

CASE SERIES AND REPORTS

Role of 3D-CT for orthodontic and ENT evaluation in Goldenhar syndrome

Ruolo della TC-3D nella valutazione ortodontica e ORL della sindrome di Goldenhar

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SUMMARY

Goldenhar syndrome is a congenital condition that includes anomalies of the derivatives of the first and second brachial arches, vertebral defects and ocular abnormalities. It is also known as oculo-auriculo-vertebrale syndrome (OAVS), hemifacial microsomia, or first or second brachial arch syndrome. It was first described by Van Duysel in 1882 and better studied by M. Goldenhar in 1952. Its treatment requires a multidisciplinary approach. Herein, we describe the value of 3D-CT evaluation in a patient with Goldenhar syndrome, with particular regard to planning diagnostic and therapeutic approach. A 7-year-old boy with Goldenhar syndrome with definite post-natal genetic diagnosis was referred to our Department of Radiology for neuroimaging of the temporal bone. By 3D-CT evaluation of this young patient we observed the asymmetry of the condyles with the right one dysmorphic, short and wide; the auricle of the right ear was replaced by a dysmorphic rough; the right middle ear had a hypoplastic tympanic cavity and the internal auditory canal of right ear was atresic. In our experience, 3D-CT is a powerful diagnostic instrument and offers many advantages: volumetric reproduction of cranium and soft tissues, no overlap of anatomic parts that limits the visibility of various structures, high precision and assurance of images, and a constant and easily reproducible reference system. In our case, 3D-CT offered a very complete evaluation of all malformations of mandibular and temporal bone that characterize this syndrome and representing an important step for ENT and orthodontic therapeutic approaches.

KEY WORDS: Goldenhar Syndrome • 3D-CT • Facial asymmetry

RIASSUNTO

La sindrome di Goldenhar è una condizione congenita associata a difetti di sviluppo del primo e secondo arco branchiale, anomalie oculari e vertebrali. Possono essere associate anche anomalie cardiache, renali e del sistema nervoso centrale. Anche conosciuta come Sindrome oculo-auriculo-vertebrale, microsomia emifacciale o sindrome del primo e secondo arco branchiale. Fu descritta per la prima volta da Van Duysel nel 1882 e successivamente studiato da M. Goldenhar nel 1952 in modo più dettagliato. Il suo trattamento richiede un approccio multidisciplinare. Nel presente articolo si descrive il valore della TC-3D in un giovane paziente con sindrome di Goldenhar, con particolare riferimento alla diagnosi e alla pianificazione dell'approccio terapeutico. Un paziente di 7 anni con diagnosi postnatale geneticamente confermata di sindrome di Goldenhar è stato sottoposto a TC-3D presso il dipartimento di radiologia del nostro Policlinico. La valutazione radiologica ha consentito di mettere in evidenza l'asimmetria dei condili mandibolari (il destro appariva dismorfico, corto e largo), il padiglione dell'orecchio omolaterale sostituito da un rudimento dismorfico, l'orecchio medio di destra presentava una cavità timpanica ipoplasica con condotto uditivo interno atresico. Secondo la nostra esperienza la TC-3D rappresenta un potente strumento di diagnosi e offre molti vantaggi consentendo una precisa rappresentazione volumetrica dei tessuti del cranio e dei tessuti molli senza sovrapposizione di parti anatomiche garantendo un'elevata precisione delle immagini. Nel nostro caso la TC-3D offre una rappresentazione completa di tutte le malformazioni sia mandibolari che dell'osso temporale che caratterizzano la Sindrome di Goldenhar presupposto fondamentale per la pianificazione dell'approccio terapeutico ortodontico e otorinolaringoiatico.

PAROLE CHIAVE: *Sindrome di Goldenhar • TC-3D • Dismorfismi facciali*

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Introduction

Goldenhar syndrome, also known as oculo-auriculo-vertebrale syndrome (OAVS), is a clinically congenital heterogeneous disorder characterized by numerous anomalies affecting the first and second branchial arches of the first pharyngeal pouch, the first branchial cleft and the primordia of the temporal bone¹. The frequency of Goldenhar

syndrome is estimated to be 1 per 5600 to 26,550 births with a male:female ratio of about 3:2, more frequently afflicting right hemifaces (right:left, 3:2)².

