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Delleman-Oorthuys Syndrome: A Neurocutaneous Syndrome with a Congenital Eye Cyst

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ABSTRACT

Delleman-Oorthuys syndrome is a rare congenital anomaly of unknown aetiology. It is also called Oculocerebrocutaneous syndrome that characteristically involves ectomesodermal tissues such as the eyes, central nervous system, and the integument. We report a 2-year-old boy with an orbital cyst in the left eye along with other manifestations of this disorder since birth.

Physical examination revealed a huge left eye cyst. An ultrasound scan of the left orbit showed a huge thick-walled cystic lesion involving the left globe with multiple internal strands and echoes. Computed tomography of the brain revealed severe atrophy of the left cerebral hemisphere in addition to characteristic cerebral malformations of Delleman-Oorthuys syndrome.

This report highlights the need for extensive neuroimaging in searching for life-threatening cerebral atrophy; unfortunately, genetic studies that could shed light on this syndrome are not available in our resource-constrained settings.

Keywords

Cerebral atrophy, Delleman-Oorthuys, Oculocerebrocutaneous, Ocular cyst.

Introduction

Oculocerebrocutaneous syndrome (OCCS) is an extremely rare multiple congenital anomaly of unknown aetiology, characterised primarily by the triad of ocular, brain, and skin malformations, occasionally associated with other features such as skull anomalies, craniofacial clefts or rib defects [1]. It is a non-neoplastic disorder that was first described by Delleman and Oorthuys in 1981 when they identified two presumably unrelated boys with an orbital cyst, cerebral anomalies, and focal dermal hypoplasia and aplasia [2]. This group of congenital abnormalities, has been named after the two authors that first described it (as Delleman-Oorthuys syndrome). Since the entity was described, several sporadic manifestations of the syndrome have been reported worldwide including reports from Nigeria [3,4]. The syndrome appears to have a male preponderance with an approximate male to female ratio of 2:1 but no known racial predilection [5]. It has no established definite pattern of inheritance however, somatic mosaicism has been postulated [5-7]. The manifestations of OCCS resemble other congenital developmental disorders such as Goltz and Goldenhar syndrome; however, the skin tags are typically in the peri-auricular

region [7]. Encephalocraniocutaneous lipomatosis and Hueber syndrome are other differential diagnoses of OCCS [3]. To the best of our knowledge this is the third case reported from West Africa. Therefore, the rarity of the syndrome prompted the need to present a brief report of this case.

Case Presentation

A 2-year-old boy, third of three siblings, born to a healthy non-consanguineous couple, was noticed to have a swelling at the left eye at birth that was about the size of the pulp of the thumb of his mother which progressively increased to about the size of a lemon as the child was growing up to the age of two years. The mass intermittently discharges clear fluid. There was an associated dark colouration of the skin of the upper eyelid and forehead of the left side of the head. There was no associated redness or itching of the eyes, and the child could see normally with the right eye. He has never convulsed in the past, has neither hearing impairment nor cardiopulmonary symptoms. The patient was a product of term gestation and there was no known history of teratogenic insult during the first trimester of pregnancy, nor previous delivery of a congenitally malformed baby. Although the patient attained normal developmental milestones, he was noticed not to be growing well like his two older siblings in the monogamous setting; his siblings are alive and well.

Physical examination revealed a small-for-age looking child, with a huge left eye cyst. Hyperpigmentation of skin over the left upper eyelid extending up the face (Figure 1). The patient was not pale and not jaundiced. There was neither cyanosis or dehydration and no peripheral lymphadenopathy. He weighed 7.5 kg, 62.5% of the expected age (underweight), height was 73cm, 82% for age (stunted), head circumference was 47cm at 10th centile for age (normal), mid-upper arm circumference of 13cm (borderline).

