Picture Story

Overlap of Delleman-Oorthuys syndrome and Goldenhar syndrome

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Introduction

Delleman-Oorthuys syndrome, also known as oculo-cerebro-cutaneous syndrome (OCCS), occurs sporadically with a triad of eye, cerebral and dermatological malformations[1]. There are patients reported with associated ear and vertebral anomalies which overlap with Goldenhar syndrome (GS)[2,3].

Case report

A baby boy was born at 37 weeks of gestation by normal vaginal delivery as the first child to non-consanguineous parents. Mother did not have any complications during pregnancy and was not exposed to teratogenic medications. Antenatal ultrasound scans, including anomaly scan, were normal. Neonatal examination showed multiple prominent craniofacial anomalies. His left palpebral fissures were fused (ankyloblepharon) with anophthalmia (Figure 1) and right eye was noted to have a significant cloudy appearance of the cornea, when opened (Figure 2). Left ear was normal. Right ear was malformed with prominent linear skin grooves on the back of the pinna (Figure 3) and the external auditory meatus was absent. A skin tag and two pre-auricular sinuses were present on the right side (Figure 4). A skin defect suggestive of cutis aplasia was noted over the vertex (Figure 5). He did not have limb anomalies and rest of the systemic examination was normal. His birth weight was 2.6 kg while his length and head circumference were recorded as 48cm and 34cm respectively.

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Ophthalmological examination showed right sided congenital cataract and left sided absent eyeball with ankyloblepharon. Hearing screening showed normal hearing on both sides. 2D echocardiogram revealed patent foramen ovale without other anomalies. Ultrasound scan of the abdomen was normal but for a small haemangioma in the liver. Ultrasound scan of the brain was normal except for left sided anophthalmia. He did not have any vertebral anomalies on radiological assessment. Baby was referred to paediatric ear nose and throat (ENT) and ophthalmology for follow up

Discussion

OCCS was first described by Delleman and Oorthuys[4,5]. It has male preponderance[2]. Some reports suggest that a defect in embryonal development during fifth or sixth week causes OCCS[2,6]. No familial tendency has been documented[6]. Common eye anomalies were orbital cysts and microphthalmia or anophthalmia. Other reported eye lesions were palpebral and iris colobomas and congenital cataract[1,3]. Common skin lesions reported were focal areas of skin aplasia or hypoplasia in the face, neck or scalp and very rarely on the trunk[1]. Skin tags, especially around the eyes, were a very prominent feature in OCCS[1].

Ventricular system anomalies, cerebral and cerebellar cysts and agenesis of the corpus callosum were reported as common associations and polymicrogyria and subcortical heterotropia were also reported with neuroimaging[1]. Most of the children with OCCS have significant psychomotor developmental delay and convulsions[2]. Some patients with OCCS have clinical features overlapping with Goldenhar Syndrome (GHS) and Goltz syndrome (GS)[1,3,6,7]. GHS, which is also known as oculo-auriculo-vertebral syndrome, is characterized by facial asymmetry, peri-auricular skin tags, dermoid cysts, iris and palpebral colobomas, cleft lip and palate, micrognathia, auditory malformations and vertebral anomalies[1,2]. Goltz syndrome, which is also known as focal dermal hypoplasia, is associated with microphthalmia, dermic hypoplasia, polysyndactyly, and affects only females[2]. Our patient has features of OCCS with some features of GHS. A comparison of the abnormalities in OCCS, GHS and our patient is shown in Table 1.[1,3,6.]

Figure 1: ***Anophthalmia with fused palpebral fissures***
**Permission given by parents to publish photograph*

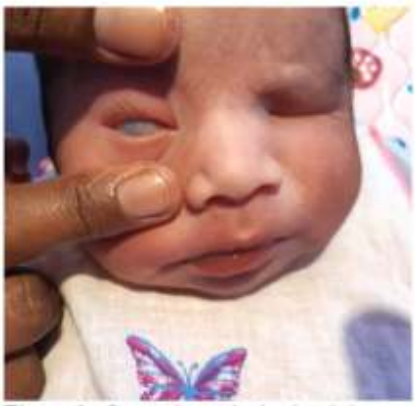

Figure 2: ***Corneal opacity in the right eye***
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Figure 3: ***Malformed pinna with grooves on posterior surface***

Figure 4: ***Absent auditor meatus. Peri-auricular sinuses and skin tag shown by arrows***

Figure 5: ***Cutis aplasia of the scalp***

Table 1: ***Comparison of clinical features***

Clinical Features	OCCS	GHS	Our patient
Neurological			
-Brain cysts	+	+	-
-Hydrocephalus	+	+	-
-Other structural anomalies	+	-	-
Eye			
- Orbital cysts	+	+/-	-
-Microphthalmia/ anophthalmia	+	+	+
-coloboma of eye lid	+/-	+	-
-coloboma of iris	+	+	-
-Epibulbar dermoid	-/+	+	-
- cataract	+	-	+
Ear			
-auricular pits	-	+	+
-malformed ears	-	+	+
-microtia	-	+	+
-Deafness	-	+	-
Oral			
Cleft lip	-	+	-
Cleft palate	-	+	-
Micrognathia	+	+	-
Skin			
Focal skin defects	+	-	+
Periauricular skin tags	+	+	+
Periorbital skin tags	+	-	-
Skeletal			
Vertebral anomalies	-	+	-
Radial hypoplasia	-	+	-
Cardiac malformations	-	+	-

OCCS: Oculo-cerebro-cutaneous syndrome, GHS: Goldenhar Syndrome

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