

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

Cherubism: a rare case report with literature review ☆,☆☆

A. Lahfidi, MD*, W.M. Traore, MD, I.D. Diallo, MD, N. Lrhorfi, MD, S. Elhaddad, MD, N. Allali, MD, L. Chat, MD

Pediatric Radiology Department, Rabat Children's Hospital, Ibn Sina Hospital, Faculty of Medicine and Pharmacy, Mohammed V University, Rabat, Morocco

ARTICLE INFO

Article history:

Received 3 July 2022

Revised 12 July 2022

Accepted 14 July 2022

Keywords:

Genetic disorder

Dysmorphism

Dysplasia

Bone abnormality

ABSTRACT

Cherubism is an autosomal dominant disorder caused by a mutation of the gene encoding the binding protein SH3BP2. However, non-hereditary forms are observed, probably related to a de novo mutation. It is clinically manifested by an enlargement or a deformation of the jaw associated with a malposition of the teeth. In severe forms, these deformations can have a psychological impact, associated with ocular complications, as well as a reduction of the pharynx lumen and phonation disorders. The appearance of the lesions on imaging is not very specific, however it remains essential for the mapping of the lesions. We report a case of cherubism in a 12-year-old boy with no family history. This case report's objective is to identify the radiological aspect of this pathology and establish an early diagnosis to oriented therapeutic interventions.

© 2022 The Authors. Published by Elsevier Inc. on behalf of University of Washington.

This is an open access article under the CC BY-NC-ND license

(<http://creativecommons.org/licenses/by-nc-nd/4.0/>)

Introduction

First described by Dr. W. A. Jones of Kingston, Ontario in 1933, cherubism has long been mistakenly considered a familial form of fibrous dysplasia. It is a rare, inherited autosomal dominant disease, which affects exclusively the mandibular and maxillary bones and is most often seen before the age of 5 [1]. Although benign, this condition, in its severe form, can lead to complications and alter the quality of life of patients. We report the clinical and radiological manifestations of a case of cherubism in a boy with no family history.

Case

A 12-year-old, without family history, with good psychomotor development. Followed for anemia since the age of 18 months and thrombopathy, he was brought to medical consultation by his family for a facial dysmorphism. The history revealed that this dysmorphic syndrome started in early childhood, with a progressive and painless deformation of the jaw during growth, leading to a tendency to isolation and loss of socialization. Subsequently, mastication disorders related to dentition disorders developed, followed by phonation and swallowing

☆ Competing Interests: The authors declare that they have no links of interest.

☆☆ Article produced in compliance with legal, ethical and institutional rules.

* Corresponding author.

E-mail address: lahfidial@gmail.com (A. Lahfidi).

<https://doi.org/10.1016/j.radcr.2022.07.063>

1930-0433/© 2022 The Authors. Published by Elsevier Inc. on behalf of University of Washington. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>)



Fig. 1 - Front view of the oral cavity of a 12 year old boy with cherubism.

