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Case Report

An unusual complex karyotype in myopericytoma

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abstract

Introduction: Myopericytoma is a perivascular neoplasm commonly found in the skin and soft tissue of extremities. These lesions often exhibit concentric vascular proliferation of spindle shaped myoid cells.

Methods/Results: We present a case of a 76-year old male who was diagnosed with myopericytoma and subsequent cytogenetic analysis found a highly abnormal karyotype. This karyotype includes cytogenetic mutations that have not been described in previous case studies of myopericytoma.

Conclusions: Some of these aberrations occur on genes that are involved in hedgehog signaling as well as pericyte proliferation, indicating a potential pericyte origin for myopericytoma tumors.

1. Introduction

Myopericytoma is a benign subcutaneous tumor with a prominent concentric perivascular growth pattern. Myopericytoma tumor cells usually exhibit oval or spindle-shaped morphology with multilayered growth surrounding blood vessels.[1] These tumors are predominately found within the subcutaneous tissues of distal extremities. Myopericytomas occur from the second decade of life onward, and with a reported slight male predilection.[2] Myopericytomas generally present as a painless slow growing nodule, less than 2 cm in diameter, and without invasion of adjacent structures.[1] On biopsy, their characteristic diagnostic feature is perivascular whorls of spindled to ovoid cells with eosinophilic cytoplasm. Subendothelial proliferation of myopericytoma cells is frequently observed, and tumor cells predominantly bulging into the lumen of a vessel can be seen. Myopericytomas are generally well-circumscribed, however in some cases the spindled proliferation may extend along blood vessels outside of the demarcated lesion.[1] Most myopericytomas are immunoreactive for smooth muscle and muscle specific actin, with patchy desmin positivity.[3] Investigators have argued that myopericytoma is part of a morphologic continuum with other perivascular tumors with smooth muscle differentiation, including myofibroma, angioleiomyoma, hemangiopericytoma, glomangiopericytoma, and glomus tumor. To our knowledge, only a single cytogenetic abnormality has been reported in myopericytoma: t(7;12) resulting in an *ACTB-GLI1* fusion product.[4] However, a complex karyotype has never been

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reported in myopericytoma. The present case is that of benign myopericytoma arising from the soft tissues of the knee. Cytogenetic analysis showed a complex karyotype with multiple balanced and unbalanced translocations, which may shed light on the biology of this relatively rare mesenchymal tumor.

2. Report of the case

A 76-year-old male presented with an enlarging mass in the anterolateral superficial soft tissue of his right knee. The lesion had been present for two years, and had recently grown in size. The patient denied any associated pain, limitations in mobility, or systemic symptoms. The patient had a history of follicular lymphoma (in remission for over four years), osteoarthritis, hypertension and hypercholesterolemia. The mass was imaged using X-ray and magnetic resonance imaging (MRI). Radiographs demonstrated a soft tissue mass without calcification. No bony destruction, erosion, periosteal reaction or joint space changes were seen. Subsequent MR imaging demonstrated a T1 isointense, T2 hypertense, well-circumscribed mass measuring 2.6 × 1.8 cm in the subcutaneous soft tissues just anterolateral to the tibial tubercle (Fig. 1). There was minimal surrounding edema and no evidence of invasion into adjacent structures.

Ultrasound guided percutaneous core needle biopsy was performed on the tumor. Hematoxylin and Eosin (H&E) stained samples of the biopsy showed a moderately cellular proliferation of relatively uniform ovoid to spindled tumor cells with plump nuclei and modest amounts of eosinophilic cytoplasm. Cells showed a solid to perivascular 'whorled' arrangement (Fig. 2**A,B**). Thin-walled vessels lined by bland appearing endothelial cells were numerous, with a sparse collagenous stroma. Mitoses were rare (1/20 high powered fields). No histologic features suggestive of malignancy were observed, including no significant cytologic atypia, no apoptotic bodies, no necrosis and no atypical mitotic figures.

Immunohistochemical staining demonstrated diffuse reactivity for α Smooth Muscle Actin (αSMA) (Fig. 2**C,D**) and focal patchy reactivity for Desmin (Fig. 2**E,F**). All other stains were negative including epithelial markers (Pankeratin cocktail), melanocytic markers (S100, HMB45 and MART1), and histiocytic markers (CD68 and CD163). Endothelial markers (CD31, CD34) were negative in tumor cells, and highlighted the numerous thin walled vessels. Based on the biopsy material, a diagnosis of myopericytoma was rendered.

