Chapter 20 / Capítulo 20

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Comprehensive approach to tuberous breasts: anatomy, diagnosis and treatment

Abordaje integral de la mama tuberosa: anatomía, diagnóstico y tratamiento

Bianca Moretti[1], Ernesto Adrián Moretti[1], Micaela Clerici[1]

[1]Universidad Abierta Interamericana, Facultad de Medicina y Ciencias de la Salud, Carrera de Medicina. Rosario, Argentina.

ABSTRACT

The study addressed the anatomy, embryological development, and main characteristics of tuberous breasts as a congenital anomaly of clinical and aesthetic relevance. It was described that the mammary gland originated in the mammary ridge during intrauterine life and reached structural maturity at puberty. It was noted that tuberous deformity was the result of a fibrous ring that restricted normal growth, causing a contracted breast base, areolar herniation, and alterations in projection. Various authors have proposed multiple classifications since 1976, reflecting phenotypic heterogeneity and the difficulty of reaching a universal diagnostic consensus. It was also noted that tuberous breasts could be unilateral or bilateral, with a high frequency of breast asymmetry, which impacted patients' quality of life. Epidemiological studies revealed that the prevalence was higher than expected and that the condition often remained underdiagnosed. It became clear that conventional mammoplasty techniques could not correct the condition, which led to the development of specific surgical procedures adapted to the severity and morphology of each case. In conclusion, it was recognised that tuberous breasts constituted a complex spectrum of malformations that required a comprehensive diagnostic and therapeutic approach. Early identification and the design of personalised surgical strategies were essential to optimise functional and aesthetic results, as well as to improve patients' self-esteem and quality of life.

Keywords: Breast; Tuberous; Embryology; Classification; Surgery.

RESUMEN

El estudio abordó la anatomía, el desarrollo embriológico y las principales características de la mama tuberosa como una anomalía congénita de relevancia clínica y estética. Se describió que la glándula mamaria se originaba en la cresta mamaria durante la vida intrauterina y alcanzaba su madurez estructural en la pubertad. Se señaló que la deformidad tuberosa era producto de un anillo fibroso que restringía el crecimiento normal, generando una base mamaria contraída, herniación areolar y alteraciones en la proyección. Diversos autores propusieron múltiples clasificaciones desde 1976, lo que reflejó la heterogeneidad fenotípica y la dificultad de alcanzar un consenso diagnóstico universal. Asimismo, se destacó que la mama tuberosa se presentaba de forma unilateral o bilateral, con alta frecuencia de asimetría mamaria, lo cual impactaba en la calidad de vida de las pacientes. Estudios epidemiológicos revelaron que la prevalencia era superior a lo esperado y que, a menudo, la patología permanecía infradiagnosticada. Se

evidenció que su corrección no podía resolverse con técnicas convencionales de mamoplastia, lo que impulsó el desarrollo de procedimientos quirúrgicos específicos adaptados a la severidad y morfología de cada caso. En conclusión, se reconoció que la mama tuberosa constituía un espectro complejo de malformaciones que exigía un enfoque diagnóstico y terapéutico integral. Su identificación precoz y el diseño de estrategias quirúrgicas personalizadas resultaban fundamentales para optimizar los resultados funcionales y estéticos, así como para mejorar la autoestima y la calidad de vida de las pacientes.

Palabras clave: Mama; Tuberosa; Embriología; Clasificación; Cirugía.

INTRODUCTION

The breast, with its complex anatomical, physiological, and embryological organization, is a fundamental element not only in female body identity but also in women's reproductive, aesthetic, and psychological health. A detailed study of its normal structure and the alterations that can affect its morphology is essential to understand both its functioning and the multiple pathologies that compromise it. Among the various anomalies described in the scientific literature, tuberous breast has become one of the most relevant, not only because of its complex clinical presentation but also because of its impact on patients' quality of life. This congenital deformity is characterized by abnormal development of the mammary gland due to a fibrous ring that limits its expansion, resulting in characteristic stigmata such as contraction of the mammary base, abnormal elevation of the inframammary fold, and areolar herniation.

A review of normal breast anatomy and its embryological origin helps us understand the extent of the morphological deviation in tuberous breasts. Likewise, the multiple classifications proposed since the first description in 1976 highlight both the phenotypic diversity of this anomaly and the difficulty of establishing a unified diagnostic criterion. This phenomenon is linked to the scarcity of large-scale epidemiological studies that would allow its prevalence in the general population to be accurately determined, reinforcing the need to continue developing systematic research. In this context, breast asymmetry, the frequency of bilateral presentation, and the coexistence of different degrees of severity are determining factors for clinical and surgical assessment. Therefore, this theoretical framework seeks to integrate the anatomical, embryological, and pathological aspects of the mammary gland, along with the historical evolution of the classification of tuberous breasts, to establish a more rigorous approach to their diagnosis and treatment.

