



Case Report

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Mammary-Type Tissues in the Labia Majora: Histological, Clinical, and Pathophysiological Insights: A Case Report



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Abstract

Background: Mammary-type tissues in the labia majora represent a rare manifestation of ectopic breast tissue, likely arising from residual mammary ridges. These tissues can undergo physiological and pathological changes similar to those of orthotopic mammary glands.

Case: We report a case of a 20-year-old woman with a chromosomal duplication who presented antenatally with a large right labial mass. The mass enlarged during pregnancy, prompting elective caesarean delivery and biopsy. Histopathology confirmed benign mammary-type tissue with lactational changes. Postpartum resolution was observed, and she remains under multidisciplinary follow-up.

Conclusion: This case underscores the importance of considering mammary-type tissue in the differential diagnosis of vulvar masses, particularly during pregnancy. Given their potential for neoplastic transformation, careful histopathological assessment and coordinated interdisciplinary management are essential.

Keywords: Mammary-type tissues; Histopathological assessment; Pathological changes; Radiological assessments; Neoplastic transformation; Physiological

Introduction

Mammary-type tissues or ectopic breast tissue of the labia majora are clinically significant, though rare, and likely arise from disrupted embryological, hormonal, and histological development. Since 1872, fewer than 40 cases of congenital, ectopic mammary tissues in the labia majora have been studied [1]. Many of these tissues exhibit functional and morphological similarity to orthotopic mammary glands, undergoing comparable physiological and pathological transformations. Their existence highlights developmental remnants of the mammary ridge, also known as the 'milk line', a ridge of ectoderm that extends from the axilla to the groin [2,3].

Ectopic mammary-like glands, particularly within the interlabial sulcus between the labia majora and minora, may often remain asymptomatic. Physiological changes during pregnancy or lactation often bring them to clinical attention [3]. They become

clinically apparent due to hypertrophic, secretory, and sometimes pathological changes under reproductive hormone influence in the ante- and postpartum period [3].

Ectopic mammary-like gland proliferation in the vulva is histologically heterogeneous, varying from simple tubular duct structures to more complex lobular arrangements reminiscent of mammary tissue [4,5]. Classification can be supported immunohistochemically using markers such as GCDFP-15 [4]. These structures are thought to arise from embryonic precursors of the milk line, although some propose an apocrine origin, as they may exhibit morphological, hormonal, and neoplastic features analogous to those of mammary gland tissue. Their specific developmental origin, however, remains unclear [6]. Clinical presentations of such tumors warrant serial histopathological and radiological assessments [2,3,7] for accurate diagnosis.

Case Report

We highlight the case of a 20-year-old woman presenting with a large vulval mass identified antenatally, which resolved in the postpartum period. She was known to have a chromosomal duplication (1q34q44) and had been socially isolated, living independently with disability support. Her history was notable for intellectual disability, speech impediment, dyslexia, and gestational thrombocytopenia.

She received shared antenatal care through Mildura Base Hospital and was referred at 36 weeks for review of a newly diagnosed vulval mass. Initially appearing small and insignificant, it grew by 38 weeks to nearly 10cm. She remained asymptomatic. Perineal ultrasound demonstrated an irregular, predominantly firm mass with increased vascularity, anterior to the anal opening. It was considered unlikely to be a Bartholin's cyst or a drainable abscess. Given its growth and the risk of complication during vaginal delivery, a collaborative decision was made for elective lower uterine segment delivery at 39 weeks, combined with diagnostic surgical evaluation.

At Caesarian section, she delivered a live female infant of 3590g (60th percentile) with an estimated blood loss of 750ml. The right labial mass was approximately 8-10cm in diameter, firm and non-fluctuant, without apparent fluid content. Vaginal and rectal mucosa appeared normal and were not tethered or confluent with the mass. A small incisional biopsy was performed by the general surgical team, with formal resection deferred pending histopathology.

