Congenital Anomalies of the Breast

Louise Caouette-Laberge, MD, FRCSC¹ Daniel Borsuk, MD, MBA, FRCSC¹

¹ Division of Plastic Surgery, University of Montreal, Montreal, Canada

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Address for correspondence Louise Caouette-Laberge, MD, FRCSC, Division of Plastic Surgery, University of Montreal, CHU Ste-Justine, 3175 Cote Ste-Catherine O., local 7907, Montreal, Canada, H3T 1C5 (e-mail: l.caouette-laberge@umontreal.ca).

Abstract

Keywords

- Poland syndrome
- breast
- pectoralis muscle
- polythelia
- polymastia athelia
- amastia

Poland syndrome is a combination of chest wall deformity and absent or hypoplastic pectoralis muscle and breast associated with shortening and brachysyndactyly of the upper limb. Clinical presentation varies widely; therefore, reconstructive procedures have to be adapted to the deformity, ranging from chest wall stabilization or augmentation, dynamic muscle transfer, nipple and areola repositioning, and breast augmentation using prosthesis or autologous tissue transfer. Other congenital breast anomalies include supernumerary nipple and areola (polythelia) and breast (polymastia), which can generally be found on the embryonic mammary ridge. Absence of the nipple, areola (athelia), or the breast tissue (amastia) is less frequent.

Poland Syndrome

Poland syndrome is named after Alfred Poland, who described the condition in 1841. Two previous descriptions of an absence of the pectoralis muscle were reported in 1826 and 1839 without associated hand deformity. Poland syndrome is a combination of chest wall and upper-limb anomalies that are generally unilateral although a few bilateral cases have been reported. It is more frequent in males and on the right side. The prevalence, approximately 1 in 20,000 to 30,000 births,¹ is thought to be underestimated. The cause of the syndrome is still controversial, but many authors support the hypothesis of an abnormal development of the fetal vasculature in the area causing the observed defects.² Other syndromes (Mobius, Klippel-Feil, Sprengel deformity) may be associated. Most cases appear to be sporadic although there are some reports of genetic transmission.

The syndrome includes deformation or absence of ribs, partial or total absence of the pectoralis muscles, particularly the sternocostal head of the pectoralis major, resulting in the absence of the anterior axillary fold. Occasionally, webbing of the axilla with an anomalous fibrous band from the thorax to the humerus may be found. Hypoplasia or absence of the breast, areola, and subcutaneous tissue is noted in most cases. The upper limb is often shorter with brachysyndactyly of the hand.³ Other muscles may also be affected on the ipsilateral side, including the serratus anterior and the latissimus dorsi muscles.⁴ Reconstructive options are numerous depending

on the presentation and the needs of the patient (► **Table 1**). A thorough analysis of the defect should be undertaken: absence or deformation of the ribs, total or partial absence of the pectoralis muscle (► **Fig. 1A**), presence of anterior axillary fold, webbing of the axilla (► **Fig. 1B**), and size and location of nipple-areola complex (NAC) and breast tissue. The shoulder and scapula may present a significant degree of asymmetry (elevation) and the hemithorax on the affected side is often narrower. The ipsilateral latissimus dorsi muscle should be carefully examined because it may also be absent. In this case, a free muscle transfer will need to be considered in the surgical planning. Anomalies of the vascular anatomy may complicate free tissue transfers.

Thoracic Wall Reconstruction

The goal of thoracic wall reconstruction is to provide a stable bony platform. Few patients present a significant defect of the rib cage.³ If the defect is large and the chest wall unstable, split rib grafts are inserted to bridge the gap. The anterior skin flap may be adherent to the pleura; caution is required during the dissection to avoid inadvertent thoracic penetration.⁵ In the majority of cases, rib flattening or single space defects do not require bone grafts. Camouflage may be obtained with a custom-made prosthesis,⁶ particularly in cases where a muscle transfer is necessary to recreate the anterior axillary fold and good coverage of the prosthesis is provided. Insertion of prosthesis under a skin flap with little subcutaneous tissue has an unnatural appearance and a higher complication rate⁷

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Table 1 Reconstructive options in Poland syndrome

