

## Delleman- Oorthuys Syndrome: A Diagnostic Dilemma!

Delleman and Oorthuys illustrated two cases of oculocerebrocutaneous syndrome (OCCS) in 1981.<sup>[1]</sup> The combination of ocular abnormalities, cerebral malformations, and focal dermal aplasia was presumed to be the result of a disturbance in early embryogenesis.<sup>[2]</sup>

A 1-year-old male child born to a non-consanguineously married couple presented with dysmorphic facies, multiple hyperpigmented lesions since birth, and an uneventful birth history.

On examination, the right eye had severe agenesis (anophthalmos), left eye was esotropic and had conjunctival congestion with discharge, a dermal tag from the lateral canthal conjunctiva and conjunctivalization of the cornea [Figure 1a]. Both upper eyelids showed a notch medially, indicative of partial coloboma. Ultrasonography (USG) showed atrophic bulbi on the right side and a possible retino-choroidal coloboma. The left eye had an ambulatory vision.

Segmental distribution of whorled hyperpigmented patches was observed over the right side of trunk [Figure 1b]. The child had a depressed nasal bridge, hypertelorism, and low set ears. The scalp showed focal alopecia [Figure 2a] and multiple well-circumscribed, atrophic patches with loss of hair, suggestive of membranous aplasia cutis with the classical hair collar sign [Figure 2b].

Two-dimensional echocardiogram done at birth showed a small ostium secundum atrial septal defect with left to right shunt. Neurosonography done at birth showed mild bilateral ventricular enlargement with no obvious brain parenchymal abnormality. MRI brain showed mild prominence of CSF spaces in both frontal, parietal and temporal cerebral convexities with MRI spine revealing no abnormalities.

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Delleman *et al.*<sup>[2]</sup> in 1981 reported two similar cases with right-sided orbital hamartoma, left orbital appendages, multiple facial focal defects, and CT brain showing agenesis of corpus callosum. They concluded that this combination of features is the consequence of a disturbance in early embryogenesis and named it the OCCS.

Moog, Jones and Bird *et al.*<sup>[3]</sup> discovered a novel mid-hindbrain malformation in a series of cases of OCCS. However, our patient had mild bilateral ventricular enlargement with no evident brain parenchymal abnormality which is comparable to a study by Moog *et al.*<sup>[1]</sup> who reported normal imaging studies of brain in 11% of the patients and ventricular dilatation in 43%.

Similar to the ocular findings of our patient, Moog *et al.*<sup>[1]</sup> also reported one of the following ocular defects to be present in a majority of the cases: Anophthalmia/microphthalmia, orbital cyst, eyelid coloboma and arrived at the conclusion that these features were stereotypical of OCCS.

The unique pattern of segmental and whorled cutaneous hyperpigmentation found in our patient was also reported in another case of OCCS by Rizvi *et al.*<sup>[4]</sup> Focal hypo-/aplastic skin lesions and/or skin appendages were considered indispensable for establishing the diagnosis of OCCS.<sup>[1]</sup>

Our patient also had cardiovascular involvement in the form of a small ostium secundum ASD with a left-to-right shunt which has not been reported in any of the previous cases of OCCS.

OCCS should be differentiated from other similar phenotypic syndromes namely Goldenhar syndrome, Goltz syndrome

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Website: [www.idoj.in](http://www.idoj.in)

DOI: 10.4103/idoj.idoj\_304\_21

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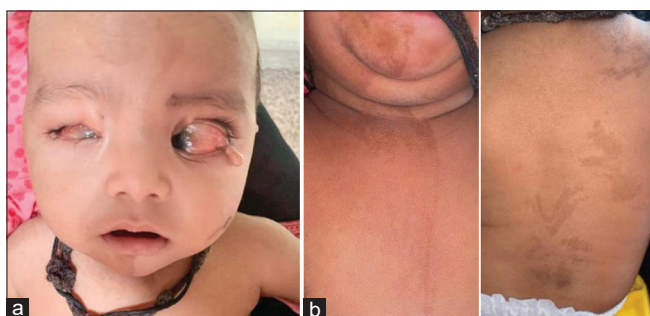


**How to cite this article:** Raju SP, Asha GS, Revathi TN, Loganathan E. Delleman- Oorthuys syndrome: A diagnostic dilemma!. Indian Dermatol Online J 2022;13:266-7.

