Case study

RARE ASSOCIATION OF AN AGGRESSIVE GIANT CELL TUMOR WITH XERODERMA PIGMENTOSUM

ABSTRACT

Xeroderma pigmentosum (XP) is a genodermatosis that manifests as photo-induced skin and ocular alterations and skin cancers. An association with a giant cell tumor is a very rare entity. We report the case of a 17-year-old patient, followed since childhood for XP, who presented with a very aggressive giant cell tumor in the wrist for which he underwent amputation. A review of the literature of these rare lesions is presented and analyzed to see if this is an association or is a chance coincidence.

KEYWORDS : Xeroderma pigmentosum , giant cell tumour,Aggressive,Distal radius.

INTRODUCTION

Giant cell tumors (GCTs) are locally aggressive benign tumors that preferentially occur in the metaphyseal area of long bones. They represent 5% to 10% of all primary bone tumors. They mainly affect young adults between 20 and 40 years of age. Their origin remains uncertain. [1]

Xeroderma pigmentosum (XP) is a genodermatosis that manifests itself by photo-induced skin and eye changes and skin cancers. An association with a giant cell tumor is a very rare entity. We report a case of a 17 year old patient with an association of XP and TCG which would be the second case reported in the literature after that of REKHA et al

CASE PRESENTATION

This is a 17 year old patient, right handed, without profession, followed for xeroderma pigmentosum since childhood, who consulted for a painful mass on the left wrist noted by the patient 2 months before.

The clinical examination revealed: tiny lenticular pigmented macules, with imprecise limits, spread over the skin and on the dorsal surface of the wrist; a shiny painful mass, hard and fixed in relation to the deep plane, circumferential, 8 cm in length (Fig.1), with limited mobility of the wrist.), with limitation of wrist mobility without downstream vasculo-nerve, nor regional adenopathy. The standard radiological workup showed a lytic epiphyseal-metaphyseal image of the lower extremity of the radius, blowing out the cortical bone in places, breaking the external cortical bone with increased soft tissue density, classified as grade III according to the Cappanacci classification (Fig. 2). Magnetic resonance imaging (MRI) revealed a large osteolytic mass, centered on the lower extremity of the radius, with irregular and heterogeneously enhanced contours, coming into contact with the radial and ulnar arteries and veins and infiltrating the digital arteries (Fig. 3). As part of the workup, a thoracic CT scan was performed, which did not reveal any other localization. The phosphocalcic assessment was normal. A biopsy of the mass performed dorsally confirmed the diagnosis of a

malignant giant cell tumor. The evolution was marked by a budding ulceration with local extension invading the two pedicles (Fig. 4).

DISCUSION:

Giant cell tumors are generally benign osteolytic lesions of relatively high frequency. They occur mainly in the epiphyses of the long bones of the limbs in 80% to 90% of cases.

of the long limbs in 80% to 90% of cases. The mechanism that triggers the malignancy of this tumor in patients with XP is not clear, but uncontrolled cell proliferation is the final carcinogenic pathway[2]. Campanacci et al. Campanacci et al [3] proposed a radiographic classification, reflecting the aggressiveness of the lesion in three grades, of which grade III is a lesion with blurred boundaries, with extension into the soft tissues that is not limited by a bony shell, which corresponds to a locally aggressive tumor. An evolution towards a cutaneous complication is certain in this guard that could be avoided by a rapid medical management or even radiotherapy, followed by surgery.

The choice of treatment is based on the local aggressiveness of this tumor and the histological result. Monobloc resection is recommended for TCG grade III [4].

It has been shown in recent studies that patients with a normal allele and a

and a common polymorphism in one of the four XP genes (XP-A, XP-C, ERCC2 or ERCC5) have an increased risk of skin and lung cancer as well as a poor response to chemotherapy [5].

Neyssa et al proved that the P53 protein mutation is involved in the pathogenesis of osteosarcoma [6].

At present there are no studies that have shown the involvement of a protein in the pathogenesis of giant cell tumors.

