Case Report

Cytodiagnosis of Chondrosarcomatous Transformation in a patient with Multiple Osteochondromas

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Abstract

Osteochondroma (Exostosis) is the most common benign tumor of the bone. It may occur as solitary or multiple lesions and most commonly affected age group is 11-20 years. Its common complications include cosmetic or osseous deformity, fracture, neurovascular compromise, bursa formation and malignant transformation. Latter is the most serious and feared complication occurring mostly in third to fifth decade. The risk of malignant progression is estimated to range from 1 to 25% in patients with multiple osteochondromas.

In this study, we present a case of high grade chondrosarcoma which was diagnosed cytologically in a 27 year old male with multiple osteochondromas. Cytological smears were cellular showing atypical cells with marked pleomorphism in a background with chondromyxoid material. Subsequent histopathology of the lesion confirmed the diagnosis. Through this case report we wish to highlight that FNAC is very effective in confirming preoperative diagnosis of chondrosarcomatous transformation in osteochondroma.

Keywords: Multiple Osteochondromas (MO); Chondrosarcoma; FNAC.

Introduction

Osteochondroma is the most common benign osseous neoplasm accounting for about 10-15% of all osseous neoplasms and 50% of all benign neoplasms. It may occur as a single or multiple lesions, the latter presenting as a hereditary syndrome [1].

The most commonly affected age group is 11-20 years, although it can present at any age between 2 to 60 years. There is no sex predilection in case of solitary osteochondromas. However, Hereditary Multiple Exostosis (HMO) syndrome commonly affects males. Caucasians are the most common race affected, approximately 0.9-2 individuals per 100,000 of population. The lesion comprises of cortical and medullary bone, which is in direct continuity with the parent bone and is covered by a hyaline cartilaginous cap [2].

Multiple Osteochondromas (MO) is an autosomal dominant disease occurring primarily in the metaphyseal regions of the appendicular long bones and the flat bones of the axial skeleton. The common complications include cosmetic or osseous deformity, fracture, neurovascular compromise, bursa formation and malignant transformation. Malignant transformation is the most serious and feared complication [3].

It has been estimated that risk of malignant progression is seen in 1 to 25% of patients with MO. Chondrosarcoma arising from the cartilaginous cap is the reason for malignant transformation. Pain,
swelling and enlargement of osteochondromas after skeletal maturity must be viewed with suspicion. The cases of chondrosarcoma reported in association with MO are typically found in patients between the third and fifth decade. Chondrosarcoma is rare in children, including children and adolescents with MO [4].

In this study we present a case of a high grade chondrosarcoma which was diagnosed cytologically in a 27 year old patient with multiple osteochondromas. It is important to diagnose malignant transformation in osteochondroma as surgery is the treatment of choice.

Case Report

A 27 year old male patient presented with tender, firm to hard swelling in right axillary region since 2 months. On clinical examination, swelling measured 3x3cm in size. There was no history of trauma although patient complained of difficulty in lifting his right hand. Relevant haematological and biochemical investigations were within normal limits.

X-ray was done which showed expansile tubulation deformity of proximal humerus involving metadiaphyseal region with cortical break. Extensive calcification was seen in overlying soft tissue showing chondroid appearance (Figure 1).

MRI revealed multiple cortical outgrowths from humerus and scapula with cartilage caps and tubulation deformity compatible with osteochondromatosis (Figure 2).

A clinical diagnosis of multiple osteochondromata with malignant transformation was made. However, there was no family history of multiple exostosis.

FNAC was done from soft tissue swelling in axillary region which yielded blood mixed aspirate. Smears were prepared and stained with Giemsa and Hematoxylin- eosin stains.

On microscopy, smears were highly cellular showing atypical cells with marked cellular and nuclear pleomorphism. The cells showed hyperchromatic irregular nuclei and abundant eosinophilic to vacuolated cytoplasm at places showing intranuclear inclusions. Many binucleate and multinucleated giant cells were also seen. Focally, background showed chondromyxoid material. (Figure 3a,b,c,d)
exostoses. A majority of lesions are asymptomatic and they are located in the bones that develop from the cartilage, such as long bones of the extremities around the knee joint, pelvis and trunk. Our patient presented with pain and motor dysfunction of the right upper limb.

Pain, swelling and enlargement of osteochondromas after the end of skeletal growth period should raise the possibility of a malignant degeneration of the benign tumor. Therefore, this complication frequently occurs after puberty and is rare during childhood. Although exostoses growth has been described as slow-growing and painless by Kivioja et al., the most frequent symptom is pain [7]. Other findings include neurovascular complications, pathological fractures and thickening > 1 cm of the cartilaginous cap in adults.

Malignant transformation is more frequent in the age range between 20 and 50 years, the increase in risk being directly proportional to the age of the patient. The exact incidence of malignant transformation in osteochondroma is not known. Authors have reported that the incidence is 0.4%–2% in patients with solitary osteochondroma and 1%–25% in patients with multiple exostoses [8].

Cytological features of sarcomatous progression displays a rich yield with plenty of tissue fragments and single cells in high grade tumors. The tumor cells have a well defined cytoplasm and rounded nuclei with 1-2 nucleoli. Binucleate cells are present and nuclear pleomorphism is of moderate degree. The chondromyxoid ground substance is usually as abundant as in chondroma. Daswani et al also reported cytological diagnoses of chondrosarcoma [9]. Additional clues, obtained through examining the clinical course and serial imaging studies, shared by the pathologist, radiologist, and clinician are essential in characterizing such tumors [4].

In adults, cartilage caps greater than 15–20 mm in thickness typically herald malignant transformation, though exceptions occur. CT, static and dynamic MRI, and angiographic studies may aid in assessing the thickness of the cartilage cap and displacement or involvement of local vasculature as well as identifying patterns often seen in cartilage tumors with malignant progression [4,10].

Thus, FNAC is very effective in confirming preoperative diagnosis of chondrosarcomatous transformation in osteochondromas, combined with radiological and clinical evaluation.

**Conclusion**

From this case report, it can be concluded that malignant transformation in osteochondroma is
diagnosed earlier by cytology, which may allow for appropriately timed wide excisions. This treatment approach has a lower risk for tumor recurrence and may also decrease the risk of unnecessary major surgery.

References