Cytogenetic analysis of the biopsy specimen was performed, as is done routinely at our institution for all soft tissue tumors without a previous diagnosis. Results showed a complex karyotype in all cells analyzed. A stemline clone, 5 of 20 cells, exhibited t(4;5)(p14;q33) (Fig. 3**A**). In addition to the stemline, 12 of 50 cells also exhibited: additional chromosomal material on the long arm of chromosome 3 (3q) (Fig. 3**B**); a balanced translocation involving the other copy of chromosome 4 and chromosome 21 [t(4;21)] (Fig. 3**C**); and a derivative chromosome 10 resulting from an unbalanced translocation between chromosomes 1 and 10 [t(1;10)] leading to trisomy 1q (Fig. 3**D**). Additionally, in 3 of 20 cells there was a balanced translocation between chromosome 13 and 22 [t(13;22)] (Fig. 3**E**). In summary the patient's karyotype was reported as: 46,XY,t(4;5)(p14;q33)[5]/45-46,idem,add(3)(q21),t(4;21)(q31;q22),der(10)t(1;10)(q11;p15)[cp12]/46,XY,t(13;22)(q22;p13)[3]. With the exception of t(4;21)(q31;q22), which has been reported in cases of acute myeloid leukemia (AML)[5] and T-cell acute lymphoblastic leukemia (T-ALL),[6] none of the above abnormalities identified have been associated with neoplastic processes. As well, none of these cytogenetic abnormalities have been previously reported in myopericytoma.

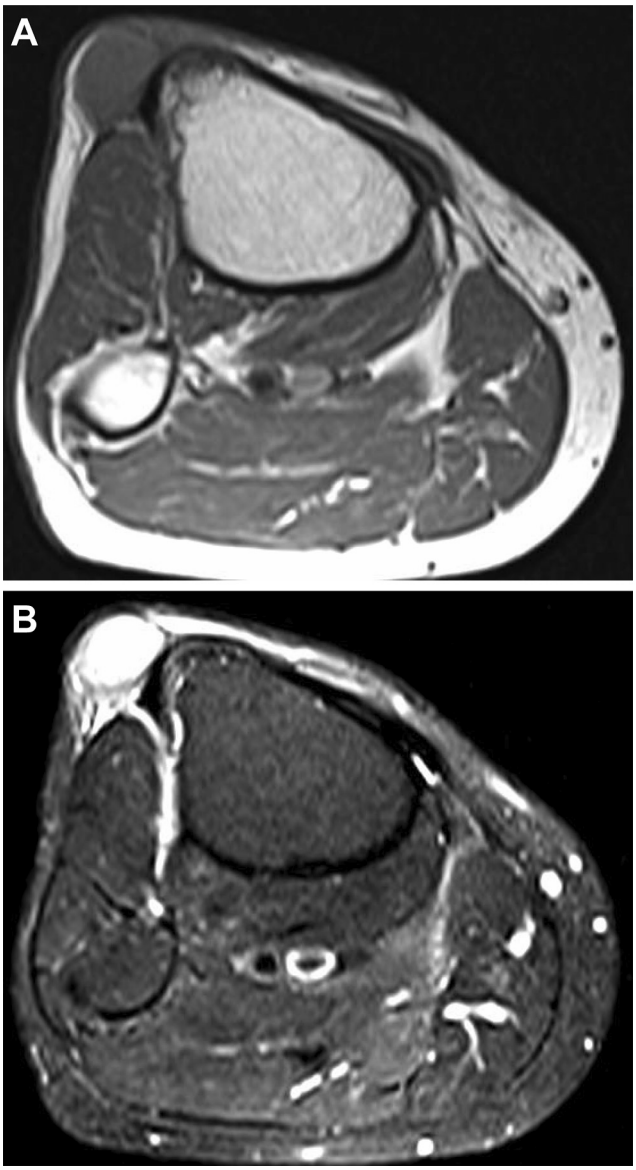

Fig. 1 – Radiographic appearance. Magnetic resonance imaging demonstrated a well-circumscribed, soft tissue lesion without connection to the underlying tibia (see upper left corner of images). (A) T1 weighted image, demonstrating isointensity. (B) T2 weighted image, demonstrating hyperintensity.

Surgical excision was performed five months thereafter. The gross specimen showed a well-circumscribed, 3.0 cm, pink to tan solid mass. Routine histologic sections demonstrated a well-circumscribed, nodular neoplasm with numerous dilated, branching, 'hemangiopericytoma-like'