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Poland's Syndrome: Current Thoughts in the Setting of a Controversy

Ashwin N. Ram, BS¹ and Kevin C. Chung, MD, MS²

¹Medical Student, The University of Michigan Medical School; Ann Arbor, MI

²Professor of Surgery, Section of Plastic Surgery, Department of Surgery, The University of Michigan Health System; Ann Arbor, MI

Abstract

Poland's syndrome is a rare congenital disorder that is characterized by hypoplasia of the pectoralis muscles and ipsilateral webbing of the fingers. The name of this condition pays homage to Dr. Alfred Poland of Guy's Hospital who in 1841 described a case of these two deformities during the autopsy of a 27-year-old convict. An exploration of the historical series reveals a clear progression of knowledge about this syndrome, accumulated by scientists across Europe and America. As such, the name "Poland's syndrome" stands as a point of contention to those who oppose the injudicious use of eponyms in medicine. An analysis of the relevant literature reveals a stepwise understanding of what has come to be known as Poland's syndrome.

Keywords

Alfred Poland; Poland's syndrome; Poland's anomaly; pectoralis hypoplasia with ipsilateral syndactyly

Current Thoughts

Poland's syndrome is characterized by an absence or underdevelopment of the pectoralis muscles in combination with webbing of the fingers in the ipsilateral hand, which can be either complete or incomplete. Other deformities can include scoliosis, hypoplasia of the rib cage and lungs, upper extremity hypoplasia, breast and nipple hypoplasia, and deficiencies of the skin, subcutaneous tissues, sweat glands, and hair of affected areas.^{1, 2, 3, 4, 5, 6, 7} Adding to the complexity of presentation is the involvement of many of the adjacent muscles including the serratus, latissimus dorsi, and the external oblique.^{1, 2, 3, 4, 5} The upper arm, forearm, and fingers also may be shortened, which is given the term brachysymphalangism.^{1, 2, 3, 4, 5}

Because of its variability in presentation, Poland's syndrome is oftentimes undiagnosed at birth. Due to the inadequacy of reporting, an accurate incidence of the disease is unknown. However, current data suggest that Poland's syndrome occurs in approximately 1 in every

30,000–100,000 live births.^{8, 9, 10, 11} Milder forms of the disease evade detection until late adolescence and puberty, when right- or left-sided differences of the body start to manifest. Irregular or asymmetric breast growth tends to be noticed more by female patients during this time and prompts them to seek treatment. In contrast, males who experience unequal chest growth typically do not seek treatment as aggressively, which contributes to the “gender predilection” of Poland’s syndrome seen in the clinics. With more severe forms of the disease, hand and arm abnormalities make the disorder more apparent early in life.

The National Human Genome Research Institute, which maintains records of specific genetic disorders including Poland’s syndrome, reported that males are affected three times as often as females with right-sided deformities twice as common as left-sided deformities.¹¹ However, many men remain undiagnosed unless they seek treatment for associated hand anomalies. This further confounds the data on the gender distribution of Poland’s syndrome. Present-day therapies have shown to be quite successful in correcting the physical abnormalities associated with Poland’s syndrome. The cosmetic concerns, if appropriately identified, can be treated at any point in life.

The etiology of Poland’s syndrome has vexed researchers for years. To date, three widely acknowledged mechanisms of pathogenesis have been described. The first focuses on genetic inheritance. Twin studies have thus far shown no evidence of a strong genetic component to the disorder, and no chromosomal abnormalities have been identified.¹² Although Poland’s syndrome is typically sporadic, familial occurrence has been observed, suggesting some degree of genetic influence. Transmission from parent to child and between distant family members has been noted.¹¹ Because Poland’s syndrome is a rare disease, the literature is mostly limited to single case studies or small sample sizes. This adds to the difficulty of determining inheritance patterns.