Goldenhar syndrome is characterized by abnormalities of the face (hemifacial microsomia, unilateral facial hypoplasia and lateral facial cleft), eyes (epibulbar dermoid or lipodermoid, mostly bilateral; colobomas of the upper eyelid, iris, choroidea and retina; and other eye anomalies),

and ears (microtia, anotia, preauricular skin tags or blind fistulas and other external ear malformations)^{13,4}. The clinical characteristics of Goldenhar syndrome vary from minor facial asymmetry to severe underdevelopment of one half of the face, with orbital deformation and microtia, or sometimes the total absence of the ear⁵. Goldenhar syndrome consists of the complete triad of epibulbar dermoids, accessory auricular appendages and pretragal fistulae. Microtia and/or auricular tags are present in 100% of cases.⁶ Combined conductive and sensorineural hearing loss is present in approximately 50% of affected individuals^{7,8}.

A number of cases have been reported in the literature, often with other malformations, such as cardiac, renal, and central nervous system disturbances and vertebral and other skeletal anomalies. A few cases with oro-dental anomalies have also been described^{1,4,9}.

Herein, we demonstrate the utility of 3D-CT in a case of Goldenhar syndrome with particular regard to study of the mandible and temporal bone.

Case report

A 7-year-old boy with Goldenhar syndrome with post-natal genetic diagnosis was referred to our Department of Radiology for imaging study of the temporal bone. The patient underwent a 3D-CT scan. Brilliance CT-64-channel (Philips Medical Systems, Amsterdam, The Netherlands) was employed. The protocol generated 120 kV and a current of 150 mA. Slice thickness of the images was 1-mm with a 0.5-mm increment. Images in DICOM format were processed on a PC using commercial software, obtaining a three-dimensional reconstruction of skeletal structures.

The 3-dimensional reconstruction of the skin plans showed hypoplasia of the auricle that on the right side consisted of a simple dysmorphic rough. The left auricle had regular dimensions and morphology. Atresia of the right auditory conduit and a normal left auditory conduit were observed. The upper images showed partial ossification of the cranium typical in paediatric age. Description of the profile: typical II class very convex profile with interposition of the inferior lip and a reduced neck-chin angle. (Fig. 1).

Skeletal studies show that the mandibular condyle, ramus and corpus were shorter on the affected side. Tilting of oral rhyme, occlusal plane and mandibular plane were evident. Number or form abnormalities were not found during mixed dentition. Mandibular condyles and temporo-mandibular joints seemed to be asymmetric, and in particular the right condyle was deformed, shorter and wider than the other side (mandibular condyle dimensions were 11 × 7 mm right and 14 × 7 mm left). The 3-dimensional reconstruction of the skeleton planes showed the different morphology of the right and left hemimandibula. Mandibular 3-dimensional reconstruction showed in detail the shift of the medial axis to the hypoplastic right side and the asymmetry of the right mandibular condyles

