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Case Report

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McCune-Albright syndrome associated with giant cell tumor and aneurysmal bone cyst: An exceptional variant

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Abstract

McCune-Albright syndrome is a rare heterogeneous genetic disorder. A case of a 25-year- old female patient with a gynecological history of precocious puberty, facial asymmetry, café-au-lait spots plus the presence of fibrous dysplasia located in the hips and iliac bones, giant cell tumor in the proximal tibia, and aneurysmal bone cyst in the tibia is reported. and contralateral fibula. Fibrous dysplasia is a rare bone disorder that consists of the replacement of normal bone by an excessive proliferation of fibrous connective tissue with non-functional bone structures, while giant cell tumor is one of the most common benign bone tumors, which is It presents in young adults between the ages of 20 and 40 with a high rate of recurrence and potential for secondary malignancy. In turn, the aneurysmal bone cyst is a benign bone tumor that represents 1 to 2% of all primary bone tumors.

Key words: McCune-Albright Syndrome, Fibrous Dysplasia, Giant Cell Tumor, Aneurysmal Bone Cyst, Bone Tumors.

Introduction

McCune-Albright syndrome is a rare heterogeneous genetic disorder characterized by a triad consisting of: polyostotic fibrous dysplasia, coffee-colored spots on the skin and multiple hyperfunction endocrinopathies such as: precocious puberty, hyperthyroidism, excess growth hormones, renal phosphate loss and Cushing's syndrome. Whose diagnosis is clinical; of the triad, 2 of the findings are enough to identify it. (1)

Fibrous dysplasia is a rare bone disorder that consists of the replacement of normal bone by an excessive proliferation of fibrous connective tissue with non-functional bone structures. It has three clinical patterns: monostotic, polyostotic and the one associated with McCune-Albright syndrome. (1)

Radiologically, fibrous dysplasia presents an aspect of expansive lesion, with endoscopic festonation, without periostic reaction and with a frosted glass density within the lesion. Fibrous Dysplasia is a rare disorder that represents 2.5% of bone tumors in general and 5-7% of all benign bone tumors and is found equally in both sexes. (2)

In turn, the giant cell tumor is one of the most common benign bone tumors, which occurs in young adults aged 20 to 40 years with a high rate of recurrence and potential secondary malignancy. It is commonly located in the metaphyseal area of the tibia, femur and distal radius. (3)

On the other hand, the aneurysmal bone cyst is a benign bone tumor that represents 1 to 2% of all primary bone tumors. They are expansive osteolytic lesions that contain blood- filled and thin-walled cystic cavities. It is commonly found in patients within the first two decades

of life and is located mainly in the metaphyses of long tubular bones, vertebrae or flat bones. (4)

This tumor can be part of other bone tumors, such as fibrous dysplasia, being located within the cell matrix of the pre-existing tumor, this coexistence is uncommon and the lesion can appear as an aggressive process that is difficult to properly diagnose. (2)

McCune Albright Syndrome is a rare pathology that, as mentioned above, is characterized by a triad in which fibrous dysplasia is present. The case of a 25-year-old patient with this syndrome is presented below, in which the association of two other bone tumors, giant cell tumor and aneurysmal bone cyst was evidenced.

Case Report

25-year-old female patient, with a personal history of childhood asthma, gynecological history of early puberty, late postoperative fixation with DPC plaque and reconstruction plate indicated by fracture of tibia and left fibula, performed in another medical center. Who is evaluated for pain in the right knee and hips. Once the physical examination has been carried out, analgesic restriction to the mobilization of hips and right knee is evident, in addition to coffee-colored spots with milk at the level of the posterior chest, neck, right wrist, facial asymmetry. (Figure 1)

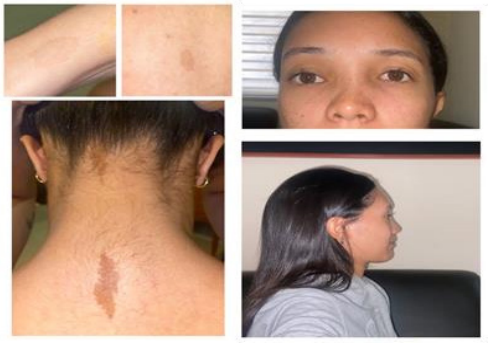

Figure 1: Coffee-colored spots with milk at the level of the posterior chest, neck, right wrist, facial asymmetry.

