CASE REPORT

Goldenhar syndrome

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ABSTRACT

Goldenhar Syndrome or Oculoauriculovertebral Syndrome is a complex syndrome characterized by an association of maxillomandibular hypoplasia, deformity of the ear, ocular dermoid and vertebral anomalies and the most severe form of hemifacial microsomia. Here we describe a 12 years female patient with Goldenhar Syndrome came to Ophthalmology department at Anwer Khan Modern Medical College & Hospital.

Key Words: Goldenhar Syndrome, Oculoauriculovertebral Syndrome, Oculoauriculovertebral Dysplasia, Hemifacial Microsomia, Limbal Dermoid.

Introduction

Goldenhar syndrome is a congenital defect characterized by asymmetrical malformations classically involving face, eyes and ears. Goldenhar syndrome was first observed and recorded by Carl Ferdinand von Arlt.[1,2] Maurice Goldenhar was the first to describe the syndrome in detail and thus the condition was called Goldenhar Syndrome.[3] The vertebral anomalies were included by Gorlin Et Al. in 1963 and then the name Oculo-auriculo-vertebral (OAV) dysplasia was suggested.[4,5] It is also associated with anomalies of CNS, cardiac and renal anomalies. The precise incidence of Goldenhar syndrome is unknown but estimated from 1 in 35,000 to 1 in 56,000 live births.[6] The male-female ratio is 3:2. Sporadic in nature occurring randomly with no apparent cause. Positive family history have been described that have suggested autosomal dominant in nature but rarely it may present as autosomal recessive and multifactorial inheritance.[6,7] Drugs like thalidomide, tamoxifen, retinoic acid and cocaine by pregnant mothers may be implicated. Heavy alcohol consumption during pregnancy and maternal infection and maternal diabetes have also been suggested as etiological factors.[8,9] The diagnosis of this syndrome is mainly based on clinical aspects. Most consider presence of ear anomalies are essential for diagnosis.

CASE REPORT

A 12 years old female came to Ophthalmology department of Anwer Khan Modern Medical College & Hospital with the complaints of Swelling over the left eye and Swelling in front of the left ear since birth. Decreased vision & hearing impairment since childhood. Initially the swelling was small and yellow-grayish in color but gradually increased in size for last 12 years. She comes from a lower socio-economic family and all her family members are in good health. According to mothers statement her maternal period was uneventful. No history of taking teratogenic drugs, pre-eclampsia, GDM, maternal infection, preterm labour or preterm birth. There is no history of neonatal infection and low birth weight of the baby.

On general examination anaemia, jaundice, cyanosis, oedema, dehydration, clubbing, koilonychia, leukonychia all are absent. Her pulse: 84 b/m, BP: 100/60 mmHg and temperature: 98^{o}F. On nervous

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system examination she was well oriented and no abnormalities were found. On Ocular examination following findings were found:

Table-1: Ocular examination findings.

	Right eye	**Left eye**
Characteristic features of swelling	No abnormality	A painless dome shaped swelling, soft in consistency, size 1/1.5cm with hair follicles over the smooth surface, immobile and adherent to the anterior & lower temporal surface of left eye touching the eyelid, conjunctiva and cornea which resembles a limbal dermoid [Fig-1].
Visual acuity	6/6	3ft finger count
Nystagmus	Absent	Absent
Ocular motility	Full in all cardinal gaze	Lateral gaze movement is restricted
Eyelid	Normal	Swelling adherent with the lower eyelid margin and at lateral canthus.
Conjunctiva	Normal	Adherent with the swelling
Cornea	Normal	Yellow-grayish swelling with hair is adherent with the temporal limbus & infero-temporal quadrant of the cornea.
Pupil	Round, regular and reacting to light	Round, regular and reacting to light
Fundus	Normal	Normal

Fig-1: Limbal Dermoid on left eye

Fig-2: Prominent hemifacial microsomia on left side of the face

On Musculoskeletal Examination, Hemifacial Microsomia is prominent on left side of the face [Fig-2]. On Otolaryngorhinological examination, preauricular skin tag of the left ear with microtia [Fig-3] and conductive type of hearing deafness evaluated by Waber and Rinne test. On cardiorespiratory, abdominal and renal system examination no abnormality were found.

Fig-3: Preauricular skin tag with microtia of left ear

We have done some investigations and they are CBC, RBS, SGPT, BT, CT, Serum Creatinine, Urine R/M/E, ECG, X-ray chest PA view and Covid 19 RT PCR test. All the test report findings are in normal range except for X-ray chest which shows scoliosis with hemivertebrae [Fig-4]. Though the patient has scoliosis we did not find any visible abnormality of her posture, gait and body movements. She also did not have any complaints of back pain or walking difficulties.

Fig-4: X-ray chest PA view showing scoliosis with hemivertebrae

Depending upon patients history, clinical examination and investigation findings our diagnosis is Goldenhar Syndrome. Diagnosis is made due to the presence of classical presentation like hemifacial microsomia, limbal dermoid, preauricular skin tag and scoliosis with hemivertebrae. After diagnosis from the Ophthalmology department we have provided surgical treatment as Excision of limbal dermoid & Biopsy for histopathological examination. On histopathological examination lipodermoid was found as the sample contains fibrous tissue, hair follicles and fat cells. Depending upon histopathological report our final diagnosis is Goldenhar Syndrome.

After 1 month patient came to us for follow up and she was doing well. Her cosmetic appearance has improved. There is no swelling over her left eye but scar mark can be seen on infero-temporal part of left side of cornea [Fig-6]. Her vision has improved to 5ft finger count and with spectacle correction it has improved to 6/60 on left eye. Her ocular motility in left eye is full in all cardinal gazes [Fig-6].

