Tuberous breast and predisposition to breast deformity in consanguineous

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Abstract
Tuberous breast deformity is a pathologic condition of the breast consisting of a constricting ring at the breast base, reduction in the volume of the breast parenchyma, and herniation of breast tissue through the nipple-areola complex with areola enlargement. This pathology is generally congenital and has an unknown etiopathogenesis. We report the first observation of tuberous breast deformity in consanguineous. This report suggests the potential role of a genetic base in the development of this deformity. Between May 2008 and March 2011, we observed six female patients from two different families, aged between 18 and 55 years, affected by tuberous breast deformity. The breast deformity was characterized by breast asymmetry in all six cases. Four patients underwent surgery to correct the deformity. Standardized objective measurements of breast and chest were taken. A Visual Analog Scale was used to evaluate patients’ and physicians’ satisfaction. The first three patients were consanguineous; two were first cousins, and the third was second cousin with one of the above. The other three patients were also from the same family: two sisters and their mother. According to Von Heimburg’s classification, the patients presented different degrees of breast deformity. In all operated cases, a good esthetic result with a high satisfaction (average visual analog scale score 9) was achieved. The results remained stable over time and no revisions were needed after the 1-year follow-up. The possibility of a parental consanguinity for breast deformities such tuberous breast has never been described in the literature. This report suggests the possible genetic role in the development of tuberous breast deformity. Further studies and genetic tests are required to prove this hypothesis.

KEYWORDS
breast deformity, familiarity, predisposition, tuberous breast

1 | INTRODUCTION

Tuberous breast deformity is a rare anomaly of breast development presenting at puberty with a noncomplete enlargement of the breast during its growth.1 This malformation was first reported by Rees and Aston in 1976, and it owes its name to the breast shape resemblance to a tuberous plant root.2 The characteristics of the tuberous breast deformity consist of a constricting ring at the breast base with minimal breast tissue that herniates through the nipple-areola complex (NAC) with areola enlargement, sagging, and associated asymmetry. It may affect just one side (unilateral) or both sides (bilateral).3

The etiology of tuberous breast remains much debated. Embryological explanations were suggested by Glaesmer, that the
deformity could correspond to a phylogenetic relapse because of the similarity of the tuberous breast to animal teats.\(^4\) Another hypothesis is an anomaly of the ensheathing structures. Some authors suggested that the tuberous breast deformity results from the strong adherence of the dermis and muscular plane occurring in puberty at the level of the superficial fascia in the lower pole of the breast. This inhibits the peripheral growth of the breast, which then expands forward, producing the tubular appearance of the breast.\(^5,6\)

Mandrekas et al. described the anatomy and embryology of the breast and suggested a possible mechanism for the development of tuberous breast.\(^7\) They pointed out that the absence of the superficial layer of the superficial fascia in the area underneath the areola may explain the invagination of the breast tissue. According to their theory, the presence of a constricting fibrous ring representing a thickening of the superficial fascia at the periphery of the NAC prevents the expansion of the breast at the lower pole and causes the breast parenchyma to herniate into the NAC.

However, despite the presence of such a constricting ring has been confirmed by histological findings,\(^8\) the authors acknowledge that their theory is pure speculation and that it is impossible to predict whether these breasts would have undergone a normal development if this fibrous ring had not been present. In addition, a recent paper by Pacifico and Kang discarded this theory and explained the etiology of the tuberous breast in view of a transient decrease in NAC skin thickness during puberty.\(^9\)

Most authors agree on the observation of a pubertal hormonal imbalance, when estrogen spurt leads to the development of secondary sexual characteristics, including the breast bud with forward growth that projects the areola forward and peripheral growth that divides the superficial fascia and enlarges the breast base.\(^10,11\) It has been suggested that the hormonal stimulus may be different in the lower quadrants which are the most affected as they have less glandular tissue and less hormonal receptors.\(^6\)

Over the past decades, several classification systems have been proposed for the tuberous breast deformity, the most accepted being Von Heimburg’s and Grolleau’s.\(^5,12\) According to the former, the deformity can be classified into four types: Type I - hypoplasia of the lower medial quadrant; Type II - hypoplasia of the lower medial and lateral quadrants; Type III - hypoplasia of the lower medial and lateral quadrants, insufficient skin in the sub-areolar region; Type IV - severe breast constriction, minimal breast base. The latter identifies only three types, without making a distinction between type II and III in terms of skin deficiency in the sub-areolar region.

With reference to the aspect of the areolar skin in patients with tuberous breast, it has been argued that it may be explained by an underlying abnormality in skin thickness, although this remains unproven. With the breast growth at puberty, the enlarging tissue pushes through the area of weakness causing the areolar skin to widen. The reasons of areolar skin lessening remain unidentified. Possibly, they are congenital or they occur only transitorily at puberty as a consequence of hormonal imbalance.

