

Gigantomastia and physical affections: a mini-review

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Gigantomastia y sus afectaciones físicas: una mini revisión

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Abstract

Gigantomastia is the excessive development of breast glandular and fatty tissues. This clinical entity carries physical suffering for women affected. Back pain is a common symptom; it is directly related to a constant mechanical spinal loading resulting from a non-physiological spinal angle and mechanical stress. This review provides a comprehensive summary of gigantomastia's main characteristics, covering its description, classification, historical background, etiology, clinical presentation, differential diagnosis, and surgical treatment. Gigantomastia brings some consequences, including pathological changes in posture and more serious affectations, such as the progression into degenerative spondylosis and disc herniations. It is necessary to consider the imaging assessment, which could initially include radiographs and MRI, which aims to identify favorable or unfavorable changes in the musculoskeletal structures.

KEY WORDS

Gigantomastia; gigantomasty; macromastia; vertebra.

Resumen

La gigantomastia es el desarrollo excesivo de los tejidos glandulares y grasos de la mama. Esta entidad clínica conlleva sufrimiento físico para las mujeres afectadas. El dolor de espalda es un síntoma común; está directamente relacionado con una carga espinal mecánica constante resultante de un ángulo espinal no fisiológico y estrés mecánico. Esta revisión resume las principales características de esta condición relacionadas con su descripción, clasificación, antecedentes históricos, etiología, presentación clínica, diagnóstico diferencial y tratamiento quirúrgico. Entre las consecuencias de este padecimiento tenemos cambios patológicos en la postura y afectaciones más graves, como la progresión a espondilosis degenerativa y hernias discales. Es necesario considerar la evaluación por imágenes, que inicialmente podría incluir radiografías y resonancia magnética, con el objetivo de identificar cambios favorables o desfavorables en las estructuras musculoesqueléticas.

PALABRAS CLAVE

Gigantomastia; macromastia; vertebra.

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Introduction

Gigantomastia is the excessive development of breast glandular and fatty tissues.¹ This hypertrophy, which occurs during pregnancy or puberty, carries physical sufferings for women affected, such as skin irritation, upper extremity neurological symptoms, and breathing problems, followed by a lower physical performance and body aches —generally located along the back, waist, and neck areas.¹⁻³ Back pain is a critical and common symptom in women with an excess of mammary tissue. It is directly related to a constant mechanical spinal loading resulting from a non-physiological spinal angle and mechanical stress.²

Whether affected women remain untreated, they will be more likely to progress into spondylosis due to the increase in thoracic kyphosis.² Current data associate gigantomastia with progressive spinal degeneration (most commonly observed in the thoracic vertebrae) and the worsening of posture.²⁻⁴

The review summarizes the main characteristics of gigantomastia related to its description and classification, historical background, etiology, clinical presentation, and differential diagnosis. It also delves into the surgical treatment of gigantomastia, a significant aspect of managing this condition. The surgical treatment options include reduction mammoplasty, mastectomy, and breast reconstruction. An updated mini review is included for each section of the manuscript.

Description

As gigantomastia has not been entirely accepted as a disease, its incidence and prevalence across countries are rarely reported. However, it is believed that 1 to 5 % of women are affected by this condition.⁵ In most articles about this disorder, authors argue that gigantomastia (also known as macromastia) is the abnormal development of mammary glands and fatty tissue, leading to physical and psychological complications. These can manifest as problems in interpersonal, sexual, and work relations. An interesting definition of this disorder is one stated by Dafydd et al.,⁶ who reported gigantomastia as an excessive volume of the breast that contributes 3 % or more of the women's body weight. Nevertheless, the term gigantomastia is not globally accepted.⁶

In terms of this paper, gigantomastia will be considered a disproportionate growth of women's breasts that carries associated physical and psychological symptoms and signs interfering with patients' daily life and their relationship with their social environment, and whose development, despite being unclear, could be attributed to hormones and its receptors, genetics, drugs, autoimmune diseases, pregnancy and infections.^{1,7,8}

Classification

Although there is no consensual classification of gigantomastia,⁹ one approach is to divide it into adolescent, gestational, iatrogenic —related to drug consumption—, and idiopathic (subdivided into juvenile and autoimmune).^{8,10} Table 1 shows a proposed contemporary classification of gigantomastia.^{1,8,10-16}

Historical background of gigantomastia

The first documented case of gigantomastia was described in 1648 in Germany by Palmuth. However, there is no information about this doctor, but it seems that this case was reported in the document "Observations medicuarum centinae tres posthumae". Regarding records, the information about the largest excised breast volume is not widely known; however, in two articles, the authors report that it rounds about 16.5-27.5 kg per breast.^{1,8,17}

Table 1
Classification of Gigantomastia

Adolescent	Progressive and slow growth of mammary glands and fatty tissue during adolescent years. Histologic findings in these patients include adipose tissue enlargement and stromal fibrosis; it rarely involves ductal or hyperplasia pseudoangiomatous stromal hyperplasia (PASH).
Gestational	Hypertrophy of the breast is more commonly presented during the first pregnancy. Histologic changes include fibrosis, collagen deposition, PASH, and edema of the interlobular stroma.
Iatrogenic	Mammary tissue hypertrophy is related to a few drugs, such as penicillamine, indinavir, and cortisone.
Idiopathic	Juvenile: Also known as virginal or puberal, juvenile gigantomastia is the most prevalent and commonly presents between 11 and 14 years of age. Autoimmune: This gigantomastia is mainly related to autoimmune abnormalities, such as myasthenia gravis, systemic lupus erythematosus, Graves' disease, etcetera.