**Received:** 17-May-2021. **Revised:** 17-Jun-2021.  
**Accepted:** 21-Jun-2021. **Published:** 03-Mar-2022.

**Table 1: Differentiating features of Delleman-Oorthuys syndrome, Goldenhar syndrome, Goltz syndrome and Haberland syndrome**

Differentiating feature	Delleman -Oorthuys syndrome	Goldenhar Syndrome	Goltz Syndrome	Haberland Syndrome
Cutaneous Appendages	Peri- orbital	Pre- auricular	Peri-oral/peri-anal	-
Focal Skin Defects	+	-	+ (Blashkoid distribution)	-
Alopecia	+	-	+	+
Psiloliparus (Nevus)	-	-	-	+
Microtia	-	+	-	-
Vertebral/Skeletal anomalies	-	+	+	+
Dental Deformities	-	+	+	-
Orbital Cyst	+	+	-	-
Micro-ophthalmia	+	+	+	-
Epibulbar Dermoid	+	+	-	-
Eyelid Coloboma	+	+	-	-



**Figure 1: (a) Dysmorphic facies with right eye anophthalmia and left sided conjunctival dermal tag. (b) Well demarcated linear and segmental nevoid hyperpigmentation over trunk and neck**

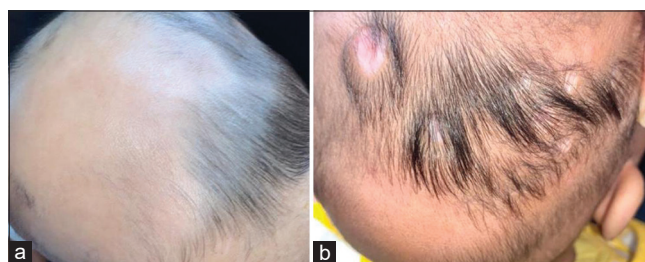
(focal dermal hypoplasia), and most importantly Encephalo craniocutaneous lipomatosis (ECCL) or Haberland syndrome. [Table 1].

Taking into consideration the Revised Diagnostic Criteria for OCCS,<sup>[5]</sup> we present a typical case of Delleman–Oorthuys Syndrome with additional involvement of cardiovascular system. OCCS belongs to the group of ultra-rare syndromes as only about 40 patients are known till date.<sup>[5]</sup>

To conclude, the above classical constellation of features makes OCCS, albeit rare, an incontrovertible diagnosis. The successful management of OCCS requires an integrated approach in the form of a comprehensive neurological assessment and a detailed ophthalmic evaluation.

### **Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The Guardian understands that names and initials will not be published and due efforts will be made to conceal patient identity.



**Figure 2: (a) Wide patches of alopecia over scalp. (b) Hair collar sign with membranous aplasia cutis**

### **Acknowledgments**

We thank Dr. Sandhya R Professor and Senior Consultant, Department of Ophthalmology, K.C General Hospital, Bangalore, India for her contribution.

### **Financial support and sponsorship**

Nil.

### **Conflicts of interest**

There are no conflicts of interest.

### **References**

1. Moog U, de Die-Smulders C, Systemans JMJ, Cobben JM. Oculocerebrocutaneous syndrome: Report of three additional cases and aetiological considerations. *Clin Genet* 1997;52:219-25.
2. Delleman JW, Oorthuys JW, Bleeker-Wagemakers EM, ter Haar BG, Ferguson JW. Orbital cyst in addition to congenital cerebral and focal dermal malformations: A new entity. *Clin Genet* 1984;25:470-2.
3. Moog U, Jones MC, Bird LM, Dobyns WB. Oculocerebrocutaneous syndrome: The brain malformation defines a core phenotype. *J Med Genet* 2005;42:913-21.
4. Rizvi SA, Siddiqui MA, Khan AA, Siddiqui Z. Delleman Oorthuys syndrome. *Middle East Afr J Ophthalmol* 2015;22:122-4.
5. Moog U, Dobyns WB. An update on oculocerebrocutaneous (Delleman-Oorthuys) syndrome. *Am J Med Genet Part C* 2018;178:414-22.