DEVELOPMENT

Normal breast anatomy

The breast (mamma, TA) (Rehnke et al., 2018) is one of the organs found in the anterior wall of the thorax in both men and women, being more developed in women due to its importance in reproduction. It is composed of glandular tissue and supportive fibrous tissue, integrated with adipose tissue, along with blood vessels, nerves, and lymphatics.

The mammary gland (glandula mammaria, TA) (Rehnke et al., 2018) is located above the subcutaneous tissue that covers the pectoralis major (m. pectoralis major, TA; Rehnke et al., 2018) and pectoralis minor (m. pectoralis minor, TA) (Rehnke et al., 2018), known as the pectoral fascia (fascia pectoralis, TA) (Rehnke et al., 2018) in two-thirds of the breast surface and the remaining third over the fascia of the serratus anterior muscle (m. serratus anterior, TA) (Rehnke et al., 2018). It extends laterally from the lateral sternal border to the mid-axillary line and vertically from the 2nd rib to the 6th rib. Between the breast and the pectoral fascia

lies loose subcutaneous tissue, or a potential space, known as the retromammary space. There is a small amount of adipose tissue there, giving the breast some mobility over the fascia. There is a small portion of the breast that extends toward the armpit, known as Spence's process/tail (processus axillaris: processus lateralis, TA) (Rehnke et al., 2018). This process tends to be the most common location for breast tumors.

On the other hand, the breast is firmly attached to the dermis by skin ligaments, known as the suspensory ligaments of the breast or Cooper's ligaments (lig. suspensorio mammaria: retinaculum cutis mammae, TA) (Rehnke et al., 2018). These are usually more developed in the upper part of the gland, thus supporting the breast.

The breast is divided into four quadrants, using the hours on a clock as a reference, with 12, 3, 6, and 9 o'clock as the reference points. From there, imaginary lines are drawn, dividing it into:

1. Upper medial quadrant
2. Lower medial quadrant
3. Lower lateral quadrant
4. Upper lateral quadrant

The nipples (papillae mammariae, TA) (Rehnke et al., 2018) are conical or cylindrical processes located at the highest point of the breast and surrounded by an area of pigmented skin, the areola (areola mammae, TA) (Rehnke et al., 2018; Kolker & Collins, 2015).

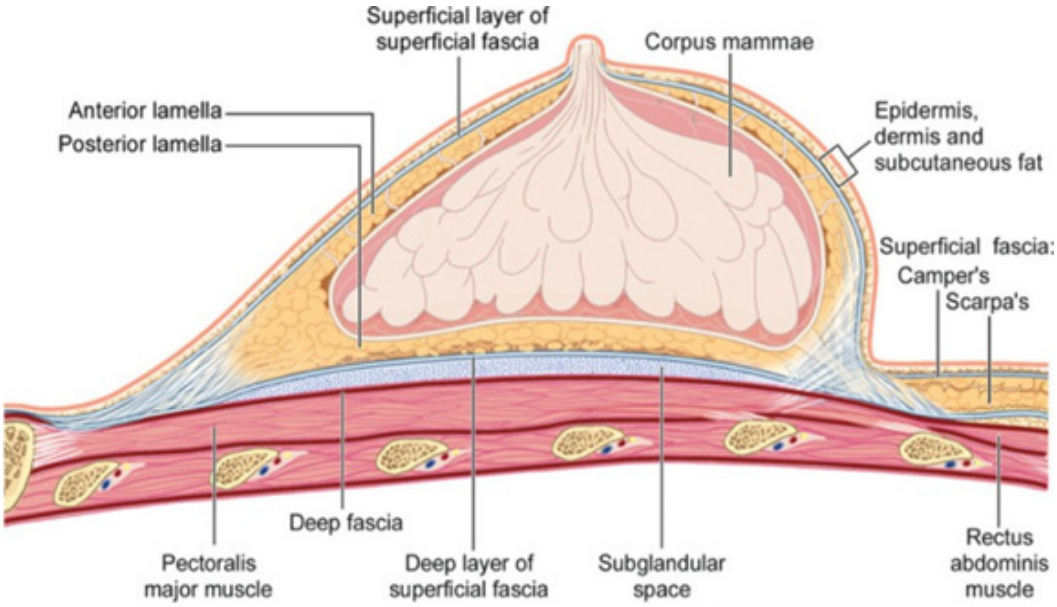
Embryological origin and development of the breast during puberty

The breast originates from the ectoderm in the mammary ridge during the fifth week of intrauterine life. In the seventh and eighth weeks, the ridge disappears, except for a small portion in the thoracic region. It then penetrates the underlying mesenchyme between the tenth and fourteenth weeks. Breast differentiation and subsequent development occur during intrauterine life and are completed at birth, with no major changes occurring until puberty.

