

## CASE REPORT



# McCune-Albright Syndrome in a 4-year-old Nigerian child: A case report from the University of Maiduguri Teaching Hospital.

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## Abstract

McCune-Albright syndrome is a genetic disorder that affects the skin, bones, and endocrine systems. It is a mosaic disease caused by mutations in the somatic-activating GNAS gene. It is a very rare disease and there are limited reports from Nigeria. We therefore present a 4-year-old girl presented with a 2-month history of cyclical vaginal bleeding, bilateral breast enlargement (Tanner stage 3, with galactorrhea), and a 2-year history of limping. Examination revealed café-au-lait macules on the left trunk, axilla, and neck, along with pre-pubertal vaginal bleeding (intact hymen, bloodstained vulva). Laboratory findings showed elevated alkaline phosphatase and suppressed luteinizing hormone (LH) and follicle-stimulating hormone (FSH) levels. A diagnosis of McCune-Albright syndrome was thus made. This report highlights an African paediatric case of this rare disorder.

## Introduction

The McCune-Albright syndrome (MAS) is characterised by a triad of poly/monostotic fibrous dysplasia (PFD), precocious puberty, and Café au lait spots.<sup>1</sup> Fuller Albright was the first to describe this syndrome in 1937.<sup>1</sup> The first case described by Donovan McCune had the above classic triad and hyperthyroidism.<sup>2</sup> However, the exact incidence of MAS remains unknown, but its prevalence is thought to range between 1 case per 100,000 population and 1 case per 1,000,000 population, making it a very rare disorder.<sup>2</sup> While MAS shows no specific ethnic or gender predisposition, reported cases occur more frequently in girls than in boys.<sup>3</sup>

Fibrous dysplasia (FD) is a rare non-malignant condition of the bone. It is characterised by the replacement of bone by dysplastic tissue in a single skeletal site (monostotic FD) or in several bones (polyostotic FD).<sup>4,5</sup> The relative incidence of monostotic fibrous dysplasia is higher than the polyostotic variants among the diagnosed cases.<sup>3</sup> While rare, fibrous dysplasia may undergo malignant transformation, most

commonly to osteosarcoma or fibrosarcoma.

The precocious puberty associated with MAS is mostly common in females, and it represents the most common endocrine abnormality seen with the disease.<sup>2</sup> Others include acromegaly or gigantism due to growth hormone excess and hyperthyroidism.<sup>1</sup> The sexual precocity is characteristically gonadotrophin-independent, resulting from excess estrogen production from ovarian follicular cysts.<sup>2</sup> Because the sexual precocity associated with MAS is gonadotropin-independent, it is more accurately described as pseudo-precocious puberty.<sup>4</sup>

Café au lait spots, the classic symptoms, often predominate on the side ipsilateral to the side with more bony features and deformity.<sup>7</sup> Given the variable expressivity of MAS, radiologic investigations play an important role in the evaluation of patients with MAS as they help confirm fibrous dysplasia and assess the extent of disease.<sup>7</sup> This case underscores the importance of a high index of suspicion in

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children presenting with café-au-lait lesions and early pubertal development, as timely diagnosis and multidisciplinary management can significantly improve outcomes.

### CASE REPORT

A 4-year-old Nigerian girl referred to the gynecology clinic of the University of Maiduguri Teaching Hospital with a two-month history of cyclical vaginal bleeding and progressive breast enlargement. Additionally, her parents reported a two-year history of limping, which had gradually worsened over time. There was no history of previous trauma. The child is the second child to the parents of a monogamous setting of seven children, and there was no history of similar illness in the family. The mother did not attend the routine antenatal clinic during the index pregnancy that resulted in the delivery of the child. She claimed to have delivered spontaneously at 9 months, and the child was healthy after birth. The child did not receive any immunisation, and there was delayed attainment of normal developmental milestones.

General examination shows that the child was not pale, anicteric, and had no peripheral lymphadenopathy. There are large, prominent hyperpigmented macules on the left side of the trunk, particularly around the axilla and nape (Fig. 1). The lesions end abruptly at the midline. On musculoskeletal examination (Fig. 2), there was genu valgum on the left and genu varum on the right, given the so-called “windswept” deformity. There was no associated joint pain or stiffness. The child displayed advanced secondary sexual characteristics, with Tanner stage 3 breast development bilaterally and expressible galactorrhea on palpation. A limited vaginal examination (inspection only) shows well-developed labia minora and majora with an intact hymen and blood-stained vulva noted.

Cardiovascular, respiratory, and neurological examinations revealed no abnormalities. Based on the constellation of findings, including precocious puberty, characteristic café-au-lait macules, and skeletal deformities, we established a provisional diagnosis of gonadotropin-independent precocious puberty, with McCune-Albright syndrome as the leading consideration. Neurofibromatosis remained an important differential diagnosis requiring exclusion.

Laboratory investigations revealed increased levels of estradiol 131.1 pg/mL (normal range 10 – 24 pg/mL), and growth hormone 19.8 ng/mL (normal range is 0.2 – 10 ng/ml) and alkaline phosphatase with both random blood FSH and LH levels <0.1 mIU/mL. Thyroid function test, full blood count, and urinalysis were all within normal limits.