Source: own elaboration, based on references 1, 8, 10-16.

Etiology

Even though gigantomastia's etiology is still unclear, some helpful studies deny or accept its correlation with various pathways: the excessive release of hormones (prolactin and estrogen),⁷ high sensitivity of estrogen, prolactin and human chorionic gonadotropin receptors,^{18,19} autoimmune diseases, such as myasthenia gravis; systemic lupus erythematosus; and rheumatoid arthritis,¹ drug consumption, as well as genetic influence. Nevertheless, they have not been tested in large groups of patients, and information about their impact on the gigantomastia pathway is somewhat diverse, but very few studies have already described critical proteins playing a role in the gigantomastia pathway.

Recent analyses explained by Kasielska-Trojan et al.¹⁵ showed that a polymorphism of the "aromatase" protein (allele G in rs749292) increased the risk of developing gigantomastia, in contrast to some polymorphisms of progesterone.^{20,21} Also, Kasielska-Trojan⁹ demonstrated that globally, the expression and the sensibility of estrogen and progesterone receptors in breasts did not have any quantity difference between control and affected women. This closes the possibility of estrogen and progesterone to protagonize the origin of gigantomastia. However, further investigations need to be focused on different polymorphisms of all these proteins to determine if they play or do not play any role in the pathway of this abnormality. In addition, scientists need to focus on the presence of that aromatase allele mentioned in these studies to determine its importance in gigantomastia.

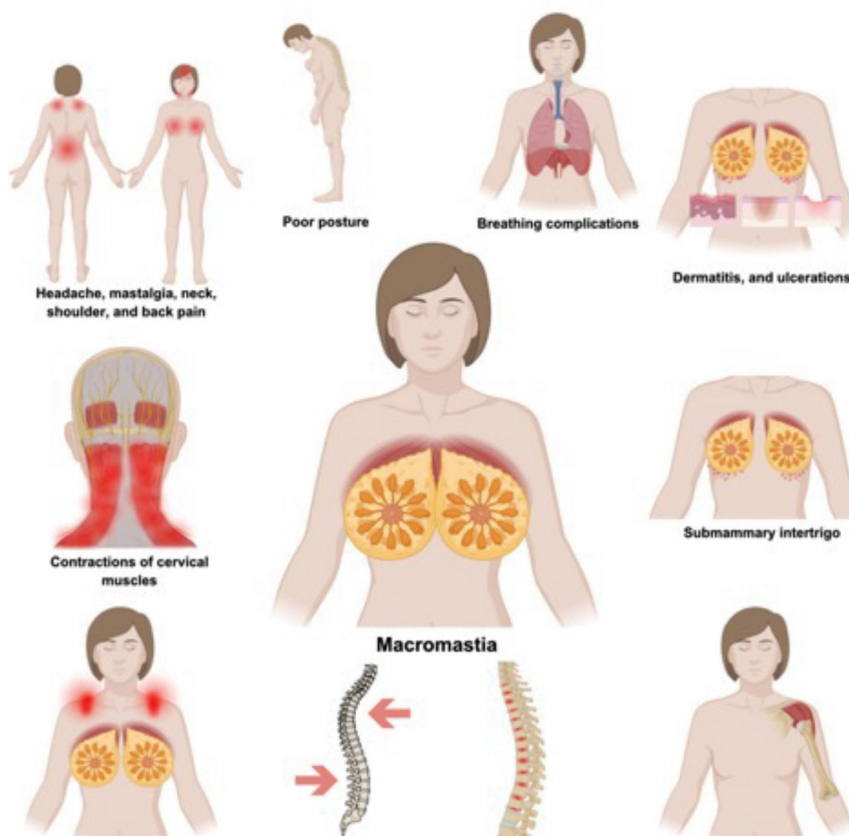
Gigantomastia seems to participate in the pathway of degenerative spondylosis because of adjacent disc injuries.² Those injuries in the vertebral endplates play an essential role in the pathophysiology of discal pain (as they carry some neural structures) because of bone alterations — osteophytes, erosions.^{22,23} The body's center of gravity is altered by moving superiorly and anteriorly during daily activities.²⁴ This alteration leads to constant paraspinal muscle contraction, which results in chronic pain.²⁴ Because of the altered gravity of the body, patients develop an altered curvature that increases lumbar lordosis and thoracic kyphosis.²⁵

Its etiology remains unknown. However, this increase in mammary tissue has genetic and hormonal elements and is mainly related to the patient's age, race, nutrition, and the number of pregnancies.^{1,4,26}

Clinical presentation

Back pain is a significant symptom in women with gigantomastia and also the one that most affects women's day-to-day. Despite being a public health issue, back pain in patients with gigantomastia has not been reported as well. Still, available data show the magnitude of this disturbance, which physicians and health systems should not ignore since most women affected with this pathology present back pain (more importantly, lower back pain) interfering in all

Figure 1
Physical perturbances of Gigantomastia



The physical afflictions of women with gigantomastia mainly affect the musculoskeletal, nervous, and tegumentary systems, resulting in impaired perception and complications when performing their daily lives

Source: own elaboration, based on references 1-3, 9, 24, 29-34.

the spheres of their lives. Clinical and statistical evidence shows that gigantomastia produces degenerative spondylosis by mechanical overload and thoracic kyphosis.^{2,27,28} Consequently, more studies should be done to seek other probable endings of gigantomastia, such as herniations and other spine affections.

