MCCUNE ALBRIGHT: A CASE REPORT

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ABSTRACT

Objective: To report a case of proximal femur fracture in a 13-year-old patient with Mccune Albright Syndrome - a rare syndrome - and to analyze the clinical outcome with surgical treatment. Materials and Methods: Patient with pain and limited movement after fracture in the right proximal femur due to pathological fracture. Radiographs and diagnoses of subtrochanteric fracture were performed on the right side. As the treatment was performed with open fracture reduction and extra-medullary tutor block with a 95-degree slide, better known as DCS. Results: After 1 month, the patient was asymptomatic, with range of motion and strength restored, comparable to the contralateral side. Conclusions: As it is a fibrous bone displasia, a fracture of the femur becomes a challenge for orthopedic treatment, as demonstrated. Nevertheless, good results are detected when treatment is individualized.

KEYWORDS: FRACTURE, SYNDROME, MACCUNEALBRIGH, ORTHOPEDICS

INTRODUCTION

Poliostotic fibrous dysplasia - a benign bone disorder with a wide spectrum of presentation - when associated with cutaneous café au lait macules and hyperfunctional endocrinopathies, such as precocious puberty and hyperthyroidism, the triad is called McCune Albright Syndrome. The syndrome is a genetic disease caused by somatic mutations in the post-zygotic gene GNAS1. The diagnosis is predominantly clinical and treatment consists of drugs such as bisphosphonate and surgery if necessary. All endocrinopathies must be treated. The estimated prevalence varies between 1/100,000 and 1/1,000,000 [1]. Its repercussions in relation to the musculoskeletal system are mostly multiple fractures resulting from low energy trauma, due to the great fragility presented by the bone of individuals affected by this syndrome. Thus, the patient often presents pathological fractures, better known for being fractures caused by low-energy trauma that would not normally result in fractures [2].

CASE REPORT

Female patient, 13 years old, attended at a tertiary emergency hospital in the city of Anápolis, diagnosed with McCune Albright Syndrome after investigation of precocious puberty with vaginal bleeding at 3 months and pubic hair at 5 months of life. She had had 13 lower limb fractures, has a left ovary cyst and irregular menstrual cycle. On physical examination, she had café au lait spots and hypertrichosis. She sought medical treatment due to a fractured right femoral neck while walking at home. The radiography confirmed a fracture in the subtrochanteric region of the patient's right femur (figure 01).

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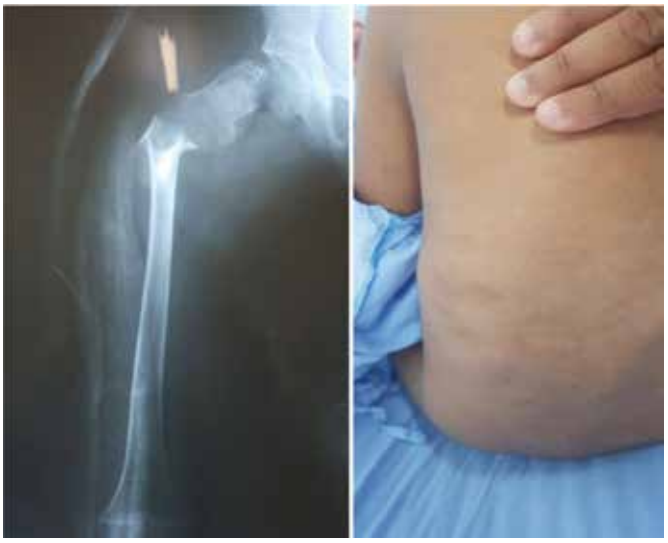

Figure 1 – A. Anteroposterior x-ray of the thigh, showing subtrochanteric fracture. B. Café au lait spots on the patient's skin.

DISCUSSION

MacCune Albright is a highly complex syndrome with several associated signs and symptoms, which makes the clinical picture quite variable. It is the result of a mosaic-type mutation that occurs early in the embryonic stage. Pathophysiology is based on an activating mutation of the gene for the Gs protein subunit, which stimulates the intracellular production of cAMP, conferring autonomous secretion of endocrine, gonadal, thyroid and adrenal tissues. This activating mutation of the Gs protein is also demonstrated in café au lait skin lesions and bone lesions of fibrous dysplasia. Endocrinopathies linked to the syndrome include precocious puberty, hyperthyroidism, diabetes mellitus, acromegaly, Cushing's syndrome, hyperparathyroidism, hyperprolactinemia and gynecomastia [3]. Although precocious puberty is the most frequent clinical manifestation, it is the bone changes that confer the greatest morbidity to the syndrome [4]. Therefore, attention should be paid to the preventive treatment of injuries with the use of bisphosphonates such as that established for the patient in question. This medication aims to reduce the causes of fibrous bone dysplasia, characterized by proliferations of spindle-shaped fibroblasts interspersed with trabeculae of immature bone tissue not surrounded by osteoblasts, causing the expansion of the areas and weakening of the involved bones[5].

