

# Tuberous Breast and Poland Syndrome: An Underestimated Association?

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Eur J Pediatr Surg 2025;35:9–14.

## Abstract

**Aim of the Study** Both Poland syndrome (PS) and tuberous breast (TB) are anomalies affecting the breast, but they are considered mutually exclusive. Our aim was to determine the possible coexistence of PS and TB and to discuss TB management when associated with PS.

**Methods** Between 2010 and 2023, 51 female adolescent PS patients were studied at our center. Among these, we evaluated those who developed TB after puberty. PS diagnosis was made based on the hypoplasia or absence of pectoralis major muscle detected clinically and confirmed radiologically. Breast anomaly on PS side was classified with Thorax Breast and Nipple (TBN) classification: B1 corresponds to breast hypoplasia, B2 to breast aplasia. TB diagnosis was made with a clinical evaluation after puberty. TB cases were classified according to Grolleau classification.

**Main Results** Among 51 postpubertal PS females, we identified 23 (45%) who developed TB. In 3 cases (13%) TB was bilateral, in 20 (87%) contralateral to PS affected side. Age at time of the first surgical procedure was 16 years. Patients were treated with breast implants, fat grafting, rigotomy, local flaps, or a combination of these. TB treatment was performed simultaneously with PS breast reconstruction when feasible, or after it.

**Conclusions** TB incidence in our series was almost twice than in general population. This is the first study demonstrating and quantifying this association. TB deformity must be considered while defining PS surgical path in adolescents. Fat grafting is the first surgical option to treat both anomalies. A multidisciplinary approach is needed to minimize number of surgeries and maximize cosmetic results.

## Keywords

- ▶ tuberous breast
- ▶ Poland syndrome
- ▶ breast malformations
- ▶ breast hypoplasia
- ▶ breast aplasia

## Introduction

Breast malformations are relatively common findings in pediatric age and are frequently associated in young females with psychological symptoms, such as social anxiety, depression, dysfunction of the sexual sphere, and lack of self-confidence that can justify an early surgical treatment. It is

often around the time of puberty that patients with congenital breast abnormalities first seek advice.<sup>1–5</sup>

Breast deformities are usually classified into four different groups: hyperplastic (polythelia, polymastia, virginal hypertrophy, gynecomastia), hypoplastic (athelia, amazia, and amastia, Poland syndrome [PS]), deformational anomalies (tuberous

received

April 17, 2024

accepted after revision

November 29, 2024

accepted manuscript online

December 3, 2024

article published online

December 20, 2024

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Georg Thieme Verlag KG,  
Oswald-Hesse-Straße 50,  
70469 Stuttgart, Germany

DOI <https://doi.org/10.1055/a-2494-7611>.  
ISSN 0939-7248.

breast [TB] deformity), and acquired anomalies (burns, tumors, iatrogenic, irradiation).<sup>5-7</sup> Among deformational deformities, TB appears to be common but often underdiagnosed. The incidence of TBs in the general population has been reported to be between 10 and 27.6%.<sup>8,9</sup> It manifests at puberty and is characterized by a conical shape of the breast due to the presence of a fibrous ring at the base of the breast itself, resulting in the absence/hypoplasia of lower breast quadrants, areolar herniation and enlarged nipple–areola complex (NAC), higher inframammary fold, and skin deficiency.<sup>10-12</sup>

PS is indeed a rare congenital condition (estimated incidence of 1:30,000)<sup>13</sup> characterized by the unilateral absence or hypoplasia of the pectoralis major muscle. PS can be associated with many other ipsilateral chests (both skeletal and soft tissue defect) and/or upper limb malformations, resulting in a wide phenotypic variability.<sup>14-17</sup>

To our knowledge, TB and PS have always been described as two different entities and considered mutually exclusive: no previous articles regarding the presence and the incidence of TB in PS patients are present in the literature. As PS manifests earlier in life compared with TB and has been shown to be responsible for significant psychological distress, we believe that the eventual development of an associated contralateral TB later in life tends to be underdiagnosed and under-investigated. The aim of this retrospective analysis is to determine for the first time the possible coexistence of these two breast deformities and to discuss TB management when associated with PS. Another endpoint is to assess the incidence of TB in this PS series and compare it to the incidence of TB in the general population: we hypothesize that TB incidence in patients affected by PS is higher than in general population.

## Patients and Methods

### Patients Selection

We retrospectively selected 51 female adolescents affected by PS evaluated at Istituto Giannina Gaslini between January 2010 and December 2023. Among these, we identified 23 patients who also developed TB at puberty.

Prepuberal girls and those classified as  $\leq$ Tanner 4 were not considered in our study.

