Bilateral Familial Polythelia Without Associated Malformations

Polietelia bilateral familiar sin malformaciones asociadas

To the Editor:

The presence of supernumerary nipples not associated with other anatomical breast structures (polythelia) is the most common malformation of accessory mammary tissue.\(^1\) As well as the nipples that normally develop on the thorax, cases have also been reported of additional nipples, areolas, mammary glandular tissue (polymastia), or any combination of these.\(^2,4\) The supernumerary elements are normally found along the mammary lines—imaginary lines running from the axillae to the groin.\(^3\) Most cases of polythelia are sporadic, although several heredity patterns have been reported.\(^5-7\) Furthermore, possible associations have been sought between polythelia and different congenital malformations, particularly malformations of the kidneys and the urinary tract.\(^4,8-10\) However, reports of familial bilateral supernumerary nipples at sites other than the mammary lines, with no underlying mammary tissue, and not associated with congenital malformations, are very rare in the literature.\(^11\)

We report the case of a 53-year-old woman who was allergic to sulfamides, had never been pregnant, and who had been diagnosed with well-differentiated intraductal carcinoma (ductal intraepithelial neoplasia stage 1c) of the left breast in 2002; the carcinoma was treated by lumpectomy, local radiation therapy, and oral tamoxifen. The patient presented no facial, cranial, palatine, ocular, or limb malformations, and no systemic symptoms. When questioned regarding her family history, the patient stated that other members of her family had bilateral supernumerary nipples: mother (I:1), 6 sisters (II:1, II:2, II:5, II:7, II:8, and II:10), and a niece (III:1) (see family tree in Figure 3). She was unaware of supernumerary nipples in the grandparents, uncles and aunts, and other nieces and nephews.

Computed tomography of the cervical, thoracic, abdominal, and pelvic regions was performed, together with an ultrasound scan of the areas on which the nipples were located; no underlying mammary tissue or associated malformations were observed. A chest x-ray, skeletal x-rays, abdominal and gynecological ultrasound scans were also performed with negative results.

The terms polythelia and polymastia are used to refer to the presence of multiple nipples and mammary glands, respectively, and are considered to be examples of atavism.\(^1\) They form part of a broad clinical spectrum, depending on which of the different anatomical structures are present: breast, nipple, areola, glandular parenchyma, or a combination.\(^4\) Nevertheless, polythelia is much more common than polymastia and is therefore considered to be the more frequent malformation of accessory mammary tissue; the incidence varies between 0.22% and 5.6% of the general population, depending on different factors (sex, ethnic group, and geographic region).\(^12\) Most cases of polythelia are sporadic, although as many as 4 forms of genetic transmission have been described\(^13,15-17\), autosomal dominant with incomplete penetrance, X-linked dominant, autosomal recessive, and paradigmont. The supernumerary nipples are most commonly found on the upper third of the mammary line, which is an imaginary line running from the axilla to the groin.\(^3\) Bilateral polythelia has been reported but is less common than the unilateral presentation.\(^6,5,12\)

Supernumerary nipples are mainly an esthetic problem of little medical significance. However, some cases of polythelia respond to hormonal changes in the same way as normal mammary tissue and are susceptible to diseases such as mastitis, cysts, abscesses, and cancer.\(^4\) Also reported in the literature is a considerable number of associations between polythelia and an increased risk of systemic diseases including cardiac arrhythmias, pyloric stenosis, epilepsy,\(^2\) Char syndrome,\(^16\) Hailey-Hailey disease,\(^7\) and different types of cancer (testicular, prostate, vesical,\(^9\) and renal cancer\(^17\)). However, most of the associations reported refer to the relationship between polythelia and the menstrual cycle. Furthermore, the patient presented...
congenital renal and urinary tract malformations, such as adult autosomal dominant polycystic disease, unilateral renal agenesis, ectopic kidney, cystic renal dysplasia, and congenital dysplasia of the pelviureteric junction.\textsuperscript{4,6,8-10}

The case of our patient is notable for the presence of bilateral familial supernumerary nipples at a distance from the typical location and apparently with no associated systemic disease or congenital malformation. Nevertheless, when polythelia is detected, a detailed family history should be taken, a careful physical examination should be performed, and a complete investigation should be undertaken using imaging studies to detect possible associated malformations and diseases.

**Conflicts of Interest**

The authors declare that they have no conflicts of interest.

**References**


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