CASE REPORT

DOI: https://doi.org/10.20453/reh.v34i1.5307

Cite as:
Ramírez R, Rivero O, Morales YR. Orofacial clinical-radiological features of McCune-Albright syndrome in an adult: a case report. Rev Estomatol Herediana. 2024; 34(1): 83-87. DOI: 10.20453/reh. v34i1.5307

Received: September 8, 2023
Accepted: December 4, 2023
Online: March 31, 2024

Conflict of interest: The authors declare that they have no conflict of interest.
Funding: Self-funded.
Ethics approval: Approval of the Ethics Committee from the Maxillofacial Surgery Service of Hospital Provincial Clínico Quirúrgico Docente Manuel Ascunce Domenech de Camagüey on April 3, 2022.
Authorship contribution:
RRS: conceptualization, data curation, formal analysis, investigation, methodology, project administration, resources, software, supervision, validation, visualization, writing – original draft, writing – review & editing.
ORP: conceptualization, data curation, investigation, methodology, resources, validation, writing – original draft, writing – review & editing.
YRMP: conceptualization, data curation, formal analysis, investigation, software, validation, visualization, writing – review & editing.

Corresponding author:
Reynier Ramírez Suarez
Address: Av. Los Ancianos E11 A29. Rpto. Previsora. Camagüey, Cuba.
Phone: +5358771830
Contact: reynierramirez93@gmail.com

Orofacial clinical-radiological features of McCune-Albright syndrome in an adult: a case report

Reynier Ramírez Suarez[1, 2, a, b, c], *Oscar Rivero Pérez*[1, 2, a, b, c], *Yanara R. Morales Paz*[3, d]

ABSTRACT

Fibrous dysplasia is a benign and progressive bone disease of genetic basis that can affect one or several bones. Its high incidence in the pediatric age served as a basis for it to be previously considered exclusive to this stage; however, there are reports to date that show its development and later growth in the second decade of life. When associated with endocrine imbalances and café-au-lait spots, it is part of the McCune- Albright syndrome. The present case report shows the clinical and radiologic features of the oral-facial region of an adult patient with McCune-Albright syndrome. Lesions in the jaws affect the morphology and cause dysfunction. At the dental level, the changes produced result in malocclusion and structural defects. Radiological studies showed changes with mixed density and ground glass pattern.

Keywords: McCune-Albright syndrome, fibrous dysplasia, café-au-lait spots.

INTRODUCTION

Fibrous dysplasia is a benign fibro-osseous lesion of bone first described by Lichtenstein in 1938 (1). It is a pathology of very low prevalence, so its diagnosis goes unnoticed in most cases (2). It is of genetic basis and slow progression, characterized by the progressive replacement of normal bone by fibrotic tissue. The process can affect a single bone (monostotic fibrous dysplasia) or multiple bones (polyostotic fibrous dysplasia) (3). The most common location of fibrous dysplasia is in the membranous bones, such as the femur, tibia, and pelvic bone. The skull is also a frequent site of involvement (4).

[1] Universidad de Ciencias Médicas de Camagüey, Facultad de Estomatología. Camagüey, Cuba.
[2] Hospital Provincial Clínico Quirúrgico Professor Manuel Ascunce Domenech. Camagüey, Cuba.
[3] Universidad de Ciencias Médicas de Camagüey, Facultad de Medicina. Camagüey, Cuba.
[a] Stomatologist.
[b] Maxillofacial Surgeon.
[c] Professor of the Facultad de Estomatología.
[d] Physician.

Traditionally, it has been considered a disease of childhood, inactive after puberty; however, the emerging concept now seems to be that fibrous dysplasia can progress into adulthood (5).

When fibrous dysplasia is associated with endocrinopathies or hyperpigmented skin lesions, it corresponds to the McCune-Albright syndrome (2). The aim of this study is to describe clinical and radiological oral-facial features of the McCune-Albright syndrome in an adult patient in order to report the increased bone growth in this stage of life and thus support the current concepts that dismiss fibrous dysplasia as an exclusive childhood disease.

CASE PRESENTATION

A 32-year-old female patient from a rural area came to the maxillofacial surgery department concerned about a facial growth in the last five years. During the interrogation, she reported that in her childhood she began to have a discrete increase in volume in this area, so she had a checkup in the pediatric services of her province of origin. In the clinical record presented by the patient, a diagnosis of McCune-Albright syndrome was registered in 2005, and it was issued by a multidisciplinary consultation and supported by a histological result of fibrous dysplasia, history of menarche at the age of 8 and café-au-lait spots in the affected region. Due to unspecified reasons, the patient did not attend her medical checkups and argued that the greatest growth occurred in the last five years.

