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Case Report

GOLDENHAR SYNDROME

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ABSTRACT

Goldenhar Syndrome also called as oculo-auriculo-vertebral dysplasia,is a rare syndrome developing from first and second pharyngeal arches during Blastogenesis.This condition was documented in 1952 by Maurice Goldenharr.The syndrome is charecterised by multiple anomalies of the ocular,auricular,cardiac,skeletal and nervous system.Pericentric inversion of chromosome 9 is one of the most common structural balanced chromosomal aberration seen in this syndrome.Multi disciplinary approach should be undertaken by several departments in evaluating such patients.

KEY WORDS: Epibulbar dermoid, Micrognathia,Preauricular tags.

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INTRODUCTION

Goldenhar syndrome (GHS) also called as Oculoauriculo vertebral dysplasia or hemi facial microsomia, was first described by Von Arlt in 1845. It was defined precisely by Goldenhar in 1952 as a syndrome characterized by anomalies like dermal & epibulbar cysts, auricular appendices & malformation of the ears. In 1963, Gorlin & Pinberg identified the vertebral anomalies associated with Goldenhar syndrome & named the syndrome as "Occuloauriculo vertebral" syndrome [1]. The incidence of this syndrome is about 1 in 5600 live births with a male preponderance of 3:2. Most of these cases are sporadic but an autosomal dominant pattern is observed. The recurrence risk is about 2 – 3 %. There is a lack of genetic linkage & the sporadic occurrence suggests a multifactorial etiology that includes nutritional & environmental factors that result in disturbance of blastogenesis [2].

Goldenhar syndrome has a wide range of manifestations including craniofacial, vertebral, cardiac, renal and central nervous system anomalies. The classic facial aspect of Goldenhar syndrome is described as hemi facial microsomia. The typical presentation of Goldenhar syndrome includes epibulbar dermoids, microtia, mandibular hypoplasia, strabismus, most probably caused by developmental defects of first & second branchial arches and vertebral anomalies [3]. Herein we describe a case of Golden Harr Syndrome reffered to our cytogenetic lab.

CASE REPORT

A boy aged about 8 years born to a nonconsanguineous couple presented to the Oph thalmology department of Sri Siddhartha Medical college and Hospital, Tumkur, India, with the

complaints of blurring of vision since one year and also had a swelling in his left eye. His mother's age at conception was 27 years. She was in good health with no history of diabetes or hypertension. She did not give any history of drug intake during pregnancy or any history of exposure to teratogens. His father's age at conception was 34 years. He was a non smoker and was in good health. The patient was born through full term normal vaginal delivery in a local hospital. His birthweight was 2.7 kgs & postnatal period was uneventful. He has an younger female sibling aged about 6 years with no obvious clinical illness. There was no h/o repeated infections or any hearing defect.Patient was reffered for genetic testing and counseling.

On examination in the Cytogenetic lab of Department of Anatomy, SSMC it was noticed that the patient had obvious facial asymmetry,microstomia as shown in Fig 1,auricular appendage of 1cm in front of left ear as shown in Fig 2. Facial asymmetry on the left side due to mandibular hypoplasia was seen.No palatal anomalies seen. Ophthalmic examina-tion revealed epibulbar dermoid in infratemporal quadrant on left side as in Fig 3 & 4. No coloboma or strabismus seen.Fundus was normal.

No vertebral or neurological anomalies seen.Echocardiogram revealed no cardiac abnormalities.

Chromosomal analysis showed a normal male Karyotype ie, 46, XY.

Fig. 1: Facial Assymetry.

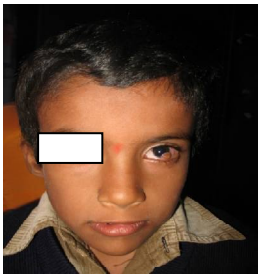


Fig. 2: Preauricular tag

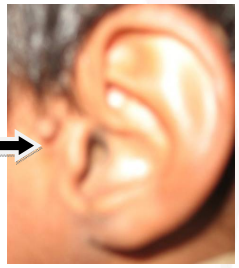


Fig. 3: Epibulbar Dermoid.

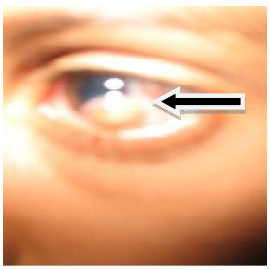
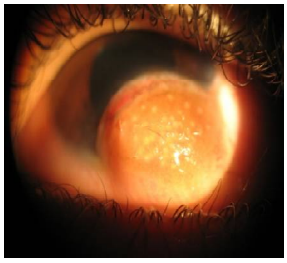


Fig. 4: Epibulbar Dermoid.



