CASE REPORT

Optic Nerve Head Drusen in Goldenhar Syndrome

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Abstract

We report a case where bilateral drusen of the optic nerve head was found in association with Goldenhar syndrome and which, to the best of our knowledge, is the only report of its kind.

Key Words

Goldenhar syndrome , optic nerve head drusen

Introduction

First described by Von Arlt in1845, Goldenhar syndrome is a rare syndrome involving defects in structures developed from the 1st and 2nd branchial arches during blastogenesis, sometimes accompanied by vertebral and / or ocular anomalies. Though the syndrome has variously been described as oculo-auriculo-vertebral dysplasia, hemifacial microsomia and goldenhar syndrome, it has been suggested that these entities may simply represent gradations in severity of an error in morphogenesis.

The triad of anomalies comprising this syndrome include accessory auricular appendages, aural fistulae and epibulbar dermoids (1). However, several other ocular manifestations have also been described.

Here we report a case where drusen of the optic nerve head was found in association with this syndrome and which, to the best of our knowledge, is the only report of its kind.

Case report

A 3 year old boy presented with pre-auricular skin tags on left side of face. Ears were normal in shape and size, with normal external auditary canal and no evidence of middle ear disease. There was partial cleft of upper lip. Eyes showed bilateral corneal opacities, and epibulbar dermoid in infero-temporal quadrant of left eye (Fig: 1). The best corrected visual acuity was 6/24 in right eye and HM (hand movements) in left eye. The intraocular pressure was 12 mm Hg in right eye and 16 mm Hg in left eye. On slit lamp examination in the right eye except for macular corneal opacity, the rest of anterior segment was normal. In left eye there was a central macular to leucomatous corneal opacity due to which the details were not very clear but the anterior chamber appeared normal at periphery. Fundus could not be examined in left eye because of central corneal opacity, whereas, in the right eye the central fundus was normal. B scan ultrasonography detected bilateral calcification (highly reflective echo) in optic nerve heads, suggestive of drusen (Fig: 2a & 2b). There was no other pathology of posterior segment. Roentgenographic examination revealed malsegmentation of posterior neural arches involving lower cervical and upper dorsal vertebrae. No neurological, cardiac, renal or limb anomalies were seen and the child was found to be of normal intelligence. There was no history of consanguinity and other members of his family were normal.

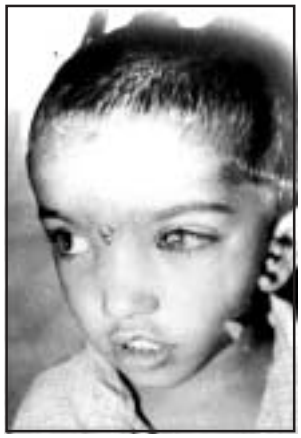

Fig. 1. Left sided pre-auricular skin tags, bilateral corneal opacities, epibulbar dermoid in left eye & partial cleft of upper lip.

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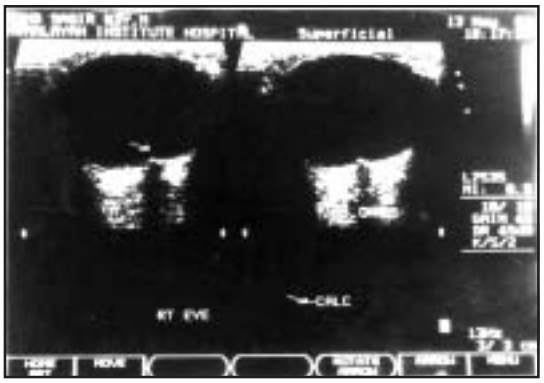

Fig. 2a. B scan of the right eye showing calcification of optic nerve head.

Fig. 2b. B scan of the left eye showing calcification of optic nerve head.

Discussion

Goldenhar syndrome is predominantly unilateral and asymmetric and is associated with a constellation of features including facial, auricular and vertebral defects. The facial defects that have been described include macrostomia, micrognathia, hemifacial microsomia. Oral defects include diminished - to - absent parotid secretion, bifid tongue, cleft lip and palate, malocclusion and dental anomalies (2,3). The partial cleft lip seen in our patient is not so common. Ear anomalies range from pre-auricular skin tags to abnormal shape and size of auricles, atresia of external auditary canal, aural fistulae, and conductive hearing loss (4).Our patient had pre-auricular skin tags occurring along a line drawn from left tragus to the angle of mouth , which is the most important and constant feature. Unilateral cases tend to be right - sided. The reported ocular manifestations of Goldenhar syndrome include choristomas (lipodermoids and epibulbar dermoids), blepharoptosis, colobomas, strabismus, micro-ophthalmia (5,6). The incidence of epibulbar dermoid (also present in our patient) varies between 32% and 92%, and in 78% cases it is found at infero-temporal limbus (5,6). No case of drusen or corneal opacity has been reported so far (2). Drusen of optic nerve are hyaline, often calcareous bodies of various sizes often situated in the prelaminar or non-myelinated portion of optic nerve head. They are mostly postnatal. Some cases, however, present at birth in which case they probably arise from degeneration of glial material towards the end of foetal life. Occasionally they occur as a dominantly inherited trait. These translucent glistening bodies resembling a heap of silver coins are usually not visible ophthalmoscopically in young children. As was the case in our patient , these lesions are usually bilateral and can easily be identified echoghraphically as they produce an echo of extremely high reflectivity (7). They are sometimes associated with retinitis pigmentosa and tuberous sclerosis.

The exact etiopathogenesis of Goldenhar syndrome is not yet clear. Most cases are sporadic although some families with either autosomal dominent or recessive inheritance have been identified . A detailed analysis of families of 74 probands found autosomal dominance as the likeliest mode of inheritance (8). A deficit of oxygen supply needed for growth and differentiation of tissues has also been proposed as a cause of abnormal development.

References

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