After the oral physical examination, facial asymmetry was observed, marked by an increase in volume of hard consistency in the right region, slight pupillary unevenness, deviation of the labial commissure towards the lower contralateral side. There were no dermatological alterations (Figure 1). Oral examination showed midline deviation to the left side during the opening movement and slight limitation to the right laterality with respect to the left. Increased gingival stippling was observed in the gingiva of the right mandibular hemiarch, as well as increased protrusion and anterior overjet. Clinical, laboratory and imaging studies were conducted, as well as consultations with other specialties, including internists, endocrinologists, radiologists, psychologists, general stomatologists, orthodontists, and periodontists.

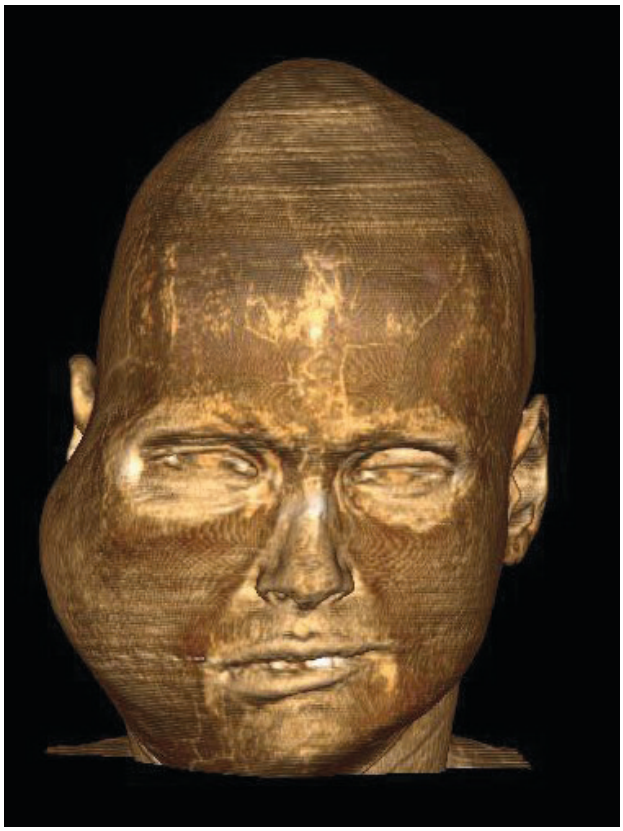

Figure 1. Volumetric reconstruction with soft tissue view showing facial asymmetry due to volume increase in the right facial region. No photograph is exposed by patient's decision in the consent form.

Panoramic radiography showed heterogeneous radiopacity in the right mandibular body and ramus region with predominant lytic areas in the angle and marked sclerotic lesions in the condyle. The roots of the lower molars were separated. At the same time, apical to them, the bone adopted the typical ground glass pattern. The lower right third molar was absent; in a superior position and distal to the apex of the second molar, it presented a radiopaque image with unclear morphological features similar to an odontoma. The left third molars did not present morphological anomalies (Figure 2).

Axial and coronal views on tomography showed lesions with thinning of the cortex, abnormal bone pattern, and loss of trabecular architecture. No alterations were found in the maxillary sinus, ethmoidal cells or orbital cavity (Figure 3).

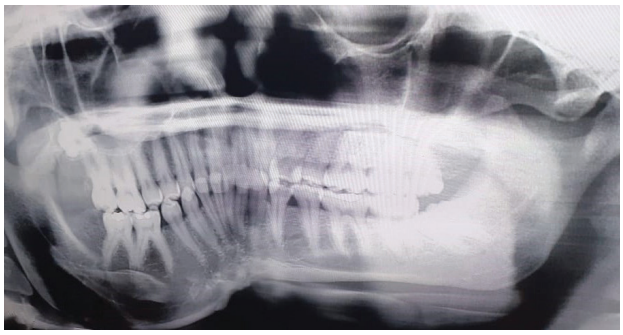

Figure 2. Panoramic radiograph showing mixed radiopacity in body and left mandibular ramus with presence of lytic areas, sclerotic and ground glass pattern in the dentate region. Radiopaque area with odontoma aspect in the region of 18 and absence of 48.

