American Journal of Clinical Neurology and Neurosurgery
Vol. 1, No. 3, 2015, pp. 147-149
http://www.aiscience.org/journal/ajcnn

Congenital Ocular Coloboma of Left Upper Eyelid Associated with Obstructive Hydrocephalus: Management

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Abstract

Coloboma literally means defect. Ocular coloboma of the eyes is an extremely rare congenital developmental defect, which constitute one of important causes of visual impairment and blindness in the pediatric age group. Reported incidence of ocular coloboma is 0.5 to 7.5 per 10, 000 live births. Authors report an interesting case syndromic variety of ocular coloboma, where the upper eyelid coloboma was characteristically associated with mental retardation. Non-contrast computed tomography head scan carried out to assess the systemic involvement, revealed obstructive hydrocephalus for which, medium pressure ventriculoperitoneal shunt was carried out. Presence of stigmata of coloboma should alert treating physician and prompt for detailed systemic evaluation, and appropriate imaging studies including ultrasonography, CT scan or MRI should be carried out to detect associated anomaly. As prompt and early diagnosis is crucial for prevention of complication, so early diagnosis followed by tailor made future therapeutic plan of management is essential for good neurological and visual outcome. So, high degree of suspicion of potentially preventable complications caused by developmental anomaly, if timely detected and early therapeutic intervention can at least delay or can almost eliminate development of complication like visual impairment and blindness.

Keywords

Ocular Coloboma, Central Nervous System Defect, Hydrocephalus, Delayed Milestones, Systemic Anomalies

Received: July 6, 2015 / Accepted: August 21, 2015 / Published online: November 11, 2015

1. Introduction

Congenital ocular coloboma (means- gap in part of the eyes structures), reported incidence is 0.5 to 7.5 per 10 000 live births. [1] In the decreasing order of predilection for involvement of component of eye in the ocular coloboma are as follow cornea, iris, ciliary body, lens, retina, choroid, and optic nerve, however, eyelid coloboma is an extremely rare occurrence. Depending upon pattern of involvement of ocular component alone or associated affection of other systemic organs, ocular coloboma can be further categorized into two types. The first type is isolated or sporadic and another being syndromic variety. In isolated type, the abnormality is only confined to orbit, may involve one or more areas of the eye; it can involve cornea, iris, ciliary body, lens, retina, choroid, and optic nerve. The Syndromic coloboma characterized by in addition to presence of typical ocular phenotype pattern, usually have associated multiple system involvement including central nervous system, hydrocephalus, cranial nerve dysfunction, seizure, malformation of gastrointestinal tract or cardiac abnormalities or renal agenesis. However, syndromic variety may have multiple systemic organ involvement. Authors present an interesting case in 6-year girl with defect of upper eyelid, for which detailed systemic evaluation by paediatrician for delayed developmental milestones including CT scan head was done, which revealed

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presence of obstructive hydrocephalus. The child underwent right sided VP shunt surgery. During follow-up period following VP shunt surgery, she had improvement in scholastic performance. There is paucity of literature regarding eye lid coloboma, so management is still debated

2. Case–Illustration

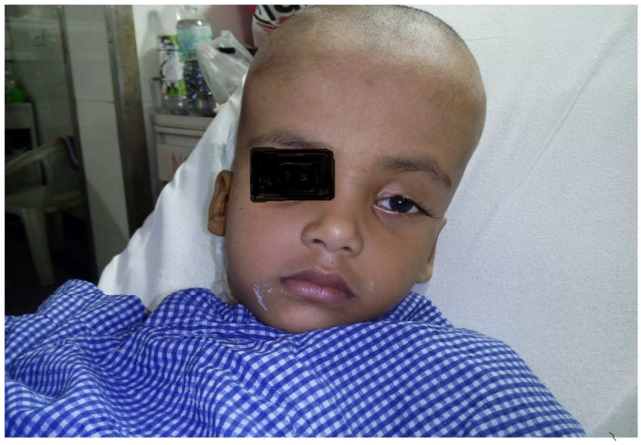

Fig. 1. Photograph of face showing small central coloboma of left upper lid.

