Polyostotic fibrous dysplasia with pseudo-precocious puberty and pathological subtrochanteric fracture of femur

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Abstract McCune-Albright Syndrome has orthopedic implications because of the potential for occurrence of pathological fractures and deformities. The case of a nine year old girl with this condition and recurrent pathological fractures of the femur is reported. We believe this is the first such case reported from the Middle East.


Keywords: McCune-Albright Syndrome, pathological fracture, polyostotic fibrous dysplasia, pseudo-precocious puberty

McCune - Albright Syndrome consists of the triad of polyostotic fibrous dysplasia, cutaneous pigmentation and endocrine dysfunction with precocious puberty. Sporadic cases have been reported from different countries. However, we were not able to locate from a literature search, any reported from the Middle East. The case of a nine year old girl with McCune - Albright Syndrome who presented to us with pathological fracture of right femur is described here.

Case Report S.S. (DOB 29-10-1985), an Egyptian girl, was first seen at this hospital at the age of four for enlargement of breast and spotting of blood per vagina. She was the second sibling in the family, born at full term by normal delivery. There was no history of maternal illness or of any medications during pregnancy. Her developmental milestones were normal. Three other siblings, a brother and two sisters were normal. Clinical examination at this age confirmed breast enlargement (B2), without any axillary or pubic hair development. No ovarian or adrenal mass was palpable. A central nervous system examination was normal. She had no symptoms or signs of hyperthyroidism and the thyroid was clinically normal.

Investigations at this stage were shown: Urine microscopy normal, Sp. Gr. 1024; Hemoglobin 12.6 gm/dl, WBC 7.5x10^9/L, MCH 27.3 pg, MCV 78 fl, MCHC 34.4%, Blood Urea Nitrogen 9.2 mg/dl, Sodium 138 meq/L, Potassium 4.1 meq/L, Bicarbonate 21.4 meq/L, Total bilirubin 0.4 mg/dl, Total proteins 6 gm/dl, Albumin 3.6 gm/dl, ALT 37 IU/L. Alk. Phos. 666 IU/L (normal <90 IU/L). Serum free T3 was elevated. Serum Oestradiol level was within normal limits. A skeletal survey showed polyostotic bone lesions and advanced bone age. Ultrasound scan of the abdomen showed that the ovaries and uterus were large for her age. There were no ovarian cysts or adrenal lesions.

She was subsequently lost to follow up until 15th November 1993 when she was readmitted with pain in the right hip and inability to walk following a fall at home. The proximal thigh and the hip were swollen and tender. A general examination showed cafe au lait spots with irregular borders over the back, abdomen, left forearm and left thigh (Fig. 1). Examination of the central nervous system was normal. No masses were palpable in the abdomen. Breasts were enlarged (B3) but she did not have any axillary or pubic hair development. Radiographs showed a displaced angulated subtrochanteric fracture and changes typical of fibrous dysplasia extending from the greater trochanter to the distal
Fig. 1 - Cafe au lait spots over anterior abdomen

Fig. 2 - Subtrochanteric fracture right femur with fibrous dysplasia

Fig. 3 - Right femur 6 months after operation
two third of shaft of femur (Fig. 2). The fracture was treated by skin traction. Skeletal survey showed similar lesions in the left femur, metacarpals, right humerus and the skull. By six weeks the fracture was clinically and radiologically united. She was mobilized on crutches and gradually allowed to weight-bear on the affected side.

She progressed well without any local pain for a further six weeks, when suddenly she felt severe pain at the right hip and could not weight-bear on that side. Radiographs showed that the proximal right femur had refractured at the same site with gross varus angulation and displacement. CT scan of the pituitary gland and the parasellar areas was normal, but the frontal bone and base of skull showed widening of the vault with opacity and small diffuse hypodensity. Serum Free T4, Free T3 and TSH levels were normal. Gonadotropin levels were subnormal. LH-RH test showed pre-pubertal pattern. The fracture was treated by intramedullary nailing to maintain satisfactory reduction and to prevent deformity. The fracture healed satisfactorily in spite of penetration of the lateral cortex of the distal femur by the intramedullary nail (Fig. 3).

Discussion McCune Albright syndrome, independently described in 1937 by McCune and Albright, is a benign disorder characterized by fibrous dysplasia of one or more bones, pseudo precocious puberty and patchy cutaneous (cafe au lait) pigmentation. The vast majority of reported cases are of sporadic occurrence. Both sexes are affected with a slight female preponderance (1:1.2). The bones frequently involved are the femur, tibia, pelvis, phalanges and metacarpals, ribs, humerus, and the base of skull. The basic pathology is fibro-osseous metaplasia. The lesions can remain static or may progressively worsen. Radiologically, the affected area of bone has a ground glass appearance. Curvature of long bones due to weakness and recurrent pathological fractures can lead to shortening and deformities like the "Shepherd's crook deformity" in the femur. Skull involvement can lead to facial disfigurement. Malignant changes can occur occasionally.

Sexual precocity occurs in the majority of females with McCune-Albright syndrome and has been described in males as well. It is often peripheral in origin (gonadotropin independent) due to autonomously functioning follicular cysts which produce sex steroids. Endocrinopathies like hyperthyroidism, acromegaly, hyperparathyroidism, Cushing's syndrome and diabetes mellitus are all considered to be due to hypothalamic hypersecretion of the releasing hormones, autonomous functioning nodular tissue or undue sensitiveness of the involved endocrine organs to the trophic hormones. The skin lesions are hyper-pigmented large macular lesions with irregular borders which occur in over 50% of patients. Histology shows melanin macro globules in the melanocytes of cafe au lait spots.

Conclusion We believe this is the first case of McCune-Albright syndrome reported from the Middle East. As the endocrine changes in this girl are nonprogressive, no active treatment is required at this stage. However because of the extensive involvement of weight-bearing bones, in future she may require either prophylactic intramedullary nailing to prevent pathological fractures or surgery to correct deformities due to weakness and recurrent fractures.

References

عرض ماكسيون - البراعم يحمل مقترحات لقسم العظام بسب عظم الدقة في حالة الكسور والتشوهات. حالة الطفلة ذات التسع سنوات وتكرار تكسر عظمة الفخذ في سجلت. وبعد هذا التقرير الأول من نوعه من منطقة الشرق الأوسط.

مفتاح الكلمات:
متلازمة ماك كيون - البراعم، كسور مرضة، شدن يبني متعدد العظام، البلوغ المبكر الكاذب.