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Case Report

Oculi auricular features of incomplete Goldenhar syndrome

Théra JP[1], Hughes D[2], Tinley C[2], Bamani S[3], Traoré L[3], Traoré J[3]
[1]Pediatric Ophthalmologist and Forensic Medicine Doctor, Faculty of Medicine / Institute of African Tropical Ophthalmology, Bamako (Mali)
[2]Pediatric Ophthalmologist, Consultant CCBRT Hospital, Dar Es Salam (Tanzania)
[3]Professor, Department of Ophthalmology, Faculty of Medicine / Institute of African Tropical Ophthalmology, Bamako (Mali)

***Corresponding author**
Dr. Japhet Pobanou THERA
Email: therajaphet@yahoo.fr

Abstract: Goldenhar syndrome is a rare congenital abnormality including a triad of ocular dermoid cyst, preauricular skin tags and vertebral dysplasia. The current case is an incomplete syndrome for the vertebral features were lacking.
Keywords: Incomplete, Goldenhar syndrome, ocular features.

INTRODUCTION

Goldenhar syndrome is a rare congenital anomaly consisting in a triad of an ocular dermoid cyst, preauricular skin tags and vertebral dysplasia [1]. It is a disorder where the patient's facial features are incompletely developed on one side, resulting in eye, ear, and jaw abnormalities. When these facial abnormalities are associated with vertebral malformations in the spine and more severe involvement of the eyes, this collection of symptoms is called Goldenhar syndrome [2].The reported incidence of Goldenhar syndrome ranges from 1: 3500 to 1: 5600. The etiology of Goldenhar syndrome is unknown. However, it is possible that abnormal embryonic vascular supply disrupted mesodermal migration, or some other factor lead to defective formation of the structures arising from the first and second branchial arches [3, 4]. It is a challenging syndrome which can call for a multidisciplinary management.

CASE REPORT

A 3-year-old male presented to the Department of pediatric ophthalmology with congenital lesions of the ears and the left eye. The child was born after a full-term normal pregnancy from non consanguineous parents. The delivery was vaginal; the prenatal and postnatal history was normal. Ocular examination noticed a left upper eyelid coloboma along with a large bulbar and corneal dermoid cyst. The right eye was normal. External examination of the ears revealed the presence of preauricular appendages (tags). No vertebral dysplasia was found as well as facial asymmetry.

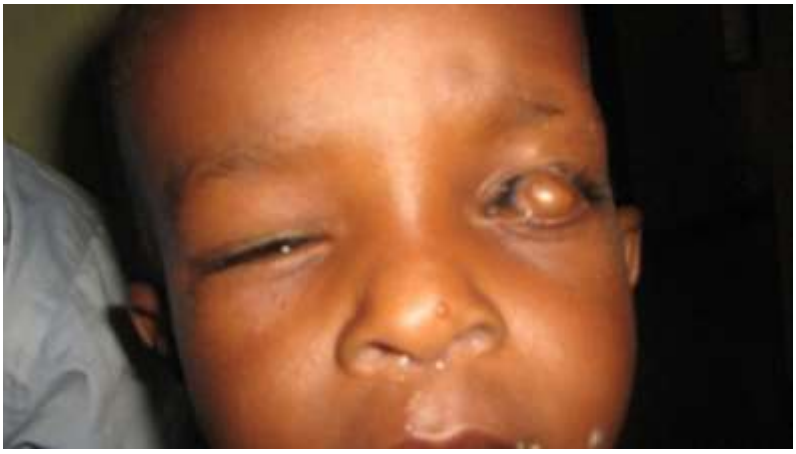

Fig.1: Anterior view of the child showing left upper eyelid coloboma along with a large corneal dermoid cyst

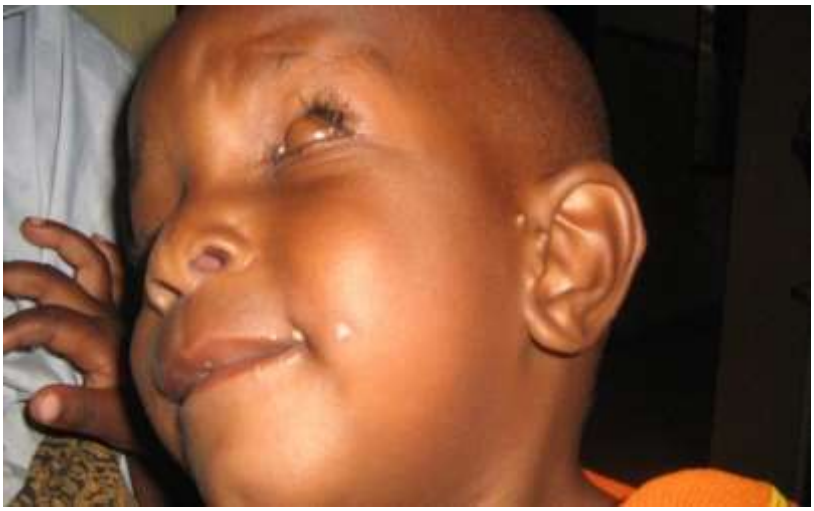

Fig.2: Lateral view of the child showing preauricular tags

DISCUSSION

Goldenhar syndrome, also known as the Oculo-Auriculo-Vertebral syndrome (OAVS), is a rare congenital malformation involving the first and second branchial arches. It was first described by Goldenhar in 1952 as a triad of craniofacial microsomia, spinal anomalies and ocular dermoid cysts [5]. It has no sex predilection and more often presents in childhood than in adulthood [6]. Goldenhar syndrome is clinically heterogeneous, and there is no agreement in the literature on the minimal diagnostic criteria. Traditionally, the presence of ear abnormalities, which leads to the search of mandibular hypoplasia and vertebral alterations, is the main clinical feature associated with Goldenhar syndrome diagnosis [7]. Ear abnormalities in Goldenhar syndrome include acrotia, microtia, preauricular tag, and auricular fistula [8]. Other ophthalmological symptoms have been also reported such as anophtalmia, microphtalmia, motility disorders, strabismus, blepharoptosis, palpebral fissure, eyelid coloboma, coloboma of the iris or choroid, iris atrophy, polar cataract, anomalies of the lacrimal drainage system, retina and optic nerve anomalies [9]. In our patient, the main features were oculo-auricular namely eyelid coloboma, corneal dermoid cyst and preauricular tags. The vertebral features were missing. Some authors found such incomplete Goldenhar syndrome [6].

Most cases of Goldenhar syndrome are sporadic. Drugs, such as cocaine, thalidomide, retinoic acid, and tamoxifen ingested during pregnancy have also been suggested as etiologic factors. Maternal diabetes and infections caused by rubella and influenza during pregnancy may be related to the development of this syndrome [10].

The management of ocular features such as dermoid cyst or eyelid coloboma is sometimes very tough. Surgical treatment of the condition related to large coloboma requires surgical repair and spectacle correction, large limbal dermoid needs excision of the dermoid with lamellar keratoplasty. Severe anomalies of the mandible require reconstruction with bone graft [11].

CONCLUSION

Goldenhar syndrome is a rare condition with possible complex abnormalities. The features may vary from one patient to another. The ocular manifestations are amblogenic. Early diagnosis and management are to be undertaken for a good rate of success.

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