www.sada.co.za / SADJ Vol. 80 No.2
https://doi.org/10.17159/sadj.v80i02.20620
The SADJ is licensed under Creative Commons Licence CC-BY-NC-4.0.

McCune-Albright Syndrome: A case report with a radiological review

SADJ MARCH 2025, Vol. 80 No.2 P114

A ASMAL[1]

ABSTRACT

A case of the rare McCune-Albright syndrome was diagnosed in a patient who presented the classical triad – fibrous dysplasia (bone), skin hyperpigmentation (skin) and precocious puberty (endocrine). The syndrome affects boys and girls equally but tends to present earlier in females. This report follows a 15-year-old male patient who presented with MAS at an oral health centre. Dental radiographic examinations were done revealing the characteristic lesions of fibrous dysplasia (FD) in the mandible and maxilla. These were critical in the diagnosis of the patient, and together with histology, confirmed that invasive surgical and prolonged orthodontic treatment were contra-indicated Common sites for FD lesions are the craniofacial and femoral regions. Baseline scans used to diagnose FD lesions are bone scintigraphy together with plain films. Further investigation may involve computed tomography (CT), magnetic resonance imaging (MRI) and ultrasound (US) scans. Follow up monitoring of the syndrome requires further imaging, contributing to the management and treatment of the patient. Malignancy associated with MAAS is rare. US scans of suspicious areas must be carried out to ensure early detection of cancer. This report has demonstrated the use of a variety of radiographical techniques to track the bony lesions, especially Cone Beam Computed Tomography (CBCT).

Keywords

McCune-Albright Syndrome; fibrous dysplasia; endocrinopathy.

Abbreviations

MAS = McCune-Albright Syndrome
FD = Fibrous Dysplasia
PP = Precocious Puberty
PAN = Panoramic radiograph
CBCT = Cone Beam Computed Tomography

CASE REPORT

A 15-year-old male presented at an oral health centre in 2009 seeking orthodontic treatment related to a malocclusion. He was short and stockily built with a slight facial asymmetry and evidence of bowing in both tibia. He complained of “bony hard lumps” on both humeri and a stiffness of the left shoulder. A routine panoramic (PAN) radiograph examination showed pockets of bony dysplasia on the mandible, prompting further investigation. Subsequently an excisional biopsy was carried out by a maxillo-facial and oral surgeon. The oral pathology report indicated the dysplasia to be a benign fibro-osseous lesion. Further investigations included plain film imaging. These studies confirmed the presence of a large fibro-dysplastic (FD) lesion on the left humerus (Figure 1), and a slight scoliosis was also observed. The patient was referred to the department of endocrinology and an overall diagnosis of McCune-Albright Syndrome was confirmed in 2010. The patient’s weight and height were monitored over time, the recording showing him to be well below the 50% growth percentile for males of his age.[1]