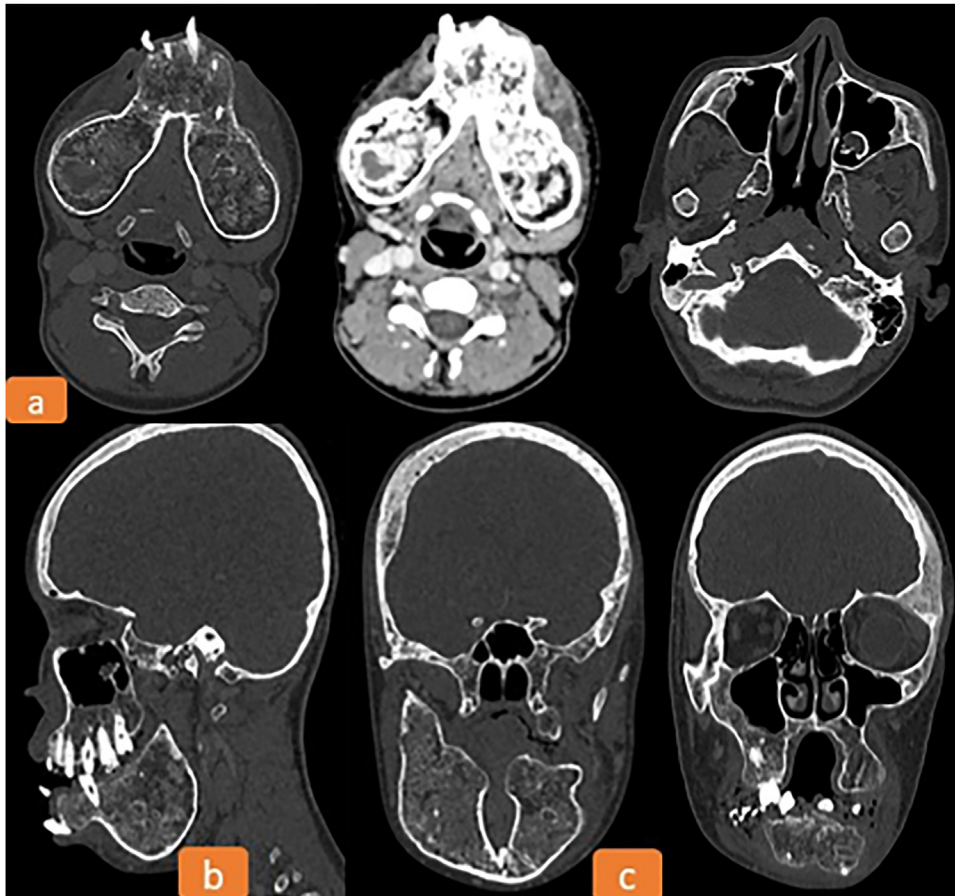


Fig. 2 - Bone window CT scan of the same patient in axial section (a), sagittal (b), and coronal (c) reconstruction, showing bilateral and grossly symmetrical enlargement of the mandibular and maxillary bone with areas of osteolysis and a ground glass matrix.

disorders. The clinical examination showed a facial dysmorphism with diffuse gingivo-mandibular hypertrophy filling the oral floor associated with dental malposition, a deformation of the lower dental arch concave upwards (Fig. 1). The gingiva was pink, firm and nodular with telangiectasias. The CT scan of the face showed bilateral and symmetrical hypertrophy of the maxillary and mandibular bones with osteolytic areas, and a ground-glass matrix of bone predominantly in the mandibular ramus and sparing the condyles, associated with an anarchic arrangement of the teeth (Fig. 2). In view of this clinical-radiological picture, the diagnosis of cherubism was evoked. The genetic testing and the bone biopsy confirmed the diagnosis. A dental extraction with orthodontic surgery and implants were indicated after correction of anemia and thrombopathy.

Discussion

Cherubism is an autosomal dominant disease secondary to a mutation in the gene encoding the binding protein SH3BP2 (SH3-domain binding protein 2) on chromosome 4. However, sporadic cases have been described with a de novo mutation [2,3]. It is a benign bone disorder that exclusively affects the jawbone and the maxilla, in which normal bone is replaced by fibrous tissue. It appears during the first decade and is characterized by a painless, progressive, approximately symmetrical tumefaction and deformation of the jaw, giving a cherubic appearance, associated with a disorganization of the teeth insertion. On X-ray or CT scan, there is an enlargement of the jawbone with sharp osteolysis and sometimes a discontinuous marginal sclerosis with multiple cystic lesions of bilateral distribution. Typically these lesions begin at the angle of the mandibular and extend to the body or ascending ramus. The maxilla can also be affected and there are associated anomalies of dental insertion. No periosteal reaction is found and extension to the mandibular condyle is rare but described [2]. The CT scan is useful for a good mapping of the lesions allowing the classification of Marck and Kudryk [4]. However, the differential diagnosis with fibrous dysplasia in its craniofacial form remains laborious. Indeed, cherubism has long been considered as a familial form of fibrous dysplasia, but molecular biology has allowed the separation of its two entities which present different genetic abnormalities. The evolution of cherubism is marked by a stabilization or

regression of the lesions after puberty. Reossification of lytic and cystic lesions is observed, giving the appearance of irregular sclerosis and a ground glass appearance of the bone structure. Therefore, surgical management is only considered after puberty by most authors. It is indicated in case of aggressive form generating ocular disorders, swallowing, elocution, or an important dysmorphic syndrome with a psychological impact. Treatment with calcitonin, an agent that inhibits bone resorption, seems to reduce the occurrence of cystic lesions of the mandible [3]. Cherubism can be associated with other genetic diseases, such as fragile X syndrome, neurofibromatosis type 1, Noonan syndrome (Turner pseudosyndrome), Jaffe-Campanacci syndrome and Ramon syndrome.

Conclusion

Cherubism is a hereditary disease that occurs in children during the first decade. However, non-familial forms can be observed. Although the radiological signs lack specificity, the diagnostic approach is possible in the presence of bilateral and roughly symmetrical lesions of the mandible and/or maxilla. The diagnosis of certitude is based on the evidence of the mutation by molecular biology.

Patient consent

The patient declares his consent for the publication of his case.

REFERENCES

- [1] Agrawal A, Gupta SK, Saxena P, Kumar P. Non-familial cherubism: clinical and radiological findings. *BMJ Case Rep* 2014;2014:bcr2013202031.
- [2] Msomi MS, Dlamini NN. Cherubism: a case report. 2017. 2017;21(1).
- [3] Wagel J, Luczak K, Hendrich B, Guziński M, Szaśiadek M. Clinical and radiological features of nonfamilial cherubism: a case report. *Polish J Radiol* 2012;77(3):53–7.
- [4] Marck PA, Kudryk WH. Cherubism. *J Otolaryngo* 1992;21(2):84–7.