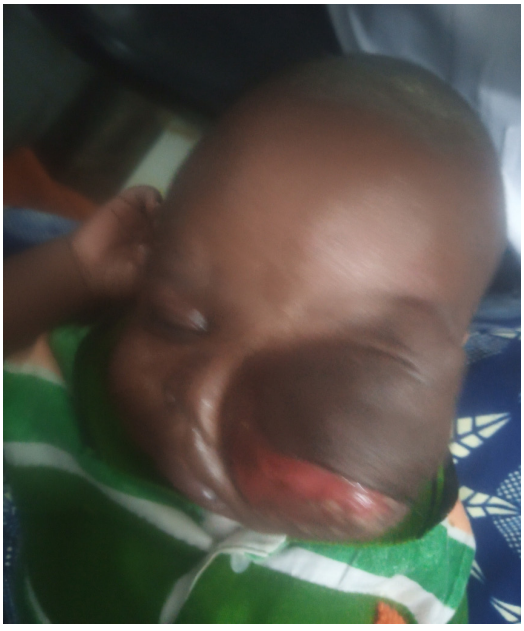

Figure 1: Clinical photograph of the patient with hyperpigmented skin over the left upper eyelid and left fronto-temporal side of the scalp and a huge left orbital cyst.

A nervous system examination revealed a conscious child with grossly intact cranial nerves and no focal neurological deficit. He had a cystic non-tender left orbital swelling measuring 4x 5x 6cm, covered by hyperpigmented skin at the upper eyelid, and has no limb or joint deformities. Other systemic examinations were essentially normal. Haematologic investigation revealed normal profile.

An ultrasound scan of the left orbit showed a huge thick-walled cystic lesion involving the bulbus oculi with many strands and internal echoic view. The anterior chamber, posterior chamber and the vitreous humour could not be unambiguously pinpointed.

Computed tomography of the brain revealed severe atrophy of the left hemispheric cerebral cortex with expanded cerebrospinal fluid space and dilatation of same side lateral ventricle, there were no areas of abnormal enhancement of the contrast medium or intracranial masses.

An cosmetic enucleation of the left eye was performed under general anaesthesia. The left upper eyelid was bluntly dissected and separated from the cystic mass. The enucleated specimen was sent unfixed immediately to the histology lab for analysis. Histopathologically, the finding on macroscopy was that of excised eyeball covered with periorbital fats measured 3x4x5cm (Figure 2).

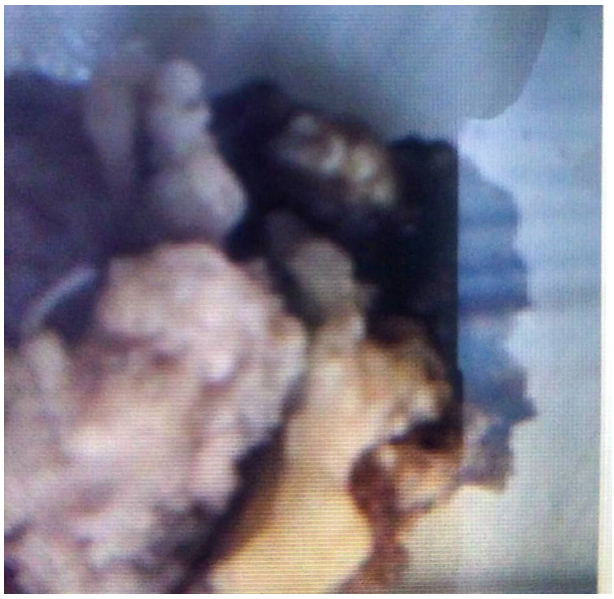

Figure 2: Image depicts excised orbital mass covered with periorbital fatty tissue.

Discussion

The brain, cutaneous and orbital malformations seen in the index case are consistent with the triad of Oculocerebrocutaneous syndrome [2,4-8]. With this classical triad of orbital cyst, cerebral and cutaneous malformations characteristics of OCCS on the side as seen in this patient, is common in males and occurs sporadically with no family history and in product of non-consanguineous

couple in virtually all documented cases as in the index patient. Recurrence in siblings have not been reported so far and no racial predilection has also been identified [2,3,9]. Some authors suggest the genetic basis of this disorder as a lethal Autosomal-Dominant mutation that enable others to survive only via mosaicism while others suggested autosomal recessive inheritance in phenotypically normal consanguineous couple parents [8-10]. Notwithstanding, genetic studies in the family is desirable for better characterization of the genetic mutation that resulted in this syndrome.

