Giant Cell Fibroblastoma in a Little Girl: A Case Report

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Abstract

Giant cell fibroblastoma (GCF) is an intermediate grade soft tissue sarcoma with a high incidence of local recurrence. It usually occurs in children during the first decade of life. This lesion is often misdiagnosed because of its variegated appearance. Here we present the case of a 2 and half -year-old girl who presented with a voluminous painless mass of the left gluteal region, previously misdiagnosed and treated as a hemangioma. CT scan showed marked peripheral enhancement and wide surgical excision with 2 cm margins was performed for final diagnosis. Slit like sinusoidal spaces lined by giant cells and staining for vimentin were highly contributive for GCF. Internal margins were microscopically unclear. No clinical recurrence has been noted within 4 weeks after surgery. Despite its rarity and non-specific findings, this lesion merits a high index of suspicion to reduce misdiagnosis and to manage it in an early stages with clear surgical margins. A best coordination between radiologists and pathologists is needed to highlight the role of imaging studies in the preoperative diagnosis of GCF.

Keywords: Giant cell; Fibroblastoma; Imaging; Surgery; Pathology

Introduction

Giant cell fibroblastoma (GCF) is a rare slow growth soft-tissue sarcoma arising from the dermis and subcutaneous layers. It is a histologic variant of DFSP and both are characterized by an intermediate grade of malignancy, local infiltrative growth, and high recurrence rate [1]. GCF occurs mostly in males during the first decade of life [2]. Radiological findings are non-specific, and the histological appearance showed pleomorphic giant cells with typical pseudovascular spaces [3]. Due to its rarity and painless character, the diagnosis of GCF is often mistaken or delayed, leading to giant forms with a high risk of relapse. Here we describe the clinic, radiologic and pathologic features of a voluminous GCF at the left gluteal region occurring in a two and half -year-old child who underwent a wide excision.

Case Report

A 2 and half -year-old female child with no personal or familiar history, presented to our hospital with a giant painless mass of the left gluteal region noted at 5 months of age. The patient’s parents reported that the lesion had grown proportionately with her and protruded into the skin


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surface over time, becoming a voluminous mass. This lesion was previously examined with ultrasonography outside the hospital. It has been diagnosed as a hemangioma and treated with beta blockers without any improvement. Physical examination revealed an about 20 cm red-purple mass at the left gluteal region, with lobular margins, compressible touch, prominent blood vessels, and ulcerative areas at the surface [Figure 1]. There was no evidence of regional lymphadenopathy. A rectal exam performed under general anesthesia doesn’t show an intraluminal involvement by the mass. A computed tomography (CT) was performed to delineate the tumor. It revealed a well-defined superficial mass of the left gluteal region, infiltrating adjacent subcutaneous adipose tissue and measuring 22x17x57mm. On unenhanced scan, the mass was homogenous and isodense without calcification. Contrast enhanced CT images showed marked peripheral enhancement in both arterial and portal venous phases [Figure 2]. No muscular invasion was observed. No lymphadenopathy or distant metastases were found. For the final diagnosis, the patient underwent a surgical excision.

Figure 1: Protuberant red-purple mass of the left gluteal region with prominent blood vessels and ulcerative area at surface.

Figure 2: computed tomographic findings of the mass. A: axial unenhanced CT image revealed a well-defined isodense mass infiltrating the subcutaneous adjacent adipose tissue, without calcification. B, C, and D: axial and coronal contrast enhanced CT images in the arterial and portal phases showed peripheral enhancement with intratumoral non enhancement areas.

Under general anesthesia, the tumor was excised with 2 cm margins on the external and superior sides. However, there was a narrow surgical margin at the medial side, due to the presence of a partial infiltration of the external anal sphincter requiring a small excision and reparation of the anus. Deeply, the fascia and a portion of the left gluteal muscle were also removed. A rotation advancement flap from the right gluteal region was used to repair the primary defect. Pathological examination revealed a soft and white mass measuring 8x6x5 cm with fasciculate containing on sectioning.
Microscopically, the mass showed wavy fascicles of spindle and stellate cell tumor admixed with multinucleated giant cells within a hypocellular and moderately myxoid stroma, infiltrating the dermal and hypodermal layers. Slit like sinusoidal spaces lined by continuous layers of giant cells were observed. Immunohistochemically, tumor cells were diffusely reactive with vimentin and CD 34 but negative for desmin, myogenin, anti AML, H, caldesmone, and CD68.

