

Poland's Syndrome: Different Clinical Presentations and Surgical Reconstructions in 18 Cases

Renato da Silva Freitas, André Ricardo Dall'Oglio Tolazzi, Vanessa Dello Mônico Martins, Breno Albuquerque Knop, Ruth Maria Graf, and Gilvani Azor de Oliveira e Cruz

Plastic and Reconstructive Surgery Unit, Hospital de Clínicas of Federal University of Paraná, R. General Carneiro, 181, 9° andar, Curitiba-PR, Brazil 80060-900

Abstract.

Background: The literature reports many variations of Poland's syndrome. This article describes 18 cases of Poland's syndrome in different stages of treatment, with variable clinical presentations and reconstructive techniques.

Methods: This study evaluated 15 females and 3 males, ages 2 to 43 years, for breast deformity, nipple–areolar complex position, pectoralis muscle malformation, thoracic deformities, and the presence of brachysyndactyly. Surgical treatment was performed for 14 patients, individualized for each case.

Results: For the women, the hypoplastic breast was treated with a latissimus dorsi muscular flap associated with silicone gel implant in five cases. Two other patients still are receiving tissue expansion for a future muscular and prosthetic reconstruction. Prosthetic implants alone were used on the affected side in four cases. The nipple–areolar complex was reconstructed for two patients. Seven women underwent contralateral breast surgery: reduction mammoplasty in three cases, mastopexy in two cases, and prosthetic implants in two cases. The only man who underwent surgery was treated with endoscopic rotation of the latissimus dorsi muscle flap.

Conclusions: This study demonstrated several breast reconstruction options for patients with Poland's syndrome, reinforcing the importance of an individualized treatment to achieve complete and adequate rehabilitation.

Key words: Breast hypoplasia—Breast reconstruction—Poland's syndrome

After Alfred Poland first published his report on the congenital thoracic and upper extremity anomaly known as Poland's syndrome in 1841 [17], many variations of the condition have been reported in the literature. In the majority of cases, Poland's syndrome is unilateral, usually characterized by breast and nipple hypoplasia, paucity of subcutaneous tissue, absence of the costosternal portion of the pectoralis major muscle, lack of a pectoralis minor muscle, deformity or aplasia of the costal cartilages or ribs II to IV or III to V, alopecia of the axillary and mammary region, and unilateral brachysyndactyly [18]. Patients have presented with many of these congenital malformations, manifested with variable severity. No relation between breast, thoracic, and upper extremity manifestations has been reported [7,19,21,22].

We describe our experience with Poland's syndrome since 1998. Our 18 cases in different stages of treatment demonstrate variable clinical presentations and reconstructive techniques.

Patients and Methods

Since 1998, we have been treating 18 patients with a diagnosis of Poland's syndrome at the Hospital de Clínicas of the Federal University of Paraná. The patients have come to us either because of their aesthetic deformities, usually noted during their teenage years, or as referrals by other colleagues, especially gynecologists. Our 18 patients comprise 15 females and 3 males ranging in age from 2 to 43 years. Every case has been evaluated for breast deformity (hypomastia or amastia), nipple–areolar complex

Table 1. Patients and clinical presentations

Patient	Gender	Age (years)	Side	Pectoralis muscle	Syndactyly	Breast	NAC	Thoracic deformity
1	F	14		Clavicular head	Synovial cyst (4th)	Hypomastia	Superior	No
2	F	23		Complete absence	No	Amastia	Superior	Pectus carinatus
3	F	36		Complete absence	No	Amastia	Superior	No
4	F	43		Complete absence	No	Amastia	Superior	No
5	F	2		Complete absence	Yes (2nd–3rd)	Hypomastia	Superior	No
6	F	25		Complete absence	No	Amastia	Superior	No
7	F	29		Complete absence	No	Hypomastia	Superior	No
8	F			Complete absence	No	Amastia	Superior	No
9	F			Hypoplasia	No	Hypomastia	Superior	No
10	F	15		Hypoplasia	No	Hypomastia	Smaller orthotopic	No
11	F	14		Complete absence	No	Amastia	Superior	Pectus carinatus
12	F	18		Hypoplasia	No	Hypomastia	Smaller orthotopic	No
13	F	22		Hypoplasia	No	Hypomastia	Smaller orthotopic	No
14	F	17		Hypoplasia	No	Hypomastia	Superior	No
15	M			Complete absence	No	X	Superior	No
16	M	18		Complete absence	Yes (3rd–4 th)	X	Superior	No
17	F	03		Clavicular head	No	Hypomastia	Superior	Thoracic asymmetry
18	M			Complete absence	Yes (3rd–4 th)	X	Superior	No

NAC, nipple–areolar complex; X, male breast hypotrophy.

position (orthotopic or superiorly sited), pectoralis muscle malformation (presence of the clavicular head and absence of the esternocostal head, whole muscular hypoplasia, or complete absence of the major pectoralis muscle), clinically evident thoracic deformity, and the presence of ipsilateral brachysyndactyly.