During puberty, the breast tissue beneath the areola grows, enlarging in size until the age of 15-16, when the breast takes on its characteristic shape. As its origin is ectodermal and its invagination is into the mesenchyme, the breast tissue is contained by a fascia, called the superficial fascia (fascia superficialis, TA) (Rehnke et al., 2018). This fascia continues with Camper's superficial fascia (panniculus adiposus, TA) (Rehnke et al., 2018). The superficial fascia is composed of two layers: a superficial layer (the outer layer covering the breast parenchyma) and a deep layer (which forms the posterior wall of the breast parenchyma and rests on the fasciae of the pectoralis major and serratus anterior muscles). Cooper's ligaments connect the deep and superficial layers of the superficial fascia (Klinger et al., 2017), as shown in figures 1 and 2.

Anatomy of the tuberous breast

The tuberous breast was first described in 1976 by Rees & Aston (Mandrekas et al., 2003). They described the anomaly because they began to notice an increase in reports of complications and unpredictable results in some complex breast configurations. This deformity is difficult to treat and is named for its resemblance to the root of a tuber. Usually, the breast should have a hemispherical shape on an elliptical base, with the horizontal diameter slightly greater than the vertical diameter. The base of the tuberous breast is deficient in both dimensions, causing it to project forward, along with significant development of the areola and nipple.



Source: Rehnke, R. D. (2018)
Figure 1. Breast anatomy

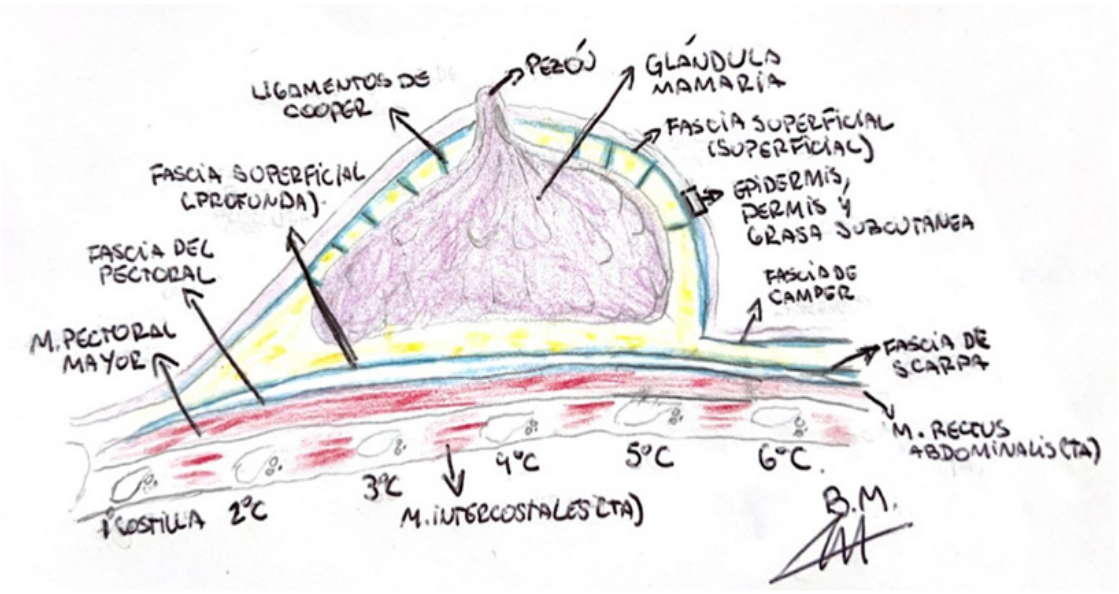


Figure 2. Drawing by Bianca Moretti inspired by the article by Rehnke et al. (2018)

The stigmata described allow the disorder to be divided into two anatomical types: . Deficiency in both vertical and horizontal dimensions, producing a small concentric base, resulting in a tuber-shaped breast with a large areola and nipple complex with herniation of the breast tissue, and Greater deficiency in the vertical dimension than in the horizontal dimension. It is described as ptotic with the nipple pointing downward but without apparent herniation of

the complex.

