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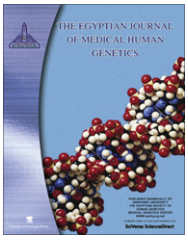

CASE REPORT

Goldenhar syndrome with skin tags on the chest wall

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KEYWORDS

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Epibulber dermoid tumor;
Goldenhar syndrome

Abstract Goldenhar syndrome is a congenital condition that is associated with abnormalities of the head and the bones of the spinal column. The abnormalities of the head can include anomalies of the eyes, ears, facial bones, and mouth. These anomalies are extremely variable in severity. The exact cause of Goldenhar syndrome remains unknown. 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 syndrome in a 1-year-old female, who presented some of the classical signs of this rare condition including Hemifacial Microsomia, epibulber dermoid tumor and preauricular skin tags. However, vertebral anomalies, deafness, renal and cardiac anomalies were absent. Skin tags on the anterior chest wall were reported in this patient for the first time.

1. Introduction

Facio Auricular Vertebral (FAV) or Goldenhar syndrome was first recorded by German physician Carl Ferdinand Von Arlt in 1845 and Goldenhar in 1952 defined the syndrome more clearly. A variety of terms have been used to describe this extremely variable disorder [1]. According to medical literature, when malformations primarily involve the jaw, mouth, and ears and, in most cases, affect one side of the body (unilateral), the disorder is often referred to as Hemifacial Microsomia. If abnormalities of the vertebrae and/or the eyes are also present, the disorder is often called Goldenhar syndrome. Within medical literature, the term Oculo–Auriculo–Vertebral (OAV) Spectrum is often used synonymously with Goldenhar syndrome and Hemifacial Microsomia. However, due to the complexity and varying severity and expression of OAV Spectrum, some researchers suggest that Hemifacial Microsomia and Goldenhar syndrome actually represent different aspects or levels of severity of OAV Spectrum. Goldenhar syndrome is also considered a variant of Cranofacial Microsomia which is the second most common facial birth defect after cleft lip and palate [2].

The incidence of this syndrome is 1 in 3500 to 5000 live births and the male to female ratio of this syndrome is 3:2 [1].

Goldenhar syndrome is caused by a disruption of normal facial development. A baby's face forms very early, normally

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between the eighth and twelfth weeks of gestation. Facial development depends on many different tissues growing together, meeting at the same time and place to form facial features. When the movement and development of these tissues are disrupted, the face may have abnormal openings, underdevelopment, and/or excess skin [3].

There is very little evidence to explain why Goldenhar syndrome occurs. In most cases, Goldenhar syndrome appears to occur randomly, with no apparent cause. However, in some cases, positive family histories have been present that have suggested autosomal dominant or recessive inheritance [4]. Family history may include cleft lip or palate, unusually shaped ears, asymmetry of face, small chin, skeletal problems, eye abnormalities, internal problems or speech and dental problems [5]. The chances of having another child with Goldenhar is less than 1% or less. Patient has about a 3% chance of passing it on to his or her children [1]. This syndrome is seen in all ethnic groups and cultures [5].

Abnormalities of chromosomes have been also identified [4]. 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 [7]. Maternal diabetes has also been suggested as an etiologic factor [8]. Also, it was suggested to be due to exposure to viruses or chemicals during pregnancy, due to abnormal vascular supply to the first arch and abnormality of mesoblastic development affecting the formation of vertebral and branchial systems [9]. Some researchers suggest that the disorder may be due to interaction of many genes with environmental factors (multifactorial inheritance) [9–11].

The symptoms associated with Goldenhar syndrome are highly variable. Some individuals with Goldenhar syndrome have many severe abnormalities, while other individuals have few minor birth defects [7]. It usually affects one side of the face only (usually the right side) [12]. Common defects include; macrostoma (the opening of the mouth is large and extended towards the ear on one side), hypoplasia (underdevelopment) of the muscles in the face, cheekbones and skin, if unilateral it leads to Hemifacial Microsomia a common physical difference seen in Goldenhar syndrome. Also, microtia (a partially formed ear) or ancrotia (a totally absent ear), preauricular (skin tags or pits), usually in front of the ear in line with the mouth opening and epibulbar dermoids are common, microphthalmia, coloboma and strabismus are also reported. Vertebral anomalies include hemivertebrae (spinal vertebrae which are small or not completely formed on one side). Other problems that may occur in some but not all cases are eye defects, deafness, cleft lip or palate, and internal problems affecting the heart, limbs or kidneys [13].

Treatment of these cases consists mainly of cosmetic surgery. The demoid tumors and the preauricular skin tags can be removed. Palatal deformities can be repaired. Early rehabilitation can be done for the mental retardation [12].