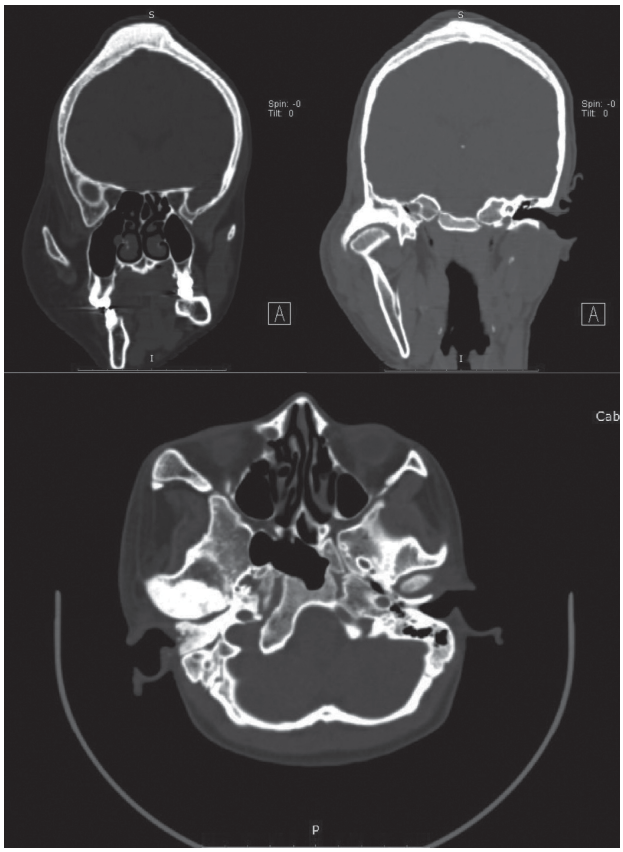

Figure 3. Axial and coronal reconstructions showing bone lesions and preservation of the maxillary and ethmoidal sinuses and the orbital cavity.

Volumetric reconstruction showed an expansive growth involving the right side of the mandible and the zygomatic bone, as well as a protrusion at the cranial upper pole. A height difference was observed between both infraorbital rims. The right mental foramen was inferior to the left one (Figure 4).

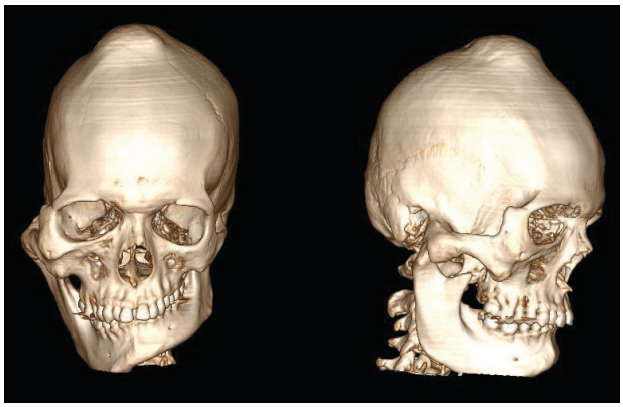

Figure 4. Volumetric reconstruction (frontal and lateral). There is an expansive growth affecting the right mandibular side and the zygomatic bone, as well as a protrusion in the upper pole of the skull.

Laboratory and other additional tests were found in normal parameters. There were no functional alterations, except for a slight restriction of lateral mandibular movement. By decision of the patient, an expectant therapeutic approach was applied.

DISCUSSION

Benign fibro-osseous lesions rarely affect the sinonasal tract and are divided into three different entities: osteoma, fibrous dysplasia and fibroma ossificans (6, 7). Fibrous dysplasia is a rare skeletal disease caused by activating mutations of the GNAS1 gene (8-10). It is known to frequently affect craniofacial bones, including the maxilla and mandible. However, its effects on dental tissues and implications for dental care are still unclear (11).

In the 1930s, McCune and, a year later, Albright presented patients with certain characteristics: fibrous dysplasia of one or more bones, café-au-lait spots on the body and precocious puberty. These patients were later identified as carriers of the syndrome that bears the surnames of these physicians: McCune-Albright syndrome (12). This disease is a rare, non-inherited condition. Although prevalence data are not available, it has been reported to occur in ranges from 1/100 000 to 1/1 000 000 000 (8, 12).

