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CASE REPORT

Delleman Syndrome: Oculocerebrocutaneous Syndrome: A Case Report

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Abstract

Delleman Oorthuys syndrome or oculocerebrocutaneous syndrome is a rare congenital disorder. It is sporadic. As the name suggests it involves integumenatry system, central nervous system and eyes. Here we present the case of a newborn male baby who presented with congenital hydrocephalous, left sided eyelid coloboma since birth along with other features of this syndrome. Goldenhar and Goltz syndrome may also present with similar features. There is need to diagnose this congenital anomaly as a separate entity for better management and prognosis of such children.

Keywords: Cystic eye, Delleman syndrome, congenital hydrocephalous, eyelid coloboma

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Introduction

The syndrome of orbital cyst and anophthalmia (microphthalmia), perioccular appendages, skin tags, cerebral cysts along with other cerebral anomalies with focal dermal hypoplasia was first described by Delleman et al. in 1981[1-4]. Based upon the combination of features of presentation it is also called as oculo-cerebro-cutaneous syndrome. It is named after the scientists who have described it.

Case Report

A male newborn was admitted on day 1 of life with antenatally diagnosed hydrocephalous. He was born out non consanguinous marriage among healthy parents. Baby had gross hydrocephalous.

On further and detailed examination patient found to have left upper eyelid coloboma with redness and a fleshy growth over the supero-temporal aspect of the bulbar conjunctiva left eye. It was also noticed that the patient had temporo-parietal alopecia on the affected side. Patient also had sacral meningocole which was already ruptured.

Birth history was normal, and he was the first child of his parents. The rest of the body had no other area of skin appendages anywhere else. Examination of the right eye was normal. Systemic evaluation revealed no abnormality (Figure 1).

Figure 1. Left upper eyelid coloboma with redness and a fleshy growth over the supero-temporal aspect of the bulbar conjunctiva with parietal alopecia

MRI of brain was done which showed multiple small gyri in left cerebral hemisphere with shallow sylvian fissures with left sided subdural collection with hemorrhages, dilated ventricles, small brain stem and cerebellar hemispheres with large retro-cerebellar cystic lesion.

MRI of spine shows low lying spinal cord; two hemicords in sacral spinal canal with separate thecal sacs and a septum is seen traversing the spinal canal along with a small collection in skin and subcutaneous fat plane in the sacral region posteriorly (Figures 2 and 3).

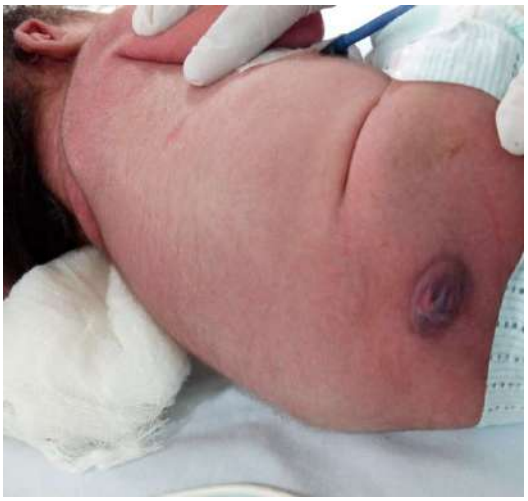

Figure 2. Ruptured sacral meningocoel.

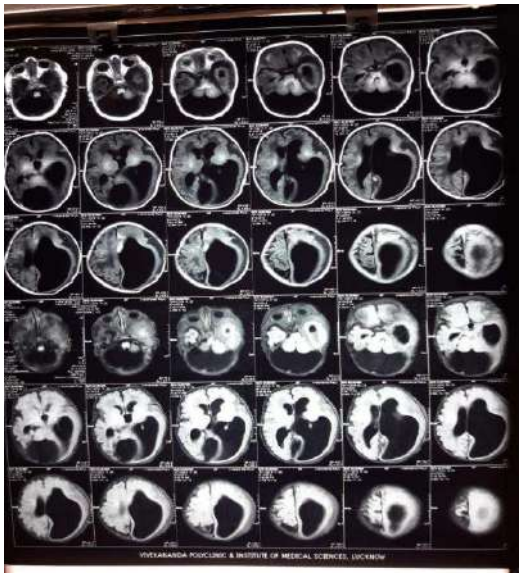

Figure 3. Left cerebral hemisphere with shallow sylvian fissures with left sided subdural collection with hemorrhages, dilated ventricles, small brain stem and cerebellar hemispheres with large retrocerebellar cystic lesion.