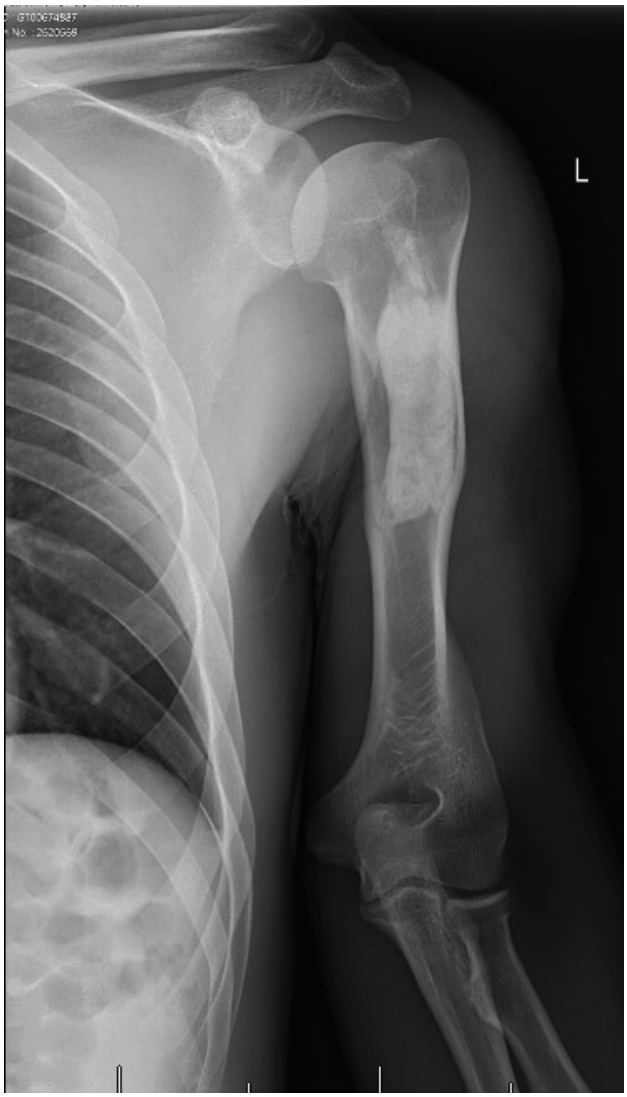

Figure 1: The radiopaque lesion in the shaft of the left humerus.

Authors’ information

1. Ayesha Asmal, Qualifications - N Dip in Diagnostic Radiography (DUT), B.Tech in Diagnostic Radiography (CPUT), Postgraduate Diploma in Health Sciences Education (Wits), Affiliations - Radiographer/lecturer, Department of General Dental Practice, School of Oral Health Sciences, Faculty of Health Sciences, University of Witwatersrand. Johannesburg.
   ORCID: https://orcid.org/0000-0003-1081-6989

Corresponding author
Name: Ayesha Asmal,
Contact Number: 076 645 7086
Email address: Ayesha.asmal1@wits.ac.za

https://doi.org/10.17159/sadj.v80i02.20620
The SADJ is licensed under Creative Commons Licence CC-BY-NC-4.0.

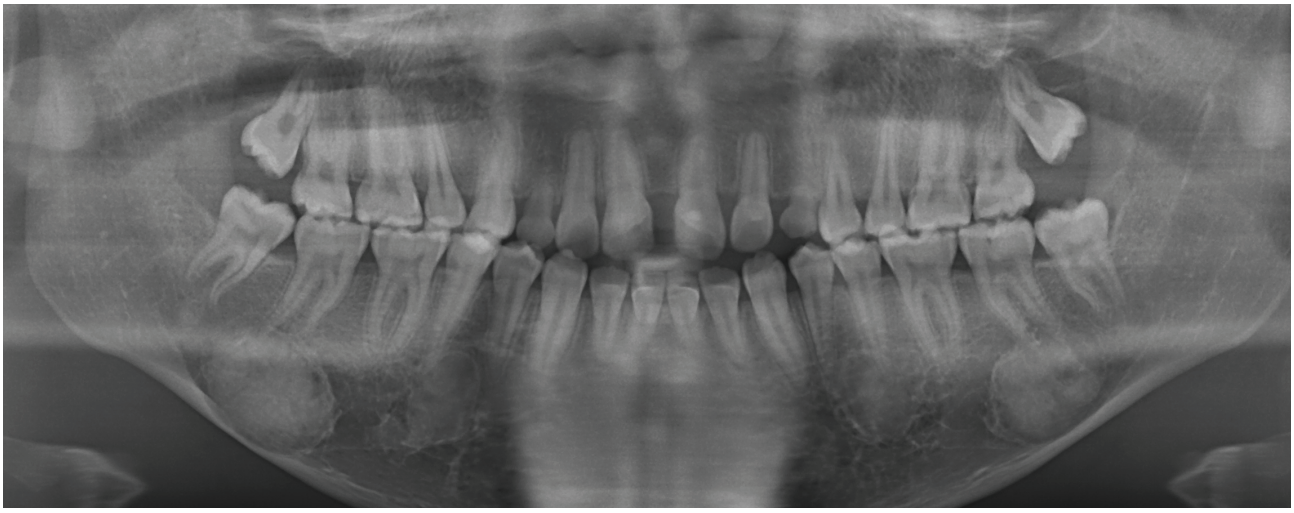

Figure 2: A PAN taken in January 2013, showing the four radiopaque lesions in the mandible. Note the slight cortical expansion on the right inferior border.