The surgical techniques were individualized for each patient, and if a wide spectrum of congenital malformations and clinical presentations was present, multiple surgical procedures were performed. Because our series included only 18 variable cases, no statistical analysis was performed.

Results

Among the 15 women treated, breast hypoplasia was found in 9 and amastia in 6 of the women. The nipple–areolar complex on the affected side was always smaller than the normal one. It was superiorly sited in 12 women, orthotopic in 3 women, and superior in all male cases. With respect to female muscular malformations, clinical and intraoperative evidence indicated complete absence of the pectoralis muscle in nine cases, absence of the sternocostal head in two cases, and complete pectoralis hypoplasia in five patients. The pectoralis muscle was absent in all the male cases. Ipsilateral syndactyly involving the second and third fingers was found in one woman. In

another woman, a synovial cyst was resected from the fourth finger of the left hand. Two male patients had brachysyndactyly, both affecting the third and fourth fingers. Regarding clinically evident thoracic deformity, six patients presented with pectus carinatum, including one man (Table 1).

Currently, the patients are found in different stages of treatment, from the preoperative stage awaiting complete breast development during puberty to the completed surgical reconstruction stage in adults. Surgical treatment was performed for 14 patients (13 women and 1 man), individualized for each case (Table 2). For the women, hypoplastic breasts were treated with latissimus dorsi muscle flaps associated with silicone gel implants in five cases. Two other patients still are undergoing tissue expansion for a future muscular and prosthetic reconstruction. Prosthetic implants alone were used on the affected side in four cases. The nipple–areolar complex was reconstructed for two patients. The contralateral breast was managed surgically for seven women, with a reduction mammoplasty in three cases, a mastopexy in two cases, and prosthetic implants in two other cases. The only man who underwent surgery was treated with an endoscopic latissimus dorsi muscle flap. Four clinical cases are illustrated in Figs. 1 to 4.

On the basis of our experience, a flow chart was developed for systematic diagnosis and surgical

Table 2. Surgical treatments for affected and unaffected breast and nipple–areolar complex (NAC) reconstruction

Patient	Surgical treatment for the affected side	Surgical treatment for the unaffected side	NAC reconstruction
1	Silicone implant	Mastopexia	No
2	Tissue expansion (not completed)	(not completed)	(not completed)
3	Tissue expansion → LDMF + silicone Implant	Mastopexia	Yes
4	No	No	No
5	No	No	No
6	LDMF + silicone implant	No	No
7	LDMF + silicone implant	No	No
8	Tissue expansion → LDMF + silicone implant	Silicone implant	Yes
9	Silicone implant	No	No
10	No	Reduction mammoplasty	No
11	Tissue expansion (not completed)	(not completed)	(not completed)
12	Silicone implant	Reduction mammoplasty	No
13	No	Reduction mammoplasty	No
14	Silicone implant	Silicone implant	No
15	Endoscopic LDMF	No	No
16	No	No	No
17	No	No	No
18	No	No	No

LDMF, latissimus dorsi muscle flap.