### Diagnostic Criteria

PS diagnosis was made based on the hypoplasia or absence of pectoralis major muscle (PMM) clinically detected and confirmed radiologically (mainly through US, CT and/or magnetic resonance imaging (MRI) were requested in select cases). A global evaluation of chest deformities involved both plastic and thoracic surgeons if necessary. We adopted the Thorax Breast Nipple (TBN) classification proposed by Romanini et al<sup>13</sup> (→Table 1) to categorize patients based on thoracic, breast, and NAC anomalies and consequently to adopt the most appropriate surgical approach to treat them.

TB deformity diagnosis was made after puberty based on the presence of one or more of its characteristic features (lower breast quadrants hypoplasia and asymmetry, conical breast shape with a constricted base, lack of breast skin, herniated NAC with enlarged areola, higher inframammary

**Table 1** TBN classification by Romanini et al

T		Thoracic
	T1	Hypoplasia or aplasia of pectoralis muscles and soft tissue
	T2	T1 and sternal deformity, pectus excavatum and/or carinatum
	T3	T1 and rib aplasia
	T4	T1, T2, and T3 (muscle, sternum, and rib defect)
B		Breast
	B1	Breast hypoplasia
	B2	Breast aplasia
N		Nipple–areola complex
	N1	NAC hypoplasia with dislocation of <2 cm
	N2	NAC hypoplasia with dislocation of >2 cm
	N3	Absent NAC

Abbreviations: NAC, nipple–areola complex; TBN, thorax breast nipple.

fold) and then classified as Grolleau I, II, or III based on clinical severity.<sup>18</sup>

### Surgical Management

Surgical treatment of PS was defined based on TBN classification derived surgical algorithm,<sup>13</sup> taking also into account the age of each patient and psychological distress. As regard to breast deformities, reconstructive breast surgery was performed at least 2 years after menarche by adopting fat grafting (FG), tissue expanders, and, in older patients, breast implants. TB deformity treatment included FG, and surgical correction of breast shape (rigotomy and reposition of inframammary fold) and volume (mastopexy, reductive mammoplasty), with or without breast implants and areolar reduction, eventually a combination of these.

### Data Collection and Analysis

Patients' demographic and clinical data, radiological findings, and pre- and postoperative pictures were evaluated by the same team of plastic surgeons and stored in a password-protected institutional Cloud Service along with informed consent for publication, in accordance with the GDPR. An approval statement for publication was obtained by our regional Ethics Committee (CER Liguria) on 5th December 2022, authorization no. 636/2022–DB id 12824.

From a statistical point of view, due to the nature of this study, descriptive measurements were most extensively adopted. Numerical variables are reported as median. Fisher exact test was used to compare categorical variables.

## Results

### Demographics and Incidence

Among 51 females with PS who satisfied inclusion criteria, we identified 23 cases (45%) who developed TB deformity at puberty: 3 (13%) were bilateral and 20 (87%) contralateral



**Fig. 1** Left Poland syndrome (PS) (B2), right side affected by tuberous breast (TB) anomaly (Grolleau I).

relative to the PS side; no TB deformity exclusively homolateral to PS affected site was registered (–Figs. 1, 2, and 3; –Table 2).

Regarding PS-related breast anomalies (B), we categorized our series according to TBN classification: 17 patients (74%) were classified as B1: breast hypoplasia, and 6 (26%) had amastia (B2) (–Table 2).

Right PS in this series was observed in 18/23 (78%) patients; consequently TB involved the left side in 18 out of 23 patients (78%) and was bilateral in 3 girls (13%), presenting a milder PS phenotype. More specifically, excluding patients with unilateral breast aplasia due to PS (B2 patients), 17 girls (74%) classified as B1 developed TB at puberty: 14 contralateral (61%) and 3 bilateral (13%); no TB deformity exclusively homolateral to PS affected site was registered.

When thorax deformity (T) in PS was considered, 15 patients (65%) were classified as T1, while a more severe



**Fig. 2** Right Poland syndrome (PS) (B2, N3), left side affected by tuberous breast (TB) anomaly (Grolleau I).



**Fig. 3** Bilateral tuberous breast (TB) and right Poland syndrome (PS) anomaly (B1).

**Table 2** TB laterality in PS B1 and B2 patients

Tuberous breast	Total (%)	PS B1 (%)	PS B2 (%)
Homolateral	0	0	0
Bilateral	3 (13)	3 (13)	–
Contralateral	20 (87)	14(61)	6 (26)
Total	23 (100)	17(74)	6 (26)

Abbreviations: PS B1, Poland syndrome with breast hypoplasia, PS B2, Poland syndrome with breast aplasia; TB, tuberous breast.

defect (T2 to T4) was present in 8 patients. No statistically significant association was found between TB presence and severity of chest defect due to PS ( $p=0.4$ ), as reported in –Table 3.