CONCLUSION

The association of an aggressive giant cell tumor with xeroderma pigmentosum is very rare. There are no arguments to explain the occurrence of severe forms of GCT on a terrain of XP. Moreover, the small number of cases reported in the literature does not allow us to validate this hypothesis. However, this association could not be a simple coincidence, which solicits a new avenue of research.

REFERENCES

1. Szendroi M. Giant cell tumor of bone. J Bone Joint Surg

Br 2004;86:5–11.

2. REKHA A,* RAI D K** Giant Cell Tumour of the Tibia

with Xeroderma
PigmentosumJournal of Clinical and Diagnostic Research. 2007Dec; 1(6):529-532

3- Campanacci M, Boriani N, Boriani S, Sudanese S.Giant-

cell tumor of bone.J Bone Joint Surg Am 1987;69:105–44.

4-Cheng JC,Johnston

JO,Giant cell tumour of bone;prognosis and treatment of pulmonary metastases Clin Orthop Relat Res 1997;May(338);205-14
PMID: 9170381

5-OTTAVIANI, Sébastien, FOREST, Anne, ESCAMPS, Vincent, *et al.* Osteosarcoma in a patient with xeroderma pigmentosum. *Joint bone spine*, 2009, vol. 76, no 1, p. 109-110.

6-Neyssa Marina, Mark Gebhardt, Lisa Teot, Richard Gorlick, Biology and Therapeutic Advances for Pediatric Osteosarcoma, *The Oncologist*, Volume 9, Issue 4, July 2004, Pages 422–44.

Conflict of Interest : The authors declare no relationship of interest.

Fig. 1: Clinical appearance of the tumor mass on the wrist and polymorphic pigmented macules

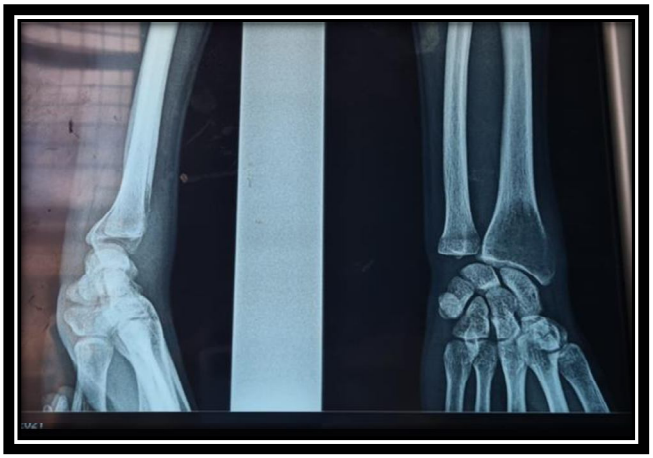

Fig. 2: Radiological aspect showing the epiphyseal/metaphyseal lytic image Grade III of Cappanacci

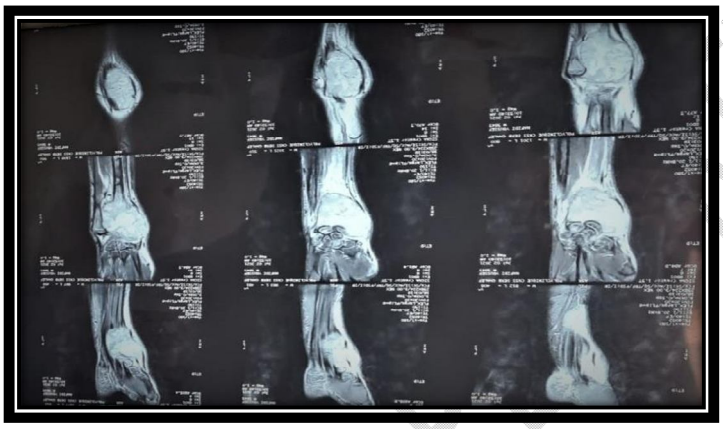

FIG 3:The MRI showing the extension of the tumor to the soft tissue of the wrist

Fig. 4: Clinical appearance showing the aggressive evolution of the tumor after biopsy.