

ANATOMICAL ANOMALIES OF THE BREAST

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ABSTRACT

Anatomic breast anomalies are alterations in the structural composition of the breast.

Objectives: The following work aims to list the main deformities, highlighting the importance of knowledge about the anatomy of this organ.

Results: The analysis of the literature makes it possible to divide, didactically, breast anomalies into numerical or structural, being generally originated during embryogenesis or pubertal development. Diagnosis and treatments are based on individual history, taking into account female anxieties and concerns.

Conclusion: anatomical abnormalities of the breasts are conditions that directly affect the physical and emotional aspects of women and should therefore be thoroughly investigated.

KEYWORDS: ANATOMICAL ANOMALIES; BREAST; NUMERICAL ANOMALIES; STRUCTURAL ANOMALIES; DIAGNOSIS.

INTRODUCTION

The breasts are external organs of the female reproductive system, whose glandular structure is responsible for the secretion of breast milk, intended for the nutrition of the offspring. Anatomically, they have a strong symbolism linked to self-image, the construction of identity and sexuality, especially for women, representing a symbol of femininity¹. In this context, it is clear that anatomical anomalies of the breasts, which may be numerical or structural in nature, and associated body distortions have a major impact on health and quality of life, especially for females, both in terms of physical as well as psychosocial aspects.

Based on this assumption, knowledge about the main anatomical abnormalities of the breasts is of paramount importance for targeted attention and comprehensive care for women, understanding the main complaints and insecurities that are often associated.

Breast Structure and Development:

The breasts are arranged bilaterally on the anterior wall of the thorax, in the adipose tissue of the subcutaneous layer underlying the skin. On its outer portion, each breast has a conical projection called the papilla and a thick, reddish-brown circular region, the areola. Internally, it is composed of a glandular parenchyma and a fibrous stroma, the first being a set of numerous secretory lobules united in lobes, which have lactiferous ducts that carry secretion to the papillary surface. The stroma, on the other hand,

consists of the suspensory ligaments of the breasts (bands of dense connective tissue originating from the dermis that surround and intersect the lobes), in addition to layers of loose connective tissue that separate the mammary complex from the underlying pectoral muscles².

From this, it should be noted that most anatomical changes in the breasts are correlated with an anomalous development at the embryonic or pubertal level:

As far as embryogenesis is concerned, in the fourth week of intrauterine development, a pair of epidermal thickenings called mammary ridges form along each side of the body of the embryo from the area of the future axilla. Although much of the mammary line disappears shortly after its formation, a small portion in the thoracic region persists and penetrates the mesenchyme. Thus, in view of the structures involved in this stage, this involution is essential for breast development, so that, if altered, it can lead to the appearance of some numerical anomalies, such as polymastia and polytelia. By the seventh week, the remnant of the mammary crest produces a well-defined primary bud of the mammary gland, which grows towards the dermis and the adipose tissue. In the tenth week, the bud starts the branching process, forming between 16 and 24 smaller units. These resulting epithelial sprouts elongate and multiply throughout pregnancy, and at the end of prenatal life, they are channeled and form the lactiferous ducts, which open into a small fossa. Shortly after birth, this pit transforms into the papilla by proliferation of the

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underlying mesenchyme. The surrounding skin also proliferates and thickens to form the areola. Thus, developmental dysfunctions at this stage can also generate anatomical anomalies of the organ, such as atelia and inverted nipple³.

At puberty, increasing concentrations of estrogen and progesterone stimulate the development of the glandular parenchyma through budding and elongation of the ducts, forming alveoli and inactive secretory cells. In addition, there is intense deposition of adipose tissue, which essentially reflects the volume of the breasts⁴. Thus, attention is needed for possible anatomical anomalies of the breasts that arise during this period, such as hypomastia, gigantomastia and breast asymmetry.

Diagnosis

The diagnostic investigation of anatomical breast anomalies must have a special concern with the anguishes and apprehensions expressed by the patient. The physician must pay attention not only to the physiological impact generated by the disease, but also to the social and emotional impacts caused.

During the anamnesis, symptoms such as pain, changes in size, presence of masses and secretions, asymmetries in shape, changes in the nipple and inflammatory signs must be well characterized and defined. Its investigation should focus on the time of emergence and the mode of evolution; its relationship to menstrual periods, pregnancy, or surgeries; menstrual history; the use of drugs, especially contraceptives; and its relationship with other symptoms or findings⁵.

The physical examination should be thorough, with breast inspection and palpation. Anatomical anomalies, such as polythelia, can be easily confused with nevi and other skin lesions, while gigantomastia and breast asymmetries can be mistakenly associated with neoplastic masses⁶.

