A Female Case of Goldenhar Syndrome with Mandibular Hypoplasia and Aural Involvement

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Authors’ contributions

This work was carried out in collaboration between all authors. All authors read and approved the final manuscript.

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ABSTRACT

Goldenhar syndrome (GS) is a poly-malformation syndrome, also defined as oculo-auriculo-vertebral dysplasia with hemifacial microsomia. It is a rare congenital defect involving first and second branchial arches. The aetiology is not known. The most supported hypothesis is based on the abnormal embryonic vascular supply after mesodermal migration. Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have been reported. We report the case of a female neonate affected by hemifacial microsomia and presence of pre-auricular tragi. Patients were subjected to computed tomography scan and MRI that revealed a mandibular unilateral hypoplasia without association of skeleton, brain and ocular alteration. The purpose of our study was to define the important role of the CT and MRI in the diagnosis of this poly-malformation syndrome.

Keywords: Goldenhar syndrome (GS); magnetic resonance; computed tomography; oculo-auriculo-vertebral dysplasia; hemifacial microsomia; pre-auricular tragi.
1. INTRODUCTION

Goldenhar syndrome (GS) is a rare syndrome, characterized by oculo-auricolo vertebral dysplasia and hemifacial microsoma. It is a congenital alteration of the first and second branchial arches. It is identified by a variable combination of defects that determine an incomplete facial development and the facial abnormalities are unilateral in many cases. The diagnosis of GS was made on clinical examination alone. In fact, a our knowledge, no diagnostic criteria have been established [1-3]. We show the case of a female neonate affected by hemifacial microsoma and presence of preauricular tragi (Fig. 1). We submitted the patient to whole body X-Ray, Magnetic Resonance (MR) and Computed Tomography scan (CT) that revealed a mandibular unilateral hypoplasia without any type of skeleton, brain, ocular and inner ear associated alteration.

Fig. 1a. Note the facial asymmetry and (b) preauricular and facial tragi

2. PRESENTATION OF CASE

A baby girl was born to a 35 years old second gravid mother by vaginal uncomplicated delivery at 41 weeks gestation. The baby was affected by a facial asymmetry and she was submitted to diagnostic study in our Radiology Department to evaluate this alteration. She had a birth weight of 3230 g; a length of 50 cm and a head circumference of 33.5 cm. Apgar score was 9 respectively at 1 and 5 minutes of life. Baby had a normal adaptation without neonatal respiratory distress. Familiar anamnesis was negative for congenital or genetic diseases. Maternal prenatal history was uneventful; pregnancy was uncomplicated and negative for drugs assumption and alcohol abuse. Maternal infectious diseases status was negative for TORCH during pregnancy. Due to the facial abnormalities the baby was moved to the neonatology department, in the third day of life. The dysmorphic features of the baby included left hemi facial microsoma, left mandibular hypoplasia and bilateral pre-aureicular tragi. Hemogram and biochemical parameters of blood did not reveal abnormalities. The cardiac, cerebral and abdominal ultrasounds were negative. Ophthalmology consult was negative for ocular abnormalities. Measurement of evoked acoustic oto-emissions recorded a conductive hearing loss. Patient underwent conventional X-ray scan, MRI and CT scan. X-Ray (Fig. 2) Whole body X-Ray scan did not show any type of the skeletal abnormalities.

The MRI control was carried out with a MRI unit (1.5 T GE- Signa) with dedicated 8 channels coil. Scan plan thickness was 4 mm and spacing 0.4 mm. The sequences acquired included axial DP-T2 FSE, axial T1-SE, axial T1-SE FAT SAT, coronal T2-FSE, axial FLAIR, sagittal T1 frFSE, axial DWI and axial HR T2 FAT SAT sequences (Fig. 3). The exam was unremarkable for acute pathology and excluded associated brain, inner ear and ocular anomalies with myelination progress normal for age and no signs of ocular dermoid cysts [2-3-4-11-12-13]. To perform CT scan of the head, it was used a 320 row-detector CT scanner (Aquilion ONE, Toshiba Medical Systems, Japan) with built-in low dose protocol for paediatrics (100 KV, 350 mAs, collimation 0.5 mm, rotation time 0.5s); further 3D reconstructions were achieved [1-10]. The most
relevant radiological evidence to support diagnosis was found at CT scan of the head, performed to assess the cause of facial asymmetry, and revealed abnormal development of right hemi mandibular, with hypoplasia of the ipsilateral ascending ramus and lack of development of the corresponding mandibular condyle. No cleft and palate was detected, no inner ear malformations were found (Fig. 4).