Radiological studies of symmetrical hip are requested, where it is evidenced: expansive lytic bone lesion, with density in frosted glass at the level of the proximal femur and iliac bones, in addition, deformity is visualized in the staff of bilateral shepherd, suggestive of Fibrous Dysplasia (Figure 2a), in the same way, a radiological study of the knee is requested where a pattern of geographical, lytic bone destruction is appreciated, with bulging of the corticals at the level of the proximal region medial face of the right tibia that impresses a Giant Cell Tumor. (Figure 2b and 2c).

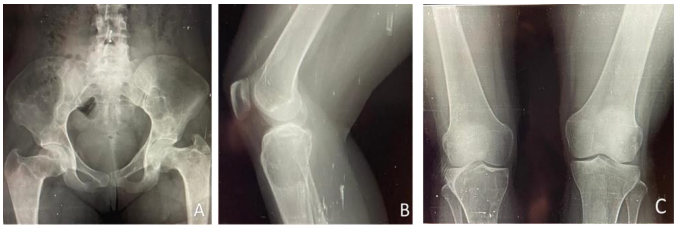

Figure 2a. Symmetrical hip X-ray with images suggestive of Fibrous Dysplasia.

Figure 2b and 2c. X-ray of the right knee in lateral projection where a giant cell tumor can be seen.

In view of the previous findings, it is presumed to be diagnosed with: McCune-Albright Syndrome associated with Giant Cell Tumor in the right proximal tibia under study.

For this reason it is taken to the operating table for the performance of intralesional resection, milling at high revolution, phenolization at 30% of the tumor bed and reconstruction with autologous graft of fibula plus ground spongy corticous bone of bone bank, with subsequent fixation with T plate of proximal right tibia (Figure 3).

A biopsy of the lesion was performed, whose description confirms the diagnosis, since it reports fragments of a lesion made up of round or oval cells arranged in random nests, ovoid nuclei with fine chromatin and without nucleolus, areas with recent hemorrhages, old and hemiderosis (Figure 3).

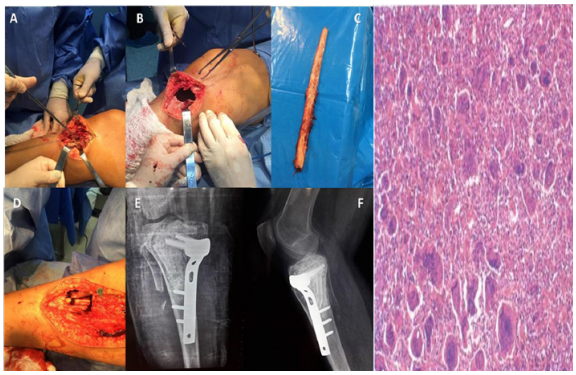

Figure 3: Intralesional resection, reconstruction with autologous fibula graft plus ground spongy cortico bone, Fixation with T-plate of right proximal tibia, Histopathological cut of giant cell tumor.

During the follow-up, the patient began to present pain in the left leg, inflammation and local heat, so an X-ray of the left leg was performed that showed an expansive injury, bulging of the corticals at the level of 1/3 diaphysiary medium of the tibia and fibula (Figure 4).

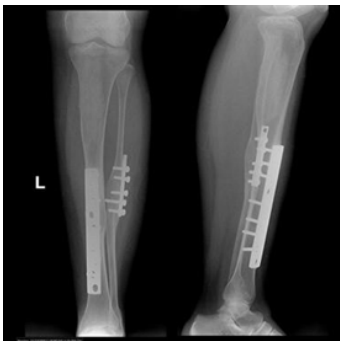

Figure 4: X-ray of left leg with osteosynthesis material, associated with image of an aneurysmal bone cyst.