Imaging exams provide great help, especially in diagnostic elucidation and malignancy exclusion. Often, mammography is the method of choice for screening, however, it has certain limitations, such as the approach to dense breasts and the young age group of patients. In addition to this exam, investigation via ultrasound is highly indicated, with several advantages for breast evaluation. It is a methodology that does not use ionizing radiation, and has better applicability in these morphological changes, considering the target audience of young patients, in the pubertal period, with parenchyma rich in fibroglandular tissue⁷.

Main Numerical Anatomical Anomalies:

Polythelia

Polythelia is defined as the presence of supernumerary nipples without the presence of associated parenchymal tissue, that is, without mammary glands. This anatomical anomaly is caused by the non-regression of the mamma-

ry line during embryogenesis³. Commonly confused with dermatological spots or nevi, supernumerary nipples occupy, as a rule, the anterior region of the thorax, lying between the axillary and inguinal regions (Figure 1). Generally, polythelia may be related to organic dysfunctions of the urogenital tract⁶.

Polythelia can be treated surgically by excision of the supernumerary nipple, especially when its presence causes aesthetic discomfort to the patient, impairing his emotional health.



Figure 1 - Polythelia. Supernumerary nipples in the right inframammary region and in the lower medial quadrant of the left breast.

Polymastia

Polymastia, also called supernumerary breast tissue, is a condition in which there is the presence of glandular parenchymal tissue in ectopic regions. Mostly found in the armpit, chest wall, inguinal region and, more rarely, in the vulva, their cause is the non-regression of the milk lines during embryogenesis³ (Figure 2).

During childhood, there are no major clinical manifestations caused by this anatomical anomaly. However, in the pubertal phase and also in the pregnancy-puerperium cycle, polymastia often begins to present itself as a region of edema with a feeling of heaviness and, sometimes, pain. Hormonal stimulation, especially due to the production of gonadotropins and sex hormones, leads to proliferation of both the stroma and the supernumerary glandular parenchyma, leading to the development of the accessory breast.

The accessory breast is made up of the same structures that make up the physiological breast tissue, therefore, it can be affected by the same pathological processes, benign and malignant, that compromise the normal breast. Thus, the diagnosis of fibroadenoma, cyst, infection and carcinoma may be present in the supernumerary breast, so that imaging investigation is essential for the correct diagnosis and treatment⁷.



Figure 2 - Polymastia. A. Photograph of a woman with polymastia in the lower medial region (LIQ) of the right breast. B. Mammography in CC and MLO views showing accessory fibroglandular tissue located in the LIQ of the right breast. C and D. Ultrasonography showing accessory fibroglandular tissue (polymastia) in the LIQ of the right breast.

Amastia

Amastia is defined as the complete absence of the glandular structure that constitutes the breasts. This condition can be present unilaterally or bilaterally (Figure 3) and have its etiology in iatrogenic or congenital causes⁵. Caused by a dysfunction during the process of breast formation in embryogenesis, such as the failure of the ectoderm to invaginate towards the mesoderm, this anatomical anomaly can be accompanied by a series of malformations of other structures originating from these leaflets, such as skin, hair and the muscles, especially the pectoralis major³.

In these cases, the use of imaging tests is fundamental for the correct diagnosis and clinical management. Magnetic resonance imaging is an interesting exam for analyzing the integrity of structures, such as the ribs and pectoral muscles; while ultrasonography is an important test for confirming cases of breast mass, attesting to the absence of underlying breast tissue and ruling out other diagnoses, such as breast hypoplasia⁷.



Figure 3- Amastia in a young patient.

Amazia:

Amazia is characterized by the absence of parenchymal tissue in the breast, with the presence of a nipple-areolar complex (Figure 4). The picture arises as a result of a defect in the formation of the mammary parenchyma and stroma during embryogenesis³. Due to the non-commitment of the nipple, the diagnosis may be impaired during childhood, only becoming noticeable with the entry into the pubertal period.

Ultrasonography reveals retroareolar tissue with a fibrotic appearance, local adipomastia, and absence of mammary glandular parenchyma.

This lack of breast growth and development, associated with the common development of the nipple and areola, is usually correlated with other congenital anomalies, such as the presence of cleft palate, saddle nose and piri-form hypoplasia⁸.

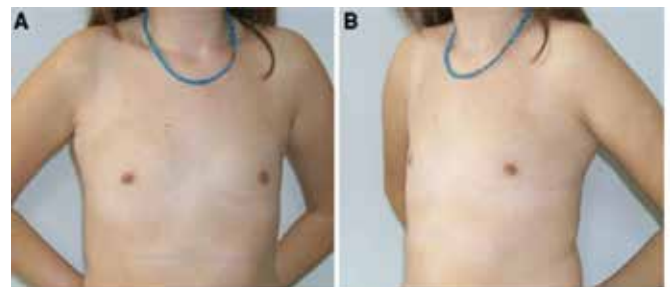


Figure 4- Amazia. Characterized by the absence of glandular parenchyma in the mammary structure.