3. DISCUSSION

Goldenhar and Treacher Collins syndrome (TCS) are included in the development anomalies of the first and second branchial arches. Characteristics of TCS are palpebral fissures, colobomas, zygomatic and mandibular hypoplasia, partial absence of the lower eyelid cilia, and abnormalities of the ears. The main difference between the two syndrome is that TCS have more often bilateral involvement [9]. Other syndromes associated with multiple pre-auricular tragi include Townes Brocks syndrome, Delleman syndrome and Wolf Hirschhorn syndrome (cervico-oculo-acoustic syndrome) [1].

The Goldenhar syndrome was first described in 1952 by Maurice Goldenhar. It has a frequency ranging from 1/35000 to 1/56000 births and affects more often the right part of the face (R/L = 3/2) and male subjects (m/f = 3/2) [2]. This pathology is characterized by ocular defects like epibulbar lipodermoids or dermoids, coloboma and microphthalmia. The aural involvement consists of pre-auricular tragi, microtia and variable grades of conductive hearing loss. The vertebral anomalies such as hemi vertebrae, cervical fusions, scoliosis, supernumerary vertebrae, occipitalization of the atlas and bifid spine are also frequent. The hemi facial microsomia is characterized by mandibular hypoplasia, macrostomia and in some cases cleft lip and palate [3]. Most common cardiovascular anomalies associated with Oculo-auriculo-vertebral spectrum (OAVS) are Tetralogy of Fallot and ventricular septal defects [4]. Furthermore micrognathia, webbing of the neck, short neck, tracheo-esophageal fistulae, abnormalities of sternocleidomastoid muscle, umbilical hernia, inguinal hernia, urologic anomalies, hypo plastic vagina may be associated [3]. The aetiology is not known. The most supported hypothesis is based on the abnormal embryonic vascular supply after mesodermal migration. According to others theories, there is some type of vascular perturbation and/or neural crestopathy or some other factor leads to defective formation of the branchial arches and vertebral systems [10]. Most of the cases are sporadic. Familial occurrences have been observed. Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have been reported. [5]. Heavy alcohol consumption, use of drugs such as thalidomide, retinoic acid, tamoxifen, and cocaine during pregnancy may be related to the development of this syndrome. Maternal diabetes, rubella and influenza are possible etiologic factors [6,7]. Monoud described a case of GS after maternal intoxication of vitamin A [8]. This pathology need to a surgery reconstruction in order to correct the mandibular and ear alteration. It can also make an enlargement of the cheeks [8].

Fig. 3. MR study: (a) coronal T2-FSE, (b) axial T1-SE, (c) axial HR T2 FAT SAT sequences and (d) axial FLAIR don’t show any abnormalities in the brain
4. CONCLUSION

Magnetic resonance imaging (MRI) is the examination of choice for CNS malformations of its high sensitivity and multi planar capability [11-13]. As reported by other authors the use of MRI in this complex pathology allows extensive research and evaluation of eventually associated malformations of the CNS frequently observed in GS cases [5]. In our case, the MR study has ruled out inner ear, vascular and ocular abnormality frequently detected in this syndrome. Specific high resolution scans planes and sequences allow evaluation of the orbits and neural structures. To assess bone involvement of the facial structures, a CT scan of head can be useful, and reasonably safe [14].

CONSENT

All authors declare that 'written informed consent was obtained from the patient (or other approved parties) for publication of this case report and accompanying images.

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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Fig. 4. CT scan: (a-c) 3D reconstructions, (b) MPR coronal reconstruction. It's evident abnormal development of right hemimandible, with hypoplasia of the ipsilateral ascending ramus and lack of development of the corresponding mandibular condyle.
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