Cornelia de Lange syndrome: A case report

Joaquim Grau Carbó [1], Julian López Jiménez [2], Mª José Giménez Prats [3], Meritxell Sànchez Molins [4]

(1) Degree in Odontology
(2) Ph.D. in Medicine and Surgery. Specializing in Estomatology
(3) Ph.D in Odontology

Correspondence:
Dr. Joaquim Grau Carbó
C/ Consell de Cent, 284 Entl.
08007 Barcelona, Spain
E-mail: joaquimgrau@eresmas.com

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ABSTRACT

Cornelia de Lange is a genetic syndrome which affects between 1/10.000 and 1/60.000 neonates, but its genetic bases are still not clear. Its principal clinical characteristics are the delay in growth and development, hirsute, structural anomalies in the limbs and distinctive facial characteristic. Dental problems are frequent and include: ogival palate, micrognathia, dental malalignment, delayed teething, microdontic teeth, periodontal disease and dental erosion produced by gastric reflux. Discussed is the case of a 29 year old patient affected by the syndrome in question, which presents the principal clinical characteristics. The patient's general state of health is acceptable, without cardiac or respiratory alterations. The intraoral exploration shows policaries, periodontal disease, persistence of the temporal teeth and ectopic molars. After completing the necessary pre-operatory preparations, the entire odontological treatment was carried out under general aesthesia, due to the patient's total lack of collaboration.

Key words: *Cornelia de Lange, special patients, odontological treatment.*

RESUMEN

El síndrome de Cornelia de Lange es un síndrome genético que afecta entre 1/10.000 y 1/60.000 neonatos, y cuyas bases genéticas todavía no están claras. Sus características clínicas principales son el retraso en el crecimiento y en el desarrollo, hirsutismo, anomalías estructurales en miembros y unas características faciales distintivas. Los problemas dentales son frecuentes e incluyen: Paladar ojival, micrognátia, malposición dental, retraso en la erupción, microdoncia, enfermedad periodontal y erosión dental producida por el reflujo gástrico. Se describe el caso de una paciente de 29 años de edad afecta por el síndrome en cuestión, que presenta las principales características clínicas. El estado general de la paciente es aceptable, sin alteraciones cardíacas ni respiratorias. La exploración intraoral muestra policaries, enfermedad peridontal, pesistencia de dientes temporales y cordales ectópicos. Después de realizar las pruebas preoperatorios necesarias, se realiza el tratamiento odontológico completo bajo anestesia general, debido a la nula colaboración de la paciente.

Palabras clave: *Cornelia de Lange, pacientes especiales, tratamiento odontológico.*

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eMail: medicina@medicinaoral.com

INTRODUTION

Cornelia de Lange Syndrome (CdLS) is relatively rare and affects, according to sources, between 1/10.000 and 1/60.000 neonates. There is no racial predilection. However, it is more frequent in women than men: 1,3 / 1 (1-3). Even though Brachman announced summarily some aspects of this syndrome (1) in 1916, it was in 1933 when Cornelia de Lange described it as degeneración amstelodamensis (4). Other less frequently used synonyms are Brachman-De Lange Syndrom and Typus degenerativus Amstelodamensis. Classically it is defined by hypo growth, mental retardation, micromelia and distinctive facial features (3).

The genetic and molecular bases of these lesions are not clear. However, it is thought to be the result of a dominant mutation. A large part of the cases diagnosed as CdLS seem to be sporadic and 10% of the cases present chromosomal alterations, translocation of the 3q 26:2-q23 (3, 5). In the existing literature, there have been two phenotypes differentiated, a classic and a milder (1, 2).

The principal clinical characteristics of this syndrome are the delay in growth and development, hirsute, anomalies in the structure of the limbs and distinctive facial characteristics (3,6,7).

