

Dental and Maxillofacial Alterations in Patients Affected From Odontochondrodysplasia: A Rare Case Report and Review of Literature

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Abstract

Introduction: This paper has evaluated the dental and or facial disorders associated to Goldblatt syndrome, also known as odontochondrodysplasia.

Aim: We report the analysis performed on a female young patient affected by this disease. We analyzed her dental and or facial features.

Materials and Methods: We adopted several diagnostic criteria: firstly, we performed radiographic investigations, followed by rhinomanometric measurements and by clinical analysis performed in order to determine the salivary flow in this typology of patients. The evidences obtained after a careful clinical, anamnestic, and radiographic analysis of our female patient allowed us to identify a number of odontostomatologic features, which are very likely to be related to this syndrome. Our patient showed some pathognomonic signs of odontochondrodysplasia already identified in the literature, that is, pectus carinatum, joint hyperextensibility, coxa valga and genu valgum, upper and lower limb asymmetry, and vertebral abnormalities. Moreover, we focused our attention on those odontostomatologic aspects that had never been analyzed by other reports in the literature: dentinogenesis imperfecta, ligamentous hyperlaxity of all joints and of temporomandibular joints in particular, poor lip competence, ogival palate, and oral respiration. Besides these, dental crowding and other important elements were identified through cephalometric measurements.

Discussion and Conclusions: In the light of all these elements and of their comparison with the existing literature, it is possible to stereotype a few recurrent odontostomatologic and systemic-generalized features in patients with odontochondrodysplasia, which can be considered as closely associated with this syndrome.

Key words: Odontochondrodysplasia, Goldblatt syndrome, Dentinogenesis imperfecta

Introduction

Odontochondrodysplasia (OCDC) is a chondrodysplastic disease. More specifically, it is a genetically transmitted, generalized Spondylometaphyseal Dysplasia (SMD). Among the clinical evidences associated to this syndrome, we can find all the clinical signs of SMD [1]: *pectus carinatum*, joint hyperextensibility, *coxa valga and genu valgum*, upper and lower limb asymmetry, and vertebral abnormalities. The association between *dentinogenesis imperfecta* and SMD was reported by Goldblatt [2], in fact, this particular form of dysplasia is also called Goldblatt syndrome. Our work was to investigate about the possible association between OCDC and orofacial features still not reported by the scientific literature.

Materials and Methods

We describe a 19-year-old caucasian female. No family history of skeletal dysplasia, joint hypermobility, or significant dental anomalies.

The patient's anamnesis revealed that from birth, she has been under medical supervision for lower-limb dysmetria, associated with short stature, mandibular hypoplasia, and bilateral epicanthus. Hematochemical examinations and hormonal values showed parathormone value which was lower than reference values.

Clinical and radiographic examinations revealed a narrow, prominent chest, signs of rachitis with *facies sui generis*, shortness of long bones, metaphyseal irregularity in the proximal femur (more serious in the proximal radius, ulna, and tibia), irregularity of the left fibula with proximal and distal metaphyseal fragmentation, irregularity of horizontal acetabulum with jagged margins, shortness of the neck of the femur, thoracic scoliosis convex to the right with square vertebral bodies, and cupping of the distal metaphyses of radius and ulna.

For all these elements, she was diagnosed with SMD [3]. We can exclude the Kozlowski type, because the genetic testing we performed on the alteration of genes *COL10A1* and *SHOX* was negative [3-7]. These genes are respectively responsible for the synthesis of proteins involved in collagen formation and for the control of the skeletal structures during the embryonic phase. The presence of *dentinogenesis imperfecta* and other facial anomalies confirmed the diagnosis of Goldblatt Syndrome [4].

We performed several clinical investigations, radiographic examinations and Nuclear Magnetic Resonance (NMR) of the temporomandibular joint (*Figure 1*), X-ray images of the chest, with details of the bones of the wrist (*Figure 2*), telecranium