Café-au-lait macules are an isolated and common dermatologic finding in the general population (13) and are due to an active proliferation of melanocytes and consequent hyperproduction of melanin (14). In a study by Akintoye et al (8), it was pointed out that 63% of patients with this syndrome had multiple dysregulated endocrine/metabolic functions. The

most common were hyperthyroidism, precocious puberty and renal phosphate wasting.

Gnatic fibrous dysplasia usually appears in the second or third decade, which may be due to initial misdiagnosis or lack of symptoms. More than 90% of lesions are monostotic and affect only one bone, which is true only for the mandible in the craniofacial region because fibrous dysplasia lesions in the maxilla can cross sutures to the sphenoid, zygoma, skull base and frontonasal bones, thus affecting more than one bone (1, 9).

In fibrous dysplasia, the bone grows in the form of a painless swelling, while the maxilla and mandible are most often affected in the head and neck area, and the ethmoid bone is rarely affected. Once the definitive diagnosis has been made through histopathological studies of fibrous dysplasia, consultation is necessary to rule out an endocrine disorder (15). General imaging studies can rule out extracranial extension of fibrous dysplasia (plain radiographs, CT scans or bone scans). Clinically, lesions can be classified as inactive (stable), non-aggressive (slow growth) or aggressive (rapid growth ± pain, pathological fractures, malignant transformations, etc.) (1).

Maxillary lesions affect both orbital morphology and content and dental occlusion. At the mandibular level, it presents with a mass at the lower border of the mandible, and later the progression of the disease will lead to dysfunction (16). Oligodontia, enamel hypoplasia, enamel hypomineralization, as well as tooth wear, rotation and displacement are examples of dental anomalies in fibrous dysplasia. The infraorbital nerve and the inferior alveolar nerve may be involved in the lesion (1).

The progression of fibrous dysplasia often decreases as patients reach puberty; however, cases with ongoing active disease have been reported (1, 5). In adulthood it can be reactivated, for example, during pregnancy (1).

Akintoye et al. (8) observed four types of radiological changes in fibrous dysplasia: ground glass (granular/condensed trabeculae), radiolucent (lytic), mixed radiolucent/radiopaque (mixed density), and radiopaque (sclerotic). The characteristic "ground glass" appearance is the result of interwoven or abnormal bone superimposed on a matrix of fibrous tissue (17). It has been observed that older subjects and patients treated with bisphosphonates have radiologically sclerotic lesions (8, 17).

Computed tomography is excellent for assessing the volume and morphology of tumors and can be helpful in determining the degree of bone involution. Magnetic resonance imaging offers a more preponderant specificity in neurovascular and ocular involution (5). A ground-glass appearance on CT scans, thinning of the cortical bone and bulging of the affected area are the distinctive and special features of fibrous dysplasia (5, 8). The indications for surgical procedures include functional and esthetic alterations of the affected area (8). In the present case, despite having marked facial asymmetry, the patient advocated a wait-and-see approach.

CONCLUSIONS

In this case report, the clinical-radiological characteristics of an adult patient with fibrous dysplasia could be appreciated, where the greatest dysplastic changes occurred after 25 years of age. The diagnosis was supplemented at the age of 15 years as McCune-Albright syndrome because it was associated with endocrine imbalances and café-au-lait spots. Deforming lesions were observed in the bones of the facial mass with marked asymmetry. Structural dental alterations and malocclusion determined the fundamental oral features. Radiological studies showed the typical ground-glass pattern and mixed density changes. A multidisciplinary approach and the use of appropriate diagnostic aids are important to establish the involvement of this disease.

REFERENCES

1. Obermeier KT, Hartung JT, Hildebrandt T, Dewenter I, Smolka W, Hesse E, et al. Fibrous dysplasia of the jaw: advances in imaging and treatment. J Clin Med [Internet]. 2023; 12(12): 4100. Available from: https://doi.org/10.3390/jcm12124100
2. Rienzi T, Silveri C, Risso M, Mendoza B, Bianchi G. Displasia fibrosa poliostótica - Síndrome de McCune-Albright. Rev Méd Urug [Internet]. 2021; 37(1): e37113. Available from: https://doi.org/10.29193/RMU.37.1.12
3. Carías A, Díaz V. Displasia fibrosa monostótica craneofacial. Acta Otorrinolaringol Cir Cabeza Cuello [Internet]. 2021; 49(4): 315-320. Available from: https://doi.org/10.37076/acorl.v49i4.545
4. Brucoli M, Garzaro M, Dosio C, Boffano P, Benech A. The surgical management of monostotic fibrous dysplasia of the inferior turbinate. J Stomatol Oral Maxillofac Surg [Internet]. 2020; 121(4): 457-459. Available from: https://doi.org/10.1016/j.jormas.2019.10.014