Researchers have explored the second cause, which may be a connection between the teratogenic effects of certain environmental factors with the development of Poland’s syndrome. One group analyzed the relationship between maternal smoking during pregnancy and Poland syndrome using national registries from Hungary and Spain. The study found a two-fold risk of developing Poland’s syndrome in children whose mothers smoked during pregnancy.¹³ This suggests there is a possible teratogenic etiology involved with the syndrome, although no follow-up work on this association has since been presented in the literature.

A more established third hypothesis focuses on vascular compromise during embryogenesis. It is thought that during the critical sixth and seventh weeks of pregnancy, a mesodermal change injures the subclavian artery, resulting in a cascade of developmental alterations in the areas supplied by the artery.^{11, 13} This event, described as Subclavian Artery Supply Disruption Sequence (SASDS), is believed to occur when the growth of the ribs drives the subclavian artery into a defective configuration.¹⁴ This, in turn, forces the artery to become hypoplastic, as noted by Dr. Alfred Poland in his original manuscript.⁷ Precipitating events for this condition can include edema of the surrounding tissue, aberrant muscle, amniotic bands, tumors, or intrinsic mechanical factors such as avascular thrombosis or embolus in the subclavian artery.¹⁴ The musculoskeletal malformations that ensue are dependent on the

degree of arterial obstruction and the location of the vessel involvement. The more proximal the occlusions occur, the more severe the nature of the resulting clinical symptoms.

The description of Poland's syndrome in this paper serves as a primer for discussion of the origin of the eponym; it is not meant to be a comprehensive overview of the disease. As we shall see, current understanding of Poland's syndrome stretches substantially beyond the initial few deformities noted over a century and a half ago. There is a rich history associated with this syndrome that centers on Poland's description in 1841. However, knowledge about this condition was collected over many decades by scientists across Europe. Even the name, "Poland's Syndrome", elicits controversy among those who feel Poland's description did not merit such recognition.

The Birth of an Eponym

One opinion has it that "the use of correct eponyms is frequently no more than a confounding exercise of one-upmanship, a practice barely to be condoned."¹⁵ This sentiment captures the essence of the debate that followed the designation, "Poland's Syndrome", to a group of congenital deformities. Uncovering the mystery behind the eponym required our extensive literature search of many of the original manuscripts that are now more than 150 years old. We distilled this information to obtain the facts that were relevant to the development of knowledge and appreciation of the disease.

In 1841, Dr. Alfred Poland of Guy's Hospital detailed an autopsy of a 27-year-old convict, George Elt. At the time of his report, Alfred Poland was a student teacher of anatomy.¹⁶ In the original document by Dr. Poland titled, "Deficiency of the Pectoral Muscles," the physical abnormalities of the autopsied man were expounded upon in great detail. Poland wrote of a man with no prior history of illness except for his inability to "draw his left arm across the chest."¹⁷ He noted that Elt's body was unusually delicate and that "the muscles of the right arm were much more developed than the left."¹⁷ According to Poland, the left external abdominal oblique muscle was alternately fleshy and tendinous, arising from the bodies of the six inferior ribs. Upon reflecting the skin, he noted that the sternal and costal portions of the pectoralis major muscle were deficient while the pectoralis minor muscle was wholly absent. He also observed that the serratus muscle was underdeveloped. In addition, he noted that the thoracic vessels looked remarkably small, and that the middle phalanges of the fingers in the left hand were all absent except in the middle finger "where a ring of bone, a quarter of an inch in length, supplied its place."¹⁷ The depiction of the cadaver in his report was created by his friend Mr. Tilston. (Figure 1).