Radiological evaluation included bilateral leg radiographs and pelvic ultrasound. Plain films demonstrated multiple expansile lytic lesions with cortical thinning and scattered sclerotic foci in both tibiae and fibulae (Fig. 3). Pelvic ultrasound revealed uterine enlargement (AP diameter: 4.0 cm) and a right ovarian enlargement (4.0×3.5 cm) with peripheral follicular development (Figs 4-5), confirming precocious puberty. These findings, combined with the clinical presentation, established a diagnosis of McCune-Albright syndrome. While arrangements were being made for specialist referral to the endocrinology team at University College Hospital, Ibadan, for expert management, the patient was unfortunately lost to follow-

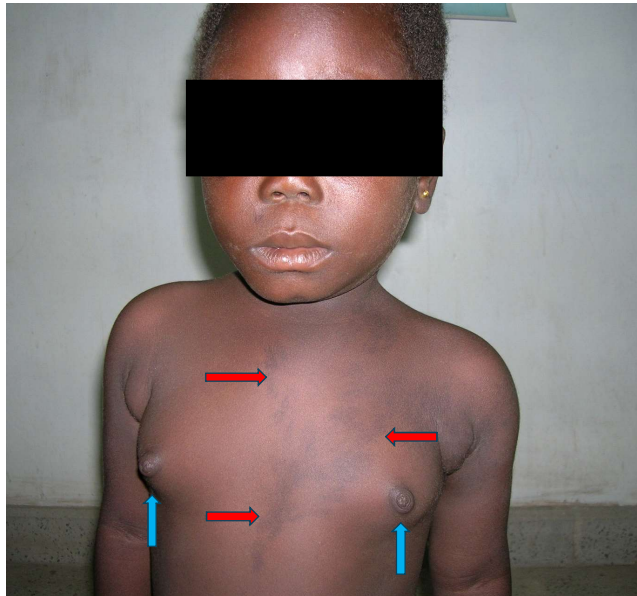


Fig. 1 A photograph of the patient showing areas of hyperpigmentation on the left side of the trunk and nape (Red arrow) and bilateral well-developed breasts, Tanner stage 3 (Blue arrows).



Fig. 2. A photograph of the patient showing wind-swept deformity and pubic hair



Fig. 3 plain AP radiograph of both legs showing multiple mixed lytic and a few sclerotic areas in the tibia and fibula bilaterally (Red arrows).

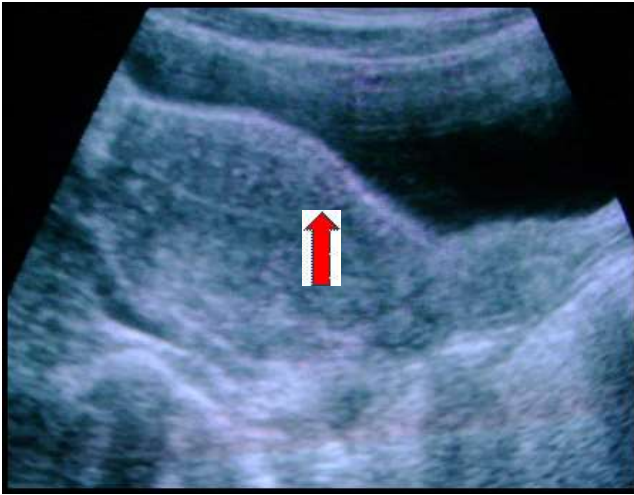


Fig. 4. A longitudinal scan of the pelvis showing an enlarged uterus (red arrow) for the patient's age.

up.

## DISCUSSION

McCune-Albright syndrome (MAS) classically presents with the triad of fibrous dysplasia (typically unilateral), café-au-lait spots, and endocrine dysfunction, most commonly precocious puberty.<sup>3,6</sup> Our case demonstrates an unusual bilateral and symmetrical presentation of the bony lesions. While MAS affects both sexes, the female predominance in reported cases likely reflects the more conspicuous presentation of precocious puberty, including thelarche and menarche<sup>2</sup>, as seen in our 4-year-old patient. The clinical presentation of the disease is variable<sup>5</sup>, and many cases remain undiagnosed until adulthood.<sup>4</sup> However, the index patient's early presentation with both skeletal (limping) and endocrine manifestations (cyclical vaginal bleeding and Tanner stage 3-breast development) informed early presentation and diagnosis.

