*The Turkish Journal of Pediatrics*
1974 / Vol. 16 / No. 3 / pp. 124 - 129

# Congenital Cardiac Anomalies in Goldenhar's Syndrome*

## (Oculo-auriculo-vertebral dysplasia)

**Muhsin Saraclar, M.D.** / Sidney Friedman, M.D.*****

The basic features of Goldenhar's syndrome (oculo-auriculo-vertebral dysplasia) are: 1) epibulbar dermoids and/or lipodermoids 2) auricular appendices and pretragal blind-ended fistulae and 3) vertebral anomalies. The initial case was described in 1845 by von Arlt;[1] in 1952, Goldenhar[2] added 3 new cases to 16 previously recorded in the medical literature. Gorlin et al[3] in 1963 suggested use of a more descriptive title, oculo-auriculo-vertebral dysplasia. In the past decade, the number of reported cases of Goldenhar's syndrome has increased to a total of approximately 150.

The purpose of this communictation is to present 7 new cases of Goldenhar's syndrome, including 6 with congenital heart disease, and to draw attention to the high frequency of congenital heart disease in this syndrome.

### *Clinical Material*

Seven children with Goldenhar's syndrome are currently under observation in the various pediatric sub-specialty clinics at The Children's Hospital of Philadelphia. Six of these patients are enrolled in the Cardiac Clinic because of associated congenital heart disease. All have the charac-

* From the Division of Cardiology, The Children's Hospital of Philadelphia; and the Department of Pediatrics, School of Medicine, University of Pennsylvania, Philadelphia, Pennsylvania, U.S.A.

** Professor of Pediatrics and Pediatric Cardiologist, School of Medicine, Hacettepe University, Ankara, Turkey; and Instructor in Pediatrics, School of Medicine, University of Pennsylvania.

*** Chief Cardiologist, The Children's Hospital of Philadelphia; Professor of Pediatrics, School of Medicine, University of Pennsylvania.

teristic physical examination and x-ray findings of Goldenhar's syndrome (Figure 1). In addition to the usual ocular, auricular and vertebral anomalies, one child has a renal anomaly, and syndactyly was present in another.

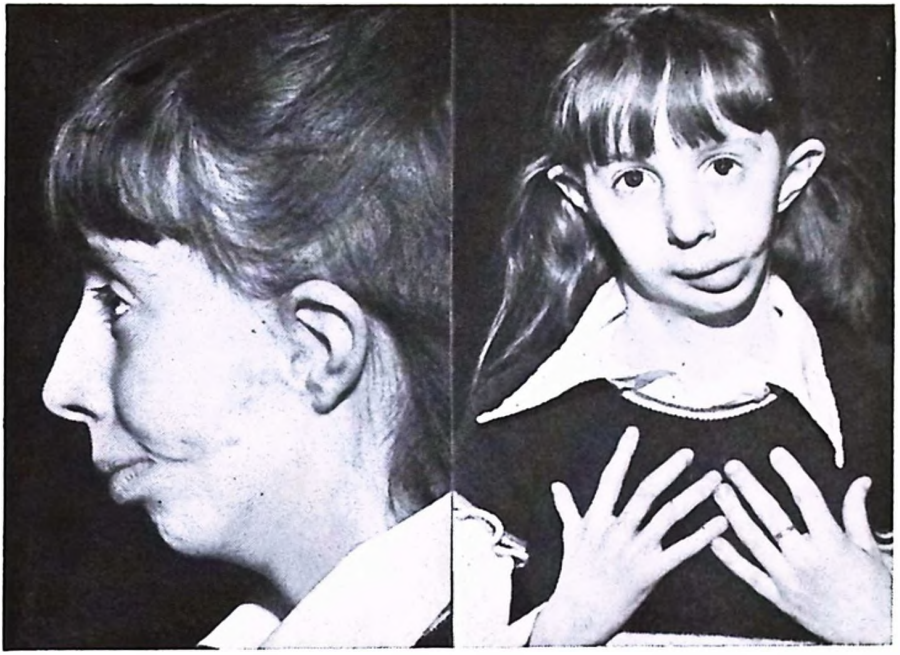

**Figure 1**

Two photographs in lateral and anteroposterior views of a 10 year old female with Goldenhar's syndrome. In the frontal view an epidermoid tumor is seen in the lower outer quadrant of the left eye. Note also the facial asymmetry, atresia of left external auditory meatus, deformity of right hand.