Following this original brief description of a single anatomic dissection, this syndrome remained obscure until Clarkson, a hand surgeon also at Guy's hospital, published a study named "Poland's syndactyly." In his report, Clarkson described the case of a child referred to him by one of the senior surgeons at the hospital. After being consulted to help out with the case, Clarkson's colleague pointed out that the complex syndactyly apparent in the child "was associated with ipsilateral absence of the sternal head of the pectoralis major."¹⁶ Clarkson then discovered that an earlier Guy's Hospital physician, Alfred Poland, had reported such a case in the Guy's Hospital Reports of 1841. Upon reviewing the literature

for this anomaly, Clarkson discovered an article by Brown and McDowell¹⁸, who had compiled the most thorough review of the paired deformities up to that point. He noticed that they too referred to Poland's case report. Clarkson then asked the Curator of the Gordon Museum to see if he could find the specimen of the hand that Dr. Poland had deposited in the museum over one hundred years earlier. The specimen was easily produced¹⁶ and printed in Clarkson's article, clearly demonstrating Poland's finding of the syndactyly between the index and middle fingers.

Clarkson's article, named "Poland's syndactyly", credited Poland with discovering a disease that is now widely known and appreciated. After exploring Clarkson's work, one can come to the conclusion that his decision to name the syndrome after Poland without a thorough investigation of previous work would be considered hasty. What exactly motivated Clarkson to name the syndrome after Poland is a matter of speculation. The authors presume that Clarkson was merely acting in the name of loyalty to his gifted predecessor and storied institution. What on the surface probably struck Clarkson as an innocent way to add to the pedigree of historic Guy's Hospital, launched a heated debate that would come to question the standing of Poland's contributions to the field, and to highlight the misleading practice of classifying disease using eponyms.

Poland's Syndrome – The Controversy

Critics, including Ravitch¹⁹, McDowell¹⁵, and Clarkson et al.²⁰, have vocalized their opinions on the "crimes involved in parroting 'false' eponyms,"¹⁵ to whom there is no more blatant example than 'Poland's Syndrome'. An examination of Poland's report makes it apparent that he indeed documented a case of pectoral absence and ipsilateral syndactyly. However, his account fell short of relating the two defects. This failure was emphasized in Dr. Frank McDowell's criticism of the eponym, in which he argued Poland's omission of the forearm and hand in the autopsy sketch proved that he "hadn't the foggiest notion that the two deformities constituted a syndrome."¹⁵ Not only that, McDowell maintains, Poland considered the hand irrelevant as evidenced by the title of his report, "Deficiency of the Pectoral Muscles." Poland's report also failed to offer a sound explanation of cause and effect of this disease. Additionally, his documentation of the case made it clear that he considered his patient to be a stand-alone phenomenon, not yet able to be categorized by a common set of deformities to comprise a syndrome.

McDowell¹⁵ (1977) and Ravitch¹⁹ (1977) sought to correct the inaccuracy in the historical record by performing a comprehensive review of the literature. Their investigations identified momentous achievements in the understanding of Poland's syndrome. A timeline of important discoveries is provided. (Figure 2).

Lallemand²¹ (1826), can be credited as the first to describe an absence of the pectoralis muscle alone. Lallemand's manuscript presented a 60-year old man with a "depression capable of receiving the fist in the upper left part of the chest."²¹ It is postulated that Sir Charles Bell²² (1835) of England may have been the earliest authority to record the condition. However, Bell's review failed to reveal any reference to combined syndactyly and absence of the sternal head of the pectoralis major. A literature search also revealed an

autopsy by Froriep²³ (1839) titled, “Observation of a Case of Absence of the Breast.” In his manuscript, Froriep described a woman with amastia, absence of ribs three and four, pectoralis major and minor, intercostal muscles, and portions of the serratus muscle. This seemed to be the first identifiable record of the observation of the paired absence of the pectoralis and ipsilateral webbing of fingers. Because Froriep published his autopsy report two years before Poland, the name “Poland’s syndrome” seemed inaccurate. Clarkson’s neglect of Froriep’s work, if unintentional, underscored the fact that these physicians were limited to the information that was accessible to them at the time.