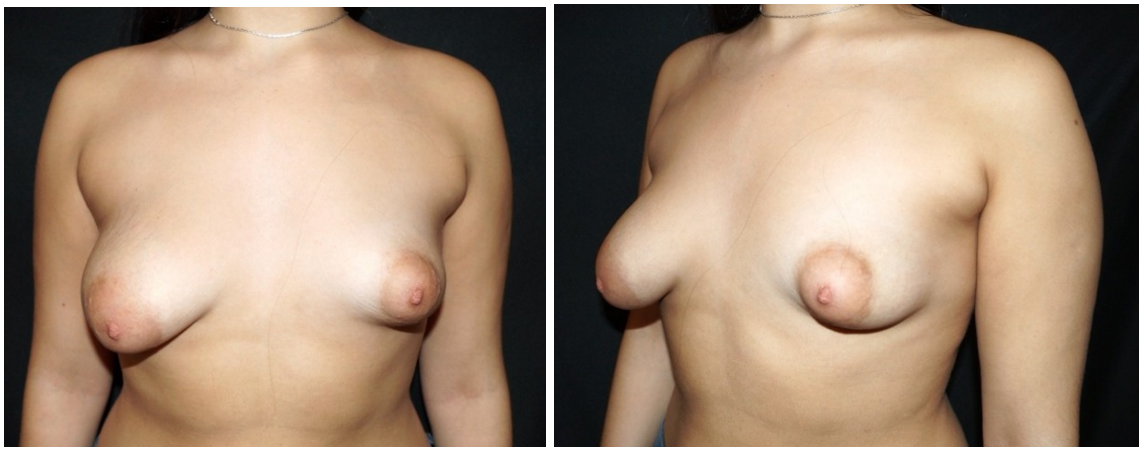


Figure 3. Patient No. 1—Patient from the Private Hospital of Rosario with tuberous breast—City of Rosario—Year 2024

Rees & Aston (as cited in Mandrekas et al., 2003) demonstrate that tuberous breasts cannot be corrected with standard mammoplasty or augmentation mastopexy procedures and describe one of the first techniques for correcting this malformation.

Klinger et al. (cited by Dessy et al., 2017), noting the difficulty of diagnosing the deformity, selected five characteristics that are key to the deformity:

1. Contracted skin envelope (horizontally and vertically)
2. Contracted breast base
3. Reduction in breast parenchyma volume
4. Abnormal elevation of the inframammary fold
5. Areolar herniation of the breast parenchyma

This condition is caused by a fibrous ring at the periphery of the nipple-areola complex, which inhibits its normal development. This constricting ring is denser in the lower breast, preventing growth during puberty. Histologically, the existence of this fibrous tissue in the aforementioned area has been confirmed. It has high concentrations of collagen and longitudinally arranged elastic fibers. It is suspected that a thickening of the superficial fascia causes this ring. Possibly, the two layers mentioned above are more closely joined than usual, or Cooper's ligaments are thicker and denser. Then, by not allowing expansion, the breast herniates toward the nipple-areola complex.

The malformation of the superficial fascia determines the severity of the deformity. It ranges from mild underdevelopment of the lower inner quadrant, but with relatively normal breast volume, to severe hypoplasia of all four quadrants. For this reason, several authors have created their own classifications, which will be discussed later (Klinger et al., 2017).

Breast asymmetry

Breast symmetry is considered an essential feature of female beauty. Any difference, whether in size or shape, causes dissatisfaction in women. Small differences are common, but in some cases, the discrepancy in size can become so great as to affect the patient's quality of life

(Lozito et al., 2022).

Tuberous breasts are often associated with breast asymmetry; however, symmetrical presentation cannot be ruled out. It should be noted that the same patient may exhibit unilateral or bilateral tuberous breasts and, in cases of bilateralism, they may present symmetrically or asymmetrically (DeLuca-Pytell et al., 2005).

Classification and frequency of tuberous breasts

According to DeLuca-Pytell et al. (2005), tuberous breast is considered a spectrum of deformity ranging from mild to severe.