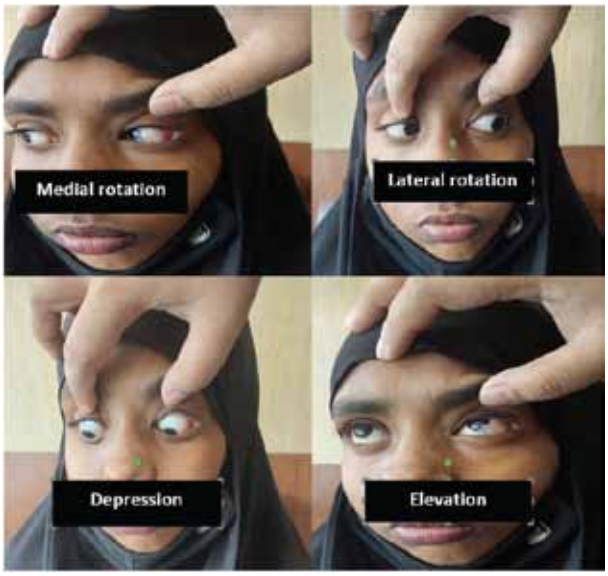

Fig-6: Ocular motility full in all cardinal gazes on left eye

DISCUSSION

The classic features of Goldenhar syndrome include ocular anomalies like epibulbar or limbaldermoids, microphthalmia and coloboma of eye lid and optic disc, ENT features such as preauricular skin tag, hearing loss, low implantation of the auricular pavilion, microtia, micrognathia, facial asymmetry like hemifacial microsomia where hypoplasia of the mandible is most common and vertebral anomalies such as scoliosis or hemivertebrae. The abnormalities are unilateral in most of the cases. Incidence of association with other body systems are cardiac (tetralogy of Fallot, ventricular septal defects and transposition of the great vessels), renal (ectopic or fused kidneys, renal agenesis, vesicoureteral reflux, ureteropelvic junction obstruction, ureteral duplication and polycystic kidney and CNS (microcephaly, encephalocele, hydrocephaly and hypoplasia of the corpus callosum).[10,11,12,13]

Goldenhar syndrome is also called 1st and 2nd Branchial Arch Syndrome (BAS) as it involves the first and second branchial arches 1st and 2nd branchial arch abnormalities manifests as combined

tissue deficiencies and hypoplasias of the face, external ear, middle ear and maxillary and mandibular arches.[14] They represent the second most common craniofacial malformation after cleft lip and palate. Hemifacial microsomia arise from branchial arch defect where preauricular skin tags are ramnents of branchial cleft.[14]

The etiopathogenesis of this Goldenhar syndrome is multifactorial, not yet fully established and involves genetic and environmental factors that cause disturbances in neural crest division, abnormal development of the first and second branchial arches during embryogenesis as well as occlusion of placental vessels.1[1,15,16]

In terms of genetic association 5p deletions, 14q23.1 duplications, or abnormalities of chromosomes 18 and 22 were observed. Families with autosomal dominant inheritance (1-2%) have shown segregation of chromosome 14q23.1 duplication inclusive of the OTX2 gene.[11,16,17]

Drugs like thalidomide, tamoxifen, retinoic acid, vasoactive (pseudoephedrine, aspirin, ibuprofen)and cocaine by pregnant mothers may be implicated. Heavy alcohol consumption during pregnancy, 5 maternal infection, maternal second trimester bleeding, gestational diabetes mellitus, multiple gestation and maternal use of assisted reproductive technology are the most common external factors involved in the occurrence of Goldenhar syndrome.[11,12]

The diagnosis of Goldenhar syndrome is clinical, however, there are some diagnostic tests that can be helpful. Prenatal ultrasonic diagnosis may theoretically be done at the 11th–15th week in utero.[18] Newer imaging modalities such as three-dimensional ultrasound may help in detection of microtia, preauricular skin tags, and asymmetry of the mandible even in mild forms. Most cases of Goldenhar syndrome occur de novo, but an autosomal dominant or recessive inheritance has also been noted. A three-generation family history has to be determined looking for cases as mild as ear tags or ear pits. Although there is not yet a specific genetic test to detect Goldenhar syndrome, array comparative genomic hybridization should be considered while testing for a possibility of recurrence.

The treatment of the patient depends on the age and systemic condition. Management is usually cosmetic. In our case we have diagnosed Goldenhar syndrome by patients history (maternal, birth and postnatal), clinical examination which shows classical presentations of Goldenhar syndrome and related investigations. Treatment was given and suggested according to patients condition.

Conclusion

Goldenhar syndrome is a rare disease and a congenital one meaning it is present at birth and it causes certain abnormalities especially craniofacial part with systemic associations. A multidisciplinary approach is necessary for the overall well-being of the patient and the treatment protocol should be determined as early in the life as possible to avoid physical difficulty and psychological stigma to the growing child. Pediatric specialists along with ophthalmologists, ENT specialists, orthopedic surgeons, neurosurgeons, orthodontists, cardiologists, urologists and maxillofacial surgeons to decide on the most appropriate treatment plan, which varies with age and systemic associations. Goldenhar Syndrome can affect the routine and social life of the patient. Early detection can help avoid complications at a later stage of life.

The prognosis for this condition is good in patients with no systemic complications.[19] They can live relatively normal lives and have a normal life span. They can get married, have children and enjoy work and recreational pursuits. Management of Goldenhar Syndrome requires long term commitment with treatment from birth to the period of growth and development.

Declaration of patient consent

We have obtained informed written consent from the patient. All the images and other clinical information to be reported in the journal with the patients consent. The patient understand that her name and initials will not be published and due efforts will be made to conceal her identity and gave her consent.

Interest of conflict: None

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