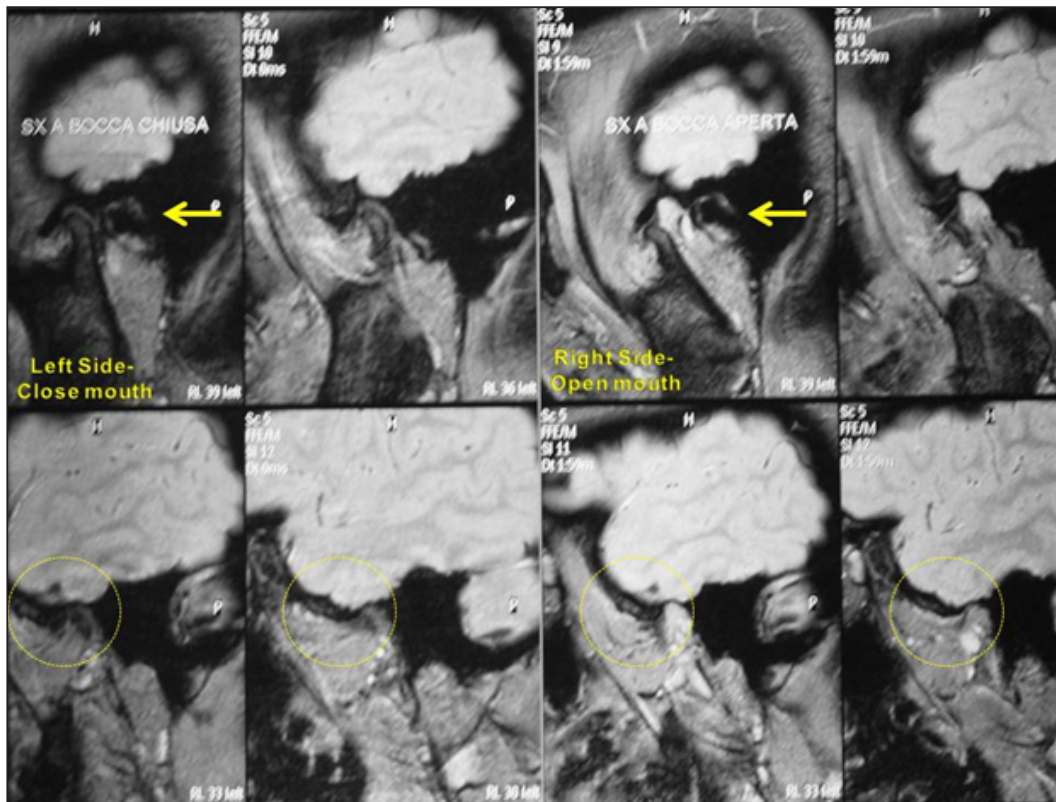


Figure 1. Open-mouth and closed-mouth NMR performed during the dynamic study of the TMJ, showing signs of ligamentous relaxation of temporomandibular, sphenomandibular and stylomandibular ligaments.

