manifestations. Disruption of the subclavian artery proximal to the origin of the internal thoracic artery and distal to the origin of the vertebral artery will lead to joint presentation of hemithorax and ipsilateral hand abnormalities. This theory of subclavian artery disruption has also been proposed for Moebius syndrome and Klippel-Feil syndrome.⁶ Interestingly, this theory may account for the aforementioned observed sidedness differences in males and females as the female embryo lags slightly behind the male embryo, resulting in a delay in defined laterality, and hence an approximately equal distribution in females and a prevalence of right-sided anomalies in males.⁵

Other etiological theories include disruption of the lateral mesodermal plate soon after fertilization,⁵ and prenatal exposure to noxious social and physical environments such as smoking,²⁶ cocaine,³⁴ misoprostol,³⁸ prenatal teratogens, and failed abortions.^{12,13,14}

Clinical Presentation

Patients with Poland anomaly are usually identified at birth if the thoracic and upper extremity abnormalities are substantial. However, multiple studies have suggested that many patients are never diagnosed due to the absence of substantial functional impairment and subtle clinical abnormalities that are unnoticed by the patient or a treating clinician.^{16,39}

Poland anomaly is defined as pectoral agenesis with or without hand anomalies. The severity of the chest wall and hand anomalies may vary substantially. The chest wall deformity not only involves hypoplasia or aplasia of the sternocostal head of the pectoralis major muscle,^{9,23,33} but may

Figure 1. Chest wall deformity in Poland anomaly.

Source. Photograph A, courtesy of Adrian E. Flatt, MD.

Note. (A) A 16-month-old male with right chest wall deformity.

(B) An 18-month-old female with right chest wall deformity. Note the hypoplasia of the pectoralis musculature and rib anomalies present in A and underdevelopment of the right areola in B, highlighting the variability in clinical presentation.

also include (1) absent or hypoplastic pectoralis minor, deltoid, serratus anterior, external oblique, or latissimus dorsi; (2) bone and cartilage defects of the ribs, clavicle, and sternum; (3) Sprengel deformity; (4) scoliosis; (5) lung herniation; (6) dextrocardia; or (7) absence or underdevelopment of the breast or areola (Figure 1).^{6,9,15,16,19,23,29,33,43,45-47}

The most common hand anomaly is partial or complete symbrachydactyly (absence of middle phalanges), but abnormalities may range from simple syndactyly to complete ectrodactyly (absence of digits) in severely affected patients^{8,23,41} (Figures 2 and 3). In addition, absence or hypoplasia of carpal bones or metacarpals; absence or attenuation of flexor or extensor tendons; shortening of the humerus, radius or ulna; constriction bands; polydactyly; and nail agenesis have been described.⁴¹

Gausewitz et al¹⁹ classified the limb abnormalities in Poland anomaly, identifying four types of upper extremity anomalies that accompany pectoral agenesis. Type 1 abnormalities are characterized by presence of all 5 digits, even if hypoplastic, encompassing the “Mitten hand”²³; type 2 deformities are characterized by functional border digits with the absence of central digits, or “atypical cleft hands”; type 3 conditions are more severe and characterized by no functional digits and therefore loss of prehensile function; and type 4 anomalies are rare and characterized by radial ray deficit with absent thumb. Type 1 deformities are the most common form of limb deficit with types 2, 3, and 4 presenting in descending frequency^{17,23} (Table 1).

Al-Qattan¹ attempted to improve the classification of hand abnormalities in Poland anomaly by adding several new types of upper limb abnormalities and suggesting a broader spectrum of limb abnormalities. In this classification, types 1 and 2, with no or minimal hand involvement, suggest that isolated

Figure 2. Symbrachydactyly in 18-month-old female with Poland anomaly.

Note. (A) Dorsal and (B) volar views of simple incomplete syndactyly of a right hand's second and fourth webspaces. Note concomitant brachydactyly with complete absence of the middle phalanx in the index and small fingers, which corresponds to the presence of a single interphalangeal crease. (C) Anteroposterior hand radiograph demonstrates the absence of index and small finger middle phalanges that reflects clinical examination.

Figure 3. Symbrachydactyly in Poland anomaly.

Source. Photograph courtesy of Adrian E. Flatt, MD.

Note. (A) Dorsal and (B) volar views of left symbrachydactyly in a 5-year-old male with Poland anomaly. (C) Dorsal and (D) volar views of left symbrachydactyly in a 4-year-old male with Poland anomaly.

hypoplasia or aplasia of the pectoral muscle is a mild form of Poland anomaly, even without detectable hand deformity. Al-Qattan extended the classification to include a broader spectrum of upper limb deformities, such as phocomelia-like upper limbs.¹ The types in this classification are as follows: type 1 is a normal hand with isolated pectoral abnormality, type 2 is defined by a hypoplastic hand without substantial deformity, type 3 is defined as symbrachydactyly with 5 potentially functional digits, type 4 is defined by aplasia of one or more rays of the hand, type 5 is defined by absence of

functional digits, type 6 is characterized by a transverse deficiency proximal to the metacarpophalangeal joints, and type 7 is a phocomelia-like deficiency. Type 3 is the most common form using the Al-Qattan classification and correlates to the type 1 abnormality according to Gausewitz et al¹⁹ (Table 1).

Diagnosis

Diagnosis of Poland anomaly may be difficult if the clinical presentation is subtle. Recent radiologic studies have

Table 1. Classifications of Upper Extremity Anomalies in Poland Anomaly.