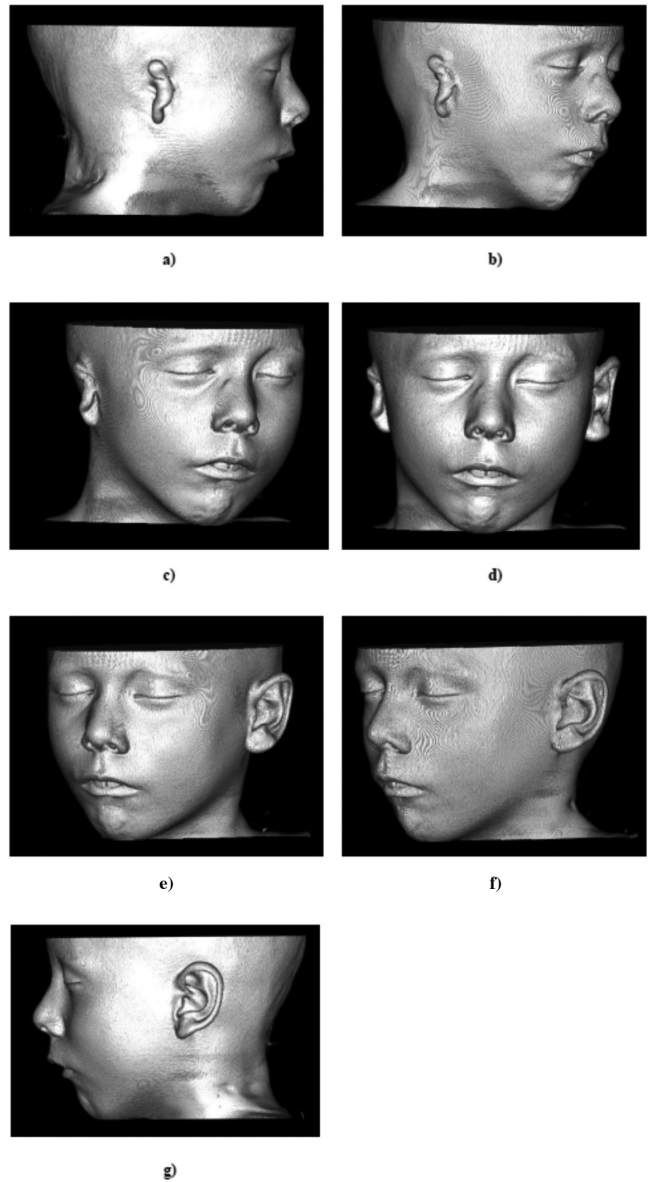


Fig. 1. (a-b-c-d-e-f-g) Three-dimensional reconstruction of the soft tissues.

that appears dysmorphic short and wide in axial and coronal scansions (Figs. 2, 3).

The study of the temporal bone showed on the right side an external auditory canal atresia, hypoplasia of tympanic cavity that was not pneumatized and occupied by dense materials from soft parts. No pneumatization was present in right mastoid cells. Left external auditory canal dimensions and morphology were normal. The left tympanic cavity was normally formed and totally occupied by dense materials from soft tissues. Both right and left ears presented minor hypoplasia of auricle bones in the right side. In agreement with the resolution of our instrument, normal representation of the malleus head that seemed articulated with a dysmorphic rough of a deformed incus was observed; it was impossible to see the staples. The left malleus and incus were visible and appeared to be



Fig. 2. (a-b-c-d) Three-dimensional reconstruction of the hard tissues.



Fig. 3. Three-dimensional reconstruction of the mandible.

articulated in an abnormal manner (reduce contact surface); the staples was difficult to observe but seemed to have regular morphology. Dimensions and morphology of cochlea semicircular canals, vestibulum and utriculum were unaltered (Figs. 4-7).

In conclusion, through 3D-CT valuation of this young patient we observed asymmetry of the condyles; in particular, the right condyle was dysmorphic, short and wide; the auricle of the right external ear was hypoplastic, only a dysmorphic rough was present; the right middle ear had a hypoplastic tympanic cavity; the right internal ear has an atresic auditory conduit.

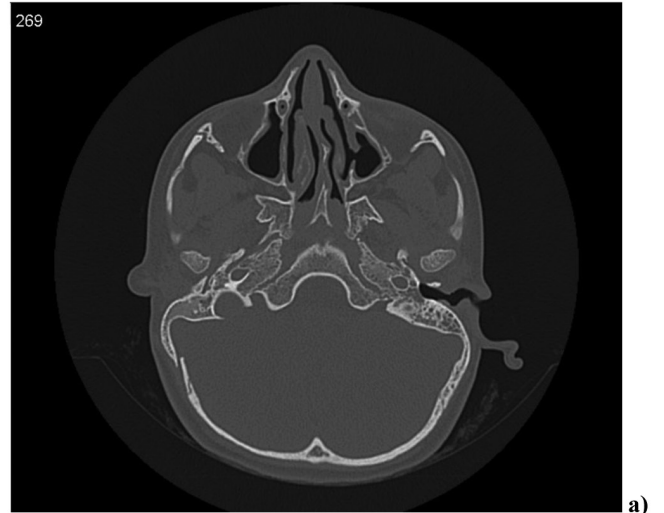


Fig. 4. (a-b) Bi-dimensional views of the external ear.