Thorax	Unstable, large bony defect	Bone grafts (split ribs)
	Stable but flat	Camouflage according to needs for muscle and/or breast reconstruction: Prosthesis Soft tissue transfer
Pectoralis	Total absence	Dynamic muscle transfer (latissimus dorsi) Additional soft tissue in subclavicular area: Myocutaneous flap Perforator flap Fat graft Custom-made prosthesis
	Partial absence Axillary fold absent	Dynamic muscle transfer: Latissimus dorsi Transverse gracilis Other myocutaneous flap
	Partial absence Axillary fold present	Soft tissue augmentation: Muscle flaps Myocutaneous flaps Perforator flaps Fat graft Prosthesis
Breast	NAC: Superior location	Tissue expansion to lower NAC prior to breast augmentation
	NAC: Adequate location	No expansion unless space needed for large augmentation
	Presence of breast tissue/ Small contralateral breast	Breast prosthesis over or under transposed latissimus dorsi myocutaneous flap Possible latissimus dorsi myocutaneous flap or other buried free flap
	Absent breast tissue/ Large contralateral breast	Tissue expansion and one of the following: Preferred: Autologous augmentation: Pedicled TRAM flap Free TRAM flap MS-TRAM flap DIEP flap SGAP flap IGAP flap Alternate: Breast prosthesis

Abbreviations: NAC, nipple-areolar complex; TRAM, transverse rectus abdominis myocutaneous; MS-TRAM, muscle-sparing TRAM; DIEP, deep inferior epigastric perforator; SGAP, superior gluteal artery perforator; IGAP, inferior gluteal artery perforator.

such as displacement, late seroma, discomfort, and extrusion. Contour restoration of the chest can also be obtained with de-epithelialized dermal-adipose flaps or with lipofilling^{8–11} without muscle transfer if the anterior axillary fold is present. Rib cartilages are often more prominent on the affected side, but reshaping is seldom necessary, particularly in women; when a breast augmentation is done, the deformation becomes less noticeable.

Pectoralis Muscle Reconstruction

The decision to transfer a functioning muscle to replace the missing pectoralis muscle depends on the presence or absence of the anterior axillary fold. When the superior portion of the pectoralis muscle and axillary fold are present, soft tissue augmentation alone is necessary and it can be provided with a muscle, myocutaneous, cutaneous, or perforator flap, with fat grafting or with a prosthesis, depending on the quality of the overlying skin flap and the amount of breast tissue present.^{8–11} The decision to harvest the ipsi-

lateral latissimus dorsi muscle for volume only has to be weighed against the possible functional loss particularly in athletes.³

When the axillary fold is missing, a dynamic muscle transfer is needed (–Fig. 2A). The ipsilateral latissimus dorsi muscle is the preferred donor. It is transposed as a pedicled flap on the thoracodorsal vessels and nerve to the anterior chest and reinserted on the humerus (–Fig. 2B). The donor site deformity is minimal because the posterior axillary fold is preserved (–Fig. 2C) with the tendon of the teres major. A midlateral thoracic incision (–Fig. 2D) is used to harvest the latissimus and elevate the anterior skin flap for muscle insertion. Another short incision is made on the inner arm to reattach the tendon on the humerus with a bone anchor device. The latissimus muscle is larger and thinner than the pectoralis muscle so it is folded on itself to provide more bulk, particularly in the subclavicular area and laterally at the level of the axillary fold. Ohjimi¹² also suggests twisting the latissimus on itself to augment the anterior axillary fold. In

Fig. 1 (A) Poland syndrome with complete absence of pectoralis muscle and breast tissue. Note the superiorly located and hypoplastic nipple and areola (reprinted with permission Caouette-Laberge L, Bortoluzzi P. Correction of breast asymmetry in teenagers. In: Hall Findley E, Evans GRD, eds. *Aesthetic and Reconstructive Surgery of the Breast*. Philadelphia, PA: Saunders Elsevier; 2010: 601–629). (B) Poland syndrome with axillary web, absent sternocostal pectoralis muscle, and breast hypoplasia. (Reprinted with permission Caouette-Laberge L, Bortoluzzi P. Correction of breast asymmetry in teenagers. In: Hall Findley E, Evans GRD, eds. *Aesthetic and Reconstructive Surgery of the Breast*. Philadelphia, PA: Saunders Elsevier; 2010: 601–629.)

cases of severe depression in the subclavicular area, a de-epithelialized skin paddle can be transferred with the muscle to increase the thickness (►Fig. 3). The arm is kept adducted for 6 weeks postoperatively. Shorter incisions have been reported with endoscopic surgery,¹³ but it makes the proce-

dure more technically challenging. Free innervated muscle transfers are needed in cases of absent ipsilateral latissimus. Alternatives are the contralateral latissimus or free transverse myocutaneous gracilis flap,¹⁴ which can provide more soft tissue bulk.