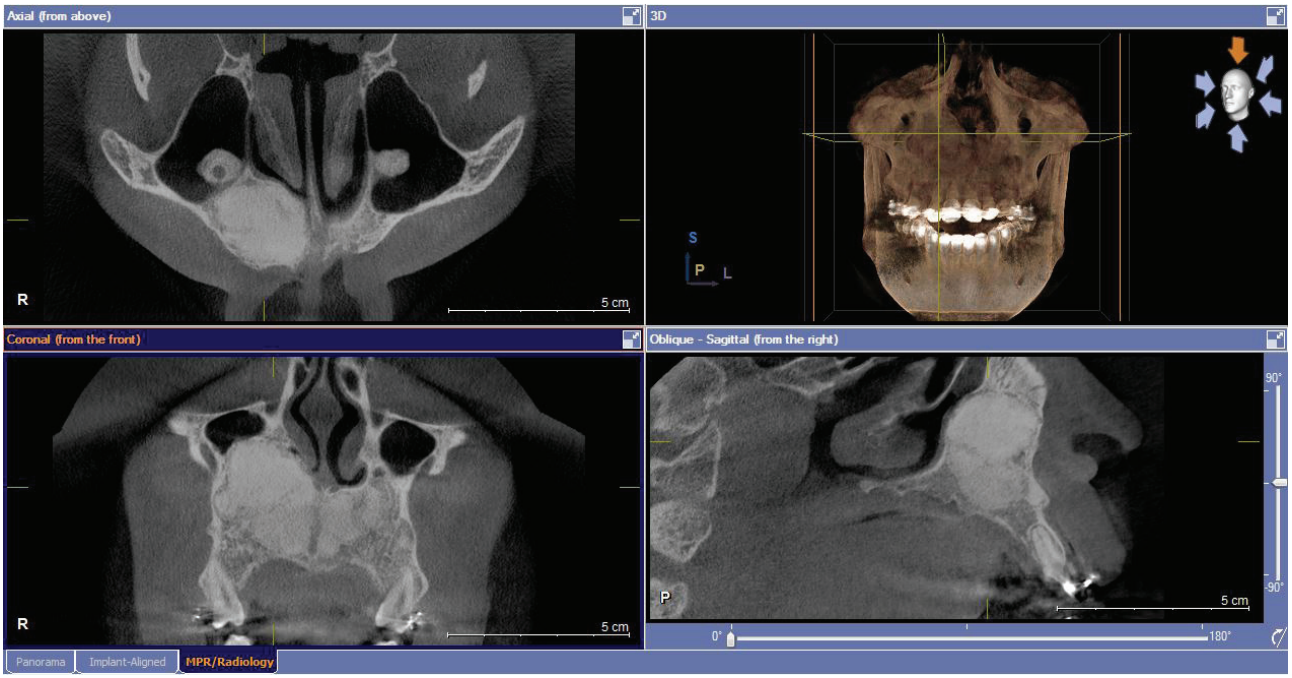

Figure 3: CBCT scan taken in 2016, revealing extensive lesions in the maxilla, encroaching into the right sinus.