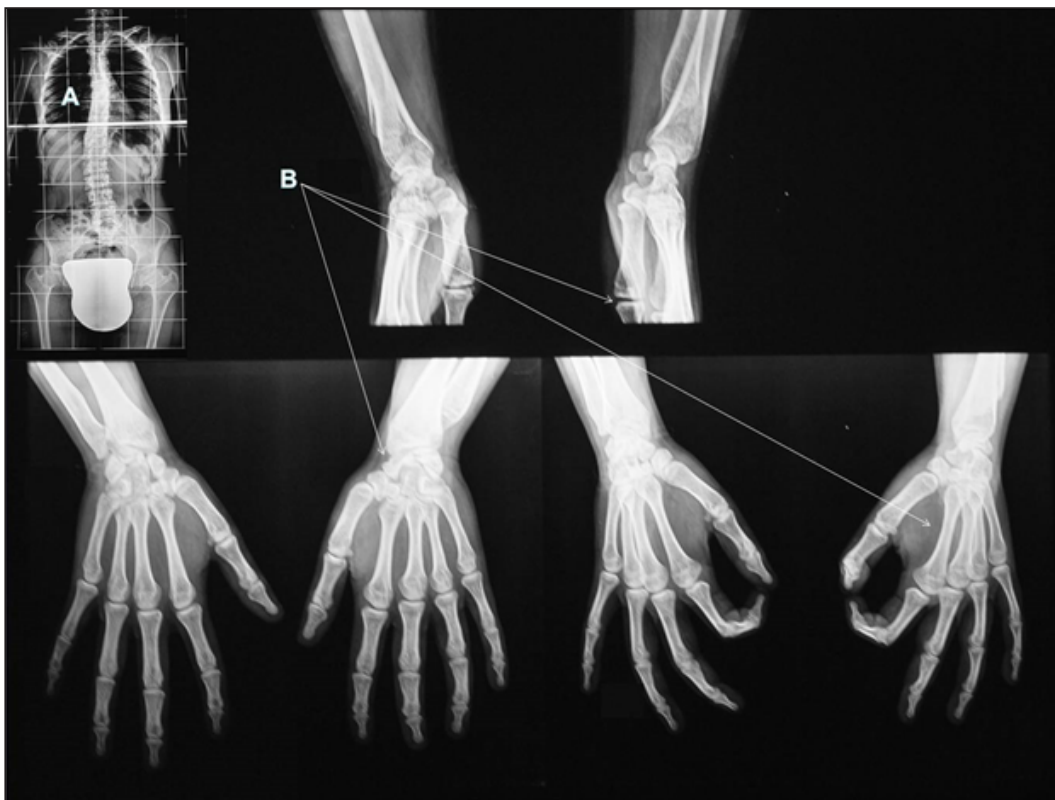


Figure 2. X-ray images of the chest, with details of the bones of the wrist.

A: right convex thoracic scoliosis
B: signs of rachitis on the articulations and on the long bones.

x-rays in sagittal view (Figure 3) and posteroanterior projection and orthopantomography (OPT) of dental arches (Figure 4).

In order to describe and detect the presence of respiratory diseases, we performed anterior and posterior rhinomanometry, and other semeiological to assess other pathological conditions. Qualitative and quantitative testing of the salivary flow was done to determine the volume and the electrochemical composition of saliva.

Results

Our observations confirmed the Goldblatt syndrome typical features; moreover, a part of the orofacial investigations, we observed a severe skin hyper-extensibility, mainly in the carpometacarpal region: our literature review did not provide any data about this alteration, which could further extend the set of signs of Goldblatt syndrome. We also performed a NMR of the Temporo-Mandubular Joint (TMJ): the dynamic

