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Goldenhar Syndrome: About A Case

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Abstract: Goldenhar syndrome is a rare disease that was first described in the early 1950s. It is characterized by a polymalformative syndrome most commonly associated with an epibulbar dermoid cyst, pretragalian diverticula and facial anomalies. We report a case of Goldenhar syndrome, where almost all the classic signs of this rare disease were present including transmission deafness. A 7-year-old girl was examined in our department of otolaryngology and maxillofacial surgery in Rabat, a facial asymmetry, associated with right facial paralysis and hypoplasia of the right mandible. At the otological examination we notice the presence of pretragian tubercles without pre-fistulae with agenesis of the right pavilion. The right external acoustic meatus is stenosed not allowing visualizing the eardrum. At the audiogram: transmission deafness more marked on the right. CT of the temporal bone shows a major aplasia on the right side and minor aplasia on the left side. Gorlin, in 1963, suggested the name of oculo-auriculo-vertebral spectrum (OAVS) and it also included vertebral anomalies as signs of this syndrome. The etiology of the oculo-auriculo-vertebral spectrum remains uncertain. However, there are important arguments in favor of the existence of multiple causes and it seems to be a heterogeneous etiologic group.
Keywords: Epibulbar dermoid cyst; Goldenhar syndrome; Oculo-auriculo-vertebral spectrum.

INTRODUCTION

Goldenhar syndrome is a rare disease that was first described in the early 1950s. It is characterized by a polymalformative syndrome most commonly associating an epibulbaric dermoid cyst, pretragial diverticula, and facial massive abnormalities.

CASE PRESENTATION

A 7-year-old girl was followed in our department of otolaryngology and maxillofacial surgery of Rabat with facial asymmetry, hypoplasia of the right hemi-mandible, associated with right facial paralysis. On otological examination we note the presence of pretragial tubercles without pre-auricular fistulas with agenesis of the right pavilion; the acoustic external meatus is stenosed, which does not allow the eardrums to be visualized. At the acoumetry we notice a transmission Deafness more marked on the right. Ophthalmological, neurological and osteoarticular examination are normal.

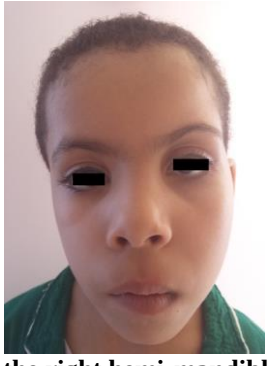

Fig-1: facial asymmetry, hypoplasia of the right hemi-mandible, associated with right facial paralysis

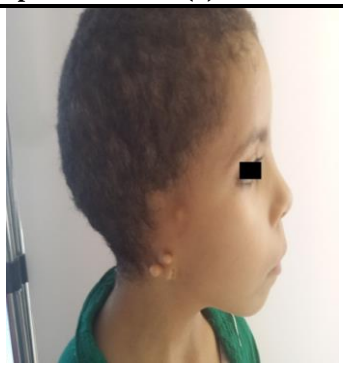

Fig-2: agenesis of the right pavilion with pretragial tubercles

A paraclinical report was made thus to the orthopantomogram (figure 3) we note the presence of hypoplasia of the mandible on the right side and a normal dental development. The child had no other malformation or clinical abnormality. The staturo-ponderal state was normal for age.

An extension assessment with X-rays of the cervical spine and thorax and an abdominal ultrasound were performed and returned normal.

The CT of petreous bone (Figure 4) shows major aplasia of the right side and minor aplasia of the left side.