Fig. 2 (A) Poland syndrome with minimal muscle in the subclavicular area, absent anterior axillary fold and hypoplastic breast in adequate location (Reprinted with permission Caouette-Laberge L, Bortoluzzi P. Correction of breast asymmetry in teenagers. In: Hall Findley E, Evans GRD, eds. *Aesthetic and Reconstructive Surgery of the Breast*. Philadelphia, PA: Saunders Elsevier; 2010: 601–629). (B) Two years after muscle transfer and breast augmentation with submuscular breast implant inserted at the time of muscle transfer, note scar on proximal arm for tendon reinsertion on the humerus. (C) Minimal deformity of the back following latissimus transfer (pronator teres remains for posterior axillary fold) (Reprinted with permission Caouette-Laberge L, Bortoluzzi P. Correction of breast asymmetry in teenagers. In: Hall Findley E, Evans GRD, eds. *Aesthetic and Reconstructive Surgery of the Breast*. Philadelphia, PA: Saunders Elsevier; 2010: 601–629). (D) Midlateral thoracic scar 2 months after latissimus transfer (Reprinted with permission Caouette-Laberge L, Bortoluzzi P. Correction of breast asymmetry in teenagers. In: Hall Findley E, Evans GRD, eds. *Aesthetic and Reconstructive Surgery of the Breast*. Philadelphia, PA: Saunders Elsevier; 2010: 601–629).

Fig. 3 (A) Poland syndrome with absence of pectoralis muscle and soft tissue deficiency in subclavicular area, well-developed breast (note lateral and superior projection of the breast due to lack of underlying pectoralis) (Reprinted with permission Caouette-Laberge L, Bortoluzzi P. Correction of breast asymmetry in teenagers. In: Hall Findley E, Evans GRD, eds. *Aesthetic and Reconstructive Surgery of the Breast*. Philadelphia, PA: Saunders Elsevier; 2010: 601–629). (B) Myocutaneous latissimus transfer with insertion of de-epithelialized skin island in subclavicular area. (C) Postop at rest (Reprinted with permission Caouette-Laberge L, Bortoluzzi P. Correction of breast asymmetry in teenagers. In: Hall Findley E, Evans GRD, eds. *Aesthetic and Reconstructive Surgery of the Breast*. Philadelphia, PA: Saunders Elsevier; 2010: 601–629). (D) Postop with muscular contraction showing the buried soft tissue in the subclavicular area (Reprinted with permission Caouette-Laberge L, Bortoluzzi P. Correction of breast asymmetry in teenagers. In: Hall Findley E, Evans GRD, eds. *Aesthetic and Reconstructive Surgery of the Breast*. Philadelphia, PA: Saunders Elsevier; 2010: 601–629).

Nipple Areola Relocation

When the location of the nipple areola complex (NAC) is too high, tissue expansion is needed to lower it to a proper level. It also provides the space for secondary breast augmentation. The tissue expander can be inserted early before contralateral breast maturation and slowly inflated over the years^{4,5} or delayed until definitive reconstruction is undertaken. It can be placed over or under the latissimus muscle⁴ at the time of muscle transposition depending on the amount of overlying soft tissue. It is important to suture the muscular pocket around the implant to prevent migration during expansion. When an autologous breast reconstruction is planned in the expanded pocket, significant overexpansion is required to allow postoperative edema of the transferred tissue and avoid a compartment syndrome within the capsule of the expander. We generally use the same mid-lateral thoracic incision for muscle transposition, expander insertion, or definitive breast augmentation with a prosthesis or free flap unless microvascular anastomosis requires exposition of the internal mammary vessels. The resulting scar is hidden under the resting arm and is well tolerated. However, preoperative marking of the inframammary crease is essential to guide the dissection and ensure correct location of the tissue on the anterior chest wall.¹⁵

Breast Reconstruction

Definitive breast augmentation is done after growth of the normal breast is completed. Augmentation with prosthesis

can provide good symmetry in cases where there is already some breast tissue on the affected side and when the normal breast is small and nonptotic. If the location of the NAC is adequate, the prosthesis can be inserted at the time of the muscle transfer, generally under the muscle if the overlying breast is small. The pocket is closed to keep the implant in the desired location and prevent superior migration in the subclavicular area. When the growth of the contralateral breast is not completed, a permanent expander/implant¹⁶ can be used for volume adjustment during final growth.¹⁷ A modest-size autologous breast augmentation can also be obtained with a buried latissimus dorsi myocutaneous flap, a buried free transverse myocutaneous gracilis flap¹⁴ or fat grafting.^{8,9} Autologous augmentation is helpful in cases of complete absence of the breast, and when the normal contralateral breast is of a large size, to obtain better symmetry and progressive ptosis with aging.¹⁵ A tissue expander is inserted at the time of muscle transposition and overexpansion is obtained prior to autologous tissue transfer. Many options are available^{18,19}: pedicled or free transverse rectus abdominis myocutaneous flap (TRAM) or deep inferior epigastric perforator flap (DIEP) or free inferior or superior gluteus myocutaneous or perforator flap (IGAP/SGAP). Our preference is for an inferior gluteus flap because of the large amount of adipose tissue that can be harvested and the easily concealed scar,^{20,21} particularly in teenagers who have little adipose tissue on the abdominal wall. Preoperative assessment of the vasculature is