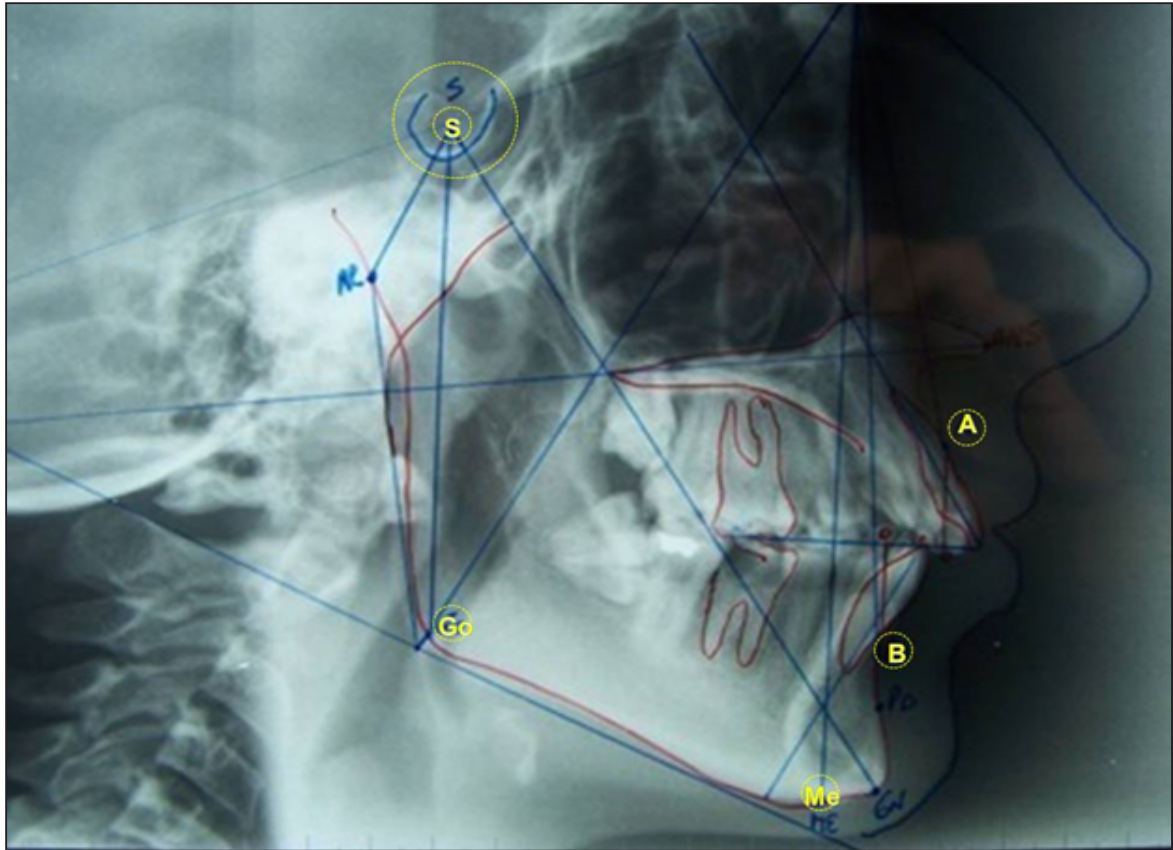


Figure 3. Cephalometric tracing performed on a latero-lateral projection teleroadiography of the skull.

Legend: According to the Steiner's analysis, the most important points in a cephalometry are:

S Sella: Mid-point of sella turcica which is a saddle-shape depression in the sphenoid bone of the human skull.

N Nasion: calculated as the intersection of the frontal bone and two nasal bones of the cranium.

Go Gonion: Most posterior inferior point on angle of mandible

Me Menton: Lower most point on the mandibular symphysis

A point: Position of deepest concavity on anterior profile of maxilla

B point: Position of deepest concavity on anterior profile of mandibular symphysis

According to the Steiner's analysis, the most important angles in a cephalometry are:

ANB: it indicates the skeletal relationship between the maxilla and mandible

SNA: it indicates whether or not the maxilla is normal, prognathic, or retrognathic.

SNB: it indicates whether or not the mandible is normal, prognathic, or retrognathic.



Figure 4. Orthopantomography (OPT) of the reported case.

analysis of the TMJ revealed bilateral signs of laxity of temporomandibular, sphenomandibular, and stylomandibular ligaments. These features could affect other joints in the rest of the body of these patients as well (*Figure 2*).

The results of odontostomatologic alterations in Goldblatt syndrome derive from the analysis of cephalometric and clinical data of our patient (*Figure 3*).

Sagittal and posteroanterior telecranium x-rays allowed us to obtain cephalometric data according to Jarabak's cephalometric analysis [8], which outlined a clinical condition of Goldblatt syndrome. The orthodontic diagnosis clearly defines a II class/II division (Angle's classification) in a hyperdivergent patient [9].

In our patient with OCDC, hyperdivergence was measured through the cephalometric values. The sum of angles at the cranial base, which is the sum of angles NSAr/SArGo/ArGoMe, was 409°; the percentage height of the facial skeleton, which is posterior facial height multiplied by 100, divided by anterior facial height, was 61.1%; the lower gonial angle was 87°; the position of the mandible compared to the cranial base (NSGn, y-axis), was 79°; the intermaxillary angle, existing between maxilla (MX) and mandible (MB) and expressed as MX/MB rate, was 30°.

All of this worsened even further because of the severe mandibular retrognathia, showed by the SNB angle, which was 72°, and characterized the anteroposterior position of the mandible compared to the cranium.

Another fundamental element of the cephalometric analysis was the patient's skeletal class II, showed by the difference $SNA-SNB = ANB$, which was 8°, as confirmed by Witz index (A0/B0) indicating the intermaxillary rate, which was 9 mm (values higher than 4 mm indicate a basal class II). Besides, the angle A-Na-Pog, indicating the protrusion of the upper jaw with relation to total profile, was 8°. The angle Ans-Na-Pog, showing the position of the body of maxilla with relation to the body of mandible, was 18°.

These results indicate an evident skeletal class II.

Linear measurements of the skeletal structures were performed, which showed that the length of the anterior cranial base was 16 mm shorter than normal values, the length of the body of mandible was 10 mm lower than n.v., and the length of the lateral cranial base was 5 mm shorter than normal values [10].

These data are absolutely innovatory, considering the total lack of cephalometric references in the literature [4].