5. AlMomen AA, Molani FM, AlFaleh MA, AlMohisin AK. Endoscopic endonasal removal of a large fibrous dysplasia of the paranasal sinuses and skull base. J Surg Case Rep [Internet]. 2020; 2020(1): rjz404. Available from: https://doi.org/10.1093/jscr/rjz404
6. Tovío Martínez EG, Urbano del Valle SE, Vergara Hernández CI, Díaz-Caballero AJ. ¿Displasia fibrosa o fibroma osificante? Caracterización histológica de dos casos inusuales. Univ Odontol [Internet]. 2019; 38(81). Available from: https://doi. org/10.11144/Javeriana.uo38-81.dffo
7. Díaz V, Carías A. Displasia fibrosa en seno etmoidal y esfenoidal: a propósito de un caso. Rev Otorrinolaringol Cir Cabeza Cuello [Internet]. 2021; 81(1): 68-71. Available from: http://dx.doi.org/10.4067/S0718-48162021000100068
8. Akintoye SO, Otis LL, Atkinson JC, Brahim J, Kushner H, Robey PG, Collins MT. Analyses of variable panoramic radiographic characteristics of maxillo-mandibular fibrous dysplasia in McCune-Albright syndrome. Oral Dis [Internet]. 2004; 10(1): 36-43. Available from: https://doi.org/10.1046/j.1354-523x.2003.00971.x
9. Jiménez C, Schneider P, Baudrand R, García H, Martínez A, Mendoza C, et al. Caracterización clínica de pacientes chilenos con displasia fibrosa/síndrome de McCune-Albright. Rev Méd Chile [Internet]. 2022; 150(10): 1275-1282. Available from: http://dx.doi.org/10.4067/S0034-98872022001001275
10. Guzmán Menco E, López Aparicio E, Harris Ricardo J. Displasia fibrosa: parámetros a considerar para la decisión quirúrgica. Rev Haban Cienc Méd [Internet]. 2018; 17(3): 451-461. Available from: https://www.revhabanera.sld.cu/index.php/rhab/ article/view/1413
11. Akintoye SO, Lee JS, Feimster T, Booher S, Brahim J, Kingman A, et al. Dental characteristics of fibrous dysplasia and McCune-Albright syndrome. Oral Surg Oral Med Oral Pathol Oral Radiol Endod [Internet]. 2003; 96(3): 275-282. Available from: https://doi.org/10.1016/S1079-2104(03)00225-7
12. Uribe González G, Sigler Morales L. Síndrome de McCune-Albright en un adolescente. Informe de un paciente. Cir Gen [Internet]. 2017; 39(1): 37-40. Available from: https://www.medigraphic.com/cgi-bin/new/resumen.cgi?IDARTICULO=74562
13. Anderson S. Café au lait macules and associated genetic syndromes. J Pediatr Heal Care [Internet]. 2020; 34(1): 71-81. Available from: https://doi.org/10.1016/j.pedhc.2019.05.001
14. Hernández L, Espinosa MAL, Méndez V, Mercado M. Síndrome de McCune-Albright: características clínicas en una población pediátrica y adulta. Rev Endocrinol Nutr [Internet]. 2012; 20(1): 11-18. Available from: https://www.medigraphic.com/cgi-bin/new/summary.cgi?IDARTICULO=36436
15. Martínez Coronel M, Rojas Mercado H. Displasia fibrosa craneofacial avanzada por remodelación ósea. Salus [Internet]. 2014; 18(3): 46-50. Available from: http://ve.scielo.org/scielo.php?script=sci_arttext&pid=S1316-71382014000300009&lng=en
16. 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 [Internet]. 2019; 14: 139. Available from: http://dx.doi.org/10.1186/s13023-019-1102-9
17. Burke AB, Collins MT, Boyce AM. Fibrous dysplasia of bone: craniofacial and dental implications. Oral Dis [Internet]. 2017; 23(6): 697-708. Available from: https://doi.org/10.1111/odi.12563