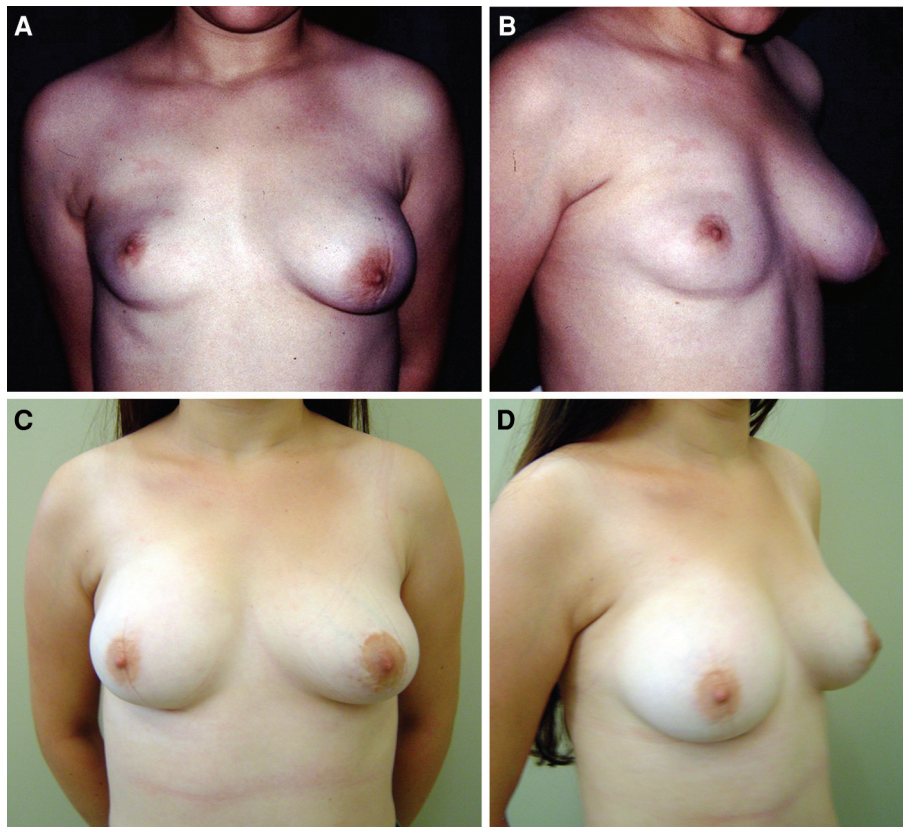


Fig. 1. Patient with moderate right breast and pectoralis muscle hypoplasia (A and B). Postoperative view after right breast silicone implantation (200 ml) and left periareolar mastopexy (C and D).

treatment. The four principal elements evaluated were the affected breast, the contralateral normal breast, the size and position of the nipple–areolar complex, and thoracic deformities (Figs. 5, 6, and 7).

Discussion

Poland's syndrome is a nongenetic congenital disorder with a low risk of recurrence in the same family

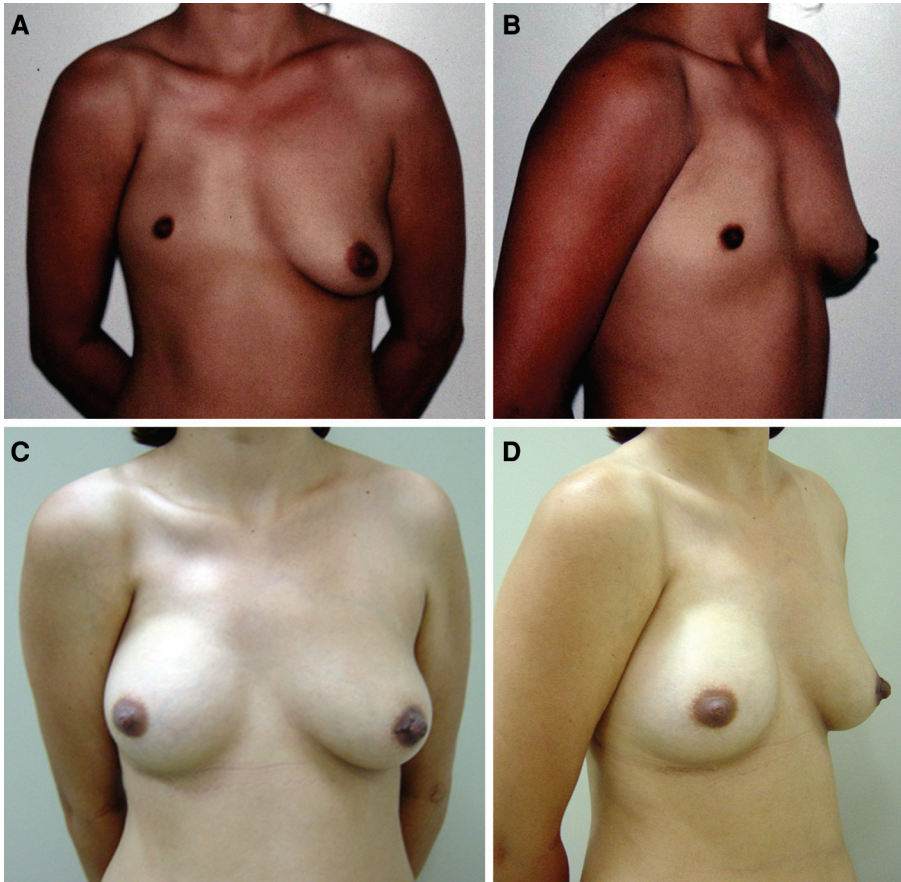


Fig. 2. Patient with severe hypoplasia of the breast and pectoralis muscle on the right side, associated with mild hypomastia on the contralateral side (A and B). Postoperative view after silicone implantation covered by a latissimus dorsi muscle flap (C and D).

