

GOLDENHAR SYNDROME

• maxillofacial rehabilitation •

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ABSTRACT

Goldenhar syndrome is one of the most common congenital anomalies of the first and second branchial arches. Its main alterations affect the eye (dermoids and/or epibulbar lipoderoids), the external ear (auricular appendages, blind-ended fistulas) and the spine (hemivertebrae and vertebral fusion, among other malformations). This work presents the maxillofacial surgical correction of a 28-year-old female with Goldenhar syndrome. Literature and etiopathogenesis are also briefly reviewed.

Keywords: Congenital abnormalities. Goldenhar syndrome. Oculoauriculovertebral syndrome. Facial bones. Surgery.

INTRODUCTION

Goldenhar syndrome is one of the most common congenital syndromes of the first and second arches. It has also been described as “First Arch Syndrome” and “Gorlin Syndrome.” Currently it is better known as oculoauriculovertebral dysplasia, name given by Gorlin et al. (1963) and Sugar (1966). Oculoauriculovertebral dysplasia is a congenital

symptomatic complex of unknown etiology, in which the primary alterations are located on the eye (dermoid and/or epibulbar lipodermoid), the external ear (auricular appendices, cervical sinus) and the spine (hemivertebrae, vertebral fusions, and other malformations).⁽¹⁾ Other anomalies have been documented in association with

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Goldenhar complex, including cardiac, pulmonary and renal defects. These occurrences have raised questions on whether the origin of the syndrome lies, in fact, on the first and second branchial arches. The literature proposes that it may result from the inadequate migration of primitive line cells, since the dysplastic organs of associated anomalies have a mesodermal origin.⁽²⁾ The use of some drugs such as cocaine, thalidomide and retinoic acid during pregnancy, as well as maternal diabetes; have been reported as predisposing factors for the development of oculoauriculovertebral dysplasia.⁽³⁾ This paper aims to report the maxillofacial surgical correction of a 28-year-old female with Goldenhar syndrome. Literature and etiopathogenesis are also briefly reviewed. This study protocols were approved by the Ethical Review Board of the Faculty of Dentistry of Federal University of Bahia, protocol number 13464613.6.0000.5024, and are in accordance to the Brazilian National Health Council Resolution 466/12.

CASE REPORT

Twenty-eight year old patient, dark-skinned, was evaluated at the Professor Edgar Santos University Hospital at Federal University of Bahia and, through clinical and radiographic exams, was diagnosed with Goldenhar Syndrome. The patient had facial asymmetry, left mandibular hypoplasia, tongue hyperplasia, left auricular appendix, scoliosis and ectopy of the left kidney; had previously undergone surgery to remove epibulbar dermoid on left eye. No mental disorders were detected during the evaluation period, nor were found signs of hearing deficiency or palatal cleft. It was not possible to carry out a satisfactory evaluation of dental

development, as the patient had lost most of her dentition throughout her lifetime.

For surgery planning purposes, several presurgical exams were carried out, among them facial and panoramic radiographs, with McNamara e Ricketts' cephalometric analysis. There was also a careful inspection of the internal and external pharyngeal structures, which guided the choice of intubation: nasal intubation was chosen. The patient initially underwent partial glossectomy, due to hyperplasia of the tongue. A temporary prosthetic device was previously prepared to establish the occlusal guide, being fixed on the maxillae with 2 mm x 6 mm titanium screws. A Risdon bilateral incision was done, and the Caldwell and Lettermann technique⁽⁴⁾ was used to correct mandibular deformation, followed by bilateral osteotomy of the mandible with rotation and fixation with steel wires. Mentoplasty was also needed to improve facial harmony.

DISCUSSION

The patient showed clinical characteristics of Goldenhar Syndrome, as described previously, including facial asymmetry, mandibular hypoplasia and a previously removed left epibulbar dermoid tumor⁽⁵⁾ (Figure 1). The patient also presented ectopy of left kidney, compatible with the description of associated renal abnormalities found in the literature.⁽⁶⁾

Despite the reported prevalence of 5-58% of congenital cardiac disorders,^(7,8,9,10) including Tetralogy of Fallot, defect of ventricular septum and a wide variety of cardiovascular malformations, these were not found on the patient.



Figure 1. Photograph of Goldenhar Syndrome patient. Note facial asymmetry, mandibular hypoplasia and auricular appendix.

The anatomical variations of skeletal structures and of cervical and craniofacial soft tissues deriving from dysmorphic syndromes may result in serious intubation problems. In case of detailed evaluations, it may not be able to safely foretell the potential difficulties of this procedure.⁽¹¹⁾ Some authors recommend the use of a laryngeal mask airway, laryngoscopy with a rigid optic automatic device and a fiberoptic bronchoscope.⁽¹²⁾ Nasal intubation was chosen, as describer previously, with no interferences in the surgery.