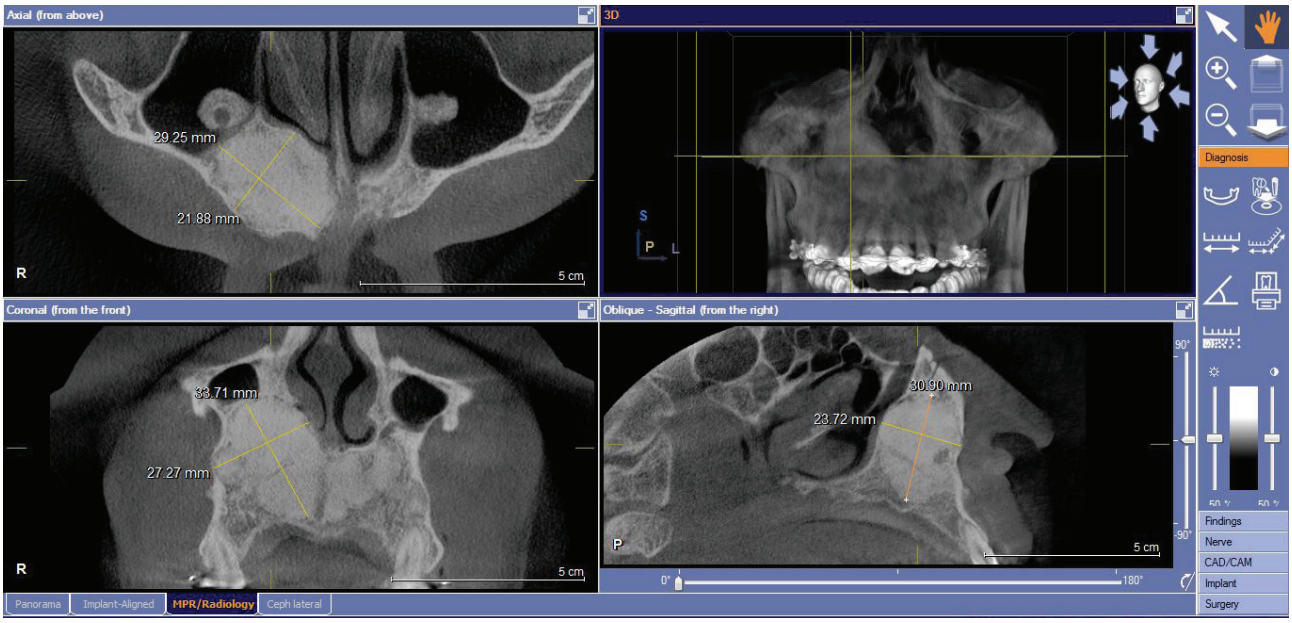

Figure 4: The technology of the Gallileos system enabled progressive measurements, allowing for monitoring of the size and extent of the lesions.

www.sada.co.za / SADJ Vol. 80 No.2
https://doi.org/10.17159/sadj.v80i02.20620
The SADJ is licensed under Creative Commons Licence CC-BY-NC-4.0.

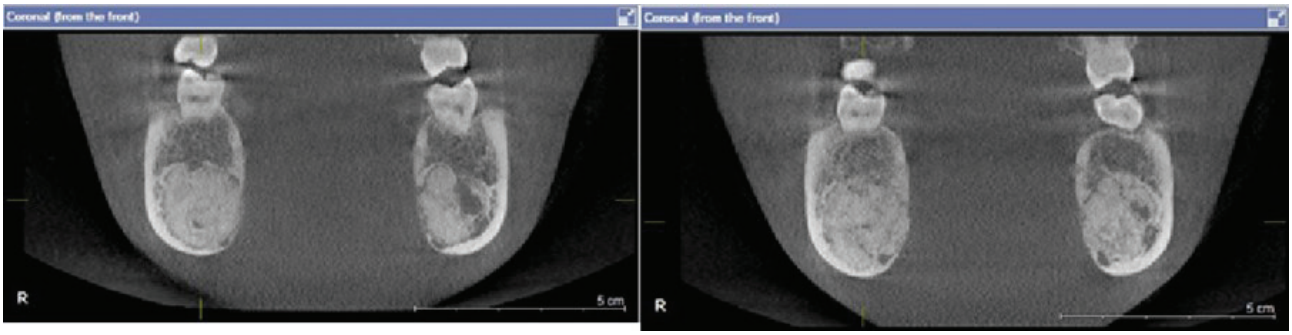

Figure 5: Comparison of consecutive coronal views showing the possible conversion of woven bone into soft tissue with accompanying increased radiolucency. Left image 2016, Right image 2022.

