CASE REPORT

Coexistence of giant cell fibroblastoma and encephalocele

Nishat Afroz,¹ Nida Shamim,² Anshu Jain,¹ Mayank Soni¹

¹Department of Pathology, Jawaharlal Nehru Medical College, Aligarh, Uttar Pradesh, India
²Jawaharlal Nehru Medical College, Aligarh, Uttar Pradesh, India

Correspondence to

Dr Nida Shamim,
 nidasham75@gmail.com

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SUMMARY

Giant cell fibroblastoma (GCF) is a rare soft tissue tumour that occurs almost exclusively in children younger than 10 years of age and is mostly located in the superficial soft tissues of the back and thighs. We present a rare case of GCF with encephalocele in a 1.5-year-old boy who presented with a swelling in the occipital area of the scalp since birth. CT scan suggested encephalocele without any suspicion of a mass lesion. On histopathology, an ill-defined proliferation of fibroblasts in a heavily collagenised and focally myxoid stroma was seen containing numerous multinucleated cells having a floret-like appearance along with mature glial tissue bordering a cystic space. Immunohistochemically, the stromal cells were positive for both, vimentin (diffuse) and CD34 (focal) thereby confirming the histological diagnosis of GCF. This case highlights the unusual coexistence of GCF with congenital defects and its histogenetic resemblance to dermatofibrosarcoma protuberans.

BACKGROUND

Giant cell fibroblastoma (GCF) is a rare soft tissue tumour that occurs almost exclusively in children younger than 10 years of age and exceptionally in adults.¹ The lesions are mostly located in the superficial soft tissues of the back and thighs. Rarely it may present in the head and neck region, however, till date only one case of encephalocele with an area mimicking GCF has been reported.² Encephalocele results from a bony defect in the skull table, which allows herniation of the meninges or brain tissue. We report a rare case of GCF in association with encephalocele in a 1.5-year-old boy who presented with a swelling in the occipital area of the scalp since birth that was diagnosed clinically and radiologically as an encephalocele.

CASE PRESENTATION

The patient was a 1.5-year-old boy who presented with a midline swelling in the occipital area of the scalp since birth. The swelling was soft, non-tender and measured approximately 2×3 cm and was slowly progressing in size. At the time of presentation there were no evident symptoms of neurological involvement. A non-contrast CT (NCCT) of the head demonstrated an occipital bone defect along with a sac herniating through it. The lesion was of 30–50 HU. MRI of the patient was not performed due to financial constraints. The patient was taken up for surgery and the excised tissue was sent for histopathological examination. During surgery the neck of the herniated sac was dissected and the sac was opened. On opening, the sac

contained degenerated brain tissue, thereby supporting the clinicoradiological diagnosis of occipital encephalocele, which was further confirmed on histopathological examination of excised sac contents.

Gross specimen comprised of a single, well-circumscribed, skin and hair covered, creamish white tissue piece measuring 3 cm × 2 cm × 2 cm with a greyish white gelatinous cut section (figure 1). On microscopy, a skin covered growth with poorly circumscribed and ill-defined proliferation of fibroblasts in a variably collagenised and focally myxoid stroma was seen along with numerous multinucleated giant cells having a floret-like appearance (figure 2A). No areas of mitoses or necrosis were seen. Formation of cystic and sinusoidal structures lined by spindle and floret cells was seen. One-third of the tumour showed mature glial tissue, bordering a cystic space (figure 2B). A histological diagnosis of GCF with an encephalocele was rendered. Immunohistochemically, the constituent cells and multinucleated cells were positive for both, vimentin (diffuse positivity; figure 3A,B) and CD34 (focal positivity; figure 4A,B). These cells were negative for keratin, S-100, desmin, smooth muscle actin, neuron specific enolase and CD68, thereby confirming the diagnosis of GCF.

DIFFERENTIAL DIAGNOSIS

The differential diagnoses of GCF include liposarcoma, myxofibrosarcoma, malignant fibrous histiocytoma and papillary intralymphatic angioendothelioma. Liposarcoma can be differentiated by identification of lipoblast, absence of slit-like sinusoidal spaces and occurrence of this lesion in adulthood. Myxofibrosarcoma can be differentiated by its deeper location and occurrence in older patients. Malignant fibrous histiocytoma is characterised by markedly atypical mesenchymal cells, frequent mitotic figures, absence of sinusoidal spaces and rarity of this neoplasm

Figure 1 Resected specimen showing skin-covered cystic structure with white gelatinous wall.