Hypo-growth is at first intrauterine and very intense, with delays in osseous maturation and grave, hypertonic mental deficiencies (100% of the cases). At birth and during the length of their life, these patients present a weight and size inferior to that corresponding to their age. The intellectual coefficient is not over 50%.

The facial features are disitincitve, with microcephaly, the eyebrows very close together (sinofria), generalized hirsute (the frontal implantation of the hair is low), the ears are implanted low, small nose, nostrils antiverities (anti spillage), full philtrum, thin lips, micrognathia (the commissural inclined downwards) and perioral cyanosis (6,7).

The extremities are also usually altered by the presence of simian palm groove, limited mobility of the elbow, micromelia, syndactyly.

Ocularly they may present palpebral ptosis, conjunctivitis or chronic blefaritis, stenosys of the palpebral canal, severe myopia, nistagmus, micro- cornea 30% suffer from cardiopathy and the hypoacousia is either at a low or moderate grade.

Finally, the dental problems include: Ogival palate (20% present associated palatal fissures), little development of the mandible, dental malalignment, delayed tooth eruption, microdontic teeth, periodontal disease and dental erosion produced by frequent gastric reflux (which can also produce esophagitis, esophageal stenosis and pulmonary problems). (6,7)

CASE REPORT

The patient is a 29 year old woman diagnosed with CdLS. She presents the morphological characteristics of the syndrome with microcephaly, hirsute, sinofria, low frontal line of hair implantation, small nose, cyanosis perioral, small ears and low implantation, along with profound mental handicap. She is 1.20 meters tall and weighs 35 Kg. (Fig. 1)

Her general health is acceptable without cardiac or respiratory alterations. She only presents esophageal stenosys with frequent spasms due to the gastro- duodenal reflux (also characteristic in this type of patient) (8), which requires esophageal dilatations using fibrogastroscopy under general anesthesia approximately once a year.

The intra-oral exploration showed the presence of polycaries (in 16 teeth), moderate periodontal illness, persistence of the temporal teeth (5.5, 6.5, 7.5, 8.5) and the presence of ectopically erupted molars. It was not possible to take an orthopantomograph due to the patient's lack of cooperation. (Fig. 2)

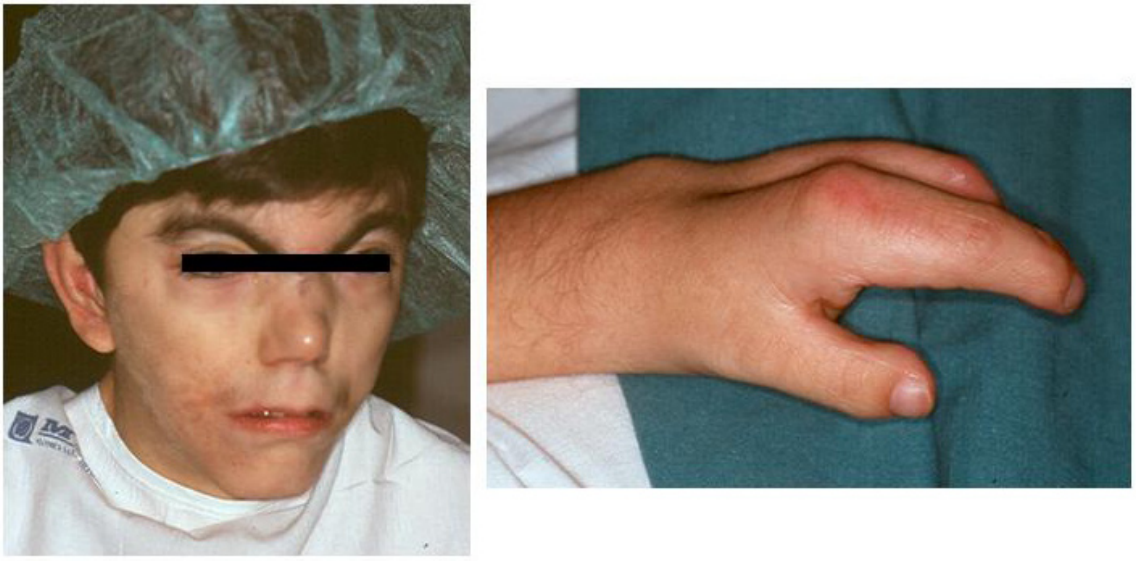


Fig. 1. Particular facial features (A), micromelia and sindactilia (B) in Cornelia de Lange Syndrome.