Gausewitz et al ¹⁹ classification		Al-Qattan ¹ classification		Al-Qattan ¹ treatment options
1	All digits present; may be hypoplastic, "mitten hand"	1	Normal hand (isolated pectoral abnormality)	No treatment necessary
2	Functional border digits with absent central digits	2	Hand appears smaller than the contralateral hand	No treatment necessary
3	No functional digits	3	Brachysyndactyly with all digits present	Surgery may improve function
4	Radial ray defects with absent thumb	A	Mild to moderate hypoplasia of hand	
		B	Severe hypoplasia of hand	Surgery may improve function
		4	Some functional rays still present	
		A	Radial club hand with floating or absent thumb	
		B	Adactyly of index finger	
		C	Adactyly of index and long fingers	
		D	Adactyly of central rays (cleft hand)	Salvage procedure possible
		E	Adactyly of ulnar rays	
		5	All digits are functionless or absent	Consider prosthesis
		6	Transverse deficiency proximal to the metacarpal phalangeal joints	
		7	Phocomelia-like deficiency	Consider prosthesis

identified previously undiagnosed Poland anomaly with the use of standard mammograms.^{3,32,39} In these cases, there was no identifiable hand deformity and thus these studies only identify isolated pectoral aplasia or hypoplasia. Although no long-term outcomes studies of patients with Poland anomaly exist, identifying all clinical manifestations of Poland anomaly is an important aspect of diagnosis, which may lead to better care through a better understanding of the condition. Furthermore, it is important to correctly identify this condition to avoid potential clinical mismanagement and possible litigation in cases of incidental trauma.²² Several authors have suggested the need for chest radiographs, complete blood count, urinalysis, and renal ultrasound in addition to complete physical exam at time of presentation,^{1,32} given reports of other systemic malformations co-existing with Poland anomaly.^{2,21,35,47}

Treatment

Treatment of Poland anomaly is determined by the severity of the defect and the resulting anatomical dysfunction. If a patient presents with minimal abnormalities or no functional deficit, then observation is prudent and surgical intervention is not warranted. If surgery is indicated, it is typically performed within the first 18 to 24 months of life, to prevent progression of the deformity with growth of the skeleton and soft tissue.¹⁹ Moreover, surgical intervention at an early date may allow the child to develop more useful functional patterns for daily activity.²³ Currently, there is no clear surgical indication based on the classification schemes of Gausewitz et al¹⁹ or Al-Qattan.¹

The surgical treatment of Poland anomaly is usually divided into 2 categories: surgical management of the

thoracic anomaly and surgical management of the hand anomaly. Surgical intervention for the thoracic abnormality is typically cosmetic (lack of the pectoralis major muscle and axillary fold in male patients and hypoplasia or aplasia of the breast in female patients).¹⁶ Surgical treatment options include reconstruction of the chest wall with myocutaneous flap coverage and breast augmentation procedure as needed. It is important to evaluate the condition of the latissimus dorsi prior to any reconstructive procedure, as this is the most commonly used flap, due to its regional locale.^{7,29,45} In addition, preoperative angiography and chest computed tomography (CT) scan should be considered to evaluate the blood supply of potential flaps and thoroughly understand the extent of the anomaly.^{4,7,16} Surgical reconstruction for children is often performed in 2 stages and 1 stage for adults.

Surgical intervention for the hand anomaly depends on the exact nature of the deformity and should emphasize reconstruction of a functional hand.⁴⁶ For patients with simple syndactyly, intervention should be limited to syndactyly reconstruction of the affected digits, especially if the thumb is involved, in an effort to improve pinch and grasp. Digital separation is recommended in all but the mildest incomplete syndactyly, particularly because most patients with Poland anomaly have notable brachydactyly and digital separation can improve functional length (Figures 4 and 5). In general, reconstruction of the second and third webspaces can be delayed until 18 months of age without adverse effect on hand function or fine motor development. Early surgery is recommended for border digits as syndactyly between digits of disparate length may result in flexion contracture or angular deformity. If there are functionless central digits, these may be excised in an

Figure 4. Syndactyly reconstruction in Poland anomaly.

Note. (A) Volar and (B) dorsal views demonstrating surgical markings for planned syndactyly reconstruction of second and fourth webspaces of right hand in a 18-month-old female. Three-flap web-plasty³¹ planned for fourth webspace, obviating need for skin grafting. (C) Intraoperative image demonstrating distal bifurcation of the common digital artery in the second webspace. The radial digital artery of the long finger was ligated after confirming perfusion with temporary application of a small vessel clamp. (D) Postoperative image demonstrating final flap position in the fourth webspace.

Figure 5. Two-year follow-up after syndactyly reconstruction for Poland anomaly.

Source. Photograph courtesy of Adrian E. Flatt, MD.

Note. (A) Preoperative dorsal and (B) volar views demonstrating symbrachydactyly of right hand in a 16-month-old male. (C) Postoperative dorsal and (D) volar views 2 years following third webspace reconstruction.

effort to improve the function of peripheral digits and grasp.²³ Non-vascularized phalangeal toe transfers, vascularized toe transfers, and distraction osteogenesis are all viable surgical options with good functional results and acceptable patient satisfaction.^{24,40,44} Corrective osteotomies may be used for malaligned or incongruent joints of the hand to improve function.²³ In addition, capsulotomies and tendon reconstructions may be performed for joint contractures.⁴¹

Ethical Approval

This study was approved by our institutional review board.

Statement of Human and Animal Rights

This article does not contain any studies with human or animal subjects.

Statement of Informed Consent

This article is a review article and informed consent was not necessary prior to compilation. However, informed consent was obtained from legal guardians for all patients depicted in figures.

Declaration of Conflicting Interests

The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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