Poland syndrome. A case report

Amador Jimenez Leyva M.D.
Elizabeth Del Carmen Rodríguez Rojas M.D.
Manuel Alfredo Calderón Vizcaíno M.D.
Alan De la fuente Malvaez M.D.
Ángel Alejandro Hernández Moreno M.D.
Ana Lourdes Pérez Vázquez M.D.
Jorge Emmanuel Maldonado Perdomo M.D.
Raúl Beder Caltenco Solis M.D.

Background:
INTRODUCTION: Poland syndrome was first described by Alfred Poland in 1841. It is a congenital disorder characterized by the total or partial absence of the pectoralis major muscle and anomalies in the ipsilateral hand.
DISCUSSION: The etiopathogenesis seems to correspond to an alteration in the mesoderm, which develops from the pectoral area to the digital extremity of the upper limb. It is caused by a primary defect in the development of the proximal subclavian artery.
CONCLUSION: Surgical timing will help improve physical and mental development issues. Surgical planning for Poland syndrome patients should begin during the growth period.

Keywords: Poland syndrome, Absence of pectoral muscle, Thoracic malformation, Breast implant, Mastopexy.

Mexico City, Mexico

Case Report
Plastic Surgery
OPEN ACCESS

Poland syndrome, also known as Poland sequence or anomaly, was first described by Alfred Poland in 1841 (1). It is a congenital disorder characterized by the total or partial absence of the pectoralis major muscle and anomalies of the ipsilateral hand. It can be associated with other pectoral, cervical, intrathoracic, and even brachial malformations (2). Other characteristics associated with this syndrome include pathological elevation of the scapula, shortening of the arm due to underdevelopment of the forearm bones, dextrocardia, diaphragmatic hernia, and renal abnormalities (3). The incidence is reported to be 1 in every 7,000–10,000 live births, with a male-to-female ratio of 3:1 (4). There is no defined inheritance pattern. Although the pathogenesis is not fully described, it is attributed to an interruption of blood flow in the subclavian artery during the sixth week of gestation (5). While syndactyly appears to be the most characteristic anomaly of the hand, as more cases have emerged, other malformations have been documented, and cases without syndactyly have even been described (6). Although extensive classifications of all types of arm anomalies in Poland syndrome have been published and are internationally recognized as a reference tool for experts in the field, thoracic anomalies still lack a complete classification. Several past classifications have attempted to establish a severity grade for thoracic malformations in Poland syndrome (7). The extent of breast and chest wall deformities varies widely in Poland syndrome due to the variable degree of association between atrophy and not only the muscular plane but also the thoracic bony and cutaneous planes (including the breast in women) (8). In 2003, Foucras et al. published a series of 10 patients with mixed results because the implants were prepared using plaster molds (9). In 2015, Majdak-Paredes et al. found that custom-made silicone implants were very useful for moderate deformities (10).

Case report

To report the pathological clinical case of a female patient with a history of Poland syndrome and her follow-up at 16 years old for breast reconstruction with a silicone implant and contralateral mastopexy. Hypotrophy of the left pectoralis major muscle was observed without arterial alterations.

The patient was reevaluated at age 15 (see Figure 1), at which time a two-stage management with tissue expansion and subsequent placement of a silicone implant was chosen (see Figure 2). During the clinical evaluation, measurements of the breast base, the diameter of the areola-nipple complex, and various

From the Plastic and Reconstructive Surgery Service, Lic. Adolfo Lopez Mateos Regional Hospital, Institute of Security and Social Services of State Workers, Mexico City, México. Received on October 8, 2024. Accepted on October 14, 2024. Published on October 17, 2024.

DOI 10.5281/zenodo.13948870

Figure 1. Preoperative frontal and lateral view.

anatomical distances, including the distance between the parasternal region and the areola-nipple complex, the midline sternum and the areola-nipple complex, as well as the inframammary fold and the areola-nipple complex, were taken (see Figure 3). It was observed that the patient did not present any deformities in the thoracic extremity or hands. A silicone implant was then placed to restore the patient's aesthetics.

Before the second surgical procedure, due to the patient's symptoms, the decision was made to perform a mastopexy on the right side along with the placement of a silicone implant (see Figure 4).

A follow-up was conducted 28 days after surgery (see Figure 5), during which the patient expressed satisfaction with the results (see Figure 6).

Discussion

Poland syndrome is a very rare congenital musculoskeletal disorder characterized by unilateral hypoplasia of the chest wall and abnormalities of the ipsilateral upper limb. In 1841, Alfred Poland described the unilateral absence of the pectoralis major, serratus anterior, and external abdominal oblique muscles associated with ipsilateral syndactyly (11).