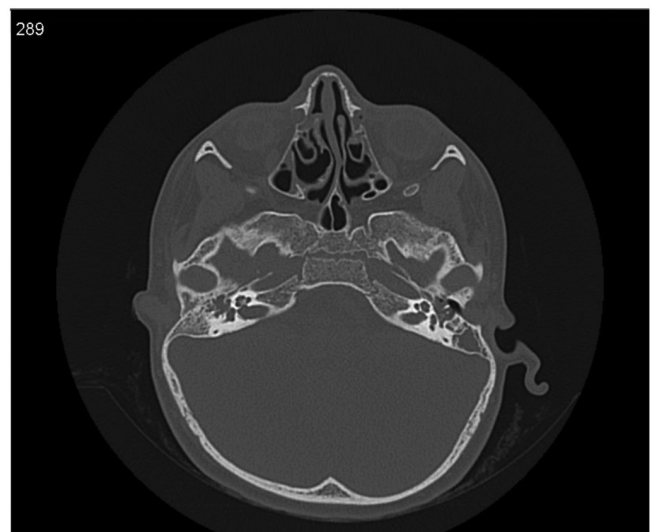


Fig. 5. Bi-dimensional views of the middle ear: ossicular chain and hypoplastic tympanic cavity.

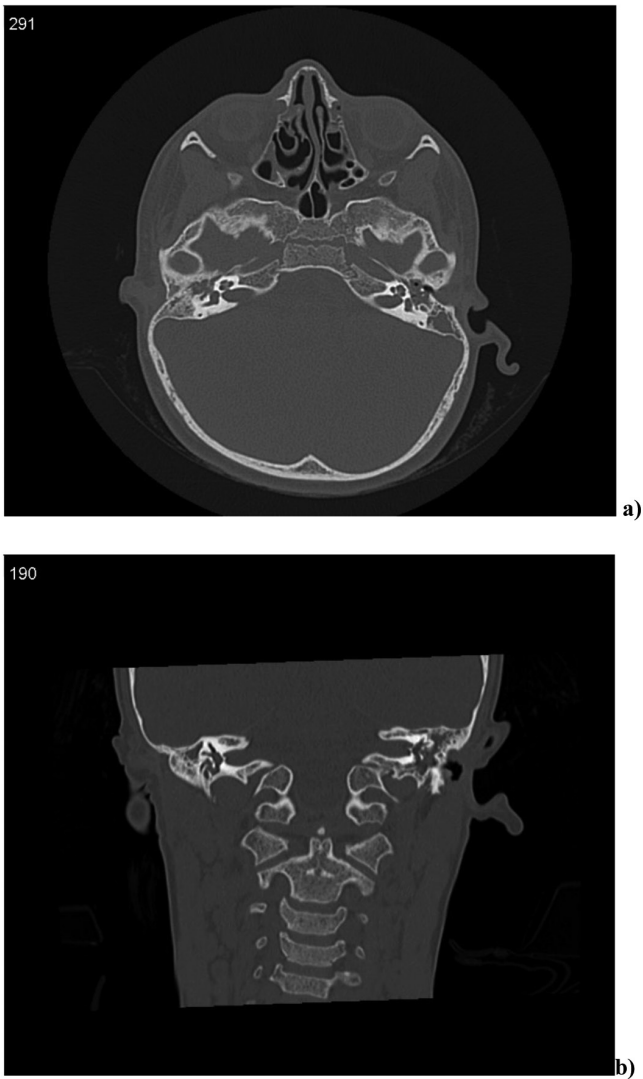


Fig. 6. (a-b) Bi-dimensional views of the internal ear.