(<1%) [10]. Also known as unilateral chest–hand deformity, it is found in 1:7,000 to 1:100,000 live births. It most commonly affects males (2:1 to 3:1) and has a right side prevalence (60–75%) [1,8,14,15].

In our series, we have been treating 15 female and 3 male cases. The preponderance of female cases could be explained by the major aesthetic deformity found in the female breast, as well as by the stigma and importance given to this fact by women in our community. The deformities in men usually present as a soft thoracic depression and a flattened or absent anterior axillary fold. These distributions have varied once the case is sporadic or familial. In sporadic cases, males are affected more frequently, especially on the right side. In female sporadic cases, however, the side distribution difference is not found in familial cases. In these cases, the gender and side predominance is almost the same [1,3,4].

Theories have been proposed to explain the etiology of the congenital malformations. The most accepted theory refers to a hypoplasia of the subclavian artery and its branches. The site and degree of flow velocity impairment determines the extent and severity of the developmental changes [4,5,15]. Other factors could contribute to an increase in its incidence. Autosomal dominant traits, single gene defects, intrauterine trauma, viral infections,

attempted abortion, teratogenic drugs, and smoking all have been reported as factors increasing the risk for Poland's syndrome [6,8,13,18].

Poland's syndrome has a great spectrum of clinical presentations, which correlates with the degree of functional impairment and cosmetic deformities. Mild forms are reported in 1:16,500 live births [1]. Despite the large variance in this syndrome, the diagnostic criteria include isolated absence of the pectoralis major muscle and breast hypoplasia [4]. We had a large variance in clinical presentations from mild hypomastia with pectoral hypoplasia to complete absence of the pectoralis muscle and amastia.

The muscle malformations involve, in almost every case, absence of the sternocostal head of the pectoralis major muscle and, in most cases, absence of the pectoralis minor muscle. Other muscle units that also can be involved at a lower frequency are the latissimus dorsi, the external oblique, and the serratus anterior muscles [2,11,12,16].

The breast involvement varies greatly. Hypomastia and even amastia are the presentations most frequently found (in more than 30% of female patients). The nipple–areolar complex usually is involved, and it can be superiorly sited, hypoplastic, or even absent. Correction of this nipple–areolar complex dystopia is one of the most difficult reconstructive stages. To

