American Journal of
Biomedical Science & Research

www.biomedgrid.com
ISSN: 2642-1747

Review Article

Poland Syndrome: About A Case of An Orphan Disease

Luis Ernesto Oliveros[1], Mauricio Uribe[2], Andres Tena Cardona[3], Paula Andrea Parra Sánchez[4*], Elia Karina Ortega Mejía[5], María Paz Navarro Arguello[6], María Alejandra Amin[7] and Francesca Ferla[8]

[1]*Physician Specialist in Plastic Surgery, Universidad del Bosque (Bogotá, Colombia)*

[2,3,7,8]*Physician, Universidad del Bosque (Bogotá, Colombia)*

[4]*Physician, resident of Internal Medicine, Universidad del Sinú (Cartagena, Colombia)*

[5]*Physician, Universidad del Sinú (Cartagena, Colombia)*

[6]*Physician, Universidad libre (Barranquilla, Colombia)*

***Corresponding author:** Paula Andrea Parra Sánchez, Physician, resident of Internal Medicine, Universidad del Sinú (Cartagena, Colombia).

To Cite This Article: *Luis Ernesto Oliveros, Mauricio Uribe, Andres Tena Cardona, Paula Andrea Parra Sánchez*, Elia Karina Ortega Mejía, et.al. Poland Syndrome: About A Case of An Orphan Disease. Am J Biomed Sci & Res. 2024 24(1) AJBSR.MS.ID.003166,* DOI: 10.34297/AJBSR.2024.24.003166

Received: August 13, 2024; **Published:** September 19, 2024

Abstract

Poland Syndrome is a condition characterized by the absence or underdevelopment of the pectoralis major muscle and other malformations on the affected side of the body, including abnormalities in the hand and ribs. The article describes the clinical presentation, diagnosis, and treatment options for the affected patient. Additionally, existing literature is reviewed to provide a deeper understanding of this orphan disease and its medical management.

Keywords: Poland syndrome, Pectoral agenesis, Breast hypoplasia, Clinical case, Malformations

Introduction

Poland syndrome is a congenital malformation characterized by hypoplasia or agenesis of the pectoralis major muscle, associated or not with ipsilateral brachysandactyly [1]. The syndrome was named in honor of Alfred Poland who described the absence of the pectoral muscle and ipsilateral symbrachydactyly , in a corpse in 1841 at *Guy's Hospital*.

It is a rare pathology, with an incidence of 1 per 30,000 live births, predominating in males with a ratio of 3:1, and being more frequent on the right side [2]. In Colombia it is on the List of Orphan Diseases covered by Law 1392 of 2010.

Although cases have been reported that show a familial association, its presentation is usually sporadic. The cause is still unknown. Bavinck and Weaver describe the theory of the "sequence of interruption of the supply of the subclavian artery" as a cause of this and other pathologies, [3]. Other mechanisms are also described such as infections, trauma to the mother's abdomen, exposure to teratogens, cocaine, tobacco, etc.

Its clinical manifestations can be diagnosed from birth if they are important. However, they can be almost imperceptible during infancy and early childhood, delaying their diagnosis.

Ultrasonographic scanning is the first-line imaging method to confirm the clinical suspicion of Poland syndrome. It is a highly accessible, radiation-free and cost-effective test that allows detecting asymmetries between the pectoral muscles on both sides and the anatomical arrangements with adjacent to the pectoralis major and minor. If deformities of the rib cage are evident during the clinical examination, contrast-enhanced computed tomography will be used to describe bone and vascular abnormalities [4].

According to the study carried out on a sample of 245 patients over 10 years, they proposed a classification system based on the most frequently found abnormalities in the pectoral muscle. This classification proposes 3 Poland syndrome phenotypes. Type 1 or minimal form is an isolated defect of the pectoral muscle (without rib or upper limb anomaly). Type 2 or partial forms are defects of the pectoralis muscle associated with abnormalities of the ribs or upper extremities. Type 2a or upper extremity variant are upper extremity anomalies without rib defects. Type 2b or thoracic variant is comprised of rib defects without upper limb anomalies. Type 3 or complete form includes defects of the pectoralis muscle associated with anomalies in the upper limbs and costal region.

