A CASE OF GOLDENHAR'S SYNDROME

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SUMMARY

Goldenhar's syndrome (Goldenhar-Gorlin syndrome, facioauriculovertebral sequence, oculoauriculovertebral dysplasia) is a variant of craniofacial microsomia (first and second branchial arch syndrome). It is generally characterized by epibulbar dermoids and/or lipodermoids, pretragal blinded fistulas, skin tags on the cheek and vertebral anomalies. A 24-year-old female patient with Goldenhar's syndrome was presented in this paper.

Key Words: Craniofacial anomalies - Facial abnormalities - Goldenhar's syndrome.

INTRODUCTION

Goldenhar's syndrome which has similar features to craniofacial microsomia (first and second branchial arch syndrome) is characterized by epibulbar dermoids and/or lipodermoids, pretragal skin tags and blinded fistulas and vertebral anomalies (1-9).

Epibulbar dermoids which are the most critical and typical features of this syndrome occur frequently at the lower temporal quadrant of the globe (1,3,6,7,9). Upper lid colobomas are also common in these patients (1,6).

Single or multiple skin tags may be seen on the preauricular or cheek region. They may be found unilaterally or bilaterally (1,3,6,7).

Vertebral anomalies are seen in 75-85 percent of the cases as vertebral fusion, scoliosis, hemivertebrae and spina bifida (1,3,6,7).

Asymmetry of the skull, mild malar hypoplasia (in about 60 percent of the cases), receding chin, aplasia of the ramus and condyle of the mandible, hypertelorism, blepharoptosis, narrow palpebral fissures, lacrimal drainage system anomalies, cleft lippalate, high-arched palate, bifid uvula, bifid tongue, double lingual frenulum, digital anomalies (clinodactyly, hypoplasia, webbing, polydactyly), urogenital anomalies and cardio-vascular system anomalies have been reported as associated anomalies with Goldenhar's syndrome (1-3,6,7,9). Facial nerve weakness is another associated finding with this syndrome (2,5).

The incidence of the syndrome was estimated one in 3500-5600 live births (6).

CASE REPORT

A 27-year-old female patient was admitted with the complaint of a furrow on the left cheek, which was present since birth.

There was no family history of congenital lesions.

During physical examination, an asymmetry was found on the face due to hemifacial hypoplasia. The patient had macrostomia and there was a furrow on the cheek extending from oral commissure to the ear on the left cheek. Hypoplasia of soft tissue (skin, subcutaneous tissue and muscle) and bone (mandible) was found around this furrow. She had no tragus on the left ear. A skin tag settled down the location of absent tragus. Another skin tag on the cheek had been excised before (Fig.1a, 1b). There was also facial nerve weakness on the affected half of the face (Fig 1c). Scoliosis was found during physical examination.

During x-ray examination, hypoplasia of body and ramus of mandible on the left side and scoliosis and spina bifida occulta in the vertebral column was found (Fig 2a, 2b,3).