Therefore, due to the particularities of this patient, treatment with Flexible Intramedullary Nails was tried as a first option for the treatment of the patient's subtrochanteric fracture with closed reduction and without opening the fracture focus. However, because a good intraoperative reduction was not achieved, a new synthesis material was chosen. The DCS - Dynamic Condylar Screw - that is, an extramedullary tutor with sliding screw with 95 degree lock. Thus, after a good open reduction, the fracture was fixed with DCS, showing good reduction and good fixation [6] (figure 2).

Early mobility, physiotherapy and weekly follow-up with the patient were released. After 4 weeks, he was released to walk with partial load and at that time he had little or no pain and range of motion similar to the contralateral limb. Greater attention was paid to the treatment and guidelines to be followed in order to avoid new fractures.

Therefore, there is a need for individualization in the treatment of patients with special demands as described in this case. This occurs since we have a syndrome with bone demands with low resistance to trauma and patients with special needs. Thus, it is necessary to draw on the experience of the team involved to provide orthopedic treatment to treat the fracture as well as to avoid or reduce the possibility of new fractures providing better living conditions for the patient. It also increases his survival by avoiding new fractures.

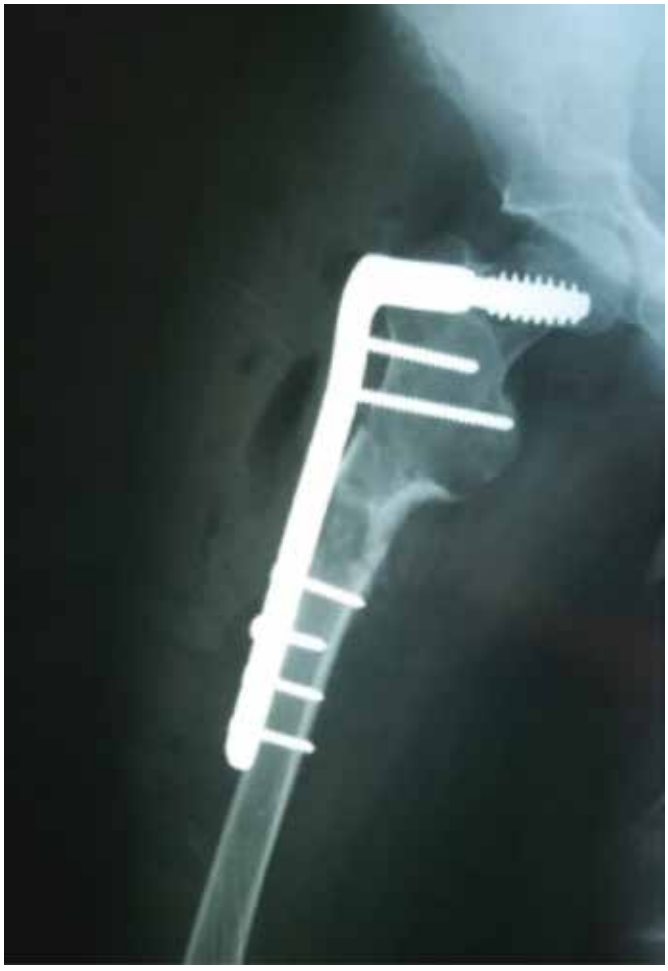

Figure 2 - Anteroposterior x-ray of the thigh, showing the result after fixation with DCS.

REFERENCES

1- Atalla Â. Displasia fibrosa: relato de caso e revisão de literatura. 20th ed. Rev Méd Minas Gerais. Belo Horizonte; 2009. p. 399-403.
2- Chapurlat RD. Medical Therapy in Adults with Fibrous Dysplasia of bone. J Bone Miner Res. 2006 Dec;21(Suppl 2):114-9.
3- Shenker A, Weinstein LS, Moran A, et al. Severe endocrine and nonendocrine manifestations of the McCune-Albright syndrome associated with activating mutations of stimulatory G protein GS. J Pediatr Oct. 1993;123(4):509-18.
4- Alves AL, Canavarros F, Viela DAS, Granato L, Próspero JD. Displasia Fibrosa: relato de três casos. Rev Bras Otorrinolaringol. 2002 mar/abr; 68(2):288-92
5- Terpstra L, Rauch F, Plotkin H, Travers R, Glorteux FH. Bone Mineralization in Polyostotic Dysplasia: Histomorphometric Analysis. J Bone Miner Res. 2002 Nov; 17(11):1949-53.
6- P. Ruedi T. Fraturas Subtrocantéricas do Fêmur. In: P. Ruedi T, ed. by. Princípios AO do tratamento de fraturas. 1st ed. Porto Alegre: ArtMed; 2009. p. 856-858.