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CASE REPORT

Carcinoma in Situ of the Breast in a Young Male: Case Report

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ABSTRACT

Introduction: Male breast cancer accounts for 1% of all breast cancers. Within this group, ductal carcinoma in situ represents 5% to 15%, making it an extremely rare entity. The median age of presentation is 60 years, therefore rare at young ages. The causal relationship with mutations in the BRCA 1 and 2 genes is well established. The most frequent clinical presentation is nipple discharge or a retroareolar mass. Histologically, the papillary subtype is the most common, with a high overexpression of hormone receptors and very infrequent HER2 positivity. Gynecomastia may coexist, although it is not considered a direct risk factor.

Clinical case: 16 year old male patient. Presented with bilateral gynecomastia and treated with liposuction. Pathology revealed low-grade ductal carcinoma in situ of the right breast, extensive loss of CK5 marker and almost 100% marking of estrogen receptor, with no invasive component. He was referred to a mastologist, who performed bilateral mastectomy without sentinel lymph node biopsy. Final pathology: residual ductal carcinoma in situ in the context of gynecomastia with associated atypical hyperplasia. No invasive neoplasia or stromal microinvasion. Genetic panel revealed CDK12 gene and NF1 gene mutation.

Discussion: The presentation of ductal carcinoma in situ in men occurs on average 5 years later than in women; its occurrence in young people and adolescents is infrequent and rarely pur but associated with invasive component. Given its low frequency, scientific evidence is limited and there are no specific management guidelines, so the approach is extrapolated from that used in adult women breast cancer. The standard treatment is total mastectomy. Axillary dissection is not indicated, as axillary invasion is exceptional. When the diagnosis is made by biopsy, the risk of underestimation is up to 26%. Endocrine or radiation may be applied to patients at higher risk of invasion. Disease-free survival rate exceeds 90% at 5 years. Local recurrence is infrequent and associated with conservative treatments.

Conclusions: Carcinoma in situ of the breast in men is a very rare entity, generally associated with a genetic mutation. Its treatment is mastectomy. Other therapies should be tailored to each individual case.

Keywords: Male breast cancer, ductal carcinoma in situ, gynecomastia.

Introduction

Breast carcinoma in situ comprises a group of atypical epithelial lesions confined to the mammary lobule or duct, surrounded by a layer of myoepithelial cells, without breaching the basement membrane. It includes several subtypes, the most frequent being ductal carcinoma in situ and lobular carcinoma in situ. Its incidence increased significantly after the expansion of mammography as a screening method[1-2].

In the United States, approximately 59,080 new cases of non-invasive (in situ) breast cancer are estimated each year. This represents approximately 20–25% of cancers detected in mammography screening programs[3].

In Uruguay, in situ tumors account for around 5.3% of all diagnosed breast cancers[4].

In women, ductal carcinoma in situ (DCIS) of the breast is not an obligatory precursor to invasive cancer; if left untreated, 40% of cases will progress to invasive carcinoma[5].

Male breast cancer accounts for less than 1% of all breast cancers worldwide[6], a figure that is also observed in Uruguay[4-7].

Clinically, the most common manifestation is the appearance of a palpable nodule, usually painless, located in the subareolar region. Other signs may include nipple retraction, discharge, skin ulceration, or changes in the skin such as redness or thickening. Due to the smaller volume of breast tissue, the tumor is more likely to infiltrate neighboring structures at early stages, which can delay diagnosis and worsen the prognosis[8-10].

Compared with breast cancer in women, male breast cancer is often diagnosed at more advanced stages. This is mainly due to diagnostic delay, low clinical suspicion, and the absence of specific screening programs. As a result, lymph node involvement and larger tumor size at diagnosis are more frequently observed[8-9]. Additionally, the association with carcinoma in situ (particularly DCIS) is less frequently identified in men than in women, likely due to the limited amount of breast tissue and the tendency for diagnosis at a more advanced stage[8-10].

Carcinoma in situ of the male breast is a rare entity, representing a small proportion of cases within male breast cancer. The most common subtype is DCIS, while lobular carcinoma in situ is extremely rare due to the limited presence of lobules in the male breast. In most cases, carcinoma in situ does not occur in isolation but rather in association with invasive carcinoma; its concomitant presence has been reported in approximately 5–15% of male breast cancer cases. Pure DCIS in men is uncommon and is usually diagnosed incidentally or at earlier stages compared with invasive disease[8-10].

Regarding prognosis, when adjusted for stage and tumor characteristics, survival is similar between men and women. However, overall outcomes tend to be worse in men, largely because they are diagnosed at later stages. Additionally, male breast cancer is more frequently hormone receptor–positive, which allows for effective therapeutic options in many cases, although this does not fully offset the impact of delayed diagnosis[8-10].

Gynecomastia is a benign proliferation of the glandular tissue of the breast in men. It is a common condition that during puberty, can be considered a physiological and usually resolves spontaneously. A hormonal imbalance between estrogens and testosterone is postulated as the cause. Evaluation should be aimed at ruling out underlying causes (paraneoplastic, endocrinopathies, medications and drugs) as well as the presence of other diseases in the breast tissue. Surgical treatment is recommended when gynecomastia does not resolve after treating the underlying cause, or when it does not resolve spontaneously after a period of observation in cases of pubertal gynecomastia[11].