It was taken again to the operating table for extensive resection of the fibula and intralesional resection of aneurysmal bone cyst through the plate without removing it at the level of the left tibia, biopsy was performed reporting a lesion composed of septa of hyalinized dense connective tissue, hypocellular, separated by spaces with hemorrhagic material, covered with elongated and flattened cells, areas of fibroconnective tissue made up of fusiform cells arranged in intertwined bundles, newly formed capillaries in concomitance abundant spaces full of hemorrhagic material and fiscally identifies newly formed and reactive bone that confirmed the presence of Aneurysmal Bone Cyst (figure 5).

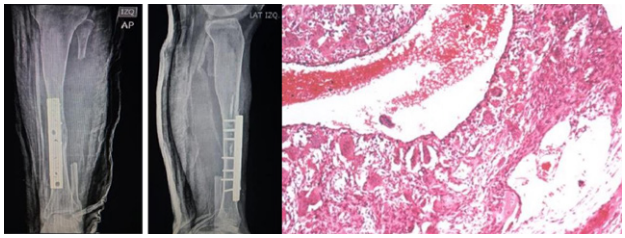

Figure 5: POM of broad resection of fibula and intralesional resection of aneurysmal bone cyst through plate and Histopathological cut.

Currently the patient is in stable general conditions, does not report pain, with walking and preserved mobility (figure 6), without relapses a year and eight months of postoperative, performing annual checks.

Figure 6: Postoperative physical examination.

Discussion

McCune Albright Syndrome (MSA) is a rare disease characterized by the presence of multiple coffee and milk spots on the skin, endocrine alterations, such as early puberty and the presence of fibrous tissue in the bones (1). In this case report, the patient presents coffee-colored spots at the level of the posterior chest, neck, right wrist, early puberty, presence of lytic lesions with frosted glass pattern in the pelvis and bilateral femur plus deformity of shepherd's staff in the right femur, subjective of fibrous dysplasia.

During our search we did not find previous studies describing the association between the proximal tibia giant cell tumor and McCune Albright syndrome (5).

The presence of dysplasia and aneurysmal bone cyst in the same patient with McCune Albright syndrome is very rare. However, these two bone lesions may be related to an alteration in the GNAS1 gene (2). Fibrous dysplasia is believed to be susceptible to the formation of bone cysts due to the vascularization of the lesion (6). In the sample obtained and proceeded for biopsy, it was possible to evidence focal cystic degeneration with hemosiderosis, foci of recent and old hemorrhage, thinning of the corticals, nests of osteoid material with active osteoblasts, confirming the presence of the aneurysmal bone cyst.

In this case, a patient is presented with McCune Albright syndrome and the presence of two bone manifestations, characterized by the presence of aneurysmal bone cyst and giant cell tumor, which is a rare situation in clinical practice, not reported in the literature. Therefore, a comprehensive focus on early detection and adequate treatment of these lesions is necessary to improve the patient's quality of life.

Conclusion

The present case highlights the clinical complexity of McCune-Albright Syndrome, an entity marked by bone and endocrine alterations that can manifest themselves in a highly heterogeneous way. The coexistence of fibrous dysplasia with an aneurysmal bone cyst and a giant cell tumor in the same patient constitutes an exceptional presentation, not previously described in the literature, which highlights the need to maintain a high rate of suspicion against the atypical progression of skeletal lesions in patients with GNAS1 gene mutation.

The histopathological analysis confirmed a secondary cystic process on a dysplastic bone, supporting the theory that the altered microarchitecture and the high intrinsic vascularity of fibrous dysplasia predispose to the development of expansive lesions such as the aneurysmal bone cyst. Likewise, the concomitant identification of a giant cell tumor raises questions about the possible influence of the anomalous bone microenvironment generated by the mosaic mutation characteristic of SMA on the appearance of osteoclastic strain tumors.

This case underlines the importance of a multidisciplinary evaluation, continuous radiological surveillance and an individualized therapeutic approach to prevent functional and structural complications in this type of patient. The rarity of this association reinforces the need to systematically report these presentations to expand the clinical and biological understanding of the bone spectrum of McCune-Albright syndrome and optimize future diagnostic and therapeutic strategies.

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