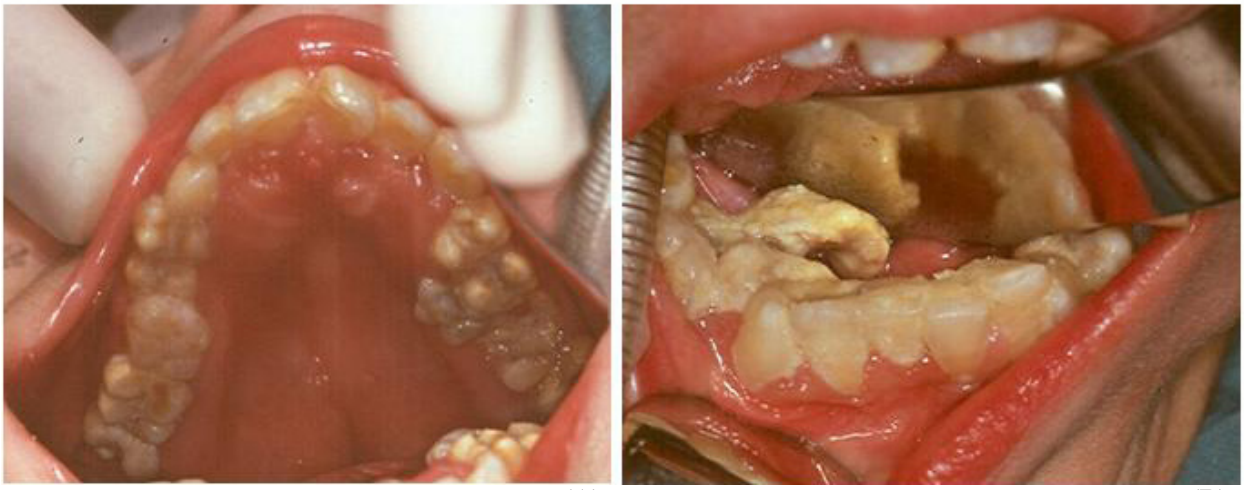
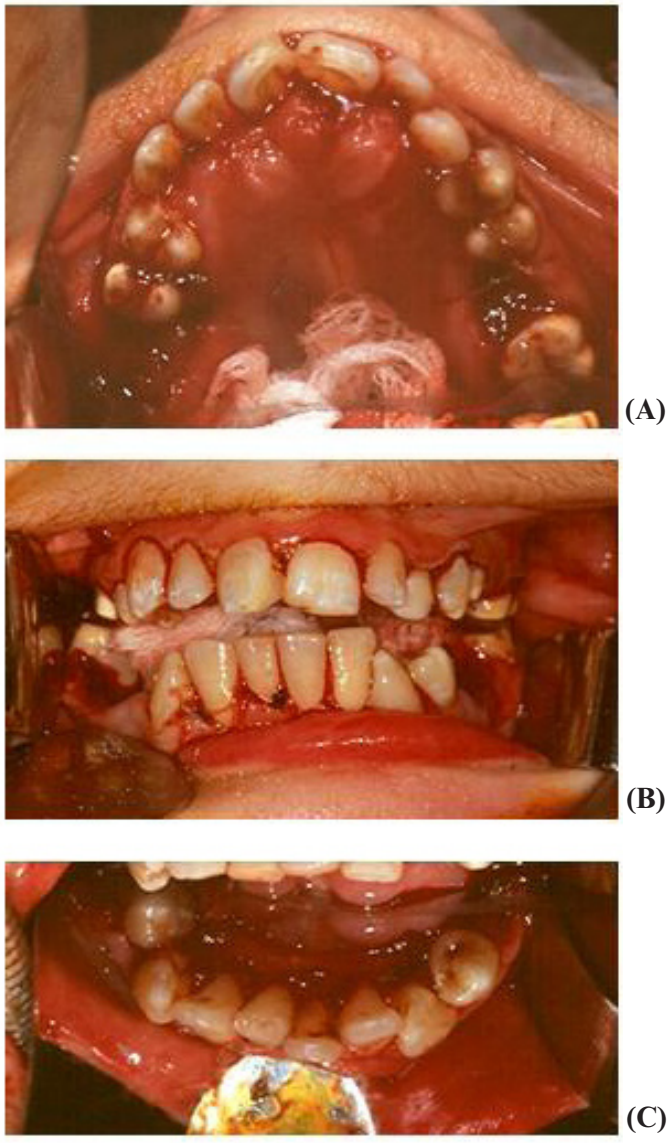