Regarding TB deformity classification, of 20 patients with unilateral TB, 8 of 23 (35%) were classified as Grolleau type I, 10 (45%) as type II, and 2 (20%) as type III.

Patients with bilateral TB were classified as follows: one had bilateral TB type II, one had bilateral TB type I, and the third had TB type III homolateral to the PS and type I contralateral to the PS side.

**Reconstruction Surgery in this Series**

Approximately half of the patients affected by PS and TB (11 out of 23, 48%) completed TB corrective surgery in our series (–Table 4). Nine patients (39%) partially accomplished

**Table 3** TB and chest deformity severity association

	T1	T2–3–4	Total	P
TB	15	8	23	0.4
No TB	14	14	28	
Total	29	22	51	

Abbreviation: TB, tuberous breast.

**Table 4** TB surgery, clinical data

ID	Side	Classification	Age at first TB surgery	No. of surgery for TB correction			Type of surgery for TB correction (referring to contralateral to PS if bilateral TB)			
				Total	Associated to PS correction	TB correction only	Glandular flap/mastopexy	Breast implant	Rigotomies + FG (volume, mL)	Areolar reduction/contralateral graft
1	Contralateral	III	16	2	2	0	No	Yes	Yes (60)	No
2	Bilateral	H: II, C: II	17	2	2	0	Yes	Yes	Yes (30)	No
3	Contralateral	II	22	1	1	0	Yes	Yes	No	No
4	Contralateral	II	14	1	1	0	Yes	No	No	No
5	Contralateral	II	15	3	2	1	Yes	No	Yes (100 + 120)	No
6	Bilateral	H: III, C: I	16	2	1	1	No	Yes	Yes (90)	Yes
7	Contralateral	II	17	1	1	0	No	Yes	Yes (66)	No
8	Contralateral	II	14	1	1	0	Yes	No	No	Yes
9	Contralateral	I	16	3	3	0	No	No	Yes (80 + 78)	Yes
10	Contralateral	II	15	2	2	0	Yes	No	Yes (30)	Yes
11	Contralateral	I	15	2	2	0	no	No	Yes (100 + 90)	No
	Total			20	18	2	6	5	8	2

Abbreviations: BI, breast implant; C, contralateral; FG, fat grafting; H, homolateral; TB, tuberos breast.

it and the last three patients (13%) refused further surgery after PS reconstruction.

The further analysis concerns only patients who completed the whole reconstructive process for TB deformity (→Table 4).

The mean age at the time of the first surgery was 16 years old.

Considering the total number of surgical procedures (20), only 2 of them (10%) were performed in a surgical session aimed at correcting TB deformity exclusively, whereas 90% of the procedures were performed during an operation aimed at treating both the PS and TB (contralateral and/or ipsilateral) breast deformities simultaneously.

TB correction required a single procedure in four patients (36%), a double step in five patient (45%), three surgical interventions in two cases (19%). Most patients (7 out of 11, 63%) required a combination of multiple procedures of FG,



**Fig. 4** Patient no. 5. Preoperative image: Right Poland syndrome (PS) (T2B1N2) + contralateral tuberos breast (Grolleau II).

surgical correction of breast shape and volume, and areolar reduction (→Figs. 4 and 5; →Table 4).

FG was the most performed technique and was adopted in eight patients (72%): in five patients a single (45%) procedure was performed, in three cases FG was repeated twice (27%). The injected volume of FG varied from 30 to 200 cc in every single procedure; the mean volume FG was 76 cc, depending on fat availability of the donor site (usually abdomen, hips, and thighs) and the entity of the defect needed to be corrected.



**Fig. 5** Patient no. 5. Result after both Poland syndrome (PS) and tuberos breast (TB) correction.

In two N3 patients (NAC absent), areolar reconstruction of the PS-affected side was performed using a skin graft from the contralateral nipple with an enlarged areola (TB).

In all patients, a contralateral breast prosthesis was implanted to correct breast abnormalities associated with PS.

## Discussion

### Incidence

PS-related breast anomalies can be suspected at neonatal/pediatric age in case of major or more clinically evident anomalies whereas other anomalies, such as TB, might not be detectable or cause any psychological symptoms until puberty, when soft tissue defects become more evident and adolescents start experiencing high level of body uneasiness.<sup>2,19</sup>

As already mentioned, the incidence of TB malformations in general population has not been determined yet and it is probably underdiagnosed, since milder forms are not brought to the medical attention.<sup>9</sup> In our series, 45% of the patients affected by PS developed TB at puberty.

