Abstract: McCune Albright syndrome is associated with patchy cutaneous pigmentation (cafe au lait spots) and fibrous dysplasia of the skeletal system. It is associated with peripheral precocious puberty and can also cause pituitary, thyroid and adrenal aberrations. We are reporting a five year old child who presented with knock knees, multiple cafe au lait spots and his whole body skeletal survey revealed polyostotic fibrous dysplasia.

Keyword: McCune Albright syndrome, cafe au lait spots, fibrous dysplasia, precocious puberty.

INTRODUCTION: McCune Albright syndrome is due to non inherited post zygotic mutation of gene GNAS1 with features of polyostotic fibrous dysplasia, multiple cafe au lait spots, precocious puberty and other endocrine problems.

CASE HISTORY: Five year old male child, first born to third degree consanguinous parents presented with knock knees of 6 months duration. Child was evaluated as a case of rickets at a private hospital and referred to ICH for further management. On examination, child had bony prominence over the left supraorbital region, cafe au lait spots extending from the nape of neck up to the gluteal region and bilateral genu valgum. His vitals revealed tachycardia (HR=150/min) with a resting heart rate of 110/min and examination of the other major systems was normal. Blood investigations were normal except for raised ALP levels (509 IU/L). Serum calcium and phosphorous levels were normal but had increased fractional excretion of phosphate. He was further investigated with whole body skeletal survey which revealed patchy areas of bone lysis and sclerosis of skull, elbow, knee and wrist, consistent with polyostotic fibrous dysplasia and his skeletal age was advanced, corresponding to 7 years. With these features of polyostotic fibrous dysplasia, multiple cafe au lait spots and phosphaturia (all typical of McCune Albright syndrome), a diagnosis of the same was made. Further endocrine work up was normal. Tanner staging was done and he had stage 1 sexual development. CT brain also showed fibrous dysplastic changes of the bony prominence over supraorbital region. Cardiology and endocrinology opinion were obtained. ECG confirmed sinus tachycardia and echocardiogram was normal. Child was treated with phosphorous supplements, Pamidronate injection and Propanolol for symptomatic sinus tachycardia. He was advised follow up after 2 months or earlier, if symptomatic.
1. Knock knees.
2. Cafe au lait spots on the nape of neck.
3 & 4. XRay of hand and leg showing fibrous dysplasia.
5. CT skull showing fibrous dysplasia of left supraorbital margin.

**DISCUSSION:**
McCune Albright syndrome (MAS) is an extremely rare disorder with an incidence rate of 1 in 100,000 to 1 in million people that classically affects the bones, skin and endocrine system. MAS is characterised by fibrous dysplasia of bone that occurs with at least two additional findings—patches of abnormal skin pigmentation (cafe au lait spots with jagged borders) and dysfunction of endocrine system. It is a result of sporadic mutation in GNAS 1 gene. Fibrous dysplasia can affect any bone, but most commonly involves skull, facial bones, long bones and ribs. Cafe au lait spots have a characteristic distribution and is termed as 'Coast of Maine'. Hyperphosphaturia is due to phosphatonin.

The most common endocrine abnormality of MAS is precocious puberty which is due to gonadotropin independent autonomous ovarian or testicular function and is more common in girls. Other features include Cushing's syndrome, hyperthyroidism and GH excess resulting in gigantism or acromegaly. Management includes intravenous bisphosphates for polyostotic fibrous dysplasia, aggressive oral phosphorous replacement for hypophosphatemia and calcitriol therapy with calcium and phosphorous for hypophosphatemic rickets. Specific treatment is indicated for associated endocrine problems too.

**FOLLOW UP:**
The child was followed up after 6 months. His endocrine workup and ultrasound abdomen were within normal limits.

**PROGNOSIS:**
It is usually not associated with increased risk of mortality except in a few patients who develop malignancies. Polyostotic fibrous dysplasia can lead to frequent fractures and very rarely malignant transformation. Long term Calcitriol and phosphorous supplementation can lead to nephrocalcinosis and loss of renal function over time.

**CONCLUSION:**
We have presented this case for its rarity in a male child.

**REFERENCES:**
1. McCune Albright syndrome: a case report in a male; Pubmed journal Nov 2010 - by Patel KB.
4. Nelson textbook of paediatrics- Endocrine system, chapter 556.6, page no 1891.
5. Nelson textbook of paediatrics-Endocrine system,chapter 556.6, page no 1891.
7. National Organization for Rare Disorders-NORD.