Molecular Particularity in Rare Tumour of Buttock: Case Report and Literature Review*

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ABSTRACT

Introduction: Hyalinizing spindle cell tumor with giant rosettes (HSCT) is a very uncommon mesenchymal tumor that has similar morphological and biological features to the low-grade fibromyxoid sarcoma (LGFMS). Case Report: Reported herein is a case of primary tumour of buttock HSCT that had rare FUS-CREB3L1 fusion transcripts, a product of characteristic chromosomal abnormality t (7, 16) (q33, p11) of HSCT and LGFMS. The patient was a 48-year-old man who had a large solitary mass in the buttock. Histologically, it was composed of bland spindle cells with variable cellularity deposited in a densely hyalinized stroma alternating with myxoid areas. Characteristic collagen rosettes were scattered in the cellular areas. Reverse transcription-polymerase chain reaction (RT-PCR) assay using formalin-fixed, paraffin-embedded tissue detected FUS-CREB3L1 fusion transcripts. In our knowledge is the second case may display a variant FUS/CREB3L1 fusion transcript in international literature. Conclusion: LGFMS and HSCT probably have a wider spectrum of morphologic features than previously thought, the awareness of which will help pathologists to avoid diagnostic pitfalls. Demonstration of the t (7, 16) translocation will help to diagnose difficult cases with unusual histologic features.

Keywords: Deep Soft Tissue; Fibromyxoid Sarcoma; Hyalinizing Spindle Cell Tumor; RT-PCR; Fusion Transcripts; CREB3L1

1. Introduction

Hyalinizing spindle cell tumor with giant rosettes (HSCTGR) is a recently described low-grade sarcoma with a deceptively benign histological appearance [1]. Because it has similar clinicopathological features and biological behavior to that of low-grade fibromyxoid sarcoma (LGFMS), HSCT might be regarded as a variant of LGFMS [2,3]. Low-grade fibromyxoid sarcoma (LGFMS) is a rare soft-tissue tumor with a deceptively benign histologic appearance affecting predominantly young adults during the fourth decade of life [1]. Low-grade fibromyxoid sarcoma has a predilection for involving deep soft tissues of the thigh, inguinal region, or chest wall, affecting less frequently the shoulder or axilla [1]. Local postsurgical recurrence and metastases to lungs and bone are frequently seen [1]. Additionally, both tumors have a characteristic FUS-CREB3L2 fusion gene transcript. A product of reciprocal translocation, t (7, 16) (q33, p11), further supporting this concept [4,5]. HSCT arises in a variety of deep soft tissues, most commonly in the extremities. We describe here a new case of primary tumour of buttock HSCT harboring a molecular particularity with a rare FUS-CREB3L1 fusion transcripts.

2. Case Report

A 48-year-old man presented with a 13-cm painful left buttock mass that had been present for 1 year. The mass was palpable by digital rectal examination but did not involve the mucosa. A superficial FNAB of the subcutaneous mass was performed and diagnosed as “spindle cell neoplasm, favor low grade sarcoma”. The lesion was resected with negative surgical margins. Macroscopically, the tumors were well circumscribed, firm, and yellow-tan. For microscopic examination, wet formalin-fixed tissue was available in our case. Microscopically, a pseudocapsule surrounded the tumors. The tumors were comprised of bland, fibroblastic spindle cells arranged in a whirling growth pattern within a hyalinized collagen bundles and a myxoid matrix (Figure 1). The myxoid areas were characterized by low cellularity with a prominent capillary network. However, in some areas the
spindle cells were concentrated around blood vessels. Alternating with the myxoid regions were fibrous areas with varying degrees of cellularity, including some hypocellular areas comprised nearly entirely of dense collagenous tissue. A large rosette-like structure with hyalinized stroma was found (Figure 2), which is characteristic of LGFMS. Immunohistochemistry in our case revealed strong positivity with vimentin only; no staining was observed with Smooth muscle actin, desmin, protein S-100, or CD34 in the sections examined. PCR amplifications were performed in a 50-μL reaction volume containing 13 AccuPrime Pfx reaction mix, 1 unit AccuPrime Pfx DNA polymerase, 0.3 μM of each of the forward and reverse primers and 200 ng template DNA. The PCR was run on a PCT-200 DNA Engine. The cycling included an initial denaturation at 95°C. For 2 min, followed by 30 cycles of 15 s at 95°C, 30 s at 58°C, and 2 min at 68°C, and a final extension for 5 min at 72°C. All ligations were performed overnight at 16°C in 10-μL reaction volume containing 13 Ligase Reaction Buffer, 5 units T4 DNA ligase, and 1:3 vector to insert ratio. When purification was required, the DNA fragments were purified using either the QIAquick gel extraction kit or the QIAquick PCR purification kit. For sequence analysis, the ABI Prism BigDye terminator v1.1 cycle sequencing kit was used and the products were analyzed on an Applied Biosystems Model 3100-Avant DNA sequencing system. RT-PCR demonstrated the FUS-CREB3L1 fusion transcripts resulting from the reciprocal translocation, t (7, 16) (q33, p11). The patient received no additional therapy. Clinical follow-up, including radiographic studies, had not detected recurrent tumor at 3 years of follow-up.

3. Discussion

The HSCTGR was first reported in a series of 19 cases culled from a large soft tissue consultation service by Lane et al. in 1997 [2]. Clinically, HSCTGR can develop at almost any age (average age, 38 year) in the deep soft tissues of the extremities, particularly the thigh [2,6]. Other sites of involvement are the chest wall, axilla, rarely buttock, and the [2]. The most common symptom is a painless, deep-seated, slowly enlarging mass [2,6]. Grossly, the tumor is an oval multilobulated mass, ranging in size from 2 to 20 cm in diameter. Although most of the lesions appear well circumscribed, they can extensively infiltrate the surrounding soft tissue [6,7]. A prior report on the imaging characteristics of LGFMS described a heterogeneously hyperechogenic multi-nodular sonographic appearance, a heterogeneous MR imaging appearance with low to slightly high SI on T1-weighted images, heterogeneously low to high SI on T2-weighted images, and heterogeneous postcontrast enhancement [3]. The cut surface has a whorled white-tan appearance. Cystic degeneration is an uncommon finding [7]. Histologically, the tumor cells have a deceptively bland appearance, consisting of short, ill-defined fascicles of fusiform to spindled cells with minimal atypia and barely perceptible levels of mitotic activity [6]. In a small number of cases there may be foci that are more cellular and atypical, which would mimic an intermediate grade fibrosarcoma [3,6]. The mitotic figures were difficult to identify, usually with less than one mitosis per 50 high-power fields [6]. The most characteristic feature was the presence of a variable number of large rosettes like structures. These structures either occupy only a small portion of the lesion or are so prominent as to obscure the other features of the neoplasm [2,6]. The rosettes, which tended to cluster, were made up of a central collagenous core flanked by an irregular rim of rounded neoplastic cells. The immunohistochemical profile, espe-
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REFERENCES