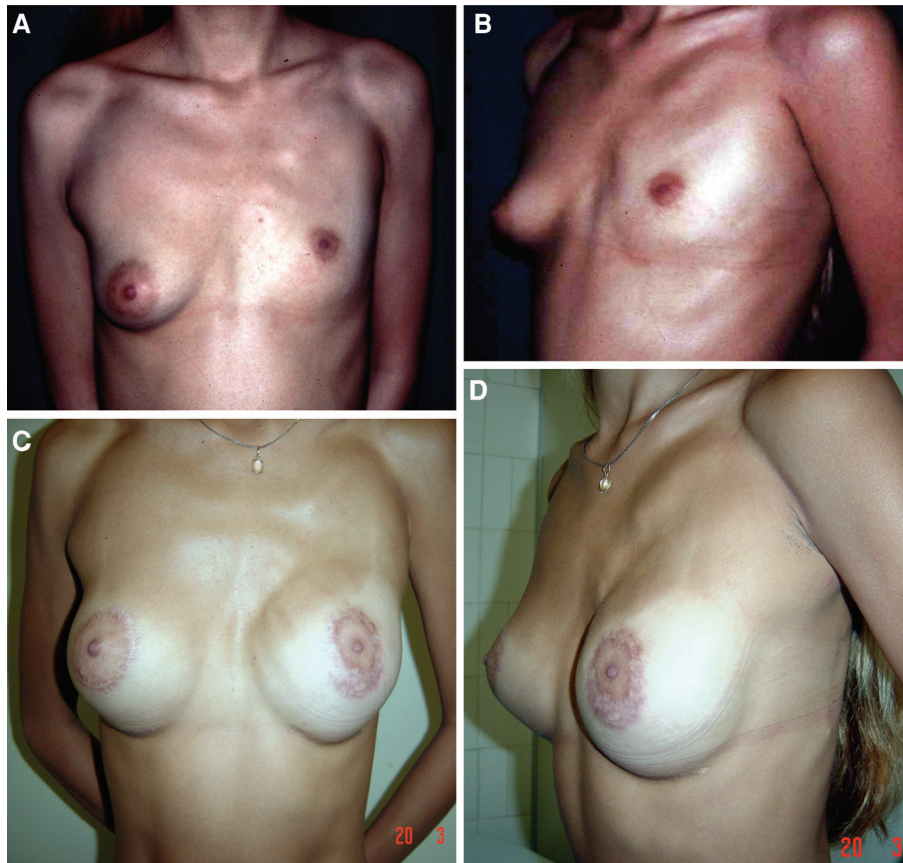


Fig. 3. Patient with severe hypoplasia of the breast and pectoralis muscle on the right side, associated with mild hypomastia and ptosis on the contralateral side (A and B). Postoperative view after bilateral silicone breast implantation, rotation of the left latissimus dorsi muscle flap, and repositioning of the left nipple–areolar complex (C and D).

achieve symmetry, we have made a bilateral periareolar incision, with skin resection on the top of the normal nipple–areolar complex and on the bottom of the affected complex. After a round-block suture, both complex positions are compensated to an intermediate site, giving a more symmetric appearance. Moreover, for some patients with a severe hypoplastic nipple–areolar complex, we have used the whole complex to reconstruct the nipple, with a peripheral tattoo performed to achieve areola symmetry.

A rib cage depression (the most common thoracic deformity), scoliosis, aplasia of some portion of ribs (usually II to IV or III to V), a contralateral pectus carinatum, or even a normal rib cage all may be found among patients with Poland's syndrome. Several surgical techniques are used to reconstruct the large spectrum of Poland's malformations. Moreover, age and sex are the other important factors in determining the most appropriate treatment. Despite the great importance of costal cage reconstruction, improvement of the respiratory function, and support to tissues and prostheses used for mammary reconstruction, we do not discuss it in detail because we have not had any cases with this severe thoracic deformity requiring this type of treatment.

Regarding mammary reconstruction, patients with mild pectoral and breast hypoplasia can be treated

simply with a silicone implant, as performed for four patients in our series. As with the 14th case in Table 1, when contralateral breast augmentation also is desired, a smaller prosthesis can be inserted in the unaffected side to achieve symmetry. The majority of patients, however, present with severe breast hypoplasia or even amastia. The condition includes not only muscular paucity, but also a scarcity of subcutaneous tissue. A silicone implant alone would result in an artificial appearance to the affected breast. In these cases, a muscular or musculocutaneous flap is necessary to cover the prosthesis [16]. A latissimus dorsi muscle flap is a good option and has been our first choice. Its large size and flattened shape covers almost the entire implant, and its cranially fixed portion outlines the anterior axillary fold. We prefer to use the latissimus dorsi flap in the second step to avoid the hypotrophy caused by tissue expansion.

Some surgeons rotate a musculocutaneous flap from the dorsum. The island of skin transferred together with the latissimus dorsi muscle allows a one-stage mammary reconstruction. Even when a two-stage reconstruction is necessary, we prefer skin expansion before the muscular flap rotation because it provides a better cosmetic result without extra breast scars and different skin texture and color [9,21,22]. Other feasible options, especially when the ipsilateral latissimus dorsi muscle also is atrophied,