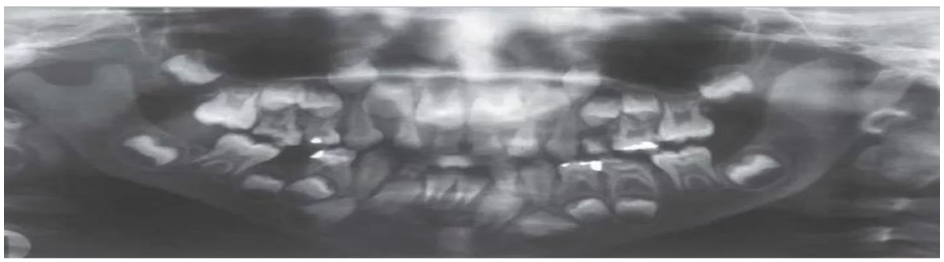

Fig-3: hypoplasia of the mandible on the right side

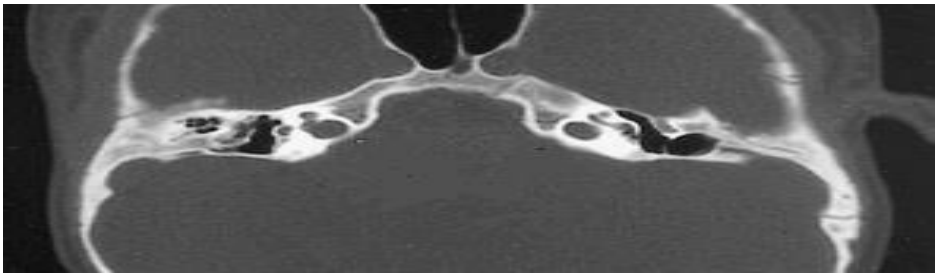

Fig-4: major aplasia of the right side and minor aplasia of the left side

DISCUSSION

Goldenhar syndrome is a malformative complex that combines a corneoscleral dermoid cyst, an upper eyelid coloboma, mandibular hypoplasia and a structural change in one or both ears, as well as vertebral abnormalities. It was first described in 1845 by Von Arlt but recognized as a syndrome by Goldenhar in 1952. Its actual incidence is not known, as most publications generally address oculo-auriculo-vertebral spectrum (OAVS). The prevalence of these is estimated at 1 in 5,600 cases to 1 in 45,000 live births, according to the authors. In the population of Tasse *et al*. [1], Goldenhar syndrome was present in 7.5% of patients OAVS carriers.

The etiological factors of Goldenhar syndrome are poorly understood. The association of Goldenhar syndrome with taking traditional medicines during pregnancy has also been described. On the other hand, Baum *et al.* [2] reported a notion of family history in two cases. Associated chromosomal abnormalities have been found by other teams, including the short arm of chromosome 5 and the long arm of chromosome 22 [3].

Clinically, our patient had an atrial abnormality chart associated with abnormalities of the mandible, consistent with the diagnostic criteria for Goldenhar syndrome. However, Goldenhar's syndrome may include ocular abnormalities such as coloboma, corneoscleral cyst, dacryocystitis, lacrimal nerve stenosis, anophthalmia, cryptophthalmia, cataract, irian abnormalities. Other malformations may be associated and sometimes worsen the prognosis; thus Furtado described a tracheal stenosis in a patient. In the

Morrison et al cohort [2] 8 out of 25 OAVS patients had cardiac abnormalities: 4 isolated inter ventricular communications (IVC), 1 IVC associated with atrial communication, 1 IVC plus pulmonary artery stenosis, 1 tetralogy of Fallot and 1 complex heart disease. For Greenwood *et al*. [3], one in seven patients has a cardiac anomaly, half of which is a tetralogy of Fallot. In the Rollnick *et al*. [3] series, in a population of 294 OAVS patients, cleft lip or labia palate was found in 22% of cases, 5% in cardiac abnormalities and 34% in skeletal abnormalities, while 52% cases had no associated abnormality. Finally, Digillio *et al*. have described in addition to the abnormalities elicited laryngomalacia in two out of three patients. In our patient, no associated abnormalities were found during clinical examination or skel *et al*. X-rays, echocardiography or laryngoscopy. Life-threatening may be involved if Goldenhar's syndrome is associated with a general abnormality, especially cardiovascular.

Other syndromes associated with multiple preauricular tragi include Treacher-Collins syndrome, Wolf-Hirschhorn syndrome, Nager's acrofacial dysostosis, Wildervanck syndrome (cervicooculoacoustic syndrome), Townes-Brocks syndrome and Delleman syndrome [4,5]. Treacher Collins syndrome is associated with maxillary and mandibular hypoplasia but is not associated with ocular and aural anomalies [6].

The care is multidisciplinary and it combines the maxillofacial surgeon, the otolaryngologist, the ophthalmologist, the stomatologist, the pediatrician, the plastic surgeon, the anesthesiologist and the geneticist [7].

CONCLUSION

Goldenhar Syndrome is a rare congenital anomaly. Its management is multidisciplinary and remains difficult in our context. The vital prognosis can be engaged in case of associated malformations that it is essential to look for.

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