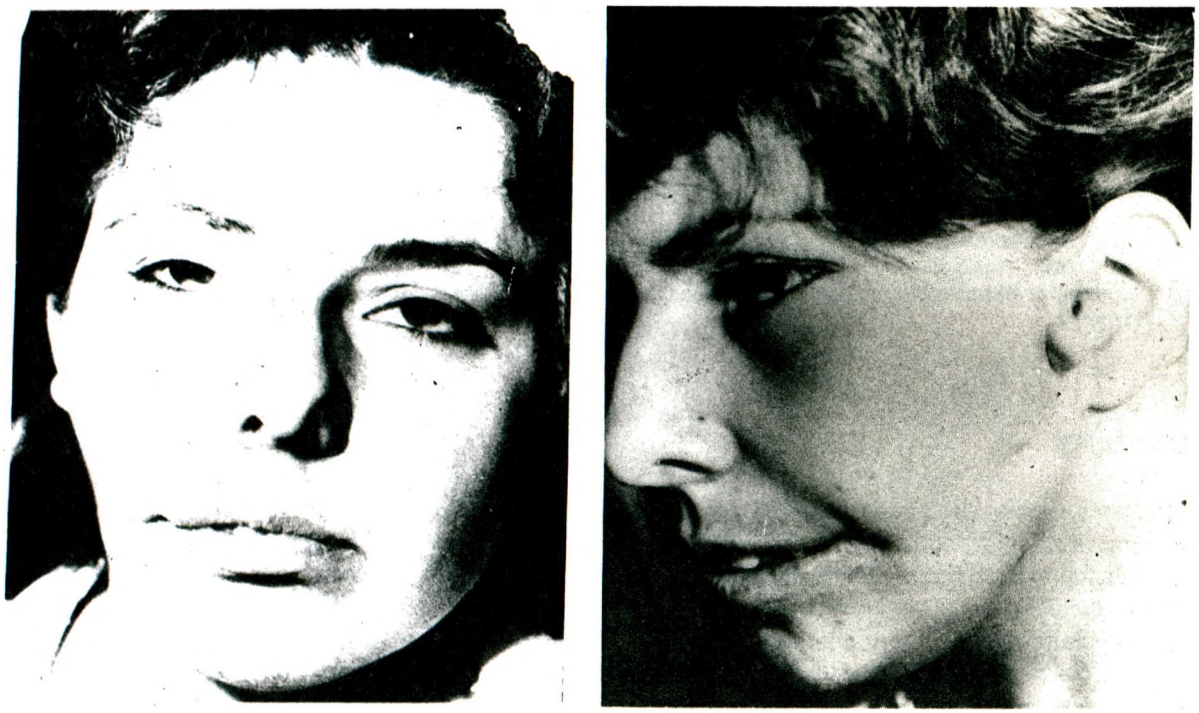

Fig 1. (a) Front view and b)lateral view of the face of the patient.

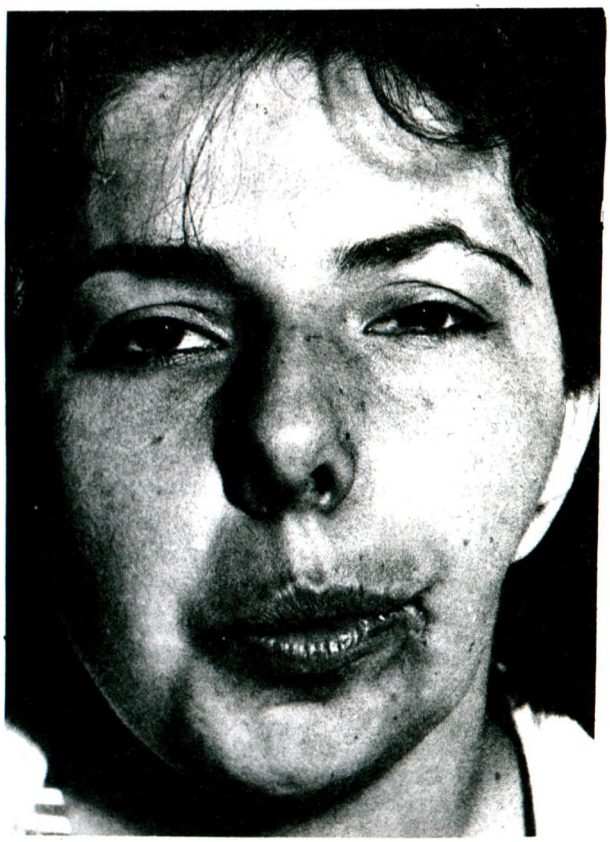

Fig 1. c) Weakness of facial nerve on the affected side.

DISCUSSION

Craniofacial microsomia is a complex malformation usually associated with abnormalities of other systems. Because of heterogenous nature of craniofacial microsomia, it is difficult to classify the individual deformity. Some authors separate the cases according to associated malformations.

