Periosteal fasciitis in a 7-year old girl – a diagnostic dilemma

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Abstract

Periosteal fasciitis, considered as a subtype of nodular fasciitis, is a rare benign soft tissue mass often misdiagnosed as a malignant lesion due to the fast and infiltrative growth pattern and histological features. Nodular fasciitis is usually found in the upper extremities of adults and in the head and neck region of children. Incorrect diagnosis may lead to over- treatment, potentially causing disturbed orofacial development in growing children. A fast growing asymptomatic lump initially suspicious for a malignant bone tumor was found in the left angular area of the mandible in a 7-year-old healthy girl. Radiographic exam revealed an exophytic expansile, destructive nodule arising from the periosteal region. A diagnosis of periosteal fasciitis was made based on the histological findings of an open biopsy and the lesion was enucleated subsequently. Fluorescence *in situ* hybridization analysis revealed a *USP6* gene rearrangement and confirmed the diagnosis molecularly. Because of the aggressive growth pattern without external provocation and the gene rearrangement test we suggest that NF should be regarded as a benign neoplasm rather than a reactive process. The patient is free of disease three years after surgery.

Keywords: Jaw tumors; neoplasms of children; nodular fasciitis; FISH, *USP6* gene

Introduction

Nodular fasciitis (NF) is a rare and benign soft tissue mass often misdiagnosed as a malignant neoplasm because of its fast and infiltrative growth. It has been considered as a reactive proliferation of fibroblasts. The lesion was first described by Konwaler et al.[1] 1955 as pseudosarcomatous fasciitis. The pathogenesis remains unknown[2-5]. Periosteal fasciitis is considered to represent a subtype of nodular fasciitis constituting about 4% of all cases[6]. The literature on periosteal fasciitis is scanty.

NF is most common in the upper extremities of adults. It can be found at any age but most of the cases are detected in adults in their 3rd to 5th decade of life[7]. NF in children is rare[3,7]. Only a few articles have been published on pediatric patients in the head and neck area which is the most commonly involved site in children[8]. Boys seem to be more often affected than girls[2,5,8].

NF is usually unilateral[2] and typically presents as a painless mass developing in the subcutaneous or deeper soft tissues within only a few weeks. It is generally well circumscribed but it can infiltrate in surrounding tissues[7].

Due to the versatile clinical and radiological appearance and sometimes worrisome histological (pseudosarcomatous) features, the diagnosis of NF can be challenging. The preferred treatment is excision and tendency of recurrence is considered low[2,4]. Metastases have not been reported. A proper diagnosis is important because otherwise patients may undergo too radical treatment.

Case report

A fast growing soft tissue mass developing within one month in the mandible of a 7-years old healthy girl was detected by a pediatrician. An ultrasound examination raised the suspicion of a non-homogenous bone tumor (diameter 1.5 cm) in her left mandibular angular area. The tumor was painless and firm on palpation.

The cone beam computed tomography (CBCT) examination revealed a 1.0 x 1.5 cm outwards growing and juxtacortically located lesion eroding the cortical bone (Fig. 1A–C). Tiny bony septae were detected inside the lesion (see video of CBCT on the web-version of the article).

Further magnetic resonance imaging (MRI) examination showed a multilocular space filled with liquid in the inferior part of the mandible. An open biopsy was performed. Microscopically the lesion was found to be well circumscribed with a surrounding fibrous capsule (Fig. 2A). Some irregular bone trabeculae were found especially at the periphery (Fig. 2B). The major part of the biopsy specimen consisted of spindle-shaped fibroblasts without striking amount of atypia or increased mitoses (Fig. 2C). After several external consultations a diagnosis of periosteal fasciitis was made.

Within three months the lesion had doubled in size. At surgery, a capsulated soft tissue mass firmly attached to the underlying bone was found (Fig. 1D). It was rather vascularized, rubbery on palpation, and had multiple thin covering layers of well vascularized soft-tissue. Enucleation supplemented by removal of a narrow bone rim surrounding the area was performed. Histological features of the surgical specimen were essentially similar to that of the biopsy. Subsequently, FISH analysis was performed and showed a *USP6* gene rearrangement in 42/50 (84%) lesional cells strongly supporting the diagnosis of a periosteal fasciitis (nodular fasciitis) (Fig. 2D). The patient is free of disease three years after surgery.

Discussion

Nodular fasciitis is a rare and often misdiagnosed tumor of soft tissues. Incorrect malignant diagnoses may lead to overtreatment, potentially causing disturbed orofacial development in growing children. Periosteal fasciitis is considered a subtype of NF with periosteal overgrowth and reactive new bone formation. Only a few case-reports have been published.

Until recently, NF has been considered as a reactive process and the diagnosis of NF has been made primarily on the basis of histological, radiological, and clinical findings. Amary et al.[9],

however, found *USP6* rearrangements in 91% of 34 NF, rendering *USP6* FISH analysis a reliable and useful ancillary test to diagnose NF. Identification of a replicable gene rearrangement strongly suggests that NF is a benign neoplastic proliferation. Furthermore, in our patient, the lesion doubled in size within three months without external provocation or in response to some identifiable stimulus speaking for neoplastic nature. Based on these findings we consider periosteal fasciitis as a benign neoplasm, most likely a subtype of NF.

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Figure legends

Figure 1.

A. Conebeam computer tomography (CBCT) shows well bordered mutilocular radiolucency with bony septae in the left mandibular angle of a 7-years-old girl. The radiolucency is centered near the plane of the original periosteum, the enlargement

with scalloped margins partly within and partly external to the mandible and the outline merged with surrounding soft tissue.

B. CBCT axial view demonstrates buccal and lingual bone expansion.

C. CBCT coronal section reveals scalloped margins of the bone lesion.

D. Clinical picture at the operation shows encapsulated soft tissue mass attached to the underlying bone.

A link to video of the CBCT examination is found on the web-version of the article.

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Figure 2.

A. Microscopy reveals an encapsulated, spindle cell nodule containing scattered osteoid (arrow) and calcified osseous trabeculae (arrow head). Hematoxylin and eosin stain (H-E). Bar = 400μm

B. Trabeculae of new bone with plump, benign-appearing osteoblasts and osteocytes within a cellular fibrocollagenous stroma. (H-E). Bar = 100μm

C. Loose fibrous stroma with benign-appearing plump fibroblasts. (H-E). Bar = 75μm

D. Fluorescence in situ hybridization (FISH) analysis showing USP6 rearrangement as separated red and green signals (arrows). The intact USP6 allele is seen as fused red/green signals (arrow heads). Bar = 20μm