

Goldenhar's Syndrome - Case Report

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Goldenhar's syndrome is a rare condition described initially in the early 1950's. It is characterized by a combination of anomalies: dermal epibulbar cysts, auricular appendices and malformation of the ears. In 1963, Gorlin suggested the name oculo-auriculo-vertebral (OAV) dysplasia for this condition and also included vertebral anomalies as signs of the syndrome. The etiology of this rare disease is not fully understood, as it has shown itself variable genetically and of unclear causes. This work reports a case of Goldenhar's syndrome in an 11-year-old female, who presented all classical signs of this rare condition

Key Words: deformity, manifestation, oculo-auriculo-vertebral dysplasia.

INTRODUCTION

Goldenhar's syndrome is a rare presumed inherited condition, which has a multifactorial etiopathology that also includes nutritional and environmental factors that can result in disturbances of blastogenesis (1). There are several terms used to describe this rare condition known as oculo-auriculo-vertebral (OAV) dysplasia, including Goldenhar's syndrome and hemifacial microsomia (2). Goldenhar first described this condition in 1952 as a disease that presents a combination of several anomalies such as dermal epibulbar tumors, peri-auricular appendices and malformation of the ears. In early 1990's, this condition was better understood and it was agreed that, besides the picture described by Goldenhar (1952) and Gorlin (1963), this syndrome may also present heart diseases and hypoplasia of the zygomatic, mandibular and maxillary bones (3). Some authors also pointed out facial muscle hypoplasia, anatomical and morphological abnormalities of the tongue, vertebral abnormalities, anomalies of the eyes (1), lip and cleft palate (3), disturbances of the central nervous system and other

visceral anomalies (4).

There is not enough information to identify its etiologic factors. Abnormalities of chromosomes have been identified (5). On the other hand, another study suggested a disturbance of the neural crest cells as the cause of the disease (6). The influence of other factors, including the environment, during pregnancy has been also blamed. The ingestion of some drugs such as cocaine, thalidomide, retinoic acid, and tamoxifen by the mother were also related to the development of the disease (4). Maternal diabetes has also been suggested as an etiologic factor (7).

There is an overall consensus that the diagnosis of this disease must not be only based upon radiologic or laboratory results. The diagnosis of Goldenhar's syndrome should be mainly based on the clinical aspect and associated with both systemic conditions and radiologic findings (7). Most authors consider the presence of anomalies of the ear (microtia) and of appendices on the ear necessary for diagnosis. Additionally, facial asymmetry or facial and/or mandibular hypoplasia, dermal epibulbar tumors, palpebral alterations, vertebral anomalies, lateral facial clefts, and renal prob-

lems are observed (8,9). Additionally both laboratory and image tests are important for the diagnosis of the disease because anomalies of the skeletal or facial bones can be diagnosed by means of several types of image exams available today. Radiographic examination of zygomatic bones shows a macroscopic deficiency and developmental symmetry. There is also a



Figure 1. Facial assymetry of the left side and dermal epibulbar cyst on the left eye.



Figure 2. Birthmark on the upper lip.



Figure 3. Scars resulting from the surgical removal of periauricular polyps.

possibility of agenesis of these bones with lack of fusion of the zygomatic arch and agenesis of the palatine bones. Palatal cleft may be observed radiographically (10). Ophthalmologic and otorhinolaryngologic examination are also important for the final diagnosis.

CASE REPORT

An 11-year-old white female was examined at the Oral and Maxillofacial Clinic of the School of Dentistry of the Federal University of Bahia. Clinical examination indicated Goldenhar's syndrome. The patient presented facial asymmetry, hypoplasia of the mandible, dermoid epibulbar tumor on the left eye (Figure 1) and birthmarks on the upper lip and palate (Figure 2). Additionally there was evidence that periauricular polyps were surgically removed when the patient was eight months old (Figure 3). No mental problem was detected during examination. There were no signs of hearing impairment or lip and cleft palate. The mother reported the use of an anti-convulsive drug (Comital®) because of epilepsy, prior to knowing of the pregnancy (about four weeks) when the drug was changed to Phenobarbital (Gardenal®) a more suitable drug for use during pregnancy. X-ray examination of the skull and vertebral column did not show abnormalities. However, orthopantomographic examination detected hypoplasia of the mandible on the left side (Figure 4), absence of the coronoid process and hypoplasia of the mandibular condyle. Dental development was normal.



Figure 4. Orthopantomography: Agenesis of the coronoid process of the mandible, mandibular hypoplasia and hypoplasia of the condyle on the left side.

DISCUSSION

The study of this condition is still controversial because of its complexity and broad clinical aspects. Microtia appears to represent its less complex manifestation, however, several facial and systemic abnormalities may also be observed (9).

The patient exhibited clinical characteristics of complex and severe AOV syndrome as described previously (1,6), including facial asymmetry, hypoplasia of the mandible, epibulbar dermoid tumor on the left eye, vestiges of cleft lip, and the presence of previously removed peri-auricular appendices. Facial asymmetry and hypoplasia of the mandible are typical features of OAV syndrome (11). On the other hand, the presence of epibulbar dermoid tumor is variable (3). Although the patient showed vestiges of a cleft lip, this alteration is observed in about 5% of the cases (3). It is important to observe that when epibulbar dermoid tumors are present there is a tendency for the development of bilateral peri-auricular appendices as observed in this specific case.

Despite the reported frequency of cardiovascular alterations ranging from 5 to 58% (12), in this patient no cardiovascular alterations were found. Hearing disturbance or malformation of the external auditory meatus were not observed (13) nor was dysfunction of the facial nerve, which prevalence is high (14). Renal problems commonly associated to malformations of the ears (15) were not diagnosed. Previous reports of 294 patients (6) showed that these anomalies are uncommon and appear in less than 10% of the patients.

It must be emphasized that there was no previous familial report of this condition and that the mother used an anticonvulsive drug early during pregnancy. This lack of familial occurrence may suggest that Goldenhar's syndrome may be a sporadic event that occurs early in embryogenesis. Some teratological agents such as vitamin A, primidone, thalidomide (16), and cocaine (4) have been associated with the development of this syndrome as well as malnutrition, tobacco, and herbicides that are able to produce free radicals which may break the DNA and consequently result in congenital malformations (1,17).

Although the OAV presents some similarities with the Treacher-Collins' syndrome, it is now considered a distinct entity due to some characteristics not found in both diseases, including the lack of evidence

of a genetic origin of the OAV syndrome (10). In this case, the substitution of Comital® by Gardenal® may explain the development of the syndrome without the presence of all of its variants.

RESUMO

A Síndrome de Goldenhar é uma condição rara que foi descrita inicialmente em 1952 como uma combinação de anomalias que incluíam tumores dermóides epibulbares, apêndices auriculares e mal-formações da orelha. Em 1963, Gorlin sugeriu o termo displasia Oculo-Auriculo-Vertebral (OAV) incluindo anomalias vertebrais nesta entidade clínica. A sua etiologia é pouco clara, apresentando-se geneticamente variável e de causa bastante heterogênea. Os autores relatam um caso clínico de Síndrome de Goldenhar numa criança do sexo feminino, com 11 anos de idade, que apresenta características clássicas dessa síndrome como tumor epibulbar dermóide, apêndices auriculares, hipoplasia mandibular e fenda labial.

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