2. Case history

The patient was a 1 year and 3 months old female child referred to the genetics clinic complaining of multiple skin tags on the face and chest wall presented at birth. The proband was the second child of healthy non-consanguineous parents. (mother is 28 years old and father was 40 years old). The child was delivered vaginally at home after a full term pregnancy. Antenatal, intranatal and postnatal periods were uneventful with no maternal history of diabetes or exposure to teratogens. The child had a normal motor development and mild delay in mental development.

Physical examination of the child showed coarse, dysmorphic features with facial asymmetry and right sided Hemifacial Microsomia was observed (Fig. 1). Features included frontal bossing, downward slant, strabismus and bilateral epibulber dermoid tumors in both eyes (Figs. 2–4). Bulbus nose with long filtrum and depressed nasal bridge, macrostomia with high arched palate, micrognathia and hypoplasia of the mandible on the right side were observed. Bilateral microtia associated with multiple skin tags in the preauricular region bilaterally were preset (Fig. 5). There were also skin tags on both cheeks and on the anterior chest wall (Fig. 6). Systemic examination did not reveal any cardiovascular or renal abnormality. X-ray of the chest revealed no cardiomegaly, x-ray of cervical spine showed no abnormalities in the cervical vertebrae and no evidence of scoliosis. CT-scan of the brain and audiogram report showed no abnormalities. Hemogram, complete urine examination, blood urea and serum creatinine were in normal levels. Karyotype done for the patient showed no abnormality. Pelviabdominal ultrasound and echocardiography of the child also showed no abnormality.

Parents consent was taken.

3. Discussion

The patient exhibited clinical characteristics of mild OAV syndrome as described previously [1,6], including facial asymmetry, hypoplasia of the mandible, epibulbar dermoid tumor on both eyes, macrostomia with high arched palate and micrognathia, and the presence of multiple preauricular skin tags. Facial asymmetry and hypoplasia of the mandible are typical features of OAV syndrome [14]. On the other hand, the presence of epibulbar dermoid tumor is variable [12]. Epibulbar dermoids and lipodermoids, coloboma of the eyelid, microphthalmia,

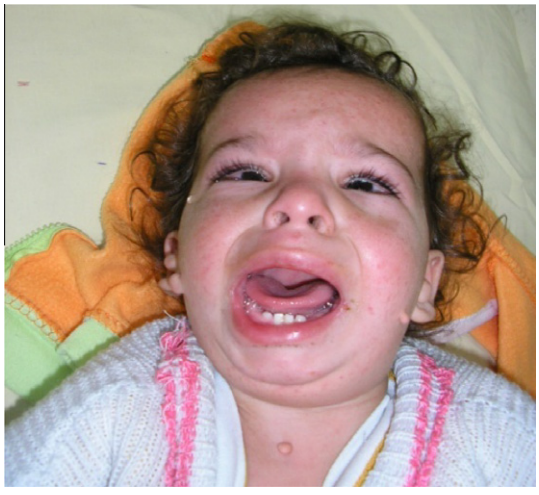
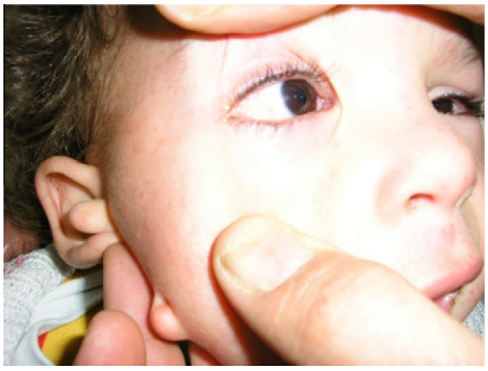
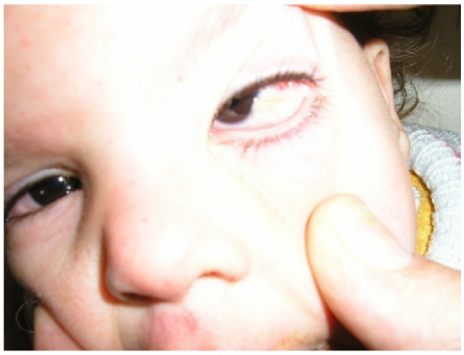


Figure 1 Dysmorphic features and Hemifacial Microsomia.





Figures 2 and 3 Bilateral epibulber dermoid tumors.