In 1952, Goldenhar considered the cases of craniofacial microsomia who had epibulbar dermoids-lipodermoids, vertebral anomalies to constitude another syndrome. On the other hand, this entity was included in the classification of unilateral craniofacial microsomia as "Goldenhar type" (=Type IV) by some authors (7). So, it is very difficult to differentiate Goldenhar's syndrome from craniofacial microsomia. However, the deformity in craniofacial microsomia has three main features: Auricular, maxillary and mandibular hypoplasia. It can widely involve external ear (various ear deformities or anotia), mastoid process, middle ear, temporal bone and base of the skull (1,7,9). Craniofacial microsomia does not include ocular abnormalities which are seen in

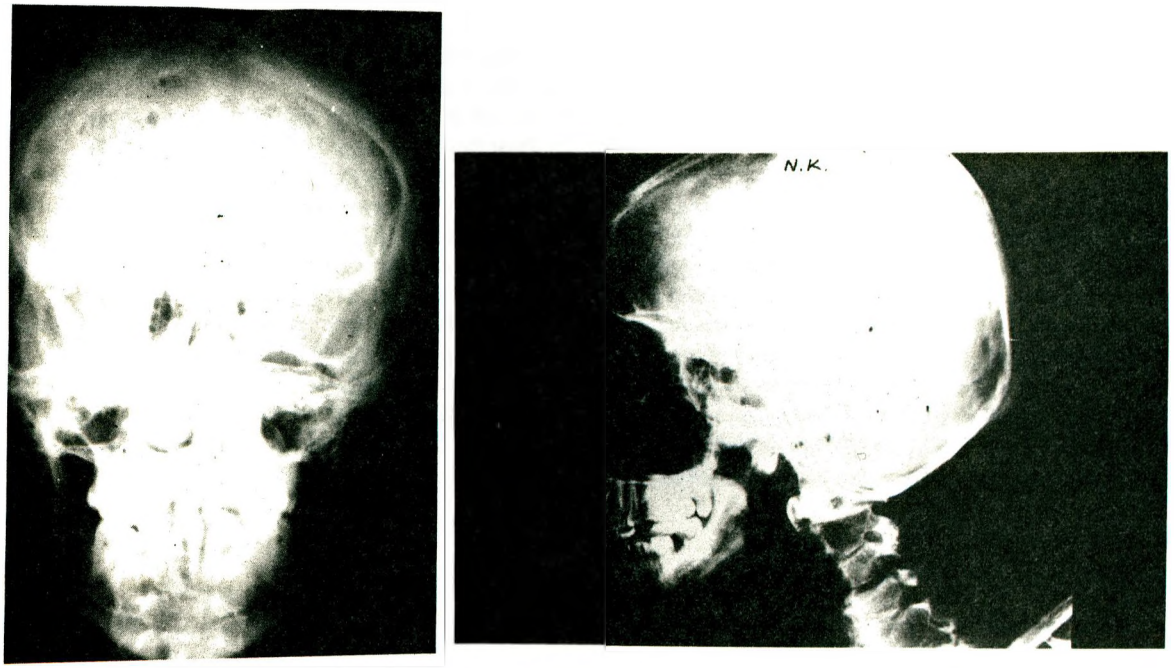

Fig. 2 (a) X-rays of anterior and b) left lateral of the head (hypoplasia of body and ramus mandible)

Goldenhar's syndrome (7,9).

Goldenhar's syndrome is closely related with no.7 rare craniofacial cleft (8). Tessier had admitted that no.8 cleft belongs to Treacher-Collins and Goldenhar's syndrome. He had also reported that soft tissue malformations are more severe than bony defects in Goldenhar's syndrome and dermatocele and true cleft of the lateral canthus are parts of this syndrome (4).