KI67 stained about 2% of tumor cells [figure 3]. The diagnosis of GCF was made with unclear microscopic internal margins for preserving functional benefit.

Figure 3: A, B: histopathological features of GCF specimen showing wavy fascicles of spindle and stellate cells tumor admixed with multinucleated giant cells infiltrating the dermal and hypodermal layers. Typical and unique finding of slitlike sinusoidal spaces lining with giant cells is very remarkable. C, D: immunohistochemical staining showed a diffuse expression of vimentin and CD34 as brown.

No clinical recurrence has been noted within 4 months after surgery [Figure 4].

Discussion

GCF is a rare indolent soft-tissue sarcoma originally described by Drs Enzinger and Schmooker in 1982, following which around 100 cases have been reported [1]. It is considered as a histological variant of dermatofibrosarcoma protuberans (DFSP) and occurs mostly in males younger than 10 years of age [2,3]. GCF and DFSP are both fibroblastic tumors previously considered in tumors of the skin and actually removed in soft-tissue sarcomas in the revised 2013 World Health Organization (WHO) Classification of Soft-Tissue Sarcomas [4]. Both lesions share common clinical, immunohistochemically, and molecular abnormalities. They are characterized by an intermediate grade of malignancy, local infiltrative growth with high recurrence rate, positive staining for CD34, and the presence of translocation t (17; 22) (q22; q13) that leads to a specific fusion of the platelet-derived growth factor beta (PDGF beta) with the collagen type 1alpha1 (COL1alpha1) gene [5]. GCF Metastasis has never been reported.

This lesion can occur on any part of the body with a predilection for the back and thigh. Other less common sites include the anterior chest, shoulder, perineum, and extremities [2,3]. In our case, the tumor was located in
the left gluteal region, which is a critical site for surgical management.

Like DFSP, GCF appears primarily as a painless skin-colored (violaceous or bluish erythematous), indurated or atrophic plaque. Later, the plaque develops confluent nodules which gradually enlarge to form a protuberant mass of varying size. Generally, this slow growing process takes several months or years to as long as 60 years, and then complications such as bleeding, ulceration, and pain may arise. GCF is often superficial infiltrating dermal and subcutaneous tissue. In advanced stages, it may invade deep structures such as the fascia, muscle, periosteum, or bone [5].

In infancy, GCF is often mis or underdiagnosed at an early stage and may be confused with other lesions such as vascular tumors, leading to delayed treatment. The little girl in this case had a protuberant painless mass of the left gluteal region with ulcerative areas and enlarged blood vessels at the surface, previously misdiagnosed as a hemangioma. Imaging studies are helpful in preoperative assessment and follow for recurrence.

They may delineate the tumor’s size and extent, and define its relationship with adjacent neuromuscular structures and bone. However, there are a little report describing the imaging findings of GCF in the pathologic literature [1, 2, 5, 6].

GCF has nonspecific imaging findings. On ultrasound, the tumor often appeared as a low echogenic mass with a rich blood supply. Computed tomography (CT) showed a well-defined superficial nodule or mass isodense without calcification on unenhanced images, with homogeneous or inhomogeneous moderate to marked enhancement[5, 6].

MRI is the preferred imaging modality for the evaluation of GCF, which generally showed low signal intensity compared to skeletal muscle on T1-weighted images, and intermediate to high signal intensity on T2-weighted images. On contrast-enhanced T1-weighted images with fat suppression, the tumor demonstrated moderate to marked contrast enhancement [1, 6].

In a recent report, Min and Al observed a marked peripheral enhancement of GCF on MRI, and related that to hypercellularity with abundant vascular channels and myxoid component of the peripheral portion of the mass visible on pathologic examination.

This intense peripheral enhancement of the mass is also remarkable on CT images in our case. That may confirm a consistent contrast behavior of the tumor on both plain CT/MRI scans, and support the suggestion of Min and al that there is different degree of cellularity and histological components between center and periphery of the mass.

For this reason, pathologists should mention the degree of cellularity between the center and the periphery of GCF, and further radio-histologic correlation studies are needed to highlight the imaging findings of GCF.

The histogenesis of giant cell fibroblastoma has been a topic of considerable debate amongst researchers. Ultrastructural studies have, however, demonstrated fibroblastic differentiation [7].

Grossly, GCF appears as an unencapsulated, gray-white, gelatinous lesion. It ranges from 1 to 8 cm in dimension with a mean of 3–4 cm [3, 7].