Table I: Measurements repeated over several years did not reveal regression or other changes of clinical relevance.

	In mm	2016	2022
Maxilla A	Axial L	27.88 mm	28.81 mm
	Axial W	23.65 mm	21.95 mm
	Coronal L	34.57 mm	36.00 mm
	Coronal W	27.49 mm	28.17 mm
	Sagittal L	30.92 mm	31.74 mm
	Sagittal W	23.38 mm	22.87 mm
Maxilla B	Axial L	18.19 mm	18.09 mm
	Axial W	12.26 mm	13.34 mm
	Coronal L	19.54 mm	22.04 mm
	Coronal W	13.08 mm	13.06 mm
	Sagittal L -	19.72 mm	18.33 mm
	Sagittal W	17.12 mm	20.33 mm
Mandible A	Axial L	20.92 mm	20.20 mm
	Axial W	17.73 mm	16.17 mm
	Coronal L -	20.53 mm	21.77 mm
	Coronal W	20.47 mm	22.01 mm
	Sagittal L	22.17 mm	22.25 mm
	Sagittal W	16.72 mm	18.25 mm
Mandible B	Axial L	16.12 mm	17.87 mm
	Axial W	12.22 mm	12.73 mm
	Coronal L -	14.86 mm	15.54 mm
	Coronal W	13.45 mm	14.15 mm
	Sagittal L -	14.42 mm	15.54 mm
	Sagittal W	14.24 mm	16.07 mm
Mandible C	Axial L	11.62 mm	13.60 mm
	Axial W	9.34 mm	9.15 mm
	Coronal L	14.14 mm	13.99 mm
	Coronal W	11.10 mm	12.37 mm
	Sagittal L	15.25 mm	16.61 mm
	Sagittal W	10.53 mm	10.60 mm
Mandible D	Axial L	14.66 mm	16.89 mm
	Axial W	13.03 mm	13.09 mm
	Coronal L	15.64 mm	16.62 mm
	Coronal W	13.66 mm	14.98 mm
	Sagittal L	22.23 mm	22.88 mm
	Sagittal W	16.20 mm	15.97 mm

PAN radiographs and Cone Beam Computed Tomography (CBCT) scans taken between 2013 and 2022 enabled periodic assessment of the lesions and assessment of the possibility of any dental treatment. The 2013 PAN revealed four radiopaque lesions in the mandible with slight cortical expansion below the 47 (lower 2nd molar) (Figure 2). Maxillary lesions were discovered when a CBCT scan was taken in January 2016 (Figure 3). The oral radiologist's report of the 2017 CBCT scan identified the lesions to be a form of fibrous dysplasia (FD), presenting variously "ground glass" and pitted "orange peel" app earances. The mandibular lesions encroached on the inferior alveolar canals on both sides. The patient, however, did not complain of paraesthesia. This may be due to the thinning of the bone and encroachment to have largely occurred lingually, as seen on cross-sectional views.

The assessment of the lesions over time revealed minor apparent changes in measurements which had no clinical relevance (Figure 4 and Table 1). Changes in the radiographic appearance in the 2022 CBCT scan may have been associated with conversion of woven bone into soft tissue (Figure 5). However, this would require confirmation by biopsy. It was clear from the radiological findings that further major dental treatment was completely contraindicated.

The patient is a healthy and active individual currently professionally employed. He maintains that he currently suffers no pain or discomfort and is satisfied with the status. He is determined to live his life to the full.

