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Case Report

Radiology Section

Unveiling McCune-Albright Syndrome in a Case of Ovarian Cyst with Bony Lesions: A Case Report

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ABSTRACT

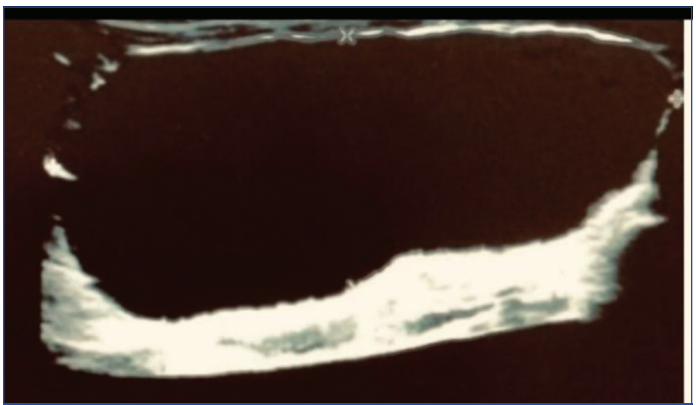
McCune-Albright Syndrome (MAS) is a non-inherited disorder caused by missense point mutations in the GNAS1 gene located on the long arm of chromosome 20. It is a sporadic disease characterised by polyostotic Fibrous Dysplasia (FD), café-au-lait lesions, and a variety of endocrine disorders, including gonadal hyperfunction, hyperfunction of the thyroid and adrenal cortex, as well as excessive Growth Hormone (GH) secretion. The diagnostic triad includes polyostotic FD, precocious puberty, and café-au-lait lesions. When at least two of these are present, the diagnosis of MAS is confirmed. We present a case of an 18-year-old girl who presented with abdominal distension and a history of ovarian cystectomy. A detailed history revealed precocious puberty with vaginal bleeding at age three. Imaging showed a large, well-defined ovarian cyst with no solid component or septation. Magnetic Resonance Imaging (MRI) and Computed Tomography (CT) revealed multiple expansile lytic lesions with a ground-glass matrix in the pelvic bones, skull, and sphenoid, suggestive of polyostotic FD. Café-au-lait macules and hormonal findings pointed towards MAS. This case highlights the importance of considering MAS in young females with recurrent ovarian cysts, skeletal abnormalities, and early pubertal changes, as ovarian cysts with bony lesions do not always indicate metastasis and require careful evaluation to avoid misdiagnosis.

Keywords: Fibrous dysplasia, Lytic expansile lesion, Recurrent ovarian cyst

CASE REPORT

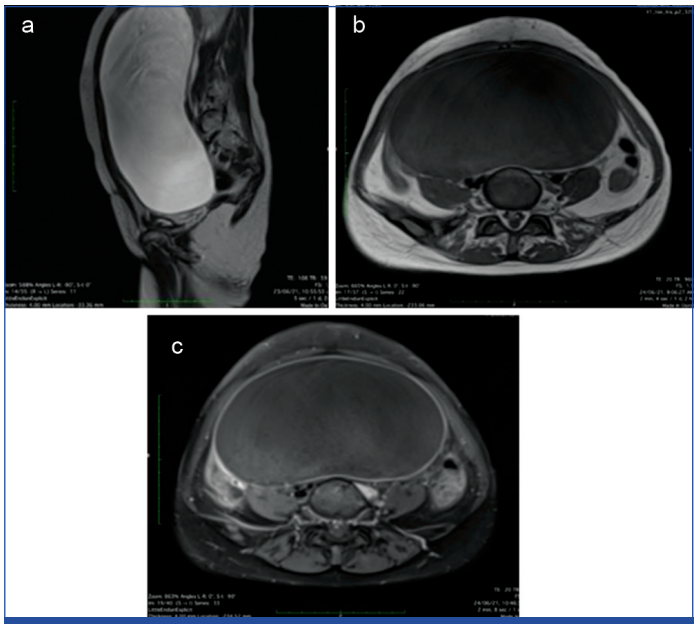
An 18-year-old girl presented with abdominal distension for three weeks, occasionally accompanied by pelvic discomfort. She also experienced menorrhagia, though her cycles remained nearly regular, with no history of bone pain or bony deformities. She had a history of ovarian cystectomy five years prior, with histopathological examination reporting a benign follicular cyst. A detailed history revealed an episode of vaginal bleeding at the age of three years. There was no history of bleeding from any other orifices, trauma, or child abuse. The gynaecologist advised an Ultrasonography (USG), for which she was referred to us.