In each of the 6 cases with congenital heart disease, the physical examination, ECG, and x-ray findings were entirely appropriate to the cardiac lesion present; cardiac catheterization and angiocardiography were performed in 3 cases (Table I). The cardiac lesion, most commonly encountered, was ventricular septal defect, found in four of the six patients. In two instances the ventricular septal defect occurred as an isolated lesion; in two others there were associated lesions. Severe pulmonary hypertension was present in the two patients with combined lesions. Spontaneous closure of a ventricular septal defect occurred in one patient. The fifth patient had coarctation of a right aortic arch with a retroesophageal left subclavian artery. The sixth patient had mild pulmonary stenosis. Congestive heart failure was present in the clinical course of three patients; in one instance this was successfully treated by division of a patent ductus arteriosus.

## TABLE I

SUMMARY OF CLINICAL FINDINGS IN 7 PATIENTS WITH GOLDENHAR'S SYNDROME

| Case No. | Age (years) Sex | Non-cardiac anomalies | Cardiac anomalies | EKG | Chest x-ray | CHF | Method of cardiac diagnosis | Cardiac surgery |
|---|---|---|---|---|---|---|---|---|
| 1 | 10 F | Ocular dermoid, preauricular tags and fistulae, deformity of external ear, facial asymmetry, syndactyly. Vertebral anomalies | VSD ASD TAPVD PAH | RAD RVH | CE, prominent PA seg. Increascreased pulm. vasc. | + | Cardiac cath and surgery | Total correction |
| 2 | 7 F | Preauricular tags and fistulae, deformity of external ear, vertebral anomalies | VSD PDA PAH | RAD RVH | CE, prominent PA seg. Increased pulm. vasc. | + | Cardiac cath and surgery | PDA division (VSD closed spontaneously) |
| 3 | 2 M | Ocular dermoid, preauricular tags and fistulae, facial asymmetry. | VSD | WNL | CE, increased pulm. vasc. | + | Cardiac cath | |
| 4 | 6 F | Microphtalmia, preauricular tags and fistulae, deformity of external ear, vertebral anomalies. | VSD | WNL | CE, increased pulm. vasc. | | Clinical | |
| 5 | 3 M | Cleft of upper eyelids, deformity of external ear, facial asymmetry, cleft palate, vertebral anomalies, absent left kidney. | Coarct. of aorta | LVH | Right aortic arch | | Clinical | |
| 6 | 1 M | Coloboma of iris and chorioid, preauricular tags, anomaly of rib. | PS | WNL | WNL | | Clinical | |
| 7 | 6 M | Ocular dermoid, preauricular tags, cleft palate, vertebral anomalies. | | WNL | WNL | | Clinicial | |

VSD: Yentricular septal defect. ASD: atrial septal defect. TAPVD: total anomalous pulmonary venous drainage. PAH: pulmonary artery hypertension. PDA: patent ductus arteriosus. Coarct.: coarctation. PS: pulmonary stenosis. RAD: right axis deviation. RVH: right ventricular hypertrophy. WNL: within normal limits CE: cardiac enlargement. PA seg.: pulmonary artery segment. Pulm. vasc.: pulmonary vascularity. Cath: catheterization. CHF: congestive heart failure.

*Discussion*

The most common ocular findings in Goldenhar's syndrome are epibulbar dermoids and/or lipodermoids. These circular lesions are milky white or yellow in color. The dermoid is usually located at the limbus or corneal margin in the lower outer quadrant, the lipodermoid in the upper outer quadrant. Coloboma of the superior lid is also a common ocular finding; others are ptosis, coloboma of the iris or chorioid, microphtalmia, microcornea, anophtalmia, and strabismus. One or more ocular manifestation was present in 6 of our 7 patients with Goldenhar's syndrome.

The characteristic auricular manifestations of this syndrome are hypoplasia and deformity of the pinna of the ears, atresia or stenosis of external auditory meatus, preauricular appendices and blind-ended fistulas, microtia, and deafness. Auricular anomalies were present in all of our 7 cases.

Oral anomalies are present in more than 50 % of patients with Goldenhar's syndrome. These include maxillary hypoplasia, micrognathia, high-arched palate, macrostomia, bifid uvula, cleft palate and lip, dental malformation and malocclusion.

Vertebral anomalies in Goldenhar's syndrome were first described by Gorlin et al[3] in 1963. Although these are common, they are usually of little clinical significance. The common vertebral anomalies are occipitalization of the atlas, cuneiform vertebrae, fused vertebrae, supernumarry vertebrae, spina bifida, hemivertebrae, and lumbarization of the first sacral vertebrae. Other skeletal anomalies described are anomalous ribs, and persistently patent anterior fontanel. Six of our seven cases have one or more of the bony anomalies.