The mandibular retrognathia and the transverse palatal hypoplasia caused an atypical deglutition and oral respiration. After these clinical outcomes, we needed to define if oral respiration was a chronic condition, or a transitory condition. In order to confirm and define the condition of oral respiration, we performed quantitative and qualitative analyses: two of these tests were well-known clinical maneuvers, which define the permeability of the upper airways [11]. The first test was a type of indirect rhinomanometry called "Glatzel mirror" test, the other test was the "nostril reflex of Gudin". The last test performed was a rhinomanometry [12]: the value of binasal resistance that we obtained was $5 \text{ cm H}_2\text{O} \cdot \text{l}^{-1} \cdot \text{s}^{-1}$ (n.v. lower than $3 \text{ cm H}_2\text{O} \cdot \text{l}^{-1} \cdot \text{s}^{-1}$); the uninasal value was $8 \text{ cm H}_2\text{O} \cdot \text{l}^{-1} \cdot \text{s}^{-1}$ (n.v. lower than $6 \text{ cm H}_2\text{O} \cdot \text{l}^{-1} \cdot \text{s}^{-1}$). About pressure, total

unilateral resistance was $1.80 \text{ Pa} \cdot \text{ml}^{-1} \cdot \text{s}^{-1}$ (n.v. lower than $0.50 \text{ Pa} \cdot \text{ml}^{-1} \cdot \text{s}^{-1}$). These results confirmed that our patient was suffered from chronic oral respiration.

Intraoral examination revealed upper dental crowding, more severe from teeth 1.3 to 2.3, lower crowding, more severe from teeth 3.3 to 4.3. Teeth 1.8, 2.8, and 4.8 were not erupted. The authors were able to diagnose a form of *Dentinogenesis Imperfecta* of mild type; in some patients with DI may be lacking the clinical and radiographic features of DI in permanent teeth, moreover, the absence of mutations on genes COL10A1 and SHOX makes us sure that the reported case was not the severe form, called Kozlowski type.

Intraoral examination also showed signs of a mild hyposalivation. For a qualitative and quantitative evaluation of the salivary flow, we used different test already described in the literature [13]. We found also a condition of salivary acidosis: it can surely contribute to quickly degrade the crown of the teeth.

Discussion

After literature review, we noticed that there are only few papers focused on facial anomalies in patients with OCDC. Particularly, these articles focus on general elements (prominent orbits, epicanthus, down-slanting palpebral fissures, short nose with flat nasal septum, midfacial hypoplasia, and prognathism), which do not include intraoral and orthodontic aspects, except for the *dentinogenesis imperfecta* [5].

Other publications on this syndrome focus on the genetic aspect [6,7]. Therefore, we have directed our evaluations on the dental and maxillofacial alterations associated with OCDC.

Our report allowed us to say that there is a correlation between this syndrome and some dental and craniofacial alterations; more specifically, patients with OCDC have a reduced mandibular and maxillary development, along with an altered growth of the bone segments. These conditions lead to mandibular retrognathia and transverse palatal hypoplasia indicating a skeletal class II, which is etiologically caused from this syndrome. The altered growth and the altered formation of ossification centers in patients with Goldblatt syndrome certainly affect and alter the development of the facial skeleton, causing asymmetries and morphostructural alterations.

These conditions are in addition to the clinical condition of oral respiration and of atypical deglutition. Besides, explaining and documenting the existing connection between OCDC and cephalometric measurements is a crucial element for the treatment of these patients, also from the dental point of view, which has not been analyzed enough so far.

About the dental and intraoral conditions, malposition of teeth, frequent dysodontiasis, and destructive caries processes can be related to *dentinogenesis imperfecta* [15]. Because of the altered ossification and formation of tissues constituted by collagen fibers, enamel, and dentine are also affected by these structural anomalies causing greater caries receptivity [16] in patients with OCDC. We can say that *dentinogenesis imperfecta* of these patients plays an important role in the

development of caries processes and dysodontiasis typical of this pathology.

Moreover, malposition of teeth and altered salivary flow or composition [17] further worsens caries receptivity of these subjects.

Conclusions

Our study review the literature on the topic of OCDC and orofacial anomalies: we have assessed that these patients could have a peculiar bone conformation of the facial skeleton, along with dental and oral conditions of *dentinogenesis imperfecta*, hyposalivation, and salivary acidosis. These informations can help the physicians to intercept other related pathologies, thanks to a proper therapeutic plan and a multidisciplinary approach to the follow-up.

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Consent Statement

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing Interests

The authors declare that they have no competing interests.

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