In the USG abdomen, the right ovary showed a well-defined, thin-walled, anechoic cystic lesion measuring 14 cm in the pelvis with posterior acoustic enhancement. There was no septation, calcification, fat density, or solid component, suggesting a likely Ovarian-Adnexal Reporting and Data System for Ultrasound (ORADS US) III lesion. The left ovary showed a small cystic lesion measuring 2 mm, with no septation, calcification, fat density, or solid component, indicating a likely ORADS US I lesion [Table/Fig-1].



[Table/Fig-1]: USG abdomen: Thin-walled, anechoic cystic lesion in the pelvis with posterior acoustic enhancement. No septation, calcification, fat density, or solid component, noted suggesting a likely ORADS US III lesion.

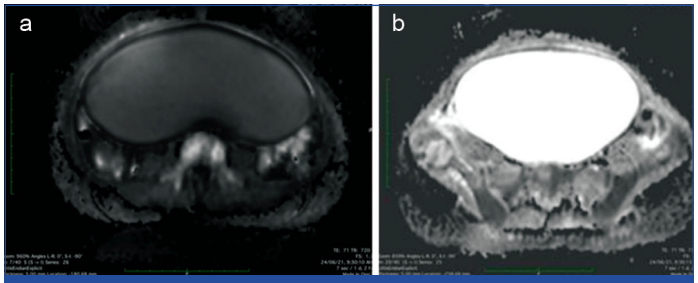
MRI was done for further characterisation. T2-weighted images (axial, coronal, sagittal sections) showed a well-defined, smooth-walled, large T2 hyperintense cystic lesion extending superiorly up to the diaphragm, anteriorly reaching the anterior abdominal wall, displacing the bowel laterally, and extending up to the prevertebral area posteriorly. The lesion appeared to originate from the right adnexa, with the right ovary not separately visualised. The left ovary showed a T2 hyperintense cystic lesion. There was no evidence of ascites [Table/Fig-2].



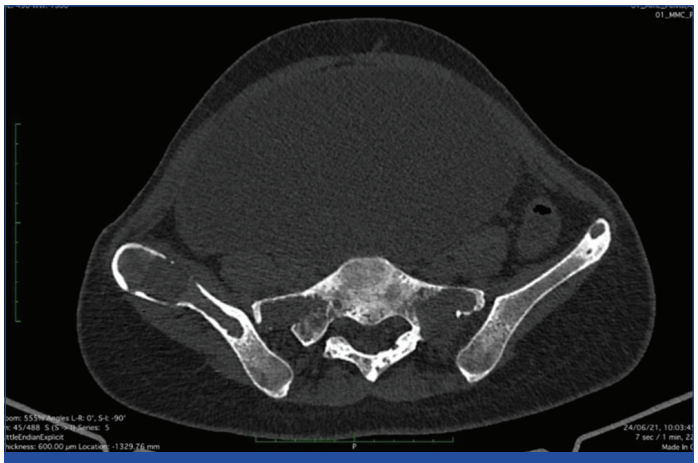
[Table/Fig-2]: Sagittal T2-weighted images showing a large well-defined, smooth-walled, T2 hyperintense cystic lesion in right adnexa, with the right ovary not separately visualised; T1-weighted MRI : Lesion appears hypointense; Post-contrast images (T1 C+(Gd)): Mild, smooth enhancement of the wall, with no enhancing septation or solid component.