Fig. 2. Oral appearance before treatment. **(A)** **(B)**



(A) **(B)** **(C)**

Fig. 3. Oral appearance after treatment.

The additional complementary tests that were solicited (general analytical, radiology of the thorax and electrocardiogram) did not show any alterations which might contraindicate the general anesthesia.

Under general anesthesia the following odontological treatment was completed: Extraction of temporal teeth and ectopic molars and the appropriate periodontal treatment; leaving the mouth, after treatment, healthy. (Fig. 3)

While the patient was still under general aesthesia and in coordination with her gastroenterology, a session of esophageal dilation using fibrogastroscopy was carried out.

Seven days after the treatment the patient was scheduled for a visit to control the state of the mouth and instruct the family members on daily dental hygiene. Likewise, they were advised that revisions every four months would enable early diagnosis of any new alterations (cavities, accumulation of plaque).

DISCUSSION

The Cornelia de Lange syndrome is a rare polimalformation genetic disease. The craniofacial structures are greatly affected (1-3). The case we have described presents the principal clinical characteristics of this syndrome. The alteration in the development and growth of the maxillas implies the presence of dental malalignments. The mental deficiency in conjunction with motor deficit, dental malalignment, the type of diet and other factors, makes the presence of periodontal cavities are frequent in this type of patient. For this reason, the development of the maxillaries and the teeth, and the presence of pathology, should be watched from early on, and implement the appropriate preventative or therapeutic methods.

The patient in our case had their first consultation with an odontologist at 29 years of age, due to the discomfort caused by the condition of her teeth. When treating a patient with a polimalformative syndrome every precaution must be maximized to ensure that any additional systematic

pathology is discarded or detected. It is necessary to request consultations from various specialists in order to evaluate the cardiopulmonary, hepatic, renal or hematological state which may condition the orthodontic treatment (9).

Patient collaboration is indispensable considering the oral manipulations, to determine the complementary techniques for handling the most ideal conduct in each case (2). Our patient's collaboration was null, for this reason we performed the orthodontic treatment under general anesthesia.

It must be remembered that when a sedative or general anesthesia is used, one must consider the possibility of performing simultaneously other procedures (endoscopys, ocular exams…). Since our patient regularly receives sessions of endoscope esophageal dilations, we contacted her gastroenterologist to take advantage of the patient being under general anesthesia.

At the end of the odontological treatment it is essential remit the patient to preventative odontological services to give them some basic rules of oral hygiene adapted to each individual case and implicating the family and care takers in the caring for the dental health.

Preventative revisions starting in infancy and in coordination with the pediatrician are necessary. Routine revisions every six months facilitates the the changes in orofacial growth, detection of pathologies and strengthen the care of teeth at home.

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