Despite the degree of the intracranial malformation detected on neuroimaging, at two years the patient has not demonstrated neurologic deficit as he had normal gross and fine motor development. This is similar to what was reported in many case reports in which the observed neurologic deficits were less severe than the demonstrated intracranial imaging abnormalities [2,3,10]. Nevertheless, a long-term follow-up is necessary as his neurologic status may deteriorate as the child grows older. Other differential diagnoses of the OCCS such as Goldenhar [12], Encephalocraniocutaneous lipomatosis and Hueber syndrome have been ruled out in our patient, clinically [13,14]. The presence of congenital cystic eye, cerebral malformations and cutaneous anomalies in this patient are further added weight to the diagnosis of Delleman-Oorthuys syndrome [2]. Oculocerebrocutaneous syndrome (OCCS) is an extremely rare multiple congenital anomaly of unknown aetiology. Genetic studies of the family were desirous for better characterization of the genetic mutation that resulted in this variant syndrome but was not available in our setting.

Conclusion

This report highlights Delleman-Oorthuys syndrome, which is a rare congenital anomaly of unknown aetiology. There is need for extensive neuroimaging in searching for life-threatening cerebral atrophy; unfortunately, genetic studies that could shed lighter on this syndrome are not available in our (resource-constrained) settings.

References

1. Delleman JW, Oorthuys JWE, Bleeker-Wagemakers EM, et al. Orbital cyst in addition to congenital cerebral and focal dermal malformations: A new entity. Clin Genet. 1984; 25: 470-472.
2. Delleman JW, Oorthuys JWE. Orbital cyst in addition to congenital cerebral and focal dermal malformations: a new entity?. Clin Genet. 1981; 19: 191-198.
3. Ugalahi M, Olusanya B, Fasina O, et al. Delleman syndrome: A case from West Africa–Features and the challenges of management. Niger Postgrad Med J. 2018; 25: 191-194.
4. Badejo OA, Fasina O, Balogun JA, et al. Delleman-Oorthuys syndrome (Oculocerebrocutaneous syndrome) in a Nigerian child: a case report. Therapeutic Advances in ophthalmology. 2018; 1-6.
5. Saatei AO, Arikan G, Saatei P, et al. Oculocerebrocutaneous syndrome. J Pediatr Ophthalmol Strabismus. 2008; 45: 181-183.
6. Hall JG. Review and hypotheses: Somatic mosaicism-observations related to clinic genetics. Am J Hum Genetics. 1988; 43: 355-363.
7. Al-Gazali LI, Donnai D, Berry SA, et al. The Oculocerebrocutaneous (Delleman) syndrome. Journal of Medical Genetics. 1988; 25: 773-778.
8. Moog U, Jones MC, Bird LM, et al. Oculocerebrocutaneous syndrome: The brain malformation defines a core phenotype. J Med Genet. 2005; 42: 913-921.
9. Arora V, Kim UR, Khazei HM. Delleman-Oorthuys syndrome: 'Oculocerebrocutaneous syndrome'. Indian J Ophthalmol. 2009; 57: 387-389.
10. Happle R. Lethal genes surviving by mosaicism: a possible explanation for sporadic birth defects involving the skin. J Am Acad Dermatol. 1987; 16: 899-906.
11. Tambe KA, Ambekar SV, Bafna PN. Delleman (oculocerebrocutaneous) syndrome: Few variations in a classical case. Eur J Paediatr Neurol. 2003; 7: 69-70.
12. Ortiz-Basso T, Vigo R, Iacouzzi S, et al. Delleman (oculocerebrocutaneous) syndrome: A case report. Indian J Ophthalmol. 2014; 62: 741-743.
13. Hunter A. Oculocerebrocutaneous syndrome: An update. Am J Med Genet. 2008; 146A: 674.
14. Rizvi SW, Siddiqui MA, Khan AA, et al. Delleman-Oorthuys syndrome. Middle East Afr J Ophthalmol. 2015; 22: 122-124.