mandatory because anomalies are frequent. When the thoracodorsal vessels provide the blood supply to a transposed latissimus dorsi muscle, the internal mammary vessels are the preferred recipient vessels.

Polythelia (Supernumerary Nipples)

Polythelia, the presence of supernumerary nipples (SNNs), is important for physicians to note in the newborn due to the preponderance for growth and change with increasing hormone levels. The SNN can develop similar pathologies as a normal breast, including breast neoplasia. Although most SNNs develop along the embryonic milk line, they may also appear on the back, shoulder, thigh, face, or vulva (►Fig. 4A).²²

The reported incidence of SNNs has varied, as high as 5.6%, and as low as 0.22% in a white European population.²³ Although there is little evidence to support association of SNNs with other congenital anomalies or syndromes, they are often familial. There is no correlation with gender and SNNs

have no predilection for the right or left side; however, there is a lower incidence of bilateral SNNs.²²

The management of SNNs has traditionally been observation. Changes to the pigmented lesion should be treated as for any melanocytic nevus, with early excision and histopathological assessment. An oncologist should evaluate nipple growth that occurs at a time other than childhood, puberty, or pregnancy.

Polymastia

Polymastia has an even lower incidence than polythelia, and also occurs along the embryonic milk line. Like polythelia, it is usually sporadic, but can have familial inheritance.²⁴ Unlike polythelia, polymastia can be associated with other congenital anomalies, particularly thoracic and renal.

Treatment of polymastia is variable and depends on the size and location of the supernumerary breast gland and nipple. Simple mastectomy is the option of choice in patients who present with a third distinct breast mound; however,

Fig. 4 (A) Polythelia supernumerary nipple inferior to the inframammary crease. (B) Polymastia in a case of severe left thoracic hypoplasia, scoliosis and Sprengel deformity. (C) Excision of nipple-areolar complex of accessory breast. (D) Accessory breast tissue preserved and mobilized on posterior pedicle. (E) Accessory breast tissue transposed superiorly and fixed on rib cage to improve support to residual breast. (F) Result after upper breast transposition inferiorly and medially on a superolateral pedicle, residual scar located in inframammary fold.

disruption of the inframammary fold and soft tissue envelope of the remaining breast should be prevented when possible. In cases where the accessory breast is adjoined to the native breast, tissue-sparing techniques with skin de-epithelialization and accessory nipple excision can usually restore the mound to a normal appearance and location (►Fig. 4B–F).

Corrective surgery for polymastia should be performed when breast development is complete and final breast tissue volumes have been achieved. It is often difficult to predict the form and position of the accessory breast mound and nipple and early excision may ultimately compromise the eventual outcome.

Athelia/Amastia

The absence of nipple and/or glandular breast tissue is a rare occurrence.^{25,26} According to Lin and Nguyen, there are different presentations of amastia and each can be attributed to different underlying pathologies. Ectodermal defects may cause bilateral absence of breasts in males and females and is related to the failure of development of the ectodermal layer and its appendages. Bilateral amastia may be an isolated occurrence or be associated with other anomalies of the palate and upper extremities. Unilateral amastia may be a variant of Poland syndrome and should be treated accordingly.

Treatment of amastia and athelia may follow the principles of postoncologic breast reconstruction with special attention paid to the placement of the inframammary fold and nipple. Tissue expansion with autologous and/or prosthetic breast reconstruction techniques is available and should be chosen based on patient and surgeon preference. Autologous reconstruction with abdominal, dorsal, or gluteal tissues may be chosen based on body habitus and patient preference. However, in the pediatric population the gluteal tissue transfers may be preferable due to the general availability of soft tissue and the less conspicuous scarring.^{20,21} Abdominal wall weakening and the frequent unavailability of adequate soft tissue make the abdomen a less-appealing donor site in adolescents and young adults as compared with the more mature adults seen with breast cancer. Prosthetic reconstruction with tissue expanders and permanent implants follows the same principles as adult reconstruction and can be employed at the surgeon's discretion.

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