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Figure 2 (A) Histology showing an ill-defined proliferation of fibroblast around a cystic space bordered by mature glial tissue (H&E×10). (B) Floret cells against myxoid background (H&E ×40).

in childhood. Papillary intralymphatic angioendothelioma can be differentiated by positive vascular markers and absence of myxoid and cellular areas in this tumour.^{3–9}

TREATMENT

The patient was taken up for surgery. The clinical picture, the radiological findings depicting occipital bone defect with herniated sac and intraoperative finding of herniated brain tissue into the sac were all in favour of an occipital encephalocele rather than a primary tumour. Excision of the swelling with repair of the defect was performed and tissue was sent for histopathological examination. However, treatment for GCF includes removal of the tumour with wide excision and these patients do not require any form of chemotherapy.¹⁰ Mohs' micrographic surgery has been advocated for recurrent GCF.¹¹

OUTCOME AND FOLLOW-UP

The patient is doing well 3 months after surgery with no residual neurological deficits. Although local recurrence is common, long-term outcome of this tumour is very good after re-excision of the tumour.

DISCUSSION

GCF is an intermediate grade, soft tissue tumour of childhood with high incidence of local recurrence. Metastasis has never been reported. It shows a preponderance in males with majority of cases seen during the first decade of life.^{3–5} Clinically, the

patient usually presents with a small, painless, slow-growing soft tissue swelling which has a predilection for the back and thighs.⁵ Other less common sites include anterior chest, shoulder, perineum and extremities.^{5,6} Thus GCF in association with encephalocele in the scalp area is notably a rare incidence.

On gross examination, GCF appears as an unencapsulated, grey-white, gelatinous lesion. Histologically, these lesions show a variable degree of cellularity with a fibromyxoid to hyalinised stroma containing bland spindle cells and multinucleated giant cells. The spindle cells may demonstrate mild to moderate pleomorphism and are arranged diffusely or in vague fascicles, their elongated nuclei have vesicular to hyperchromatic chromatin. The cytoplasm is usually scanty and eosinophilic. The giant cells contain a variable number of round to oval vesicular nuclei and have abundant amphophilic cytoplasm with irregular cytoplasmic contours. Slit-like sinusoidal spaces lined by either a continuous or discontinuous layer of multinucleated giant cell is a unique and diagnostic feature of GCF.^{3,4}

The histogenesis of GCF has been a topic of considerable debate among researchers. Ultrastructural studies have, however, demonstrated the fibroblastic differentiation.⁹ Previously, GCF were considered as a juvenile form of dermatofibrosarcoma protuberans (DFSP) but recent studies suggests that both these lesions are on a spectrum of same entity as confirmed by clinical, morphological, immunophenotypical and molecular studies.^{3,9} Both lesions share common clinical features of male predominance, slow growth, painlessness and anatomical

Figure 3 (A and B) Vimentin immunostain showing diffuse cytoplasmic positivity (×4 and ×10).

Figure 4 (A and B) CD34 immunostain showing focal cytoplasmic positivity ($\times 100$ and $\times 40$).

location on mainly the trunk. Morphologically, GCF and DFSP shows predominantly dermal or subcutaneous location and rarely superficial skeletal muscle involvement, honeycomb and parallel growth patterns, sparing of adnexa, myxoid changes and prominent vasculature.⁹ By using immunohistochemistry, both lesions show positive staining for CD34 and negative reaction to keratin, S-100, HMB-45, smooth muscle actin and desmin. Cytogenetically, t(17;22) translocation is seen in GCF and DFSP.⁹ Although GCF and DFSP share several clinical as well as morphological features, we can still differentiate them solely on the basis of histological findings. The presence of pseudovascular spaces lined by giant cells, solid areas with stromal giant cells, consistent haemorrhage, perivascular and onion skin-like chronic inflammation without storiform pattern favours the diagnosis of GCF.⁹ Vimentin is the only immunohistochemical detection marker that has been found to consistently stain GCF.⁴

Learning points

- ▶ A rare childhood tumour of intermediate grade malignancy, giant cell fibroblastoma may present at unusual sites and may coexist with other congenital defects.
- ▶ Owing to its close resemblance to dermatofibrosarcoma protuberans and misinterpretation as sarcomas, it is imperative to accurately diagnose this lesion.
- ▶ Thorough histopathological examination of the entire tumour mass along with supportive immunohistochemistry is all that is needed for a definitive diagnosis.

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Competing interests None.

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