Atelia:

Atelia is characterized by the absence of the nipple-areolar complex in the presence of mammary parenchyma and stroma (Figure 5). Although normally this anatomical anomaly is associated with trauma or iatrogenic events, its congenital form can also appear, especially when associated with amastia.

Currently, there are specific surgical techniques for the construction of a new structure of the areola and papillary structure, through the implantation of dermal flaps. Furthermore, alternative reconstitution through tattooing or micropigmentation is not uncommon⁶.



Figure 5- A) Atelia. Represented by the absence of nipple-areolar structure, with stroma and glandular parenchyma present. B)

Main Structural Anatomical Anomalies:

Hypomastia:

Hypomastia, also called mammary hypoplasia, is an anatomical breast anomaly noticed after the onset of puberty and characterized by poor development of the glandular parenchyma and stroma that make up the organ⁶. (Figure 6)

Hypomastia is caused by a decrease in estrogen production by the body or the insensitivity of the breast tissue to recognize and respond to the produced and circulating hormones. Thus, it is, as a rule, an anomaly secondary to other dysfunctions such as gonadal dysgenesis or pituitary hypogonadism⁴.

Ultrasonography is the preferred method for evaluating hypomastia, especially due to the reduced size of the breasts, making it difficult to use mammography. This anomaly is diagnosed, as a rule, under the age of 30 years. The examination reveals breasts with poorly developed glandular parenchyma and little support tissue⁷.



Figure 6- Hypomastia in a young patient. Areopillary development considered normal, with small parenchymal and stromal proportions.

Breast hypertrophy:

Mammary hypertrophy is an abnormal increase in the size of the breasts (Figure 7). Its extreme form, gigantomastia, is defined as dysfunctional and disabling breast enlargement that requires an average reduction of 1500 grams per organ for correction. Its occurrence is more common in pregnant women and in women in the pubertal period, and may even have an idiopathic or pharmacological character⁶.

The pathophysiology of this anomaly is generally associated with an increase in circulating levels of steroid hormones, such as occurs in pregnancy, puberty and the use of medications; or to an increased sensitivity of breast tissue to physiological levels of these substances. Howev-

er, regardless of the etiopathogenesis, the condition must have a clinical investigation based on differential diagnoses, such as lipoma, fibroadenoma, phyllodes tumor and cancer. In this context, the use of imaging tests is fundamental for the search for tissue alterations that help to rule out the hypothesis of malignancy. Breast pain, nipple-areola complex ulcerations, low back pain, inflammatory abscesses and psychological disorders associated with self-image are common⁹.



Figure 7 - Bilateral breast hypertrophy in a postpartum woman.

Asymmetry:

Breast asymmetry must be differentiated from a purely physiological condition that is common to women, after all, the majority of the female population has some degree of variation in the size of the breasts. However, in some cases, it can be configured as pathological⁶. (Figure 8)

When the discrepancy in the size of the breasts becomes large, it can become an aesthetic problem that impacts the quality of life of the woman, requiring a medical approach. Yet another reason that leads to the need for better clinical investigation is the fact that voluminous breast asymmetries may be a manifestation of other diagnoses that are causing the observed size variation, such as fibroadenomas, cysts and carcinomas.

The expected increase in breast volume at puberty can be affected by a series of factors that lead to asymmetry, such as genetic influences on the higher concentration of hormone receptors in one of the breasts, trauma, infections and iatrogenic or growing nodules and masses¹⁰.



Figure 8- Physiological breast asymmetry.

Inverted Nipple:

Nipple inversion is an anatomical anomaly, congenital or acquired, characterized by the transposition of the entire nipple towards the subareolar region, inside the breast. Developed due to a proliferation of fibrous tissue between the nipple and the subareolar parenchyma, this inversion can have different etiologies.

Although congenital presentations (Figure 9) of inverted nipple do not have major clinical consequences beyond the aesthetic deformity, this anatomical anomaly, when acquired, must be thoroughly investigated and detailed, seeking to identify the benign or malignant nature of the lesion. Other manifestations such as eczema, pain, secretions, loss of sensitivity and nipple stiffness may be associated with the condition.

It is relevant to analyze the presence of bilaterality of nipple inversion: bilateral inversion is a finding suggestive of congenital anomaly and benignity, whereas unilateral inversion is an indication that there may be an associated malignant tumor lesion. In this same analysis, the time elapsed by the inversion indicates that acute anatomical alterations generally occur with benign processes, with an infectious aspect, while chronic variations are more suggestive of neoplasms¹¹.