A wide variety of visceral anomalies may be encountered in patients with Goldenhar's syndrome. Several recent reports have emphasized the high frequency of congential heart disease in this syndrome. Friedman and Saraclar[4] reviewed 84 cases of Goldenhar's syndrome, the majority found in journals of special interest to surgical subspecialists, and found only 32 instonces in which information was provided concerning the cardiac status of the patient. In 17 (53 %) of these 32, cases, a statement was made indicating the absence of heart disease. In the other 15 cases (47 %) congenital heart disease was recognized. The distribution of the cardiac anomalies in this group was as follows: 3 with ventricular septal defect, 3 with tetralogy of (Fallot, 2 of patent ductus arteriosus, 2 with atrial septal defect, 1 coarctation of the aorta, 1 L-transposition of the great

arteries with dextrocardia 2 with; 1 right bundle branch block, and in two cases a specific diagnosis was not stated). Subsequent to this review the following additional instances of heart disease in Goldenhar's syndrome have been reported: 2 cases by Labrune et al,[5] one with ventricular septal defect; 13 cases by Greenwood et al,[6] 6 with tetralogy of Fallot, one with ventricular septal defect; and 2 cases by Comas and Gonzales,[7] one with patent ductus arteriosus and aortic stenosis.

Of the total of 56 reported cases of Goldenhar's syndrome with a definitive cardiac description, 30 (54 %) had congenital heart disease. The cardiac anomalies encountered conformed to the conventional distribution of anatomic lesions in the general population for the age group observed. Ventricular septal defect and tetralogy of Fallot were the most common lesions. Patent ductus arteriosus, coarctation of the aorta, and atrial septal defect ranked second. Several children had multiple anomalies. The severity of the lesions varied over a wide range.

Visceral anomalies, other than congenital heart disease, reported to occur in Goldenhar's syndrome are inguinal hernia, anal atresia, rectal prolapse, renal anomalies, and hypoplasia of the lungs. Mental retardation has been observed in 10 % of the cases.

Cosmetic improvement in the various facial deformities of Goldenhar's syndrome can be achieved by surgical means. Ocular dermoids may be removed; hearing may be improved by amplification techniques. Vertebral anomalies usually require no treatment. Surgical treatment of heart defects in patients with Goldenhar's syndrome provides no unusual problems, nor do the oculo-auriculo-vertebral anomalies complicate the cardiac care. A variety of elective plastic surgical procedures were performed in our patients; 2 have tolerated cardiac surgical procedures without complications.

*Summary*

Seven new cases of Goldenhar's syndrome (oculo-auriculo-vertebral dysplasia) including 6 with congenital heart disease have been presented and the literature reviewed. Of 56 patients adequately described, 30 (54 %) had congenital heart disease. The cardiac anomalies encountered conformed to the conventional distribution of anatomic lesions in the general population for the age group observed.

*REFERENCES*

1. Von Arlt, F.: Klinische Darstellung der Krankheiten des Auges, vol. 3 Wien, 1845, p. 376, cited by Gorlin et al.[3]

2. Goldenhar, M.: Associations malformations de l'oeil et de l'oreille en particuler le syndrome dermoide epibubaire-appendices auriculaires-fistula auris congenita et ses relations avec la dysostose mandibulo-faciale, J. Genet. Hum. 1: 243, 1952.
3. Gorlin, R. J., Jue, K. L., Jacobsen, U., and Goldschmidt, E.: Oculoauriculovertebral dysplasia. J. Pediat. 63: 991, 1963.
4. Friedman, S., and Saraclar, M.: The high frequency of congenital heart disease in oculo-auriculo-vertebral dysplasia (Goldenhar's syndrome), J. Pediat. 85: 873, 1974.
5. Labrune, B., and Choulot, J. J.: syndrome oculo-auriculo-veretbral. Syndrome de Goldenhar. Ann. Pediat. 21: 713, 1974.
6. Greenwood, R. D., Rosenthal, A., Sommer, A. M., Wolff, G., and Craeenn, J.: Cardiovascular malformations in oculoauriculo-vertebral dysplasia (Goldenhar syndrome) J. Pediat. 85: 816, 1974.
7. Comas, A. P. and Gonzales, A. L.: Congenital heart disease in Goldenhar syndrome. J. Pediat. 86: 989, 1975.