Specific scientific literature on in situ breast cancer in men is scarce. Only one published case of DCIS in adolescence has been reported, coinciding with gynecomastia and treated with subcutaneous mastectomy[12].

We present a similar and rare case, of an adolescent male in whom a pure ductal carcinoma in situ was found in the context of treatment for pubertal gynecomastia.

CASE REPORT:

16-year-old male patient, with a family history of a grandmother who died of breast cancer at the age of 40, and no other significant medical history. Presented with bilateral pubertal grade I gynecomastia, with no other symptoms associated such as nodules or nipple discharge.

On physical examination, he was found to have breast enlargement greater on the right side, with a palpable, well-defined, subareolar disc. No redundant skin.

Breast ultrasound reported: increase in retroareolar soft tissue bilaterally, more prominent on the right side, with features consistent with gynecomastia. No nodules or other abnormalities are identified. Hormonal laboratory tests were within normal limits.

After one year of surveillance with no remision, and considering self - steem repercussion, it was decided to proceed with surgical treatment. Plastic surgeon performed partial excision on the right breast and liposuction of the left breast. Pathology revealed low-grade DCIS of the right breast. He was referred to a mastologist, and underwent completion bilateral subcutaneous mastectomy through bilateral periareolar incision, without sentinel lymph node biopsy. The resected tissue measured 31x21x10 mm on the right side and 25x20x16 mm on the left side.

The final anatomopathological study showed residual DCIS in the context of gynecomastia, with associated atypical hyperplasia. No invasive neoplasia or stromal microinvasion was found (Figure 1). Margins of resection were free of DCIS.

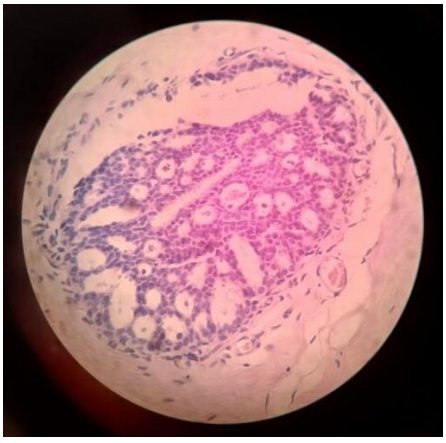

Figure 1: Microscopy, ductal carcinoma in situ, H&E 10x. The duct is expanded by a rigid cribriform structure, composed of cells with monomorphic nuclei, without a "streaming" phenomenon.

Immunohistochemical analysis revealed almost 100% expression of estrogen receptors (ER), and extensive loss of CK5 marker (Figures 2, and 3).

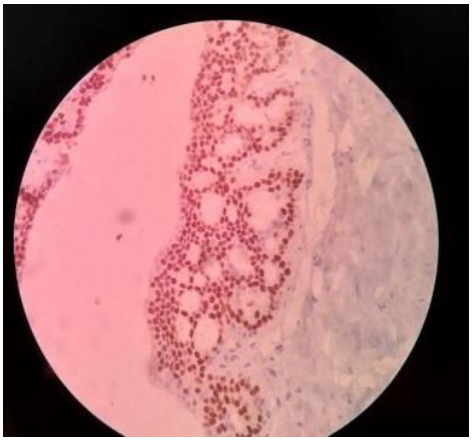

Figure 2: Estrogen Receptors: intense, nearly universal staining.

A genetic panel revealed two variants of uncertain significance:

- CDK12 gene c.4146C>G p.His1382Gin: reported as an autosomal recessive missense mutation. Pathogenic, involved in the development of hereditary breast and prostate cancers.
- NF1 gene c.2188A>T.p.Asn730Tyr: reported as a missense mutation not associated with breast cancer.

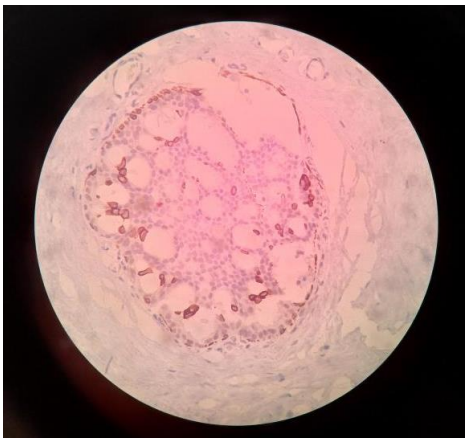

Figure 3: Near total loss of CK5 expression, except in basal cells.

Gene sequencing was performed on the brother and mother to determine whether the mutation is de novo or inherited. The patient's brother presents similar variants in the CDK12 and NF1 genes. The mother does not present pathogenic variants.