There are other classification mechanisms that take into account all presentations of thoracic anatomy and score them with natural numbers according to their severity. The surgical techniques reported in the medical literature are diverse and offer procedures with autologous tissue or the use of synthetic materials. One of the first techniques used was the pedicled latissimus dorsi flap. In addition, it is possible to perform procedures using microsurgical techniques, such as the Deep Inferior Epigastric Artery Perforator (DIEP) flap, the Anterolateral Thigh Artery (ALT) flap or the Transverse Myocutaneous flap (TMG). Other variants are also available, such as reconstruction with breast implants. This article aims to describe the case of 2 patients with Poland syndrome and their surgical approach, with successful resolution.

Case Presentation

25-year-old patient, with a history of Poland Syndrome, who attended a plastic surgery consultation for the reconstruction of her hypoplasia in the right breast. He reports secondary psychological involvement and muscular imbalance. The physical examination reveals hypoplasia of the right breast with severe asymmetry and malposition of the nipple areola complex (dislocation of how many cms to establish classification), located in the superolateral region of the right hemithorax, near the anterior axillary fold 12.5 cm from the sternal fork. It presents excavation at the level of the anterior axillary fold with a flange to the extension of the arm. No contraction of the pectoralis major is palpable.

The left breast is normotrophic, with a distance between the fork and nipple of 18 cm and the distance from the nipple to the sulcus of 8 cm . In the extremities, hypoplasia of the right forearm was observed due to radial agenesis and absence of the wrist and ipsilateral hand. Simple chest magnetic resonance imaging is indicated to confirm the presence or hypoplasia of the latissimus dorsi muscle and define surgical technique. The simple chest MRI reports asymmetry of the anterior chest wall due to a decrease in the breast parenchyma, the thickness of the fatty tissue and the absence of the muscle fibers of the pectoralis major and minor on the right side. The right latissimus dorsi muscle has the usual morphology, arrangement and signal intensity; there are no asymmetries with respect to the contralateral muscle. Therefore, in the surgical meeting, the reconstruction of the right breast with a latissimus dorsi muscle flap (remote pedicled) is proposed in combination with an expander prosthesis and lipograft in a first surgical stage and in a second stage, symmetrization of the contralateral breast with a breast prosthesis and lipograft in affected region. The patient had an adequate postoperative evolution, without complications, obtaining a symmetrical result.

Technique Used: Augmentation Mammoplasty with Implants in Retromuscular and Retrofascial Planes

In the present case, an augmentation mammoplasty technique was implemented by inserting round implants with a nanotechnological texture, supplied by the commercial manufacturer Motiva. On the unaffected side, that is, the right side, implants were placed in the retromuscular plane . Meanwhile, on the affected side, the placement of implants in the retrofascial plane was chosen.

Justification of the Technical Choice in this Patient

The indication for this specific technique is based on the presence of breast asymmetry in the patient, attributable to agenesis of the upper pectoralis muscle and atrophy of the mammary gland. It is relevant to highlight that no deformity was evident in the upper limb. The selection of this strategy was based on the presence of adequate glandular thickness, despite atrophy. As validation of this observational hypothesis, a pinch test was carried out with a thickness greater than 3 centimeters in both the lower and upper pole. Furthermore, asymmetry was detected in the size and position of the areolas, which motivated the implementation of a lower areolar technique on the affected side and a superior areolar technique on the contralateral side.

Complications

Postoperatively, a partial dehiscence was recorded in the left peri areolar incision, which was managed by dressings. The attending surgeon used hyperbaric oxygen therapy and state-of-the-art dressings, thus avoiding the need for reoperation.