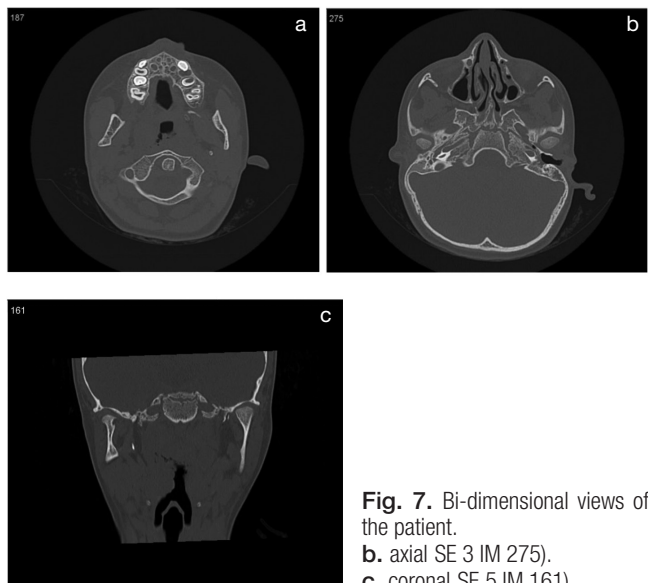


Fig. 7. Bi-dimensional views of the patient.
 b. axial SE 3 IM 275).
 c. coronal SE 5 IM 161)

Discussion

Goldenhar Syndrome is a very rare pathology that was first described in 1952 by Maurice Goldenhar and is characterized by microtia, hemifacial microsomia, dermoids and vertebral anomalies¹⁰. The cause of this condition remains unclear¹¹, but seems to be heterogeneous¹². Most cases are sporadic. However, familial cases and concordant monozygotic twins have been reported, suggesting a genetic origin¹³. In addition, several chromosomal abnormalities have been documented in affected individuals¹⁴. Multiple chromosomal anomalies have been linked to this complex, the most significant of which are deletion of 5q, trisomy of 18, and duplication of 7q. Oculo-auriculo-vertebral spectrum has also been observed in the children of diabetic mothers, and include those exposed to retinoic acid, thalidomide, and primidone¹⁵. Recent research has investigated the potential interaction of environmental factors with genes, and suggest the possibility of multifactorial inheritance. Genetic diagnosis is recommended to understand the type of abnormality.

Goldenhar Syndrome is clinically heterogeneous and there is no agreement in the literature on minimal diagnostic criteria; no clinical exam is 100% diagnostic so that it cannot be used for pregnancy screening, except in case of malformation of other organs. It is difficult to examine the temporal mandibular joint using radiographs due to the superimposition of neighboring structures, such as the petrous region of the temporal bone, the mastoid process, and the articular eminence. CT imaging has become an alternative to conventional radiographic methods because it provides a high quality image without superimposition, as well as 3D reconstruction and analysis of the joints for determining the actual dimensions of the structures¹⁶. The accuracy of 3D-CT reconstructions has been amply demonstrated. 3D-CT is a powerful diagnostic instrument and offers many advantages: volumetric reproduction of cranium and soft tissues, no overlap of anatomic parts that limits the visibility of the structures, high precision and assurance of images, and a constant and easily reproducible reference system. The only disadvantages are the horizontal position of the patient during the exam that can distort the location of the soft tissues of the face (because of artifacts there is a lack of detail about the occlusion), the expensive cost of the procedure and the time of exposure, which are higher than in other imaging techniques. Cavalcanti¹⁷ studied the accuracy of 3-CT by comparing the results of linear measurements on 3D images with physical measurements taken on skulls. They concluded that the difference between the two measurements was minimal and that the 3D images were of high precision. 3D-CT enables visualization of specific tissues in several sequential planes without the problems of superimposition and magnification and provides the possibility to

make an accurate diagnosis of anatomic structures and malformation associated with this syndrome.