DISCUSSION

Goldenhar syndrome (also known as Oculo-Auriculo-Vertebral Syndrome (OAV syndrome) is a rare disorder charecterised by incomplete development of the ear, nose, soft palate, lip, and mandible. It is associated with anomalous development of the first and second branchial arches during blastogenesis. Classical triad of syndrome includes Hemi facial microsomia, Epibulbar dermoid and Deformity of ears with pre auricular appendage [1,2]. Tsai and Tsai reported this syndrome in three consecutive generations in a family [1]. In our case all the features mentioned above were seen.

Ocular manifestations include epibulbar dermoid in 75% of cases,bilateral at infratemporal quadrant [2]. There are choriostomas (nest of normal tissues in abnormal places). Dacryocystitis has been reported in some cases. Other ocular manifestations are coloboma, microopthalmos, cataract,iris anomalies, anopthalmos, optic nerve hypoplasia and squint [1]. In our case left unilateral epibulbar Dermoid in the infra temporal quadrant was seen.

Auricular manifestations are preauricular skin tags,accessory auricle [1]. Anotia is rare and has been reported by Jaison and Batra.(2) Left Pre auricular tag was seen in our case.

Involvement of vertebra and ribs are observed in 24%of cases [3]. Spina bifida is the least severe of all vertebral anomalies. Hemivertebral loss of vertebral arch and fusion of posterior elements of cervical vertebrae have been reported [1]. Association of post axial polydactyly with GHS is described [4].

Central nervous system manifestations are seen in 46% of cases.Diffuse cerebral hypoplasia, Dilated lateral cerebral ventricles (asymptomatic Hydrocephalus), Carpus callosum dysgenesis and frontal hypodensities were the most frequent abnormalities.Arnold chiari malformation and facial nerve palsy have also been reported [5].

Cardiac defects include Ventricular septal defects,Patent ductus arteriosus,Fallot's tetrology,Coarctation of Aorta and pulmonary stenosis [6]. The reported prevelence of cardiovascular anomalies is 5-58%. The other cardiac anomalies reported are Ventricular

inversion associated with double outlet Right ventricle, Pulmonary atresia with VSD, Double outlet right ventricle and infra diaphragmatic total anomalous pulmonary venous connections [7]. In our case no cardiac anomaly was reported.

Uro genital anomalies include Renal agenesis, hydronephrosis,multicystic dysplastic kidney, ectopic kidney and malformed or absent uterus [6,7].

Facio dental anomalies include hypoplasia of malar bones, zygomatic arch and mandible, Macrostomia, Micrognathia, high vaulted cleft palate, bifid tongue and malocclusion [1]. GHS has also been associated with Cleft lip, cleft palate and also Temporo mandibular joint malformation with malocclusion [8].

Oculo auriculo vertebral dysplasia represent the mildest form of the disorder, GHS presents as most severe form and hemifacial microstomia appears to be intermediate form. 10-30% have bilateral manifestations [9]. Family history suggest Autosomal dominant or Reccessive inheritance.Some researchers suggest that the disorder may be caused by the interaction of genes in combination with environmental factors. It has been suggested that there is a defect in branchial arch development late in the first trimester [10].

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. Maternal diabetes has also been suggested as an etiologic factor [11].

The phenotypic findings of this syndrome is variable due to heterogenous etiology. Preauricular skin tags seen in 90% of cases, Microtia in 52%,Hemifacial microsomia in 77%, Epibulbar Dermoid in 39% of cases. Vertebral anomalies were noted in 7% of cases. Cardiac manifestation are found in 39% of cases ,while Genitourinary anomaly was noted in 23% and various central nervous system anomalies are seen in 47% of cases [11].

The diagnosis of Goldenhar's syndrome should be mainly based on the clinical aspect and associated with both systemic conditions and radiologic findings. 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 imaging techniques available today. Radiographic examination of zygomatic bones shows a macroscopic deficiency and developmental asymmetry.

Pericentric inversion of chromosome 9 is one of the most common structural balanced chromosomal abberation seen with incidence of 15% to 25% [3,11].

Multidisciplinary approach should be undertaken by departments Paediatrics, Cardiology, Audiology, Ophthalmology and Plastic surgery in evaluating the patient. Chromosome analysis should be performed in every patient of GHS [11,12]. GHS is also noted in families who had Hydrocephalus, Meningomyelocoele and neural tube defects [5].

The outlook for children with Goldenhar syndrome varies, but is generally very positive. Most children can expect to live a healthy life once treatments have been administered. The majority can expect to have a normal lifespan and a normal level of intelligence.

Conflicts of Interests: **None**

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