

GOLDENHAR SYNDROME: OCULAR FEATURES

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ABSTRACT

Goldenhar syndrome is a rare congenital anomaly which consists of a triad of an ocular dermoid cyst, preauricular skin tags and vertebral dysplasia. We report two cases of Goldenhar syndrome, diagnosed in a 4-year old girl and in a 20-year old young adult. The dermoid cyst is a benign tumour with serious ophthalmologic complications.

KEYWORDS

amblyopia, dermoid cyst, goldenhar syndrome, Oculo-Auroculo-Vertebral syndrome

INTRODUCTION

Goldenhar syndrome is a rare congenital anomaly consisting in a triad of an ocular dermoid cyst, preauricular skin tags and vertebral dysplasia (1,2).

Early diagnosis and treatment of this tumor prevent the development of secondary amblyopia. Hence, we report these two cases of Goldenhar syndrome, diagnosed in a 4-year-old girl and in a 20-year-old young adult.

CASE REPORT 1

A 4-year-old female, born of a non-consanguineous marriage, presented to the Department of Ophthalmology with congenital lesions of the child's face and eyes.

The child was born after a full-term normal delivery and there was no history of any maternal illness during pregnancy.

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Ocular examination revealed a dermoid nodule located on both the bulbar conjunctiva and the inferotemporal side of the limbus of the right eye, associated with esotropic strabismus (Figure 1).

Refraction identified against the rule astigmatism of 3 diopters.

Best uncorrected visual acuity was 20/40 in the right eye and 20/20 in the left eye.

There was neither evidence of any coloboma nor microphthalmia, and ocular motility was normal.

However, the girl had decreased eyelid motility leading to exposure keratopathy.

On facial examination, there were two right-sided preauricular tags along the line joining the tragus with the angle of the mouth (Figure 2).

Skeletal examination revealed unilateral preaxial polydactyly of the left hand (Figure 3). These symptoms were consistent with the diagnosis of Goldenhar syndrome.

Complete excision of the dermoid cyst was achieved. Correction of astigmatism errors and orthoptic treatment for amblyopia with occlusion was started early to correct the amblyopia and strabismus. The postoperative course was uneventful and the child was transferred to Paediatrics Department for multidisciplinary management of skeletal abnormalities. The child made a good recovery and the visual acuity was 20/125 in the right eye after 9 months of follow up.



Fig. 1: Bulbar conjunctival dermoid tumor invading the inferotemporal side of the limbus



Fig. 2: skin coloured papules in the preauricular area



Fig. 3: Unilateral pre-axial polydactyly in the left hand

CASE REPORT 2

A 20-year-old girl presented, for the first time, to the outpatient department with complaints of strabismus in her right eye. Her family history was negative.

Ocular examination revealed a superotemporal epibulbar limbal mass in the right eye associated with esotropic strabismus (Figure 4). The fibrovascular ingrowth involves the cornea upon 3 mm and caused an against the rule astigmatism of 4 diopters. Visual acuity was 20/20 in the left eye and reduced to counting fingers in the right eye. Anterior segment and fundus examinations were all normal.

Two right-sided, accessory pre-auricular appendages, were noticed in front of the external ear (Figure 4). The level of intelligence was normal and additional systemic examinations revealed no abnormalities.



Fig. 4: Bulbar conjunctival dermoid tumor invading the superotemporal side of the limbus in the right eye

The diagnosis of Goldenhar syndrome was made on the basis of these features. A complete cosmetic excision of choristoma was performed. No treatment of the amblyopia was attempted because it's late discovery.

DISCUSSION

Goldenhar syndrome, also known as the Oculo-Auriculo-Vertebral syndrome (OAVS), is a rare congenital malformation involving the first and second branchial arches (3). It was first described by Goldenhar in 1952 as a triad of craniofacial microsomia, spinal anomalies and ocular dermoid cysts.

As far as we know, only few articles focus on amblyopia and strabismus in Goldenhar syndrome. Nevertheless, if the limbal dermoide lesions are detected and excised early, amblyopia and strabismus may be prevented. Consequently, this improves the visual prognosis of Goldenhar patients.

There is no evidence of a clear inheritance pattern and no chromosomal anomalies have been described up to now (4). In our cases, there was no maternal illness during the pregnancy.

The main ocular feature of Goldenhar syndrome is the epibulbar choristoma (30-60%) (2,6). It consists of a dermoid or a lipodermoid benign tumor located on the inferotemporal or superotemporal part of the limbus. The corneal and scleral invasion by the tumours is rare and leads to against the rule astigmatism. Associated features caused by eyelid mobility disorders such as irritation and exposure keratitis can lead to

central visual axis obscuration. In both cases, the dermoid mass invaded the cornea and caused an important astigmatism with consequent amblyopia.

Other ophthalmological symptoms have been also reported such as anophthalmia, microphthalmia, 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 (5).

No anomalies other than choristoma and amblyopia were found in our cases. This syndrome can be associated with cardiac, pulmonary, renal and skeletal malformations in 50% of cases. An association of intracranial dermoid cyst and Goldenhar syndrome has been reported (6).

Aural features such as considerable preauricular hearing loss have been described in 40% of cases (2,7).

Management of ocular dermoid tumor consists of surgery.

Goldenhar syndrome requires a multidisciplinary approach and the treatment of the condition varies with age and systemic associations. The dermoid cyst is a benign tumour that causes serious ophthalmologic sequels: astigmatism, amblyopia and strabismus. The ophthalmologist should focus on visual consequences, treat early and follow up the patients closely.

Even if the central visual axis is not obstructed by the dermoid tumour, a decrease in visual

acuity may result from acquired astigmatism and amblyopia.

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