In our patient the main event is no. 7 cleft with macrostomia, a furrow on the cheek and skin tags on the soft tissue and hypoplasia of mandible on the bone framework. The other anomalies associate this cleft. Thus, these three main lesions form Goldenhar's syndrome and she clearly shows a distinct entity from craniofacial microsomia.

In conclusion, we agree with the authors who believe that no. 7 cleft belongs to Goldenhar's syndrome and it is considered as a distinct entity (1,3,6,8,9)

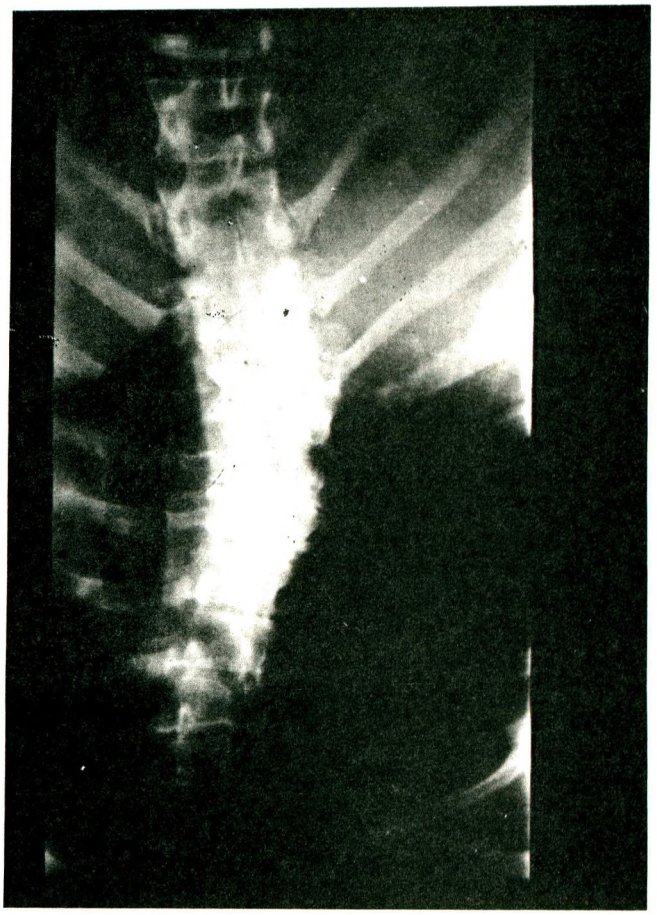

Fig. 3) X-ray of the vertebral column (scoliosis and spina bifida)

REFERENCES

1. *Converse JM. Reconstructive Plastic Surgery. 2nd ed. Philadelphia: W B Saunders, 1977: 2146, 2360, 2408.*
2. *Tessier P, Tulasne JF. Treacher-Collins Syndrome. In: Marchac D, ed. Craniofacial Surgery. Berlin: Springer-Verlag, 1987:372.*
3. *Goldenhar M. Associations malformatives de l'oiel et de l'oreille en particulier le syndrome dermoide epibulbare-appendices auriculares-fistula auris congenita et ses relations avec dysostose mandibulofaciale. J Genet Hum 1952;1:243.*
4. *Tessier P. Anatomical classification of facial, cranio-facial and latero-facial clefts. J Maxillofac Surg 1976; 4: 69-92.*
5. *Powell W J, Jenkins H P. Transverse facial clefts. Plast Reconstr Surg 1968; 42(5): 454-459.*
6. *Mansour A, et al. Ocular findings in the facioauriculovertebral sequence. Am J Opthalmol 1985; 100:555-559.*
7. *Mc Carthy JG. Plastic Surgery. Philadelphia: WB Saunders Co, 1990:2491, 3054.*
8. *Serafin D, Georgiade NG. Pediatric Plastic Surgery. C.V. Mosby Co 1984: 390-403.*
9. *Feingold M, Baum J. Goldenhar's Syndrome. Am J Dis Child 1978; 132:136-138.*