The classic musculoskeletal manifestations of MAS typically include bone pain, pathological fractures, and gait disturbances, particularly limping secondary to leg length discrepancies caused by the characteristic "shepherd's crook" deformity of the proximal femur.<sup>3,6</sup> In severe polyostotic fibrous dysplasia (PFD), recurrent pathological fractures in early childhood may lead to progressive gait abnormalities, visible skeletal deformities, and secondary osteoarthritis with associated joint stiffness and pain.<sup>7,8</sup> Our case presented an atypical musculoskeletal picture, lacking the classic shepherd's crook deformity, likely due to the patient's young age at presentation. Instead, she exhibited a distinctive "windswept" lower limb deformity

While café-au-lait spots typically localise ipsilateral to the more severely affected skeletal side, our case demonstrated left-sided predominance despite symmetrical bilateral bone involvement.<sup>9</sup> The classic triad of cutaneous, skeletal, and endocrine manifestations typically renders diagnosis unambiguous, as in our patient. However, atypical presentations may mimic systemic mastocytosis or neurofibromatosis type 1<sup>2</sup>, particularly when marked skeletal deformity is present. Additional differential diagnoses include ossifying fibroma and Paget's disease of bone, the latter distinguished by its adult onset, contrasting with the pediatric presentation of MAS.<sup>2</sup>

If precocious puberty predominates, as in this case, the

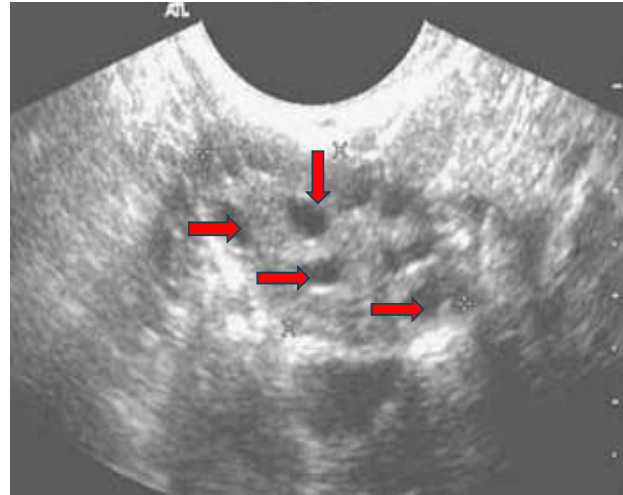


Fig.5. A transverse pelvic scan showing an enlarged right ovary with multiple growing follicles (arrows)

differential diagnosis becomes even wider and includes congenital adrenal hyperplasia. Vaginal bleeding in neonates and infants is one of the typical presentations of MAS as demonstrated in this case presented. Rhabdomyosarcoma of the vaginal tract is an important differential. This typically appears as a bunch of grapes, hence the name sarcoma botryoides.<sup>10</sup>

Furthermore, plain radiographs in McCune-Albright Syndrome typically reveal multiple patchy areas of bony lysis and sclerosis, consistent with bone dystrophy, which is evident in the present case. Characteristic features include hypertrophic regions, geodes with fine sclerotic rims, and a mix of radiopaque and radiolucent areas with thin or thickened cortices.<sup>9</sup> Although virtually any bone can be involved, frequently affected sites include the femur, tibia, ribs, and facial bones, with hand and foot involvement seen in up to 50% of cases.<sup>4</sup> In this case, both the tibia and fibula are affected, indicating polyostotic disease. In addition, bone lesions tend to localise to the metaphyseal and diaphyseal regions of long bones and may appear trabeculated, with thin cortices and a classic ground-glass appearance.<sup>2</sup> The current case shows a predominantly diaphyseal lesion. While radiographic features such as osteolysis and cortical destruction can raise concern for malignant transformation, this could not be established in the index case.

Ultrasonography can aid in evaluating soft tissue swelling. In MAS, myxomas typically appear as well-defined hypoechoic masses with occasional central fluid-filled cavities<sup>10</sup>, these features were not observed in this case. CT scans may reveal hypodense areas suggestive of myxomas and help assess facial bone deformities;<sup>9</sup> Also, on MRI, myxomas appear hypointense or isointense on T1-weighted images with gadolinium enhancement, and hyperintense on T2-weighted sequences.<sup>2,9,11</sup> MRI also provides a detailed assessment of bony involvement, comparable to bone scans. However, both investigative modalities were not performed due to financial constraints. Radionuclide bone scans using technetium-99m-labelled methylene diphosphonate can highlight areas of increased uptake in polyostotic fibrous dysplasia, useful for determining disease extent post-diagnosis.<sup>2,11</sup> This investigation was also not done, as such facilities were not available at our centre.

## CONCLUSION

This case highlights the clinical presentation of McCune-

Albright Syndrome in a 4-year-old Nigerian child, a condition rarely reported in Nigeria. It underscores the value of pelvic ultrasonography, plain radiographs, and accessible hormonal profiling in supporting diagnosis, particularly in resource-limited settings. Early recognition using these readily available tools is crucial for timely management and improved outcomes.

#### Availability of data and materials

All data relating to this study are presented within the manuscript.

#### Ethics approval and consent to participate

Written informed consent for publication was obtained from the parent of the patient.

#### Consent for publication

Written informed consent for publication of their clinical details was obtained from the relative of the patient.

#### Competing interests

All authors of this manuscript declare no competing interests.

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NMT conceptualized the report and led the manuscript drafting. AMS and SSG contributed to literature review and manuscript revision. NMT and YMD critically reviewed and edited the final manuscript. All authors approved the final version of the manuscript.