The therapeutic strategies are complex because of the variability in patient conditions depending on the important involvement of body areas that requires multidisciplinary competences (otorhinolaryngology, oculist, orthopaedic surgery, maxillofacial surgery, orthodontic). For this reason, it is very important to have a diagnostic approach that can study different body structures. Furthermore, according to previous reports, we suggest always an accurate radiological study that shows all the anatomic and functional cranial alterations. In our case, 3D-CT imaging offered a very complete evaluation of all malformations of mandibular and temporal bone that characterize this syndrome, and represents an important step for ENT and orthodontic therapeutic approaches.

References

- 1 Kokavec R. *Goldenhar syndrome with various clinical manifestations*. Cleft Palate Craniofac J 2006;43:628-34.
- 2 Goldenhar M. *Malformative associations of eye and ear, especially the dermoid syndrome and the epibulbar dermoid-auricular appendix-preauricular fistula auris congenital syndrome and its connections with mandibular-facial-dysostosis*. J Genet Hum 1952;1:243-82.
- 3 Garcia de Paula e Silva FW, de Carvalho FK, Diaz-Serrano KV, et al. *Solitary median maxillary central incisor in association with Goldenhar's syndrome: a case report*. Spec Care Dentist 2007;27:105-7.
- 4 Vinay C, Reddy RS, Uloopi KS, et al. *Craniofacial features in Goldenhar syndrome*. J Indian Soc Pedod Prev Dent 2009;27:121-4.
- 5 Bielicka B. *Interdisciplinary treatment of patients with Goldenhar syndrome - clinical reports*. Dent Med Probl 2006;43:458-62.
- 6 Jena AK, Duggal R. *Atypical Goldenhar syndrome: a case report*. J Clin Pediatr Dent 2006;31:118-22.
- 7 Ottaviano G, Calzolari F, Martini A. *Goldenhar syndrome in association with agenesis of the internal carotid artery*. Int J Pediatr Otorhinolaryngol 2007;71:509-12.
- 8 Paludetti G, Conti G, DI Nardo W, et al. *Infant hearing loss: from diagnosis to therapy Official Report of XXI Conference of Italian Society of Pediatric Otorhinolaryngology*. Acta Otorhinolaryngol Ital 2012;32:347-70.
- 9 Pinheiro AL, Araújo LC, Oliveira SB, et al. *Goldenhar's syndrome - case report*. Braz Dent J 2003;14:67-70.
- 10 Goldenhar M. *Associations malformatives de l'oeil et de l'oreille, en particulier le syndrome dermoïde epibulbaire-appendices auriculaires fistule auris congenital et ses relations avec la dysostose mandibulofaciale*. J Genet Hum 1952;1:243-82.
- 11 Gorlin RJ, Cohen MM Levin LS, et al. *Syndromes of the Head and Neck*. New York: Oxford University Press; 1990. p. 641-6.
- 12 Rollnick BR, Kaye CI. *Hemifacial microsomia and variants: pedigree data*. Am J Med Genet 1983;15:233-53.
- 13 Kobrynski L, Chitayat D, Zahed L, et al. *Trisomy 22 and facioauriculovertebral sequence*. Am J Med Genet 1993;46:68-71.
- 14 Kaye CI, Martin AO, Rollnick BR, et al. *Oculoauriculovertebral anomaly: segregation analysis*. Am J Med Genet 1992;43:913-7.
- 15 Cohen MM, Rollnick BR, Kaye CI. *Oculoauriculovertebral spectrum: an updated critique*. Cleft Palate J 1989;26:276-86.
- 16 Yañez-Vico RM, Iglesias-Linares A, Torres-Lagares D, et al. *Diagnostic of craniofacial asymmetry*. Literature review. Med Oral Patol Oral Cir Bucal 2010;15:e494-8.
- 17 Cavalcanti MG, Haller JW, Vannier MW. *Three-dimensional computed tomography landmark measurement in craniofacial surgical planning: experimental validation in vitro*. J Oral Maxillofac Surg 1999;57:690-4.

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