There may also be an absence of axillary hair and apocrine sweat glands, axillary webbing, hemivertebrae, Sprengel deformity or scapular elevation secondary to the lack of the pectoralis minor, scoliosis, dextrocardia, pulmonary hypoplasia, pectoral hypotrichosis, and cartilaginous defects of the

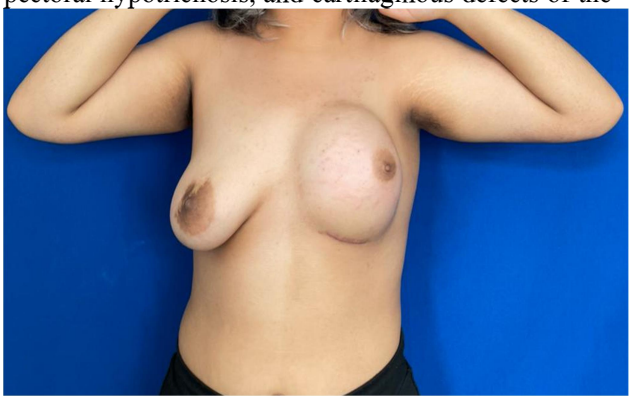

Figure 3. Postoperative, frontal view (with tissular expander)

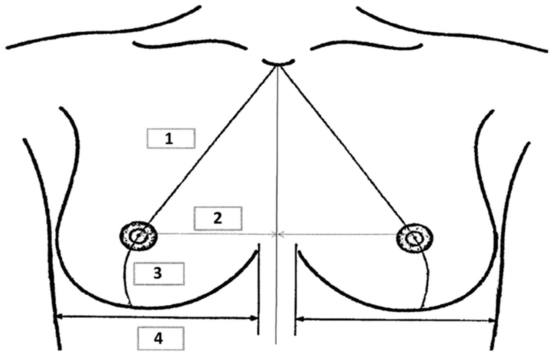

Figure 3. Anthropometric measurements.

second to fifth ribs. The hands typically show various degrees of syndactyly, brachydactyly, or even oligodactyly on the ipsilateral side, with the thumb usually being unaffected. From the literature described, the patient only presented with ipsilateral pectoral muscle hypotrophy, without any hand deformity or other abnormalities.

Numerous authors agree that the etiopathogenesis seems to correspond to an alteration in the mesoderm, the embryonic tissue from which the pectoral area to the digital extremity of the upper limb develops, due to a primary defect in the development of the proximal clavicular artery, resulting in partial tissue loss around the sixth week of gestation (12). However, cases have been described where the related arteries were normal, raising doubts about the true pathogenesis, as was corroborated in this patient (13).

When Poland syndrome occurs on the left side of the chest, some authors believe it is secondary to the thoracic deformity caused by rib malformations and their effect on heart formation during the embryonic period (14).

Our case is of particular interest due to the unusual manifestations of the syndrome. In the clinical examination, the agenesis of the mammary gland, but

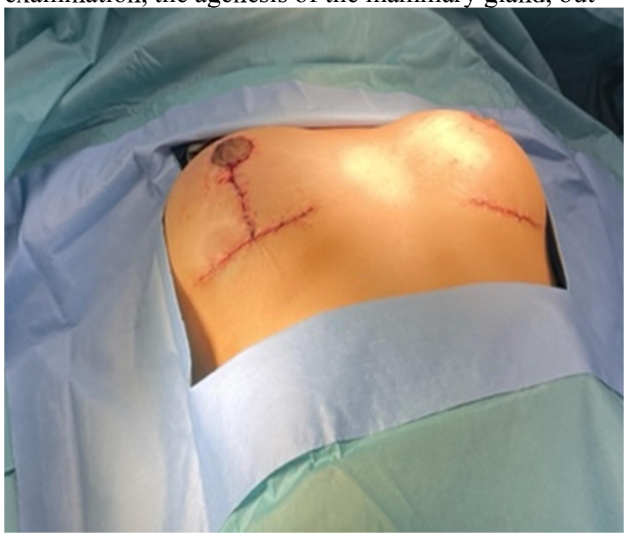

Figure 4. Immediate postoperative.