Discussion

Poland syndrome is a pathology that was described many years ago and to date has not been associated with a specific etiology. It is characterized by hypoplasia or agenesis of the pectoralis major muscle and can be associated with a fairly wide and variable spectrum of anomalies; Defects have been described that include agenesis or hypoplasia of other muscles (such as the serratus anterior, supraspinatus, pectoralis minor), anatomical alterations in the breast, to cardiac, respiratory, and renal defects, among others [5]. Currently, there is no clear consensus regarding the epidemiological magnitude of the pathology. Well, some research suggests an incidence of 1 per 30,000 live births [6,7] while others report an incidence of 1 per 100,000. With respect to Colombia, for the year 2019, 4 cases of Poland Syndrome and 1 of Poland Anomaly were reported according to data from the National Institute of Health. No reports were found after this date [8].

There are various classifications in the literature to establish the degree of severity of the thoracic malformations of Poland Syndrome. However, the TBN classification by Romanini and colleagues is the only one that includes most of the possible phenotypes [9,10]. The correction of thoracic volume through the insertion of breast

implants is positioned as one of the preeminent techniques. One of its most notable attributes lies in obtaining pleasant aesthetic results. It should be noted that among the potential complications associated with this surgical technique, capsular contracture stands out in the long term .

In this context, it is pertinent to highlight that, in the previously outlined case, the indication for the use of breast implants is based. This approach finds justification in the patient's youth and in her categorization of mild severity according to Poland syndrome .

Conclusion

Poland syndrome, although described many years ago, remains a pathology of uncertain etiology, characterized by a wide variability in its clinical manifestations. The lack of consensus on its incidence and the scarcity of epidemiological data, especially in countries like Colombia, highlight the need for more detailed investigations and a more exhaustive registry of cases. Surgical correction of the syndrome, particularly through the insertion of breast implants, has proven to be an effective option to improve the aesthetic appearance and quality of life of patients, although it is not free of complications such as capsular contracture. The choice of treatment must be individualized, considering factors such as the severity of the syndrome and the characteristics of the patient, which underlines the importance of precise classifications such as that of *Romanini, et al.* [1]. to guide appropriate therapeutic intervention. This comprehensive approach is crucial to optimize outcomes in the management of Poland syndrome.

Acknowledgement

None.

Conflict of Interest

None.

References

1. Romanini MV, Torre M, Santi P, Dova L, Valle M, et al. (2016) Proposal of the TBN Classification of Thoracic Anomalies and Treatment Algorithm for Poland Syndrome. Plast Reconstr Surg 138(1): 50-58.
2. Bavinck JNB, Weaver DD (1986) Subclavian artery supply disruption sequence : Hypothesis of a vascular etiology for Poland, Klippel-Feil , and Mobius anomalies. Am J Med Genet 23(4): 903-918.
3. De Stefano MT, Divizia F, Feletti M, Formica M, Lando M, et al. (2020) Consensus-based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet J Rare Dis 15(1): 201.
4. Hashim EAA, Quek BH, Chandran S (2021) A narrative review of Poland's syndrome : theories of its genesis , evolution and its diagnosis and treatment . Transl Pediatrician 10(4): 1008-1019.
5. Al Qattan MM (2001) Classification of hands anomalies in Poland's syndrome . Br J Plast Surg 54(2): 132-136.
6. Gozna ER, Stokes I (1979) Poland syndrome : clinical and pathomechanical considerations. Clin Orthop Relat Res (144): 25-33.
7. David TJ, Winter RM (1988) Poland's syndrome and the subclavian artery supply disruption sequence. J Med Genet 25(1): 42-48.
8. Fokin AA, Robicsek F (2002) Poland's syndrome revisited. Ann Thorac Surg 74(6): 2218-2225.
9. Martínez Frías ML, Bermejo E, Rodríguez Pinilla E, Frías JL (2016) Poland sequence : Further evidence for Poland sequence as a vascular disruption defect. Am J Med Genet A 170(6): 1340-1346.
10. Al Basti HA, El Khatib HA (2019) Poland syndrome : An update on the classification and management. Can J Plast Surg 27(1): 23-26.