The first classification was created in 1996 by von Heimburg et al. (Rehnke et al., 2018) based on four main types:

1. Type I: Hypoplasia of the lower medial quadrant
2. Type II: Type I with sufficient skin in the subareolar region
3. Type III: Type I without sufficient skin in the subareolar region
4. Type IV: Severe contraction with a small breast base

The proposed classification was illustrated, as shown in Figure 4.

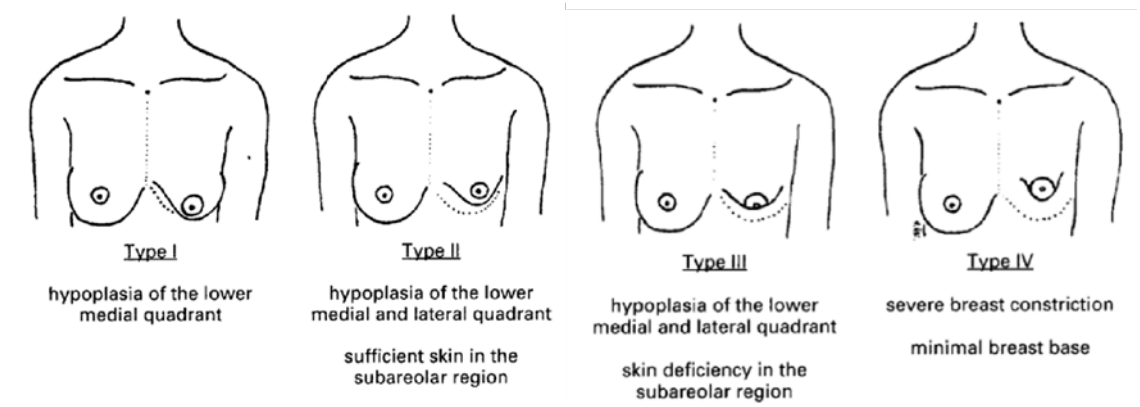


Figure 4. Classification by von Heimburg
Source: von Heimburg D (1996)

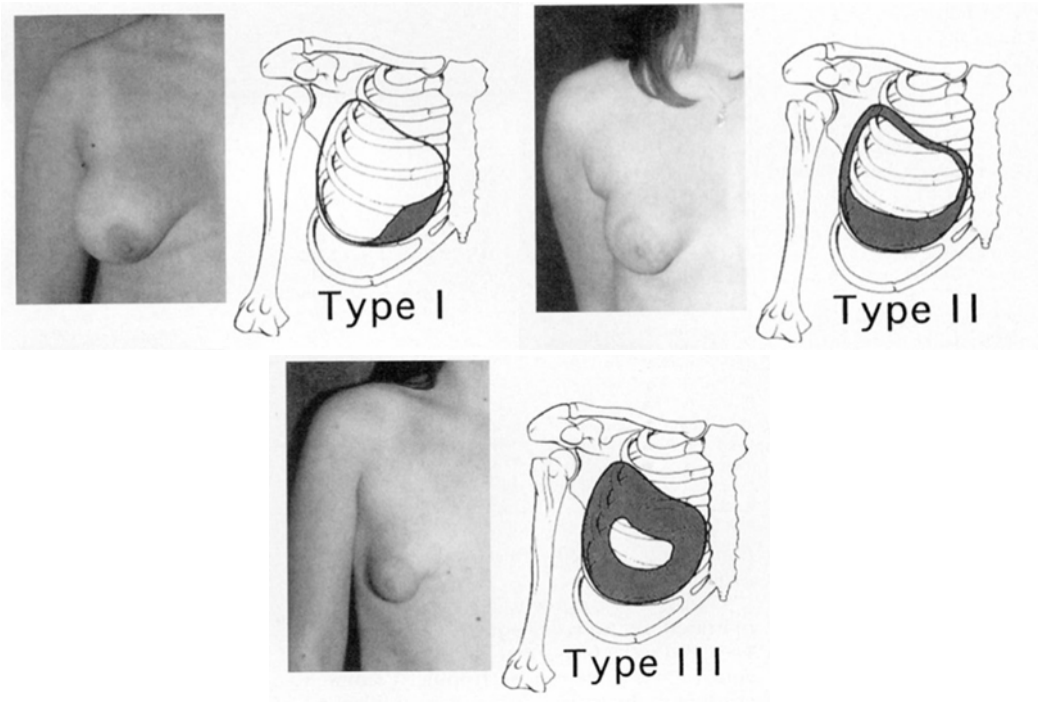
In their publication, von Heimburg et al. (as cited in Rehnke et al., 2018), with 40 patients with a total of 68 tuberous breasts, classified each one with the following results:

1. Type I: 19 breasts (28 %)
2. Type II: 18 breasts (26 %)
3. Type III: 12 breasts (18 %)
4. Type IV: 19 breasts (28 %)

The next classification proposed was that of Grolleau et al. (as cited in Mandrekas & Zambacos, 2010) in 1999, based on the previous classification. This classification uses defects in the breast base as a reference, as shown in figure 4. This results in three types:

1. Type I: Deficiency of the lower medial quadrant. Regardless of breast volume, the lower medial angle forms an italic S shape, and its contralateral side appears larger in comparison.