Discussion

Delleman syndrome is a rare genetic disorder characterized by cutaneous and ocular abnormalities as well as areas of focal alopecia along with hydrocephalus [4].

It is seen more commonly in males and occurs sporadically. There is no risk of recurrence in the siblings and with no known etiological cause [5].

Asymmetry in features is characteristic of OCCS [2]. Our patient too had his ocular and dermatological features limited to only the left side of his face. Dilatation of ventricles was also more on left side as suggested by MRI findings. Our patient had periorbital skin appendages along with parietal alopecia. Overlying skin on these alopecia regions showed patches of focal dermal hypoplasia. The skin features feature are consistent with the previously described characteristics of OCCS [2].

In our patient, ocular features consisted of a supero -temporal epibulbar dermoid on left side along with upper eyelid coloboma. Other ocular features associated with OCCS like

orbital cysts, microphthamos or anophthalmos were absent.

Affected children may present with psychomotor retardation, seizures, and/ or developmental delay. Literature has also reported of a case with hyperactive deep tendon reflexes throughout as well as a positive Babinski's sign opposite to the side with the ocular findings [6].

The etiology for the same is not known. It has been hypothesized that a sporadic mutation occuring in the fifth or sixth week of development may lead to such condition. It states that it is associated with a somatic mosaicism, which adversely affect the development of the ectodermal structures [7].

Delleman syndrome may have some characteristics which overlaps with other syndromes such as Goldenhar or Goltz syndrome. The former one is also known as oculo-auriculo-vertebral syndrome, which is characterized by a dermoid cyst, auditory and vertebral anomalies. While the latter one presents with microphthalmia, dermal hypoplasia, polysyndactyly and bad dentition [8,9,10]. These syndromes are usually differentiated by the patterns of brain malformations. Those with deformations involving midline and mesencephalon are characteristic of Delleman syndrome [7].

Conclusion

We recommend a detailed neurological workup in all such cases; and if a cerebral or cerebellar abnormality like hydrocephalus is detected as it was present in our case, it necessitates an urgent neurosurgical opinion, to prevent any further damage.

For the effective management, all such children we recommend a combined, closely coordinated approach by the ophthalmologist, the neurosurgeons, the neurophysicians as well as the paediatricians

Conflicts of interest

The authors declares that they do not have conflict of interest.

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References

1. Delleman JW, Oorthuys JWE. Orbital cyst in addition to congenital cerebral and focal dermal malformations: A new entity? Clin Genet 198;19:191-8.
2. Delleman JW, Oorthuys JWE, Bleeker-Wagemakers EM, Ter Haar BGA, Ferguson JW. Orbital cyst in addition to congenital cerebral and focal dermal malformations: A new entity. Clin Genet 1984;25:470-2.
3. Ferguson JW, Hutchison HT, Rouse BM. Ocular, cerebral and cutaneous malformations: Confirmation of an association. Clin Genet 1984;25:464-9.
4. Wilson RD, Traverse L, Hall JG, Flodmark CO, Rootman J. Oculocerebrocutaneous syndrome. Am J Ophthalmo 1985;99:142-8.
5. Patil SJ, Phadke SR. Delleman syndrome. Indian Pediatr 2006;43:173-4.
6. McCandless SE, Robin NH. Severe oculocerebrocutaneous (Delleman) syndrome: Overlap with Goldenhar anomaly. Am J Med Genet 1998;78:282-5.
7. Sujit Kumar GS, Haran RP, Rajshekhar V. Delleman syndrome with Goldenhar overlap. J Pediatr Neurosci 2009;4:53-5.
8. Moog U, Jones MC, Bird LM, Dobyns WB. Oculocerebrocutaneous syndrome: The brain malformation defines a core phenotype. J Med Genet 2005;42:913-21.
9. Roszkowski M, Dabrowski D. Encephalo-cranio-cutaneous lipomatosis (ECCL) :Haberland syndrome. A case report withreview of the literature. Neurol Neurochir Pol 1997;31:607-13.
10. Chen D, Heher K. Management of the anophthalmic socket in pediatric patients. Curr Opin Ophthalmol 2004;15:449-53.