The exact incidence of the tuberous breast is probably underestimated as numerous women with mild degrees of deformity may not seek consultation or even be conscious of the deformity.

A recent study suggested a higher incidence of tuberous breast deformity in women presenting breast asymmetry.\(^13\)

In our report, the observation of consanguinity in patients affected by tuberous breast highlights the potential role of genetic transmission of some unknown character responsible of the development of tuberous breast.

### 2 | MATERIAL AND METHODS

In this study, all patients affected by tuberous breasts observed in our Department between May 2008 and March 2011 were checked for consanguinity. In case of a positive correlation, a family tree was obtained.

Clinically, the breast deformity was classified according to Von Heimburg’s classification. Volume asymmetry was assessed by using the statistical analysis of objective breast and chest measurements.\(^14\)

The technique used to correct the deformity was the technique described by Muti, transferring glandular tissue from the upper glandular quadrants to the inferior ones.\(^15\)

Four patients underwent surgery to correct the deformity. The other two patients, namely the sister and the mother of the fourth patient, did not want to be operated on.

Standard digital photograms (frontal, oblique, and lateral views) were taken during follow-up visits, at 1, 3, 6, and 12 months after surgery.

Patients subjectively evaluated breast appearance 1 year after surgery. Their global impression of morphological improvements was recorded using a visual analog scale (VAS), based on a score from 1 to 10 (1=no correction of breast asymmetry, 10=no residual difference between breasts).

In addition, the degree of breast symmetry achievement was evaluated by an external panel of three physicians 1 year after surgery.
surgery on a subjective impression, using the same VAS by comparing preoperative and postoperative photograms.

3 | RESULTS

We observed six female patients with positive family history for tuberous breast deformity, aged between 18 and 55 years, affected by this malformation. The first three patients were consanguineous: two were first cousins (Figures 1 and 2), and the third was second cousin with one of the above (Figure 3). The other three patients were also consanguineous: two sisters (Figures 4 and 5), and their mother (Figure 6). Clinically, the breast deformity was characterized in all patients by similar features: breast asymmetry with volume discrepancy and different grade of tuberous breast. Volume discrepancy

**FIGURE 2** Preoperative appearance of a 30-year-old female patient (first grade cousin of patient in Figure 1) with bilateral tuberous breast deformity of type 2 in the right breast and type 1 in the left breast [Color figure can be viewed at wileyonlinelibrary.com]

**FIGURE 3** Preoperative appearance of a 28-year-old female patient (second grade cousin of patients in Figures 1 and 2) with bilateral tuberous breast deformity of type 2 in the right breast and type 1 in the left breast [Color figure can be viewed at wileyonlinelibrary.com]

**FIGURE 4** Preoperative appearance of a 26-year-old female patient with bilateral tuberous breast deformity of type 4 in both breasts [Color figure can be viewed at wileyonlinelibrary.com]

**FIGURE 5** Preoperative appearance of a 18-year-old female patient (sister of patient in Figure 4) with bilateral tuberous breast deformity of type 1 in both breasts [Color figure can be viewed at wileyonlinelibrary.com]

**FIGURE 6** Preoperative appearance of a 55-year-old female patient (mother of patients in Figures 4 and 5) with bilateral tuberous breast deformity of type 2 in the right breast and type 1 in the left breast [Color figure can be viewed at wileyonlinelibrary.com]
ranged between 30 and 70 cc. According to Von Heimburg’s classification, the patients presented different types of deformity (Table 1).

Surgical correction of tuberous breast deformity was achieved in all four cases. The surgical procedure took an average of two hours (time range: 1.40-2.20 hours).

The operations resulted in a normal size of the areola and in a natural breast shape. No residual evidence of tuberous deformity was observed.

During follow-up, no complications were encountered, nor revisions have been needed.

According to the VAS, good to high patient satisfaction was observed (VAS score ranging from 7 to 10, average VAS score 9), which was confirmed by the impressions of the physicians’ panel (VAS score ranging from 7 to 9, average VAS score 8).

4 | DISCUSSION

Thus far, the relationship between tuberous breast deformity and genetic predisposition has never been reported in the literature. The incidence of this deformity is usually reported to be sporadic with little risk of occurrence in relatives.16,17

The observation of consanguinity among patients affected by tuberous breast let us suppose the possible role of genetic transmission of this deformity. To date, no etiologic theory for the development of tuberous breast has been proved.18 We believe that focusing on the possible role of a genetic predisposition could help understanding a potential etiologic factor of this deformity.

Further observations of consanguinity among patients affected by tuberous breast deformity and eventual genetic studies are required to support this theory.

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