Patients with gigantomastia are widely affected in different spheres of their lives, starting from physical and psychological symptoms and concluding with complications in their lives. They suffer from low physical performance, depression, sociophobia, low self-esteem, work absenteeism, uncomfortable feelings about body image and sexual relations, difficulties exercising, and finding well-fitting clothes. Figure 1 exposes some of the most commonly reported physical affections in women with gigantomastia.^{1-3,9,24,29-34}

Imaging assessment in Gigantomastia

Imaging assessment in patients with gigantomastia is essential, as it will provide context to medics to better understand how this excessive spinal load affects axial bone structures. Some degenerative imaging findings reported in women with gigantomastia are osteophytosis, anterior sclerosis of the vertebral plates, osteoarthritis, and narrowing of the disc space.² In the study elaborated by Benditte-Klepetko et al.,³⁵ they found in MRI that an overload due to hypertrophy breast drives degenerative diseases, as well as some alterations that can be evaluated as signs of anterior disc protrusions, narrowing of the disc space, and foraminal stenosis.³⁵ Papanastasiaou²⁴ made a comparison between the different imaging technologies to quantify the pain and discomfort of breast hypertrophy. A study showed a direct correlation between the amount of mammary tissue and the worsening of posture.³⁶ This study was performed by using radiographs.³

Differential diagnosis

Although there have been so many years since gigantomastia was reported for the first time, the cause of this physical condition remains unclear.¹ Then, physicians need to make an accurate differential diagnosis, which must necessarily consider breast masses, breast abscesses, breast cancer, benign breast pathology, and the enlargement of mammary glands as a consequence of pregnancy in women who do not know their pregnant status, as well as others. A prompt differential diagnosis will help physicians to classify patients into their pathologies rapidly and to provide them assertive management, which in the case of women suffering from gigantomastia will be reduction mammoplasty.

Surgical treatment

Breast reduction surgery, also known as reduction mammoplasty, is the most effective treatment for patients with gigantomastia, and its objective is to minimize the impact of the excessive mammary tissue on the vertebral axis at the time of maintaining a pleasant aesthetic.^{2,24,29} Some studies have shown that breast reduction surgery decreases the angles of thoracic kyphosis and lumbar lordosis, mitigates their associated symptoms, and improves women's posture—directly related to the amount of excised breast tissue.^{2,24,37,38} These effects result from reducing the excessive spinal load.³

The main indications for breast reduction surgery are physical pain as well as correcting the asymmetry hyperplasia to achieve symmetry.²⁹ In several countries, reduction mammoplasty is considered an aesthetic procedure.³ However, affected women undergo this surgical operation because of the constant back pain and psychological impact on their lives.^{29,38} It is essential to know that, in the UK, the National Institute for Health and Clinical Excellence (NICE) does not consider body mass index (BMI) as a feasible indication for reducing mammoplasty in patients with back pain.³⁹ That would result from the statement that obese women will not be considered directly affected by

this pathology because of their overweight excess.

Concerning reduction mammoplasty goals, when it comes to patients, it is expected that women excised will feel relieved since their physical symptoms will gradually disappear, improving not just their day-to-day performance but also their self-esteem, because their physical appearance will be different—it is essential to have on mind that as a surgical excised treatment, scars will be present, but surgeons will do the best for patients to avoid significant or hypertrophic scars—, and finally, this will benefit patients to evade further operations.⁴⁰

Before surgeons effectuate a reduction mammoplasty, certain information about the patient must be obtained: a thorough clinical history, general medical status, pregnancy background, mammary gland development, weight changes, smoking history, family history of cancer, and the patient's morphology, such as breast size, the degree of nipple displacement, and the BMI.^{29,40}

Conclusions

As reviewed, gigantomastia brings some consequences, including pathological changes in posture and more serious affectations, such as the progression into degenerative spondylosis. Subsequently, it is necessary to consider the imaging assessment, which could initially include radiographs and, more importantly, MRI, which aims to identify favorable or unfavorable changes in the musculoskeletal structures. Given the current lack of information regarding gigantomastia, it is vital to delve into this disease's mechanisms to start describing its epidemiology, genetics, and variants of clinical presentations. This will not only help physicians understand gigantomastia but also improve the management of the disease and culminate in an improvement in daily women's lives. The need for more information is urgent, and further research is crucial.

Conflict of Interest

Conflict of interest: The authors declare they have no conflict of interest.

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