DOI 10.5281/zenodo.13948870

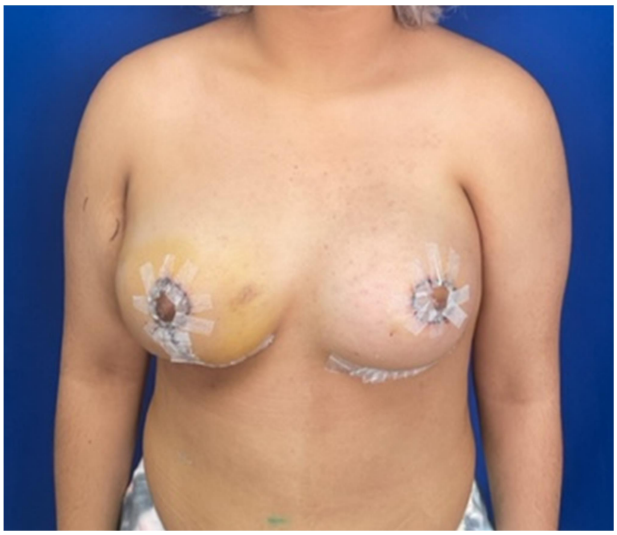

Figure 5. 28 days follow up.

with the presence of the areola-nipple complex, appeared to be the only manifestation of the Poland anomaly.

In 1989, Fraser et al. suggested that although the isolated defect of the pectoralis major is rare in Poland syndrome, it should be included among the characteristic anomalies (15). Later studies revealed that hand malformations associated with Poland syndrome are less frequent than initially assumed, occurring in only about 12% of cases. Cases like ours, where only the pectoralis muscle is affected, have been described (16).

Regarding treatment, fat transfer techniques are useful for minor deformities, significantly improving patients' conditions, but in most cases, the limits of this treatment are quickly exceeded (17). Many procedures have been proposed to correct deformities. For some, the latissimus dorsi flap remains the gold standard (18), sometimes used in women with breast prostheses. Additionally, tissue expansion, transverse rectus abdominis myocutaneous flap (19), omentum flap, thoracic remodeling surgery, or a combination of these techniques are used (20). Achieving perfect symmetry and an aesthetic outcome is extremely difficult for patients with Poland syndrome. This should be explained to the patient and family from the outset, taking into account the expectations and the reality of the results.

The technical and surgical requirements for reconstruction differ between men and women. Reconstruction with silicone implants allows for satisfactory results in females (21).

A recent study by Baldelli et al. emphasizes that patients should undergo surgery during the growth period to allow proper stabilization of body image and improved quality of life (22). When surgery is delayed until adulthood, there is no association between corrective surgery and the reduction of clinically significant body dysmorphia. This is likely due to the

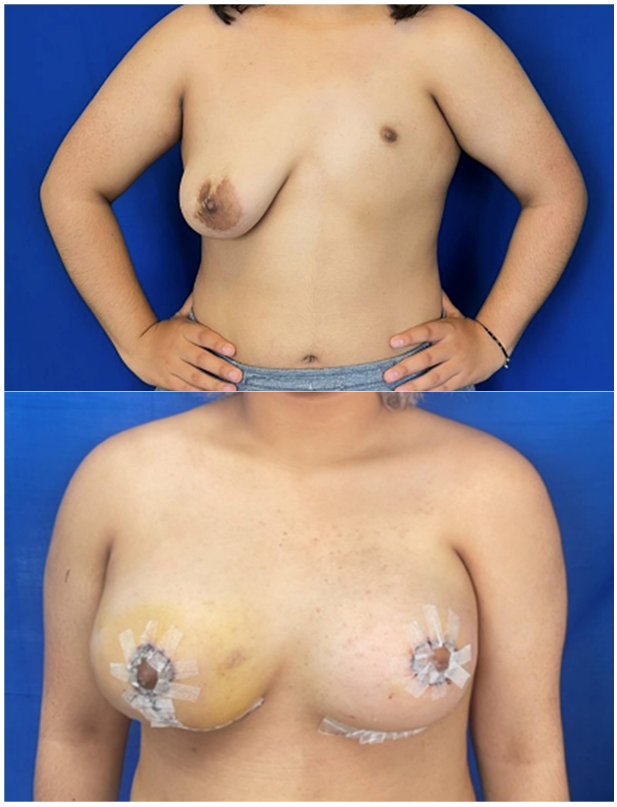

Figure 6. Preoperative and follow-up comparison.

fact that body image perception reaches a critical phase during adolescence, which consequently improves quality of life (23).

Conclusion

Soft tissue tumors, in this case masseter muscle sarcoma, is a rare and infrequent entity, with little potential for malignant transformation.

However, the reconstruction of large soft tissue defects in the maxillo-malar region is a complex task, which merits early and meticulous planning for an adequate resection with free surgical margins, as well as a total coverage of the lesion, with expected functional and esthetic results.

Conflicts of interests

There was no conflict of interest during the study, and it was not funded by any organization.