After Poland’s description of the condition in 1841, several other physicians across Europe made notable discoveries that contributed to our understanding of the disease. Jon Thomson²⁴ (1895) at the Royal Hospital for Sick Children in Edinburgh made the next noteworthy contribution in his report titled “On a Form of Congenital Thoracic Deformity.” Thomson’s report demonstrated the best, most accurate understanding of the clinical manifestations. He correctly identified the salient features of the syndrome and hypothesized that “it is a common a congenital unilateral deficiency of the structures of the front of the thorax.”²⁴

In 1900, Furst²⁵ of Sweden thoroughly described a cadaver dissection in which he proposed that this was a true syndrome, possibly for the first time in history. Not only did Furst recognize the complete defect of the pectoralis muscle with associated ipsilateral syndactyly, he also suggested that “the two groups of malformations of the chest and of the hand are so often present together that they must certainly stem from the same cause.”²⁵ Furst also suspected that “both groups of deformities are embryonal and arise from an early embryonic time.”²⁵ Furst’s observations indicate that he understood that the muscle defect and malformations of the chest must be connected to the deformities of the hand. The clarity and precision of his work merits recognition.

Nearly simultaneously, Robert Bing²⁶ (1902) listed the presence of syndactylism in 14 out of a large series of 102 patients he had collected with pectoral absence. He noted that, “In the commonest muscle defect, that of the pectorals, there is, in the first place, a clearly marked malformation of the hand, namely syndactyly with webbing, and shortening of the phalanges.”²⁶ He had found 14 cases already, including Poland’s, and was also aware of the bony defects in the thorax which “often lead to lung hernia”²⁶ in association with defects of the pectoral muscles.

In later years, physicians started to reliably associated pectoral hypoplasia with ipsilateral syndactyly, and the notion that these two deficiencies comprised one syndrome became widely acknowledged. After 1962, the literature suggested that the combination of the two hallmark deformities of “Poland’s syndrome” was accepted as such. Eventually, it would also become widely criticized for its disputed eponymous origin.

Conclusion

Poland’s syndrome is a classic example of how some inaccurate eponyms get perpetuated because of inadequate scrutiny of historical records. Even with a comprehensive literature

review, it is nearly impossible to ascertain that all the records have been exhaustively searched. For this reason, it is problematic to employ eponyms to classify medical diseases. Furthermore, the medical community would be remiss to credit one man for the achievements made by dozens of minds over many years. Nevertheless, many eponyms including “Poland’s syndrome” serve a valuable purpose by grouping together a number of related symptoms under one common name.

The challenge posed by balancing the pros and cons of using eponyms brings up a point that we cannot overlook. At the essence of the controversy lays a simple reality: medicine is a human endeavor. Honoring physicians for notable achievements in the form of eponyms can be viewed as a harmless way to bring a little bit of warmth to an otherwise cold world of facts. The least we can do, though, is to recognize the contributions of those who endeavored to shape our current understanding of disease. Perhaps if history took another course, Poland’s syndrome would instead be called Froriep’s syndrome or Furst’s syndrome. Or perhaps it might simply have been called “pectoral-aplasia–dysdactylia syndrome”^{20, 27}

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Figure 1.
Sketch of George Elt, autopsied by Dr. Alfred Poland in 1841.
Found in: Poland, A. Deficiency of the Pectoral Muscles. *Guy's Hospital Reports* 6: 191, 1841.

Table 1

Poland's Syndrome - A Timeline of Historical Achievements

1826	• Lallemand is the first to describe the absence of the pectoralis
1835	• Bell is the first to record the absence of the pectoralis
1839	• Froriep is the first to describe the paired absence of the pectoralis and ipsilateral syndactyly
1841	• Poland is the second to describe the paired absence of the pectoralis and ipsilateral syndactyly
1895	• Thomson is the first to document an understanding of the deformities accompanied one another
1900	• Furst is the first to propose that the anomalies constituted a syndrome
1902	• Bing is the first to present a case series of patients with the syndrome
1940	• Brown and McDowell are the first to document a thorough review of the syndrome
1962	• Clarkson is the first to propose the name "Poland's syndactyly" to the syndrome