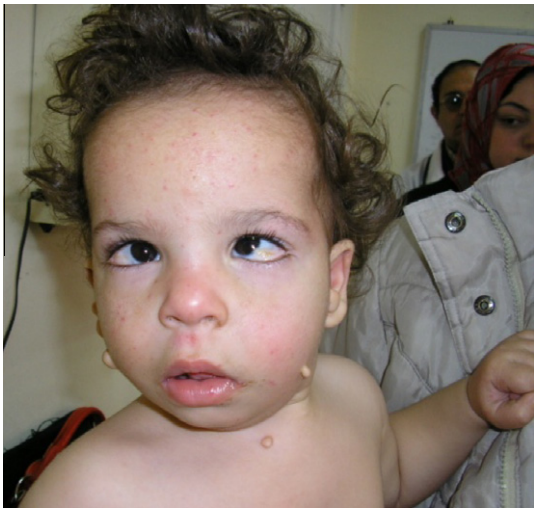


Figure 4 Right Hemifacial Microsomia, skin tags on both cheeks and the anterior chest wall. The eyes show Strabismus.

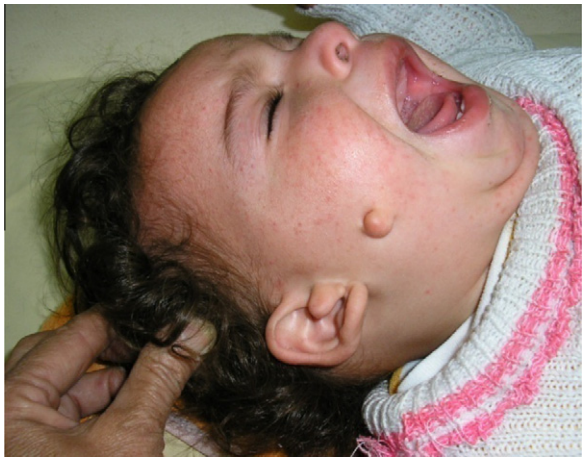


Figure 5 Deformity of the ear lobule with preauricular and cheek skin tags.

strabismus and retinal anomalies are the associated ocular problems in Goldenhar syndrome [15]. It is important to observe that when epibulbar dermoid tumors are present there

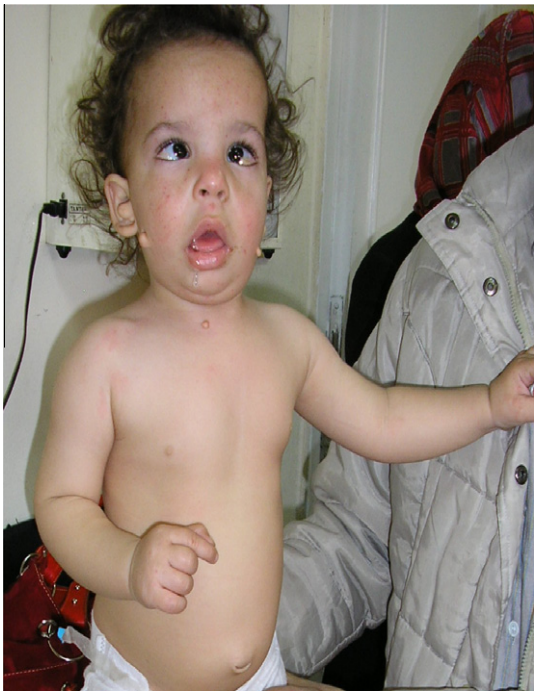


Figure 6 Skin tag on the chest wall.

is a tendency for the development of bilateral preauricular appendices as observed in this specific case.

The ear anomalies in Goldenhar syndrome include anortia, microrticia, external ear tags and aural fistulae [16–18]. The syndrome may or may not be associated with hearing loss [19]. Conductive and / or sensorineural hearing loss is present in 50% of the patients with this syndrome. The etiology of the hearing loss is varied and may include missing or malformed outer ear (anotia and microtia), narrow or missing ear canals (atresia), abnormal skin tags on or infront of the ears (preauricular tags) obstructing the external auditory meatus and abnormalities in the middle or inner ear [20]. In this case, the child showed microrticia associated with malformation of the right pinna and preauricular skin tags bilaterally. Hearing disturbance was not observed nor was dysfunction of the facial nerve, which has a high prevalence [21]. Audiometery done showed no hearing loss.

Our child showed a single skin tag on the upper part of the anterior chest wall (Fig. 6). To our knowledge no repots of

similar skin tags in this site have been reported in a case of OAV syndrome in the literature. No vertebral abnormalities were detected in our patient. A small percentage also has mental retardation associated with this syndrome. [15] Our patient had a mentality slightly below her age.

Despite the reported frequency of cardiovascular alterations ranging from 5 to 58% [22], in this patient no cardiovascular alterations were found. Renal problems commonly associated to malformations of the ears [23] were not diagnosed in our patient. 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 in our patient there was no previous familial report of this condition and there was no consanguinity between the parents suggesting a sporadic event that occurs early in embryogenesis.

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 syndrome should be mainly based on the clinical aspect and associated with both systemic conditions and radiologic findings. There is not a genetic test that can diagnose Goldenhar syndrome. Pre-natal diagnosis by scanning may identify the condition in certain cases where facial or skeletal anomalies are present [8].

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