2. Type II: Both lower quadrants are deficient. The areola points downward, and the subareolar skin segment is short.
3. Type III: All four quadrants are deficient, and the breast base is contracted horizontally and vertically. In typical cases, the breast has a tubular shape. However, the gland may be so underdeveloped that it appears severely hypoplastic, as evidenced by the contralateral breast being typically tuberous.



Source: Grolleau JL et al. (1999)
Figure 5. Classification by Grolleau

In their publication, Grolleau et al. studied 37 patients who had some tuberous breast tissue in both breasts. As some patients had combinations, when establishing the frequency of each type of tuberous breast, they considered each breast individually.

1. Type I: 40 breasts (54 %)
2. Type II: 19 breasts (26 %)
3. Type III: 15 breasts (20 %)

They then analyzed the frequency of combinations, yielding the following results:

1. Type I+I: 10 cases (27 %)
2. Type I+II: 10 cases (27 %)
3. Type I+III: 10 cases (27 %)
4. Type II+II: 3 cases (8 %)
5. Type II+III: 2 cases (5,5 %)
6. Type III+III: 2 cases (5,5 %)

They highlight that at least 81 % of patients (30 cases) have type I in one of their two breasts.

They also mention that 70 % of patients had breast asymmetry of more than 100 g. The remaining 30 % did not.

DeLuca-Pytell et al.(2005) in 2005 adopted Grolleau's classification. They reported that type III is the most common, accounting for 60,3 % (320 of 531 breasts), and type I is the least common, accounting for 15,6 % (83 of 531 breasts). Finally, type II represented 24,1 % (125 of 531 breasts).

In 2005, Persichetti et al.(2005) reported differences between tuberous and tubular breasts. A reduction in vertical and horizontal diameters characterizes tuberous breasts. Tubular breasts, on the other hand, have a long vertical diameter in the upper quadrants, extending beyond the inframammary fold, and a normal or relatively reduced horizontal diameter. The volume is normal or increased.

In their publication, they classify the degree of asymmetry according to the presumed difference in breast volume resected in the symmetrization process, as follows:

1. Grade A: Mild (-200 g)
2. Grade B: Moderate (200-400 g)
3. Grade C: Severe (+400 g)

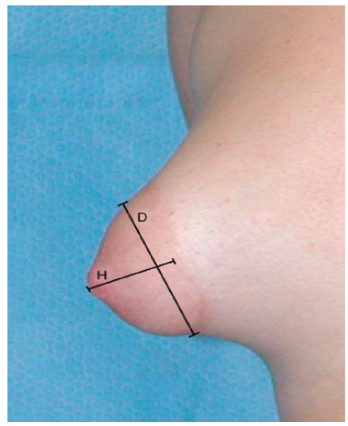
The study includes 30 patients: 11 with tuberous breast, 18 with tubular breast, and 1 with both. Table 1 shows the results, considering each breast separately:

Table 1. Incidence rate of tubular and tuberous breasts

	Grade A	Grade B	Grade C
Tubular breast	4 (22,2 %)	9 (50 %)	1 (5,5 %)
Tuberous breast	6 (54,4 %)	2 (18,8 %)	0

Source: Persichetti, P et al. (2005)

In 2007, Pacifico et al.(2007) proposed the Northwood Index. This is a standardized tool that establishes the relationship between the herniation measured from the side (H) and the areola diameter (D) measured in centimeters, as detailed in Figure 5. They conclude that if the division result is greater than 0,4, the patient has tuberous breasts (table 2).