Postoperatively, tumor markers showed elevated CA-125 (77U/ml) and normal CEA (0.9U/mL) and CA 19-9 (5U/mL). Pelvic MRI of the pelvis demonstrated a 6.7cm well-defined mixed solid-cystic mass arising from the posterior aspect of the right labia majora, inconsistent with a labial hematoma and without inguinal lymphadenopathy. A contrast-enhanced MRI was declined to allow breastfeeding.

Histopathology reported a "dermoid proliferation of naevoid cells with nests and cords of bland epithelioid cells without significant mitotic activity, with marked maturation with depth and no evidence of malignancy". Notable were proliferated glands lined with vacuolated epithelial cells surrounded by myoepithelial cells and larger ductular structures lacking mitotic activity. Immunohistochemistry was positive for cytokeratins AE1/AE3, and transcription factor GATA3 (commonly expressed in breast epithelium and urothelium), alongside p63 and SOX10 staining typical of myoepithelial cells. This confirmed benign mammary-type tissue with lactational changes.

The core biopsy site later became infected, associated with erythema and drainage of pus. Swabs grew enteric flora and *Pseudomonas aeruginosa*; treatment involved removal of sutures and wound care.

The patient was referred to the Royal Women's Hospital in Melbourne for further evaluation by gynecological oncologists. She chose to stop breastfeeding, and further assessment was planned after lactational involution. Psychological support and ongoing monitoring were arranged. At last contact, the mass had resolved and was no longer palpable.

Discussion

The labia majora, a functionally dynamic part of the external genitalia, responds to physiological stimuli such as age, hormonal fluctuations, and parity. In reproductive periods, particularly lactation, hypertrophy and hypervascularisation of mammary-type tissue can occur in response to prolactin and oxytocin [3,8].

Mammary-like tissue masses in the labia majora may present as asymptomatic nodules or cause pain and progressive enlargement, typically during pregnancy or lactation. Although generally benign, they may undergo changes resembling those seen in hidradenoma papilliferum, Bartholin's cysts, or other neoplastic disorders of the vulva [8]. Published literature reports the specialisation of these tumors, typical of breast tissues, including fibroadenomas [9], as well as neoplastic transformations into ductal carcinomas and adenoid cystic carcinomas [4,10-12].

A comparable case study by Sharma et al. [13] describes a 24-year-old primigravid woman with a vulval mass at 14 weeks identified as a lactating adenoma. They noted an incidence of ectopic breast tissue of 2-6%, often diagnosed during pregnancy as labial enlargement. Complete excision is recommended to reduce recurrence and mitigate malignant risk, supported by broader literature on the neoplastic transformations of these tissues [14]. Thus, symptomatic or enlarging lesions require excisional biopsy and interdisciplinary management involving gynecologists, pathologists, and oncologists [3,9].

This case underscores the importance of considering ectopic, mammary-like tissue in the differential diagnosis of vulvar masses, especially during pregnancy. Prompt recognition helps avoid misdiagnosis, enables appropriate interdisciplinary planning, and ultimately supports safer maternal and neonatal outcomes. Immunohistochemistry remains a cornerstone for differentiating mammary-type tumors from other vulvar neoplasms employing markers such as cytokeratins, estrogen, and progesterone receptors [2]. Notably, positivity for both cytokeratins and GATA3, as observed in our case and others [15], highlights the breast-like differentiation of these tissues [16,17].

Conclusion

Mammary-type tissues in the labia majora, though rare, represent a clinically significant anatomical variant. Their dynamic responsiveness to hormonal stimuli, potential for neoplastic change, and implications for vulvar health highlight the importance of multidisciplinary care. In our case, additional complexities includ-

ed multidisciplinary planning to support informed decision-making and postpartum care, given the patient's intellectual disability and social circumstances. Future research focusing on genetic and molecular profiling may help refine diagnostic criteria and clinical management and shed light on whether chromosomal anomalies may influence ectopic mammary tissue development.

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