Figure 9- A and B. Right inverted nipple. Benign congenital anomaly.

Tuberous breast:

The tuberous breast is a congenital anatomical anomaly

ly that becomes noticeable at puberty, through the neuroendocrine stimulus on the breasts. This deformity is characterized by hypoplasia and tissue narrowing at the base of the breast, constricting the site and leading to vertical and horizontal underdevelopment of the entire structure. Thus, the breast acquires a tubular cylindrical format anteriorly, at the same time that the glandular parenchyma undergoes herniation in the area of the nipple-areolar complex, leading to areolar dilation⁶ (Figure 10).

As for its pathophysiology, the exact mechanism is still uncertain. It is believed that the deep fascia of the breast, located between the dermis and the pectoral musculature, has greater thickening and a more resistant adherence to the regions at the base of the breast, inducing the observed narrowing and hypoplasia. Additionally, the fibrous ring formed restricts the radial growth induced during puberty, favoring development towards the nipple, where the absence of resistance and adequate support on the part of the superficial fascia causes an areola enlargement, typical of the tuberous breast¹².

Present, as a rule, in young women, although it may also be associated with cases of gynecomastia in the male population, the main negative impact of tuberous breasts is on aesthetic discomfort. Due to the narrowing of the local parenchyma and the alteration of the nipple, this condition can still impair the breastfeeding process. The proposed treatment for this anatomical anomaly is surgical correction¹².

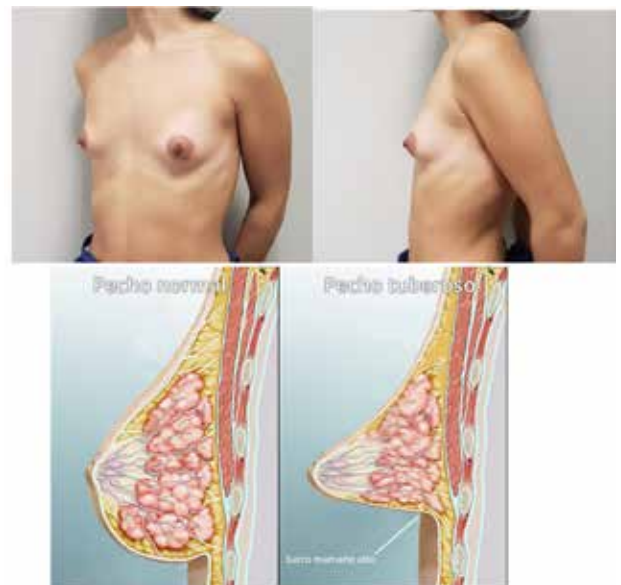


Figure 10- A) Tuberous breast. B) Schematic representation of the typical mammary structure of the tuberous breast.

Symastia:

Symastia is an anatomical anomaly characterized by the medial confluence of the breasts. It is a condition of congenital or acquired origin, in which a medial connec-

tion of the breasts appears, with the presence of an intermammary membrane on the mid-sternal line, uniting both structures⁶.

The most common form of this anomaly is iatrogenic (Figure 11). It is not uncommon for patients to develop symptoms of symmastia after breast augmentation, with the placement of breast implants, or reduction, with the removal of local tissue excess. In this scenario, the dysfunction is linked to the dissection of the midsternal fascia, responsible for joining the skin to the bone, allowing the healing process to penetrate the created space, uniting the breasts.

In the background, with rare cases recorded, simastia can also be configured as a congenital anomaly. In this condition, a malfunction in mammary embryogenesis leads to the formation of a web of supporting glandular parenchymal and connective tissue, uniting the breasts medially over the sternum region.

The therapeutic approach is surgical and is based on the removal of the tissue that composes the intermammary web, with the fixation of the dermis to the sternal periosteum and with the postoperative use of compressive pads in the region of the intermammary fold created on the sternum¹³.



Figure 11 - Iatrogenic symmastia resulting from mammoplasty and exeresis of cutaneous melanoma in the region of the intermammary sulcus.

CONCLUSION

The anatomical integrity of the breasts is an aspect of great physical and emotional importance for women, considering the strong biological and feminine symbology that these organs have. The main breast anomalies, which may be numerical or structural in nature, commonly derive from embryological inconsistencies or hormonal dysregulations, especially during puberty. Thus, a thorough investigation of cases of breast deformities is relevant in order to determine an accurate diagnosis, rule out possible malignancies and ensure adequate treatment. Furthermore, ac-

tive and comprehensive listening to women's insecurities and fears about such anomalies are essential behaviors for effective care.

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