Source: Pacifico et al. (2007). D: diameter. H: herniation

Figure 6: Northwood Index

Table 2: Northwood Index in normal breasts and tuberous breasts	
Breast type	Northwood Index (range)
Tuberous breast	0,54 (0,41-0,66)
Normal breast	0,19 (0,07-0,35)
Source: Pacifico, M. D., et al. (2007)	

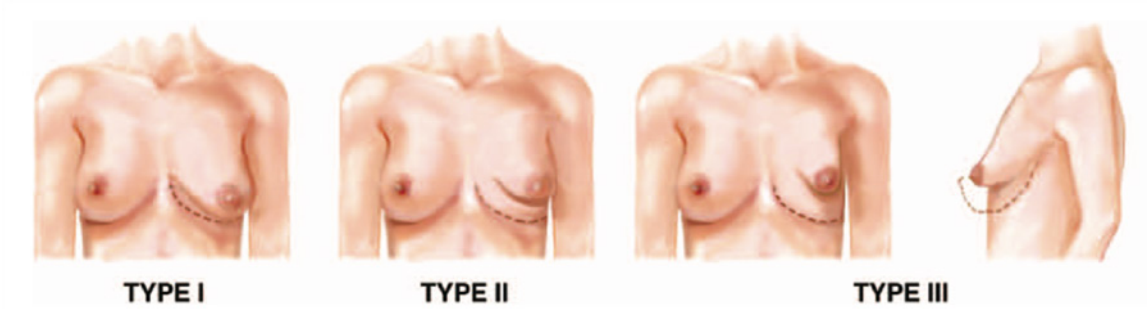
Costagliola et al. in 2013 (as cited in Lozito et al., 2022) delve deeper into the classification and characterize it according to the herniation of the nipple-areola complex. Manual compression is applied to the nipple-areola complex, and herniation is classified as permanent or intermittent. In addition, these authors argued that a fourth type, called type 0, should be added to the classification of Grolleau et al. (2005) to describe cases with a normal breast base and isolated herniation of the nipple-areola complex.

In 2015, Kolker & Collins (2015) created a more detailed classification with three types, as shown in the following table and figure 7:

Table 3. Kolker & Collins classification

Type	Breast base	Inframammary fold	Skin envelope	Volume Breast	Ptosis	Areola
I	Mild contraction	Normal laterally, Slight medial elevation	Sufficient	Deficiency Minimal or Hypertrophic	Mild, moderate, or severe	Enlarged
II	Moderate contraction	Medial and lateral elevation	Insufficient inferior	Moderate deficiency	There is no or mild	Normal, slightly or moderately herniated
III	Severe contraction	Elevation Complete of the sulcus or absence of the sulcus	Globally insufficient	Severe deficiency	Mild or Moderate	Severe herniation

Source: Kolker A. R., (2015)



Source: Source: Kolker A. R., (2015)
Figure 7. Tuberous Breast Deformity: Kolker & Collins classification

The study includes 26 patients (51 breasts) as a sample. They classify each breast, giving this result:

1. Type I: 12 (24 %)
2. Type II: 26 (51 %)

3. Type III: 13 (25 %)

Klinger et al.(2017) consider the anatomical characteristics of breast stenosis, the trophism of the mammary glandular parenchyma, and breast ptosis as the basis for their classification. They identify two types of stenosis: vertical glandular stenosis and vertical-horizontal glandular stenosis. They analyze the trophism of the mammary glandular parenchyma and classify it as hypoplastic (insufficient glandular tissue) or non-hypoplastic. Finally, they analyze whether the breast is ptotic, meaning the areolar position is below the intramammary fold. This gives eight types:

1. Type I: Vertical stenosis – hypoplastic – non-ptotic
2. Type II: Vertical stenosis – hypoplastic – ptotic
3. Type III: Vertical stenosis – non-hypoplastic – non-ptotic
4. Type IV: Vertical stenosis – non-hypoplastic – ptotic
5. Type V: Vertical-horizontal stenosis – hypoplastic – non-ptotic
6. Type VI: Vertical-horizontal stenosis – hypoplastic – ptotic
7. Type VII: Vertical-horizontal stenosis – non-hypoplastic – non-ptotic
8. Type VIII: Vertical-horizontal stenosis – non-hypoplastic – ptotic

This publication recruited 246 patients. The patients were classified according to each type as follows:

1. Type I: 58 patients
2. Type II: 41 patients
3. Type III: 22 patients
4. Type IV: 9 patients
5. Type V: 41 patients
6. Type VI: 19 patients
7. Type VII: 52 patients
8. Type VIII: 4 patients

In 2017, Innocenti et al. (Innocenti et al., 2018) proposed a new classification that includes mild forms, based on breast volume (hypoplastic or normoplastic) and tissue quality (soft or hard). The proposed classification is as follows:

- *Hypoplastic: subclassified as:*
    - Soft: This is the most severe form. The skin is very thin and folds easily due to the lack of suspensory ligaments.
    - Hard: in contrast, this has a larger breast base, a smaller areola, and thicker skin firmly attached to the parenchyma through compact unions by Cooper's ligaments. Normally, the inframammary fold is flat or concave.