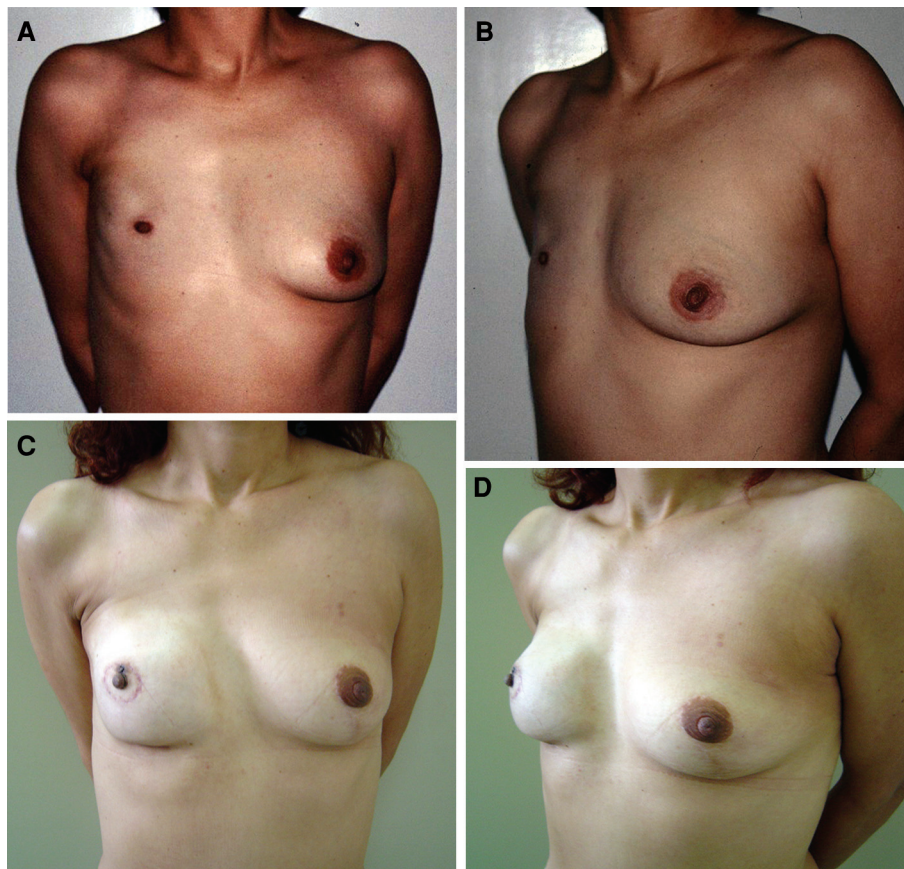


Fig. 4. Patient with severe hypoplasia of the breast and pectoralis muscle on the right side, associated with mild hypomastia on the contralateral side (A and B). Postoperative view after right side silicone implantation covered by a latissimus dorsi muscle flap, and nipple reconstruction (C and D).

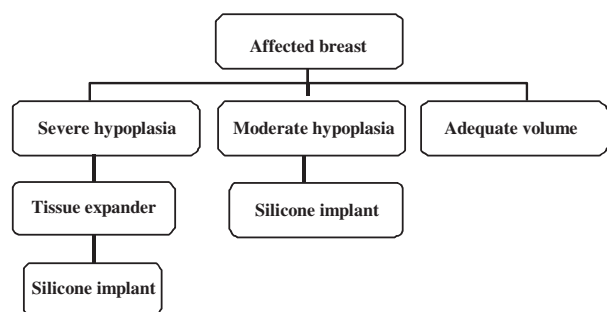


Fig. 5. Systematic scheme for treatment of the affected breast.

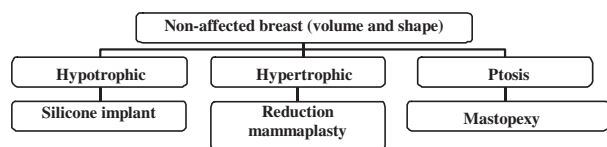


Fig. 6. Systematic scheme for treatment of the unaffected breast.

include free microvascular flaps of the contralateral latissimus dorsi, transverse rectus abdominis muscle, and upper gluteal muscle [20].

Contralateral symmetrization usually is necessary, and it can be achieved with a reduction

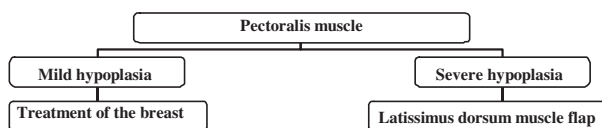


Fig. 7. Systematic scheme for treatment of the hypoplastic pectoralis major muscle.

mammoplasty (the most frequent procedure), a simple mastopexy to correct different grades of ptosis, or even an augmentation mammoplasty, as performed in the 14th case. We performed nipple–areolar complex reconstruction or symmetrization (superiorly or inferiorly sited) as a third-stage procedure. We prefer to wait for the edema to decrease and to observe any postoperative ptosis in the reconstructed breast before trying to define the differences in nipple–areolar complex size and position.

This article demonstrates several breast reconstruction options for patients with Poland’s syndrome. As observed, the spectrum of malformations is large, reinforcing the importance of an individualized treatment to achieve complete and adequate rehabilitation. On the basis of this experience, a flow chart was developed to guide the diagnosis and treatment of this rare malformation.

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