DISCUSSION

Fibrous dysplasia (FD) is an erratic, uncommon skeletal disorder where normal bone is replaced with fibro-osseous tissue.[2] FD can be monostotic or polyostotic leading to pain, deformity, fractures and/or functional impairment.[3] FD lesions are not evident at birth clinically developing within the first 10 years of life. The lesions progress during childhood, becoming static by adulthood with possible improvement in late adulthood.[4] Radiographically, FD lesions in children younger than two years appear radiolucent/cystic. The classic "ground glass" appearance of the lesions develops with age.3 There has been an association between FD and scoliosis. Plain film imaging plays a role in diagnosing and monitoring scoliosis as it can be progressive.[2]

FD commonly affects the craniofacial and proximal femur regions. Craniofacial FD can lead to facial asymmetry – which can result in malocclusion. The proximal femur region characteristically displays the classic *coxa vera* deformity (commonly known as "shepherd's crook") resulting from mechanical stress in the area.[5] Craniofacial FD is generally an incidental finding on computed tomography (CT) or magnetic

https://doi.org/10.17159/sadj.v80i02.20620
The SADJ is licensed under Creative Commons Licence CC-BY-NC-4.0.

resonance imaging (MRI) scans as it can be asymptomatic.[3] Research indicates a full body scan (bone scintigraphy) should be used for early detection and evaluation of the extent of FD lesions, followed by plain film imaging in two planes on clinically significant areas identified from the scintigraphy scan.[2,6,7] CT scans will best demonstrate the extent of craniofacial FD and stress fractures, with MRI being useful for suspected bone cysts.[8]

First described in 1937 as a triad of FD [bone], skin hyperpigmentation (café-au-lait spots) [skin] and precocious puberty (PP) [endocrine] became known as the McCune-Albright Syndrome (MAS) which affected both genders equally.[6,8] The syndrome has a prevalence estimated between 1:100 000 and 1: 1 000 000.[6,8,9] It is a non- inherited genetic disease and is not associated with environmental factors or ethnic groups.[4,5,10] This article is a follow-up of a paper published in *The South African Radiographer*[1], and investigates the role of radiology in the diagnosis and management of such cases.

Later, it was found that many other endocrinopathies are associated with the syndrome making it a much more complex syndrome than originally thought.[4,9] A finding of FD associated with café-au-lait spots raises the possibility of a diagnosis of MAS.9 The café-au-lait spots are present at birth (or shortly after), but the definitive diagnosis of MAS is made only once other symptoms manifest.[8]

PP is sometimes the presenting symptom in females (occurring in 80-85%) with vaginal bleeding occurring as early as three or four years of age.[1,6,8] Pelvic ultrasound (US) assessments may identify unilateral or bilateral ovarian cysts varying in size and complexity or, in males, testicular lesions and microlithiasis.[3,9]

There is a rare association between MAS and malignancies – these may occur in breast, thyroid, testicular, bone and pancreatic tissues.[5] US scans should be considered if there is any indication that lesions may be present.[4] A study carried out by Robinson et. al. (2018) indicates that gastro-intestinal (GI) manifestations are common in patients with MAS. Using contrast-enhanced abdominal MRI and magnetic resonance cholangiopancreatography (MRCP) in MAS patients with a history of pancreatitis, the authors found the presence of GI abnormalities in 56% of their patients with 46% having intraductal papillary mucinous neoplasms (IPMNs) – the most common type of neoplastic pancreatic cancer.[11] IPMNs can lead to invasive adenocarcinoma and usually occur at a mean age of 63 years. MAS patients recorded an earlier incidence, and it was found that the mean age dropped to 35 years.[11] Osteonecrosis of the jaw (ONJ) is another risk associated with FD/MAS as malocclusion has a negative impact on basic oral hygiene.[5] Panoramic radiographs provide this assessment of the maxillary and mandibular regions, giving an overall assessment of oral hygiene status.[7] Therefore, regular dental visits to monitor and manage any association with ONJ is crucial.[5,7] Other manifestations associated with MAS are platelet dysfunction and a spectrum of gastrointestinal disease, such as polyps, adenomas and cysts.[3]