References

1. Poland A. Deficiency of the pectoral muscles. Guy Hosp Rep. 1841;6:191-3.
2. Cruz M, Bosch J. Hipoplasia. Agenesia del músculo pectoral con sindactilia (Poland). En: Cruz M, Bosch J, editores. Atlas de síndromes pediátricos. Barcelona: Espaxs; 1998.

 DOI 10.5281/zenodo.13948870

3. Lacorte M, Marsella P, Guerrini P. A case of Poland syndrome associated with dextroposition. Ital J Pediatr. 2010;36:21.
4. Fraser FC, Teebi AS, Walsh S, Pinky L. Poland sequence with dextrocardia: Which comes first? Am J Med Genet. 1997;73:194-6.
5. Bavinck JN, Weaver DD. Subclavian artery supply disruption sequence: hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. Am J Med Genet. 1986;23(4):903-18.
6. Eugber WD. Cleft hand and pectoral aplasia. J Hand Surg. 1981;6:574-7.
7. Catena N, Divizia MT, Calevo MG, et al. Hand and upper limb anomalies in Poland syndrome: A new proposal of classification. J Pediatr Orthop. 2012;32:727-31.
8. Foucras L, Grolleau-Raoux JL, Chavoin JP. Poland's syndrome: Clinic series and thoraco-mammary reconstruction. Report of 27 cases. Ann Chir Plast Esthet. 2003;48:54-66.
9. Majdak-Paredes EJ, Shafighi M, Fatah F. Integrated algorithm for reconstruction of complex forms of Poland syndrome: 20-year outcomes. J Plast Reconstr Aesthet Surg. 2015;68:1386-94.
10. Majdak-Paredes EJ, Shafighi M, Fatah F. Integrated algorithm for reconstruction of complex forms of Poland syndrome: 20-year outcomes. J Plast Reconstr Aesthet Surg. 2015;68:1386-94.
11. Poland A. Deficiency of the pectoral muscles. Guy Hosp Rep. 1841;6:191-3.
12. Poullin E, Toussirot A, Schiano G, Serratrice G. Complete and dissociated forms of Poland's syndrome (5 cases). Rev Rhum Mal Osteoartic. 1992;59:114-20.
13. Ferraro GA, Perrotta A, Rossaun F, D'Andrea F. Poland syndrome: Description of an atypical variant. Aesthetic Plast Surg. 2005;29:32-3.
14. Galvagno G, Marra A, Ghiotti MP, Cattaneo G. Poland's syndrome. Presentation of a case of probable vascular origin. Pediatr Med Chir. 1988;10:119-21.
15. Fraser FC, Ronem GM, O'Leary E. Pectoralis major defect and Poland sequence in second cousins: Extension of the Poland sequence spectrum. Am J Med Genet. 1989;33:468-70.
16. Minguella-Solá M, Cabrera-González M. Síndrome de Poland. Revisión de 38 casos. An Esp Pediatr. 1998;48:143-7.
17. Pinsolle V, Chichery A, Grolleau JL, Chavoin JP. Autologous fat injection in Poland's syndrome. J Plast Reconstr Aesthet Surg. 2008;61:784-91.
18. Watfa W, di Summa PG, Raffoul W. Bipolar latissimus dorsi transfer through a single incision: First key-step in Poland syndrome chest deformity. Plast Reconstr Surg Glob Open. 2016;4:e847.
19. Tvrdek M, Kletenský J, Svoboda S. Aplasia of the breast: Reconstruction using a free TRAM flap. Acta Chir Plast. 2001;43:39-41.
20. Dos Santos Costa S, Blotta RM, Mariano MB, Meurer L, Edelweiss MI. Aesthetic improvements in Poland's syndrome treatment with omentum flap. Aesthetic Plast Surg. 2010;34:634-9.
21. Chavoin JP, Taizou M. Correcting Poland Syndrome with a Custom-Made Silicone Implant: Contribution of Three-Dimensional Computer-Aided Design Reconstruction. Plast Reconstr Surg. 2021.
22. Baldelli I, Santi P, Dova L, et al. Body image disorders and surgical timing in patients affected by Poland syndrome: Data analysis of 58 case studies. Plast Reconstr Surg. 2016;137:1273-82.
23. Urschel HC. Poland's syndrome. Chest Surg Clin North Am. 2000;10:393-403.

Manuel Alfredo Calderón Vizcaíno
Plastic and Reconstructive Surgery Service
Lic. Adolfo Lopez Mateos Regional Hospital
Institute of Security and Social Services of State Workers
Mexico City, México.

 DOI 10.5281/zenodo.13948870