This analysis provides us important information:

1. TB incidence in our PS series is almost twice with respect to the general population (45% vs. 27%).
2. Almost one in two girls affected by PS will develop TB at puberty. The high frequency of TB deformity in females affected by PS must be considered while defining PS surgical path.

To the best of our knowledge, no previous work has been reported in the literature describing the incidence of TB deformity in patients with PS. The small sample size and its retrospective nature are the main limitations of this study. In addition, as the real incidence of TB in the general population is not clearly available, further investigations are needed to define whether the incidence of TB in patients with PS is actually higher than in the general population.

### Surgical Treatment

Since TB deformity worsens as the breast continues to develop, a spontaneous resolution is not possible and surgical correction is required. Rees and Aston in 1976 first tried to correct this deformity and proposed radial scoring as a method of increasing the diameter of the breast.<sup>20</sup> In the following years, several techniques have been proposed<sup>20–27</sup> to correct TB deformity including musculocutaneous flaps, glandular remodeling, breast implants, and more recently FG.<sup>28–33</sup> Nowadays, the latter two procedures are the most appropriate options.<sup>1</sup> As shown in **Table 4**, half of the patients were treated with breast implants (5 out of 11, 45%) and FG was performed in 8 out of 11 patients (72%).

The innovative surgical algorithm for PS-related anomalies proposed by the last author has been adopted by our institute and applied for the first time in a pediatric/adolescent setting.<sup>34</sup> Correction of TB deformities has been included in the PS surgical pathway, which is becoming increasingly tailored. As already mentioned, 90% of procedures to treat TB took place in a surgery session that combined procedures aimed to treat both PS and TB (contralateral and/or ipsilateral) defects simultaneously. The

purpose of this approach is to reduce the number of operating room access and to correct both hemithorax defects at the same time, thus harmonizing the aesthetic result. For instance, bilateral FG was performed 6 times. In two patients, areolar reconstruction affecting an N3 (NAC absence) patient was performed using a skin graft obtained from contralateral TB-enlarged areola. In one of them, during the same operation, Poland hypoplastic breast (B1) was treated with a tissue expander and a contralateral mastopexy was performed to correct TB deformity. The other patient had simultaneous bilateral FG and areolar grafting.

### Specific Considerations

At first glance, our approach to breast reconstruction seems to partially contradict the latest evidence. In fact, numerous studies in the last years approved FG alone as a possible first choice in TB surgical treatment. In 2019, Gutierrez-Ontalvilla et al<sup>31</sup> underlined the applicability of LF alone compared with other more invasive techniques (breast implants and flaps) in teenage girls. As reported by the Spanish surgeon, the young age and the still developing breast must be considered when choosing the most appropriate surgical technique. More specifically, breast implants and flaps are associated with major surgical complications while LF offers good natural aesthetic results with minimal functional and aesthetic sequelae and few minor complications (e.g., oil cysts). Even though at our Institute we chose to adopt FG as the first-choice procedure in adolescent females affected by TB deformity alone (not associated to PS), some considerations need to be made. First, FG alone may not always be able to provide a good aesthetic result in severe cases and other surgical techniques may be required. Second, FG procedure usually needs to be repeated several times to achieve a satisfactory result and young girls might not have enough fat deposits. Moreover, since FG is also fundamental for many PS defects correction, FG is frequently performed bilaterally (PS and TB side), reducing the amount of fat available for each side. Finally, a global approach considering both thorax and bilateral breasts malformations is needed. In our series, every adolescent was treated with breast implants to correct breast defects caused by PS. Implants are in fact the first choice in PS breast hypoplasia or aplasia correction since volume defect is not only due to an abnormal breast development but also muscular absence in the pectoral region and an eventual associated ribcage malformation. In addition, the patients themselves requested bilateral breast implants because they felt uncomfortable with the difference in consistency and shape between the “Poland breast” and the TB.

### Conclusion

In our experience, TB incidence in female teenagers affected by PS is 45%. It is usually a unilateral defect (contralateral to PS) and the left side is more frequently affected.

This is the first study demonstrating and quantifying this association. Although PS is a major congenital malformation responsible for psychological distress from an early age, the possible development of ipsilateral or, more commonly,

contralateral MT should not be underestimated. The possible development of a contralateral TB defect in female teenagers affected by PS must be considered and investigated to define an increasingly global chest reconstructive plan with less access to the operating theater. FG is the first surgical option to treat both the anomalies, but it does not always provide a good aesthetic result in severe cases and other surgical techniques are required.

Further studies are needed to confirm our findings and to improve the surgical management of these two coexisting defects.

#### Conflict of Interest

None declared.

#### Acknowledgments

The authors would like to thank the Italian Association of Poland Syndrome and the Rare Disease Unit of the Giannina Gaslini Institute.

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