Microscopically, it shows a diffuse infiltration of the mid-and lower dermis with spindle cells and multinucleate giant cells arranged diffusely or in vague fascicles within a fibromyxoid to hyalinized stroma. The spindle cells have elongated nuclei with 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. The degree of cellularity varies from hypocellular with abundant myxoid stroma to moderately cellular with less myxoid material. Giant cells line gapping and branching slit like sinusoidal spaces exclusively seen in GCF and that seem to reflect a loss of cell cohesion [7,8].

Vimentin is the only immunohistochemical detection marker that has been found to consistently stain GCF [8]. In this case, typical pathologic findings and positive staining for vimentin were very suggestive for GCF.

GCF and DFSP stained both positive for CD4 but negative for other immunohistochemical markers such as keratin, S-100 protein, HMB-45, smooth muscle actin, and desmin. Furthermore, they present unique cytogenetic features, such as the reciprocal translocation t (17; 22) or more commonly supernumerary ring chromosomes containing sequences from chromosomes 17 and 22 [5,8]. Hybrid lesions, with features of both DFSP and giant-cell fibroblastoma, have also been reported.

Differential diagnosis includes three main lesions: DFSP, hemangioma and epidermal inclusion cysts. DFSC had nonspecific imaging findings, however, the presence of pseudovascular spaces lined by giant cells and vimentin marker favors the diagnosis of GCF.

Hemangioma often occurs in the limb muscles and is a painless soft tissue mass, which shows calcification on CT, low signal intensity on T1-weighted images, and high signal intensity on T2-weighted images with dots or lines of low signal, which signifies calcification or venous stones, and slowly marked enhancement. Epidermal inclusion cysts can be easily distinguished from GCF because it shows no contrast enhancement on imaging studies.
Surgical resection is the main treatment for GCF and the most significant prognostic factor is proven to be the extent of resection. Adequacy of surgical excision remains central to minimize disease recurrence. One large case series reported a local recurrence rate of approximately 50% [9]. The recommended surgical options include wide local excision (WLE) with clear margins and Mohs micrographic surgery (MMS).

The direct compromise for WLE is tumor-free margins, however, adequate margins have not been defined according to rigorous clinical trials. NCCN guidelines (National Comprehensive Cancer Network) (version 1.2020) suggest 2–4 cm lateral margins of the tumor and the excision of the investing fascia to remove any infiltrating tumor in WLE. This procedure is generally applied for primary GCFs on the trunk or extremities. It allows immediate wound repair following tumor removal and it is cost effective for patients and medical resources. However, it is unable to evaluate surgical margins during the operation and exposed to high rate of local recurrence if negative margins were not obtained [5, 9].

Mohs micrographic surgery is a stepwise procedure of tumor removal permitting a meticulous microscopic evaluation of the margins during excision. This is the ideal technique for GCFs in cosmetically and functionally sensitive regions including vulva, penis, scrotum, breast, peri-anal region, and inguinal fossa, and it is also recommended for recurrent lesions [5, 9].

Local recurrence rate after MMS is significantly lower than WLE. Despite all these advantages, Mohs surgery has been performed in few reports because it needs specialized training of surgeons, high coordination with histopathologists, and intensive cost.

Following tumor resection, Soft-tissue defect may be primarily closed, especially in regions with redundant soft tissue such as chest, abdomen, and back or reconstructed by using local or free fasciocutaneous flap or split-thickness skin grafting.

In our case, the little girl underwent a wide excision with narrow margins at the internal side to preserve anus function and the defect was reconstructed with a flap rotated from the right gluteal region.

Long-term outcome of GCF is very good especially after MMS, however, regular follow-up should be continued to detect disease recurrence [9].

**Conclusion**

GCF remains a diagnostic challenge due to its rarity and similarities with other lesions such as DFSP and vascular tumors. However, clinicians should adopt a high index of suspicion in case of superficial soft tissue mass in children under the age of 5 years. On imaging studies, peripheral contrast enhancement may presume a preoperative diagnosis of GCF; therefore, the final diagnosis consists of typical pathologic features and immunohistochemical staining.

The delay in diagnosis leads to the considerable growth of the lesion requiring extensive surgery and subsequently a large scars to be reconstructed. In cosmetic and functional regions, Mohs surgery is the ideal technique to preserve sparing tissue and to minimize the rate of local recurrence.

**References**