- *Normoplastic: This is subclassified into three types:*
    - Type I-II: Breast volume deficit in the lower pole, with only the lower medial quadrant missing in type I and both lower quadrants missing in type II. Both types show fullness in their upper poles because most of the parenchyma is displaced upward by the crease and the lack of skin covering the lower pole. They usually have different types of ptosis, and their nipple-areola complex is located closer to the inframammary fold, pointing downward.
    - Type III: Permanent or intermittent glandular protrusion within the areola

The publication includes 78 patients (145 tuberous breasts). First, it classifies them as bilateral (67 patients) and unilateral (11 patients). Then, each breast is classified as:

Hypoplastic: 62
- a) Soft: 22
- b) Hard: 47

Normoplastic: 83
- a) Type I: 48
- b) Type II: 21
- c) Type III: 14

Despite all these new classifications, the one by Grolleau et al. (2005) continues to be the most widely used in the vast majority of published studies and is the one that a large number of plastic surgeons use to define the treatment to be performed for surgical correction (Lozito et al., 2022).

Presentation of tuberous breasts

A bibliographic search was conducted on the prevalence of tuberous breasts in the last 5 years, and no specific studies on this topic were found. As a result, the search was extended to the last 10 years, and only the 2016 publication by Klinger et al. (2017) was found. The authors mention that this is the first study of its kind.

This study involved 1,600 female patients who consulted for breast surgery, either augmentation or reduction, between January 2009 and July 2014. Twenty-five percent of the patients wanted breast augmentation (AUG). Twenty-five percent wanted reduction mammoplasty (RED). Fifty percent were admitted for different breast conditions (POP).

In the AUG group, 48,5 % showed at least one of the characteristics of tuberous breasts, the most common being a contracted breast base (82 %), followed by a reduction in breast parenchyma volume (77,8 %), and the least common being herniation of the breast parenchyma into the areola (8,8 %).

In the RED group, 47,3 % were diagnosed with tuberous breast, the most common being contraction of the inframammary fold with a short lower pole (77,2 %), followed by reduction in breast parenchyma volume in the inferolateral and inferomedial quadrants (52,9 %).

Finally, in the POP group, 27,6 % showed at least one characteristic of tuberous breast, with reduction in breast parenchyma volume in the inferomedial and inferolateral quadrants being the most frequent (67,8 %), followed by a contracted breast fold with a small lower pole (52,9 %) and, lastly, herniation of breast parenchyma in the areola with a contracted breast base (6 %).

Mandrekas & Zambacos (2010) reported treating 22 patients with tuberous breasts over 10 years, of whom 19 had bilateral tuberous breasts, and 3 had unilateral tuberous breasts (41 breasts in total). They stated that in 86,3 % of cases, tuberous breast was bilateral and in 13,6 % of cases, it was unilateral.

CONCLUSIONS

Comprehensive analysis of tuberous breasts, grounded in detailed knowledge of breast anatomy and embryology, reveals that this deformity constitutes a complex clinical spectrum that requires a multidimensional approach for identification. The presence of a fibrous ring that restricts glandular growth, together with variability in the degree of hypoplasia and possible areolar herniation, means that each patient presents a unique picture that challenges diagnostic standardization. The diversity of classifications proposed over the last few decades reflects the scientific community's attempts to establish uniform parameters. However, the heterogeneity of clinical manifestations continues to limit the adoption of a universal consensus.

Clinically, tuberous breasts impact not only physical appearance but also patients' self-esteem, body image, and quality of life, correcting a cosmetic, functional, and psychological necessity. Prevalence studies, although scarce, have shown that this malformation is underdiagnosed, highlighting the importance of raising awareness among healthcare professionals about its clinical characteristics and the need for timely diagnosis. Similarly, the close association with breast asymmetry and the frequency of bilateral cases reinforce the need for individualized surgical strategies capable of restoring symmetry and anatomical proportion.

In summary, tuberous breast should be understood as a developmental anomaly whose complexity requires a comprehensive approach that combines an understanding of its etiopathogenesis with the application of specific surgical techniques adapted to each case. Only in this way will it be possible to advance the construction of a more robust knowledge base that will improve aesthetic and functional outcomes while promoting greater physical and psychological well-being in affected patients.

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