Discussed in the tumor board, there were no clear evidence in this case to propose adjuvant treatment, considering the risks and benefits, patient's age, reproductive status, low frequency of the pathology, and the patient's concern about adverse effects. Therefore no adjuvant treatments were performed. Six months after surgery, the patient's course has been favorable, with no evidence of recurrence. Follow-up is carried out jointly with oncology, including a clinical examination every 4 months.

Discussion:

While DCIS is a well-documented entity in women, its low frequency in men makes the available data much more limited for both invasive and in situ carcinoma, with most information coming from case series or isolated reports. The presentation of DCIS in men occurs on average five years later than in women; occurrence in young people and adolescents is infrequent[13-18].

In a French case series, ductal carcinoma in situ in men represented 5% of all male breast cancers. In this series of 31 patients, the median age at

presentation was 58 years; 6 patients were under 40. Eleven patients had gynecomastia, and 10 had a family history of breast cancer[19]. A more recent series from the European Institute of Oncology collected 20 cases of in situ carcinoma in men over 19 years, with a median presentation age of 62 years[20].

Regarding clinical presentation, series report that 55–58% present as a breast mass, and 35–45% with nipple discharge[20-21]. The most common histological subtype is papillary, or intracystic papillary carcinoma. Grade 3 DCIS or comedocarcinoma is rare in men. This is consistent with the fact that men lack mammary lobules, but do have a ductal system where papillary lesions can develop. Regarding molecular subtype, as in the presented case, most show strong estrogen receptor positivity and are HER2 negative[21].

RELATIONSHIP WITH GYNECOMASTIA

The association between gynecomastia and breast cancer remains controversial. They can coexist, although gynecomastia is not considered a direct risk factor. Hormonal imbalances with increased estrogens and reduced androgens, which cause gynecomastia, may play a role in the development of male breast cancer, as may regulation of aromatase expression[22]. Several studies have attempted to measure this association. In a series of 268 patients who underwent mastectomy for gynecomastia, pathology revealed a single case of unilateral DCIS (0.4%), and no cases of cancer. The authors concluded that, although proliferative lesions are rare, histopathological analysis should not be omitted in these patients[23]. Another series in Saudi Arabia found 5 cases of DCIS in 74 patients operated on for gynecomastia between ages 17 and 22 years. In the IEO case series, 25% of patients had gynecomastia[24].

GENES DETECTED

The causal relationship with mutations in the BRCA 1 and 2 genes is well established.

In the presented case, a CDK12 gene mutation was found. Unlike classic predisposition genes such as BRCA1/2, germline mutations in CDK12 seem to have limited impact on breast cancer risk; however, somatic alterations—including loss of function, overexpression, or co-amplification with HER2—have greater biological and clinical significance. CDK12 dysfunction is associated with genomic instability and impaired DNA repair pathways, which may confer sensitivity to targeted therapies such as PARP inhibitors and platinum-based agents. On the other hand, its overexpression has been linked to increased tumor aggressiveness, activation of oncogenic pathways, and resistance to anti-HER2 therapies, particularly trastuzumab[25-29].

This patient was also found to have an NF1 (neurofibromatosis type 1) gene mutation: pathogenic germline variants in NF1 are associated with the autosomal dominant disorder neurofibromatosis type 1, with an incidence of 1 in 2,700. Its characteristic phenotype includes café-au-lait macules. It predisposes to neurofibromas and other tumors, and is associated with up to a 20% risk of breast cancer, especially before age 50[30].

TREATMENT

Mastectomy is the treatment of choice for DCIS. Since the male breast has no function and the psychological impact of losing the nipple-areola complex is minimal for men, mastectomy is an accessible option. Regarding histology, Hittmar argues for radical resection, because papillary lesions can extend intraductally beyond what is visible macroscopically[21].

Axillary lymphadenectomy is not indicated for pure DCIS, given the very low risk of axillary metastasis. However, Cutuli recommends it in comedocarcinoma-type lesions or those larger than 2.5 cm, where the risk of invasion is higher[19]. Pappo and colleagues recommend sentinel lymph node biopsy (SLNB) for extensive DCIS presenting as a mass, high-grade lesions, or those containing areas of microinvasion, similar to recommendations for women[31]. In the IEO series, 13 of 20 patients underwent SLNB; no axillary lymphadenectomy was performed. There were no cases of axillary metastasis[20].

When diagnosis is obtained by biopsy, the risk of underestimation is up to 26%; thus, the IEO group recommends SLNB at the time of mastectomy[20]. In this case, the histological diagnosis was made from a partial resection, which we believe minimizes the risk of underestimation.

Endocrine or radiation therapy is not usually recommended, although, after tumor board discussion, it could be considered for higher-risk patients (intraluminal necrosis or high Ki). The disease-free survival rate exceeds 90% at 5 years. Local recurrence is rare and associated with conservative treatments. Recurrences are linked to local resection[20].

Annual follow-up with mammography is suggested, similar to what is done in women.

Conclusions

Pure ductal carcinoma in situ in men is a very rare entity, especially in the context of gynecomastia and at a young age. Therefore, information on its management is limited and not standardized. We present this clinical case to help provide information that may assist in standardizing the management of this pathology.

Conflict of Interest Statement:

None.

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