

Case Report

Breast granular cell tumour: a case report and literature review

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ABSTRACT

Granular cell tumour of the breast, being a rare disease entity poses diagnostic challenges as it often mimics carcinoma. The mainstay of treatment for GCT is wide local excision and prognosis is good with negative margin. We hereby report a case of breast granular cell tumour presenting as a palpable breast lump with positive margin involvement after wide local excision, managed by margin re-excision. We also review existing literature on the clinical presentation, histopathological and radiological description and the of the disease.

Keywords: Breast granular cell tumour, Wide local excision, Margin involvement, Margin re-excision

INTRODUCTION

Granular cell tumour (GCT) of the breast is a rare disease entity with a prevalence of 6.7 :1000 cases of breast malignancy.¹ Despite being a largely benign tumour, GCT poses diagnostic challenges as it often mimics carcinoma clinically, radiologically and microscopically.² The mainstay of treatment for GCT is wide local excision (WLE) and prognosis is good with negative margin.³ We hereby report a case of breast granular cell tumour (BGCT) with positive margin involvement on WLE managed by margin re-excision.

CASE REPORT

A 40-year-old lady was referred to our clinic for a self-detected right breast mass. She first noted the mass 1 year ago which progressively increased in size. She had no personal or family history of malignancy and enjoyed good past health apart from an episode of gestational diabetes mellitus. Physical examination showed a 2 cm hard mass with skin tethering at the inferomedial aspect of the right breast. The mass was not attached to

underlying muscle and there was no nipple retraction or palpable axillary lymph node. The mammogram showed a superficial density in the lower part of right breast with focal skin thickening. Supplementary ultrasound breasts showed an oval shaped hypoechoic lesion (~2×0.9×1.3 cm in size) at the cutaneous and subcutaneous interface in R5H ~6 cm from nipple. There was no abnormal internal vascularity. Imaging concluded that the lesion was likely skin appendage in nature. Ultrasound-guided core biopsy was performed under local anaesthesia.

Histological evaluation demonstrated syncytial nests of polygonal tumour cells with abundant granular eosinophilic cytoplasm and small uniform round nuclei with inconspicuous nucleoli. There were no atypical or malignant features such as tumour cell spindling, marked nuclear pleomorphism, prominent nucleoli, mitosis or necrosis. Immunohistochemically, the tumour cells were positive for S100 and CD68 whilst negative for cytokeratin. Findings were conclusive of granular cell tumour. Wide local excision of the tumour was performed with 1cm margin circumferentially and

posteriorly in July 2023. Specimen ultrasound showed clear gross margin.



Figure 1: BGCT presenting as a superficial density with focal skin thickening in the lower part of right breast on MLO view of mammogram.

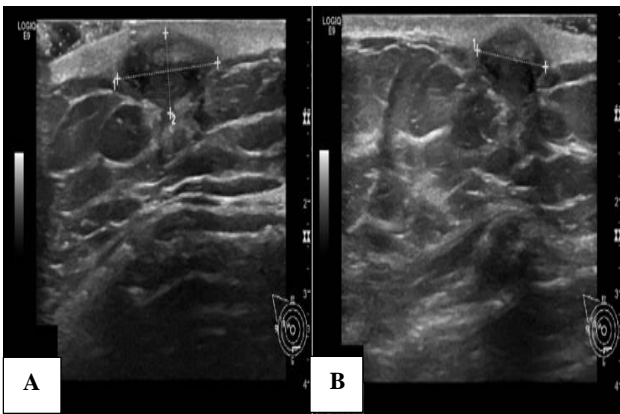


Figure 2: (A) BGCT presenting as a ~2 cm oval shaped hypoechoic lesion at the cutaneous and (B) subcutaneous interface in R5H ~6 cm from nipple.

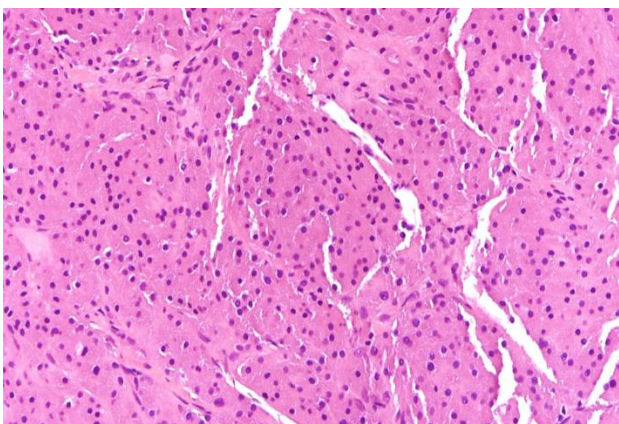


Figure 3: Core needle biopsy showing nests of polygonal cells with small centrally located nuclei and abundant, eosinophilic, granular cytoplasm. Hematoxylin and eosin staining, 200X.

