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Case Report
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Atypical Presentation of Goldenhar Syndrome in a Child: A Case Report with Review

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Abstract

Oculoauriculovertebral Spectrum (OAVS) or Goldenhar syndrome is a wide spectrum of congenital anomalies that involves the structures arising from the first and second branchial arches. The etiology of this syndrome is unclear since it varies genetically and is linked to multiple reasons. It is constituted by a wide set of symptoms and signs. Commonly, this syndrome has been mentioned to have eye, ear, facial and vertebral abnormalities. Here we report a child who has facial asymmetry with iris coloboma and ear abnormality.

Keywords: Goldenhar syndrome; Anotia; Facial palsy

Introduction

Goldenhar syndrome has an incidence of 1:35,000 to 1:56,000 with a male to female ratio of 3:2 [1] and is considered as a result of blastogenesis defect that particularly involves the structures originating from the first branchial arches [2]. In 1952 Dr. Maurice Goldenhar first reported these features together as a syndrome [1]. It was described as a triad of epibulbar dermoid, mandibular hypoplasia and pre-auricular skin tags [3]. This syndrome has commonly ocular, auricular and facial defects [3]. This is a child who has facial asymmetry with iris coloboma and an ear abnormality.

Case Presentation

A 5-year-old female child born out of a non-consanguineous marriage was brought with complaints of abnormal development of right ear since birth. She did not have any complaints of decreased hearing. She was born full term, appropriate for gestational age via normal vaginal delivery. No history of maternal illness during pregnancy. Her motor milestones were normal but mild mental retardation was present. On physical examination, the child was conscious and cooperative. On general examination, she was pale, anicteric, no sign of cyanosis, clubbing, lymphadenopathy, and pedal edema. At the time of examination, the patient's body temperature was 37.2°C with a pulse rate of 106 beats/min, with good volume, which was regular and synchronous. Respiratory rate was 26 cycles/min, oxygen saturation was 98% and blood pressure was 102/64 mmHg. Patient was of short stature with a height of 102 cm, which appears to be less than -3(Z) SD score compared to same age group individuals.

She had microtia with right sided facial asymmetry with short neck. There was decreased prominence of nasolabial fold with drooping of eyelids on the right side, with her mouth deviated to the left side (Figure 1). Left ear was normal. Right side iris coloboma was present. There was no spine and chest deformity. Neurological examination revealed normal motor and sensory system. On facial nerve examination, there was difficulty in wrinkling of forehead with inability to close the eyelids tightly with difficulty in holding air in mouth on the right side which was suggestive of Infra nuclear facial nerve palsy on the right side. Vestibulocochlear nerve examination was normal. Other cranial nerve examination was normal. There were no cranio-spinal, ribs or limb anomalies. Other systemic examination did not reveal any abnormality. Complete blood count was showed hemoglobin of 12.6 gm %, TLC-6900, platelet 3.8 lakhs. Other cranial nerve examination was normal. Her investigations revealed a normal hemogram, normal blood urea nitrogen and serum creatinine. X-ray skull, spine and chest were normal. 2D echocardiography was suggestive of 65% ejection fraction with no cardiac defect. Thyroid function was within normal limits. Neuroimaging of the brain was normal. The diagnosis of Goldenhar syndrome was established based on the constellation of the classical clinical signs.

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Figure 1: Images showing facial features of the Goldenhar syndrome: Facial asymmetry and un-lateral microtia with right ear.

Discussion

Goldenhar syndrome is characterized by a variable degree of uni-or bilateral involvement of the craniofacial structures involving the first and second branchial arches causing ocular anomalies and a vertebral defect [2]. Etiology of this condition is not yet fully established. Various factors like Chromosomal abnormality, abnormal development of neural crest cells, ingestion of drugs-cocaine, thalidomide, retinoic acid, alcohol during pregnancy lead to the development of this disease [4]. It has a multifactorial inheritance. External ear anomalies and ipsilateral facial underdevelopment is the most important feature of Goldenhar syndrome, which was present in our case [5]. There are no particular diagnostic criteria for Goldenhar. However this syndrome is considered when it includes two or more of the following: Hemifacial microsomia (including micrognathia), epibulbar dermoids and/or coloboma, ear malformations (including microtia and accessory tragi), and vertebral anomalies (fused or cervical hemivertebrae) [6]. Other abnormalities reported in Goldenhar syndrome include ophthalmic malformations, cardiovascular, central nervous system, and genitourinary malformations 6.5% to 15% of the cases are reported to have mental retardation [7]. Unilateral involvement in 85% cases, with involvement of right side to left side in a ratio of 3:2.

Various craniofacial abnormalities like incomplete development of the muscles of the face, macrostomia, cleft palate, cleft lip, abnormalities of the teeth, malar hypoplasia, maxillary and mandibular hypoplasia, and temporal hypoplasia are seen [2]. 33.3% of the patients had facial nerve palsy along with craniofacial abnormalities [8]. There was no evidence of congenital heart disease in our patient though it is seen in 40 to 60% of patients with Goldenhar syndrome [2]. Asymmetric, unilateral renal anomalies seen in 70% of the cases [2]. Ultrasonography did not reveal any anomalies like renal agenesis, ectopic kidney, urethral duplication, and vesicoureteric reflux. No vertebral anomalies (hemivertebrae and cervical vertebrae hypoplasia and cervical fusion) were seen in this case. It can be diagnosed on antenatal USG by looking for any obvious defect. The diagnosis of this syndrome is usually only dependent on the sign, symptoms and the clinical features. A integrative approach is required for the management of this disease. It involves the ophthalmologist, ENT specialist, pediatricians, orthopedician and dermatologist for a successful outcome. All infants suspected to have Goldenhar syndrome should have a diagnostic hearing evaluation- Brainstem Auditory Evoked Response (BERA) within the first six months of life. Plastic surgery is done to correct the structural anomalies of the eyes and ears. Orthodontic treatment could be required for maloccluded teeth [9,10].

Goldenhar syndrome can adversely affect the patient's life even after prompt intervention is done6. Eyelid abnormalities can lead to vision problems, while patients with microtia and atresia may require hearing assistance or surgical reconstruction [11]. Mainly cosmetic treatment is requires in uncomplicated cases. Timely detection and intervention can lead to satisfactory surgical outcomes.

Conclusion

In conclusion, timely detection and intervention can lead to satisfactory surgical outcomes. Our case has some rarely reported features such as facial asymmetry with congenital facial nerve palsy, iris coloboma and ear abnormality and short stature prompting us to report this case.

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