There are reports of authors who chose rib graft remodeling with the objective of recreating normal mandibular structures in single-stage surgical procedures to correct a defective hemimandible,

glenoid and zygomatic fossae.⁽¹³⁾ Currently, widening the human mandible by osteogenic distraction has been accepted as one of the treatments to correct severe hypoplasia. Osteogenic distraction is a method of bone formation by osteotomy, followed by the distension of the regenerating callus. This process is carried out with the help of a distension device that is fixed with screws placed directly on the bone to pre-determine the length.⁽¹⁴⁾ On the Brazilian public health service, however, this technique is very costly. Thus, it was decided to fix the mandible with aciflex, in a variation of the Caldwell and Letterman⁽⁴⁾ technique, used to correct mandibular deformities (Figure 2).

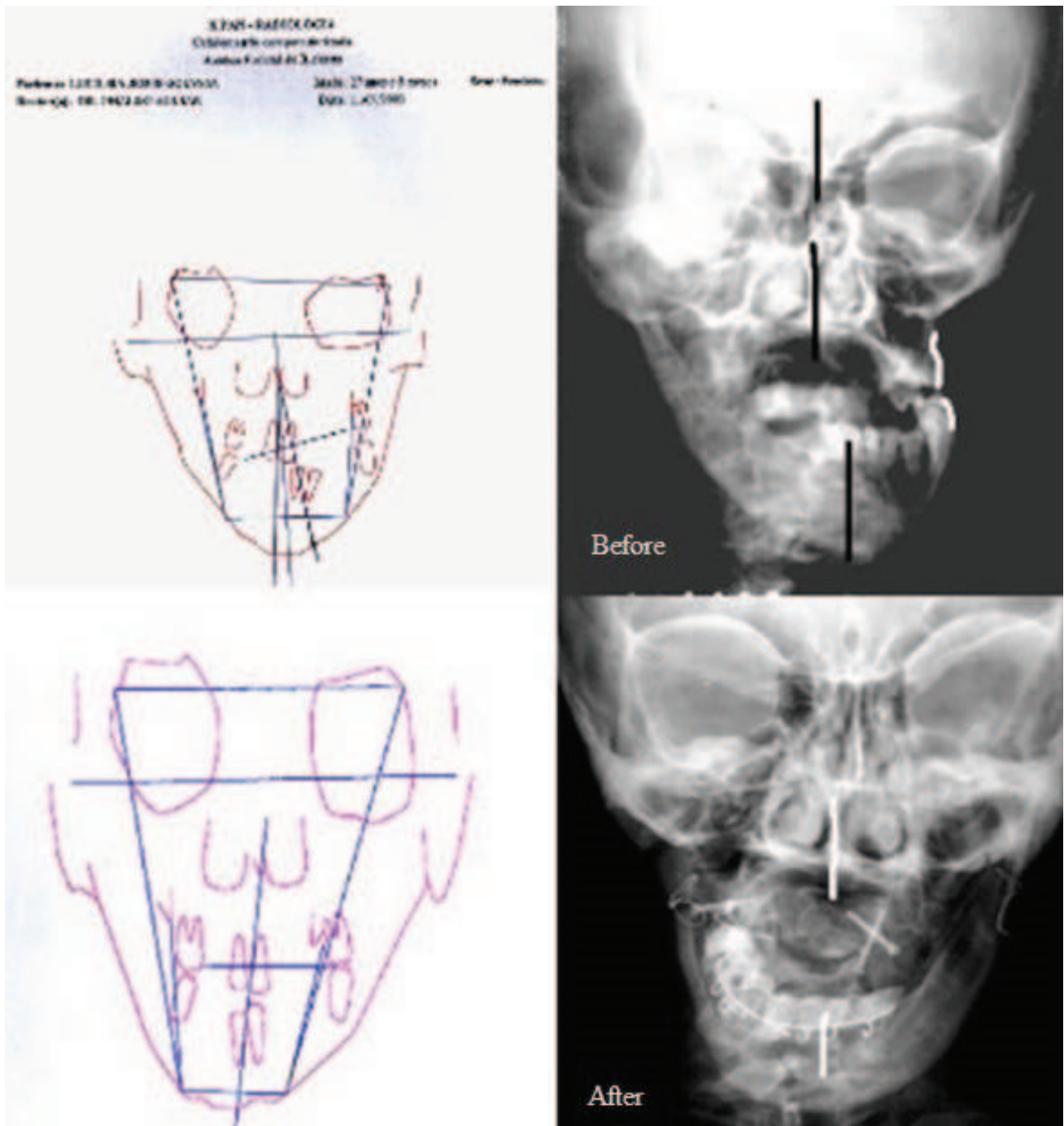


Figure 2. Panoramic and facial radiographs with McNamara and Ricketts cephalometric analysis, used for surgical planning.

It is believed that the current facial aspect of the patient contributes to his/hers biopsychosocial well being⁽¹⁵⁾ (Figure 3). Several authors^(16,17,18,19) defend Jaw Functional Orthopedics as a treatment method with good results, although many times limited,

requiring complementary surgery. However, the effects of functional orthopedic treatment, mainly if an early one, would be an important contribution. However, in other more serious cases, such as the one presented here, surgery is paramount.



Figure 3. Photograph of patient after surgery. Note the current facial aspect of the patient, more symmetrical and harmonious.

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