T1-weighted MRI showed the hypointensity of the lesion. Post-contrast images (T1 C+(Gd)) showed mild, smooth enhancement

of the wall, with no enhancing septation or solid component [Table/Fig-2]. Diffusion-Weighted Imaging (DWI) and Apparent Diffusion Coefficient (ADC) sequences showed no evidence of diffusion restriction in the ovarian lesion. Multiple bony lesions with diffusion restriction were noted [Table/Fig-3]. CT correlation was done, which showed a well-defined large cystic lesion measuring 20×7 cm in the right adnexa. There was no evidence of calcification, septation, solid component, or papillary excrescence. Multiple expansile lytic lesions with cortical destruction and a ground glass matrix were noted in the pelvic bone [Table/Fig-4]. The right ovary was not separately made out.

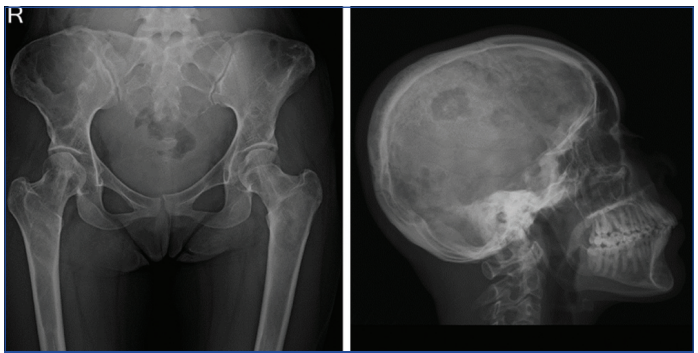


[Table/Fig-3]: DWI & ADC: No evidence of diffusion restriction in the ovarian lesion. Multiple bony lesions with diffusion restriction noted.



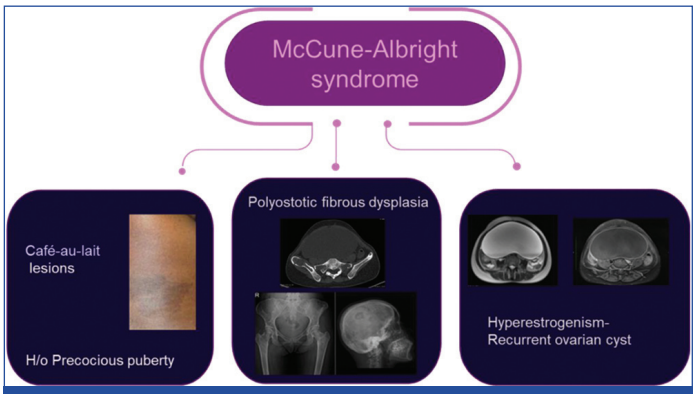
[Table/Fig-4]: CT pelvic bone: Multiple expansile lytic lesions with cortical destruction and a ground glass matrix noted in the pelvic bone.

Pelvic X-ray showed multiple lytic expansile lesions with a ground glass matrix. Skull X-ray showed multiple lytic expansile lesions with a ground glass matrix [Table/Fig-5]. CT brain bone window showed bony expansion with cortical destruction of the right squamous temporal bone and left sphenoid with a ground glass matrix. No soft-tissue component was noted, with features suggestive of polyostotic FD.



[Table/Fig-5]: X-ray pelvis: Multiple lytic expansile lesion with ground glass matrix noted. X-ray skull: Multiple lytic expansile lesion with ground glass matrix noted.

The patient had a history of vaginal bleeding at the age of three years. On examination, the patient had a normal vaginal orifice with an intact hymen, bilateral enlargement of the breasts with no pubic hair and café-au-lait macules son the trunk. History, physical examination, and imaging features suggested café-au-lait lesions, polyostotic FD, and hypoestrogenism, which manifested as precocious puberty and recurrent ovarian cysts, pointing toward a diagnosis of MAS [Table/Fig-6]. The patient was advised a multidisciplinary approach for management. Hormonal therapy with aromatase inhibitors was recommended to regulate ovarian function and prevent recurrent cyst formation. Bisphosphonates were suggested to improve bone health and reduce the risk of fractures. Regular imaging follow-up, including pelvic ultrasound and bone scans, was advised to monitor disease progression. Surgical intervention was advised for ovarian cyst as the patient was symptomatic.