Macroscopic evaluation showed a 3.8 cm infiltrating cream-coloured solid tumour in the dermis/ breast tissue abutting the superior and inferior skin margins. Microscopically, the tumour has an ill-defined infiltrating border. It is comprised of the same nests of large bland polygonal cells with abundant granular eosinophilic cytoplasm seen in the core biopsy. No malignant feature is present. It is present at the superior margin while the rest of the margins are clear. The final pathology confirmed the diagnosis of granular cell tumour.

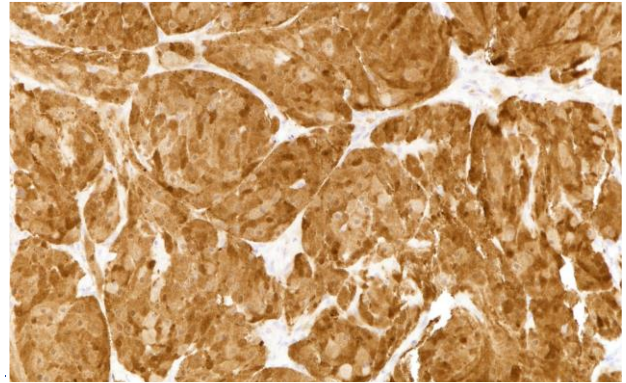


Figure 4: Tumour cells with strong and diffuse positivity for S100, 200X.

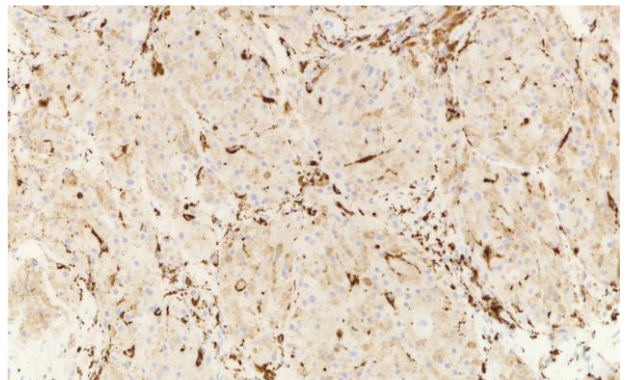


Figure 5: Tumour cells positive for CD68, 200X.

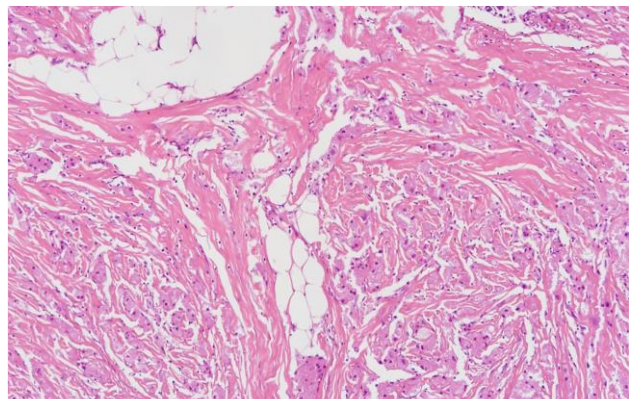


Figure 6: Excision specimen showing the tumour with infiltrating growth pattern. Hematoxylin and eosin staining, 100X.

Re-excision of superior margin was offered and accepted by patient, and was performed in August 2023 under local anaesthesia. Elliptical incision over the scar with more skin over cranial aspect of the wound was included, with full skin thickness and underlying scar tissue excised en-bloc. Final pathology showed scar with foreign body reaction and no residual tumor. Postoperative course was uneventful and last follow-up visit at 2 months showed no signs of recurrence.