As a result of the complexities and challenges with which each patient may present, the best approach in managing MAS patients would be a multidisciplinary team effort as there is no specific approved treatment.[4] Patients and their families must be supported throughout this journey making it emphatically known that this is a non-inherited disease with the rare development of malignancy.[2] Optimal management should ensure that the patient carries out regular physical exercise and attend routine and regular medical and dental visits to monitor any progress of the condition.[2,7]

The identification of the syndrome has affected and restricted the dental management of the patient. In an event of remission, perhaps some minor orthodontic treatment can be explored. Any treatment which would normally require surgery will most likely be contraindicated given the condition of the bone.

Radiological investigations play a crucial role in detecting MAS and may lead to symptomatic management for patients who suffer early pain. Baseline scans used to diagnose FD lesions are bone scintigraphy together with plain films. CT and MRI scans can be further investigations used. Additionally, baseline pelvic and testicular US scans must be carried out when PP is suspected. Monitoring the signs and symptoms of the syndrome require regular follow up imaging, playing a role in the management and treatment the patient will receive. Malignancy associated with MAS is rare, but US scans of suspect lesions must be considered to ensure early detection of cancer.

REFERENCES

1. Asmal A, Evans WG. McCune-Albright syndrome: a case report and review. The South African Radiographer [Internet]. 2020;39–42. Available from: www.sorsa.org.za
2. Hartley I, Zhadina M, Collins MT, Boyce AM. Fibrous Dysplasia of Bone and McCune–Albright Syndrome: A Bench to Bedside Review. Vol. 104, Calcified Tissue International. Springer New York LLC; 2019. p. 517–29.
3. Boyce AM, Collins MT. Fibrous dysplasia/McCune-albright syndrome: A rare, mosaic disease of Gαsactivation. Vol. 41, Endocrine Reviews. Endocrine Society; 2020. p. 345–70.
4. Robinson C, Collins MT, Boyce AM. Fibrous Dysplasia/McCune-Albright Syndrome: Clinical and Translational Perspectives. Vol. 14, Current Osteoporosis Reports. Current Medicine Group LLC 1; 2016. p. 178–86.
5. Spencer T, Pan KS, Collins MT, Boyce AM. The Clinical Spectrum of McCune-Albright Syndrome and Its Management. Vol. 92, Hormone Research in Paediatrics. S. Karger AG; 2020. p. 347–56.
6. Zhang R, Wang P, Wang Y, Meng Z, Jia Q, Wang K, et al. McCune-Albright syndrome confirmed by 99mTc-MDP. Eur J Nucl Med Mol Imaging. 2020 May 1;47(5):1326–8.
7. Javaid MK, Boyce A, Appelman-Dijkstra N, Ong J, Defabianis P, Offiah A, et al. Best practice management guidelines for fibrous dysplasia/McCune-Albright syndrome: A consensus statement from the FD/MAS international consortium. Orphanet J Rare Dis. 2019 Jun 13;14(1).
8. Van Doan N, Minh Duc N, Kim Ngan V, Van Anh N. McCune-Albright syndrome onset with vaginal bleeding. BMJ Case Rep. 2021 Jul 19;14(7).
9. Dumitrescu CE, Collins MT. McCune-Albright syndrome. Vol. 3, Orphanet Journal of Rare Diseases. 2008.
10. Leet AI, Collins MT. Current approach to fibrous dysplasia of bone and McCune-Albright syndrome. Vol. 1, Journal of Children's Orthopaedics. British Editorial Society of Bone and Joint Surgery; 2007. p. 3–17.
11. Robinson C, Estrada A, Zaheer A, Singh VK, Wolfgang CL, Goggins MG, et al. Clinical and radiographic gastrointestinal abnormalities in McCune-Albright syndrome. Journal of Clinical Endocrinology and Metabolism. 2018;103(11):4293–303.