[Table/Fig-6]: Features suggestive of McCune-Albright syndrome.

DISCUSSION

The MAS is characterised by the triad of polyostotic FD, precocious puberty, and café-au-lait lesions. When FD of bone with at least any one of typical endocrinopathies and/or café-au-lait spots is present, the diagnosis is confirmed [1].

The FD is defined as 'a pathological condition in which normal bone is altered by abnormal fibro-osseous tissue, causing distortion and overgrowth of the affected bone.' It can present with bone pain, fractures, and deformities. FD manifests in bone lesions categorised as cystic, sclerotic, or mixed patterns. Axial skeleton lesions typically exhibit smooth, homogeneous, radiolucent ground glass matrices, with craniofacial FD appearing dense and sclerotic [2]. Lesions vary in size and may cause cortical thinning, though maintaining a smooth outer contour. CT is preferred for assessing morphological changes, defining lesion anatomy, and detecting soft-tissue masses or bone destruction. MRI shows variable signal intensities on T1- and T2-weighted images, aiding in lesion evaluation and detecting malignant transformation and soft-tissue extension [3].

Numerous case reports have highlighted the challenge of differentiating FD from bone metastasis, as it is often misinterpreted on both MRI and PET imaging [4,5]. MRI characteristics in patients with histopathologically confirmed Fibrous Dysplasia of Bone (FDB) typically show low-intensity signals on T1-weighted sequences and high-intensity signals on T2-weighted sequences [6], consistent with our case. The signal intensity on T1- and T2-weighted images, along with the degree of contrast enhancement on T1-weighted images, may aid in differentiating FD from metastasis [7]. However, bone metastases also present with low-intensity signals on T1-weighted sequences and a mix of high-intensity signals on T2-weighted sequences, similar to FDB and our findings. Consequently, relying solely on MRI for distinguishing FDB remains challenging [4].

It is important to note that a CT scan is the most effective technique for visualising the radiographic features of FDB, with its most common pattern being a characteristic ground-glass appearance on imaging [8].

Approximately, 2-3% of FD patients have extra-skeletal manifestations, known as MAS. In MAS, bone alterations are typically more severe than in polyostotic FD without extra-skeletal involvement, often resulting in multiple fractures that necessitate surgical intervention [9]. Eighty-five percent of female MAS patients develop functionally active ovarian cysts, leading to gonadotropin-independent precocious puberty

[10]. Ultrasound typically reveals large unilateral ovarian cysts, which may be haemorrhagic and display both cystic and solid elements. Recurrent ovarian cysts cause intermittent oestrogen production, resulting in breast development, growth acceleration, and vaginal bleeding; between cyst formations, breast tissue regresses, and oestrogen levels return to prepubertal levels [10]. These cysts may persist into adulthood, causing irregular menstruation and potentially affecting fertility [11]. Pituitary disorders are seen in 10-15% of MAS patients, frequently involving autonomous growth hormone production associated with skull base FD, and many display pituitary adenomas or diffuse somatotroph hyperplasia [12]. Gastrointestinal abnormalities, including Intraductal Papillary Mucinous Neoplasms (IPMN) in the pancreas, affect 15% of patients and can progress to malignancy [13]. Skin abnormalities, such as café-au-lait spots with jagged borders, are among the earliest manifestations of the disease [14].

Mazabraud syndrome is a rare condition characterised by the presence of FD lesions and myxomas, which are typically located near the bone lesions [15].

CONCLUSION(S)

The MAS, a sporadic disorder, can also present later in life as recurrent ovarian cysts. High suspicion should be maintained whenever a young girl presents with recurrent ovarian cysts and skin pigmentation of café-au-lait spots. Exploring differentials like this can alter treatment strategies.

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