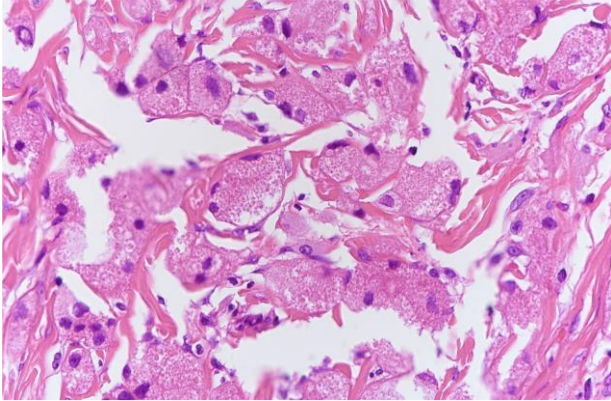


Figure 7: Excision specimen showing infiltrating polygonal tumour cells with small nuclei and abundant granular eosinophilic cytoplasm. Hematoxylin and eosin, 400X.

DISCUSSION

GCTs are soft tissue tumours arising from Schwann cells and they can appear at any part of the body.² BGCT is a rare condition which contributes to 5-15% of all GCTs.¹ The majority of BGCTs were benign and malignant characteristics were only detected in 1-2% of the disease population.¹ Clinical presentation of BGCTs is highly variable, while most palpable tumours were described to be firm, painless and mobile, some could present as the opposite. There could also be skin involvement such as thickening, retraction, dimpling or tethering as seen in our case. Associated lymphadenopathy is uncommon.¹ Imaging provides limited diagnostic value in BGCTs as they are often indistinguishable from malignant lesions. Mammographic features of BGCTs include irregular margins, spiculation, stellation and isodensity while microcalcifications are not typical.² On ultrasound, the appearance of BGCTs varies with degree of tumour infiltration and reactive fibrosis. They can present as hypoechoic to anechoic mass with intense posterior shadowing. Their margins are usually spiculated, angular or indistinct while some are circumscribed.⁴ BGCTs have vastly varying presentation on both mammogram and ultrasound, rendering radiological diagnosis difficult. As seen in our case, the patient presented with a breast lump resembling skin appendage on imaging rather than the classical description of malignant looking lesion. According to literature, MRI may be useful in determining the extent of disease and presence of

aggressive features, however, no specific characteristics of BGCTs have been identified.¹

Histopathological analysis along with immunohistochemical staining is cardinal in the diagnosis of BGCTs. Macroscopically, BGCTs are ill-circumscribed and have an infiltrative border, attributing to their resemblance to carcinomas. Microscopically, they have an infiltrating growth pattern with tumour cells arranged in sheets, cords or nests surrounded by sclerotic stroma. The tumour cells are large, polygonal to spindle in shape, characterised by abundant granular eosinophilic cytoplasm and small nuclei.⁴ They stain strongly for S100 and CD68 on immunohistochemical study. S100 is a protein found in neural cells, Schwann cells and melanocytes. It is a sensitive but non-specific marker for BGCT as 10% of breast malignancies are also S100 positive.

CD68 highlights the abundant phagolysosomes in the tumour cytoplasm in 90% of cases. BGCTs are negative for cytokeratins which are typically positive in breast carcinomas. Combinatory analysis of S100, CD68 and cytokeratin can help distinguish BGCTs from breast carcinomas.^{1,5,6} The mainstay of treatment for BGCTs is wide local excision. Surgical excision with negative margins is considered curative and is generally associated with excellent prognosis.⁷ However, in case of margin involvement, there has been conflicting opinion on margin re-excision. Papalas et al reported that even in case of positive margin or close margin on excision, recurrence is rare and hence do not warrant for margin re-excision.¹⁰ However, in a study conducted in 2018, Corso et al verified a case of local recurrence in a patient with positive margins at the first histopathological analysis at 2 year follow up.³ Similar results had been reported by Moubarki et al on examining 42 cases in their series and another 1499 cases reported in literature, where 20% of the cases with positive margins developed local recurrence.⁸ In our case, we offered re-excision of margin in view of possibility of local recurrence. Long term follow-up is recommended as 8% local recurrence rate in 8-10 years post excision has been reported.^{3,9} However, the modality of surveillance has yet to be standardised. Ghannam et al have suggested annual follow-up with clinical breast examination and mammography as the majority of BGCTs present as palpable masses that are usually well seen mammographically.⁴

CONCLUSION

BGCT is a rare soft tissue of the breast which can be difficult to diagnose due to its resemblance to malignant lesions on clinical presentation and imaging. Histological analysis along with immunohistochemical staining are essential for its diagnosis. BGCT is primarily treated with wide local excision, but its management remains controversial in case of margin involvement. Further study with long term follow-up would be helpful to assess the risk of recurrence.

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