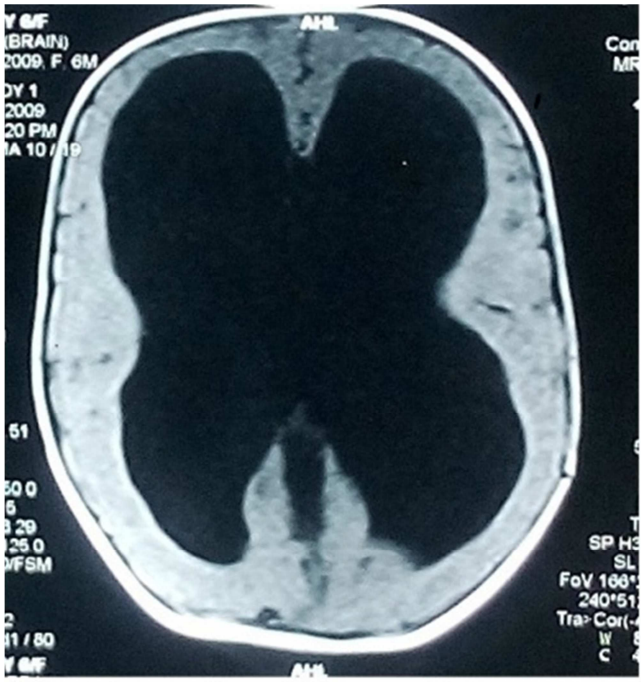

Fig. 2. MRI brain T2WI showing gross hydrocephalus with dilatation of lateral ventricle and third ventricle with prominent temporal horn.

A 6 - year - girl of non-consanguineous parents was delivered by normal spontaneous vaginal delivery at term, with no history of maternal intake of drug, alcohol or exanthematous rashes during pregnancy. At birth, parent noticed asymmetry of upper eyelids, with incurving of left upper eyelid causing improper eye closure and redness of eye associated with watery discharge. On physical evaluation by paediatrician, a 0.8x0.7 cm semicircular defect was noted in he left upper eyelid, (fig-1) however, eye brows were normal, no ulceration of eyelid was present. Her head circumference was 54 cm. She was further evaluated by ophthalmologist and found to have ocular left upper eyelid coloboma. Routine hemogram and biochemistry was normal within normal limit CT scan head revealed obstructive hydrocephalus. (Fig-2) So, provisional diagnosis of syndromic left upper eyelid coloboma with obstructive hydrocephalus was made. She was planned for C.S.F. diversion surgery.

She underwent right sided VP shunt for management of obstructive hydrocephalus under general anaesthesia, and tolerated surgical procedure very well. At last follow-up at one-year interval following VPshunt surgery, she was doing well with improvement in scholastic performance. She was also advised regular follow-up with the ophthalmology department for coloboma. Currently she is awaiting surgical repair of upper eye lid defect under occuolplasty surgical unit. As coloboma size is small, surgical reconstruction by direct approximation utilizing semicircular flap technique (Tenzel's) surgery is awaited.

3. Discussion

Ocular coloboma of the eyes is congenital developmental defect, which constitute one of important causes of visual impairment and blindness in the pediatric age group. Coloboma accounts for about 3.2–11.2% of childhood blindness and impaired vision. [1, 2, 3-6] Depending upon stand alone ocular components pattern involvement or predilection for other systemic organs, ocular coloboma can be classified into two variety; first type is isolated or sporadic form and another being syndromic variety. In sporadic variety, the developmental abnormality is only confined to adenexa or component of the orbit, may involve one or more areas of the eye; with decreasing order of involvement within the eye-globe are as follow cornea, iris, ciliary body, lens, retina, choroid, and optic nerve are affected, however, eyelid coloboma is extremely rare occurrence, but our case had upper eyelid coloboma. Another variety being syndromic coloboma, which is characterized by in addition to presence of ocular phenotype stigmata, also usually has associated multiple system involvement i.e. central nervous system, dysfunction or dysgenesis of multiple cranial nerve, hydrocephalus, seizure, intracranial lipomas and mental retardation, severe craniofacial anomalies , gastrointestinal tract or cardiac abnormalities, renal agenesis or hypoplasia [2, 3, 5, 6, 9]

Ocular coloboma development is caused due to failure of fusion of the mesodermal folds at about 7–8 weeks of gestational life. [1, 3] Exact etiopathology is still debate, however, commoner aetiology of coloboma include genetic

inheritance, environmental influences maternal exposure to teratogenic substances i.e. Thalidomide, alcohol, LSD and carbamazepine and environmental influences. [1, 2] Recent genetic study to identify genes responsible for causation of underlying inherited coloboma, significant progress are made in understanding of molecular events controlling closure of the optic fissure. However, severity of disease manifestation is related to temporal expression of the gene. Eye lid coloboma is reported to associated with chromosomal disorder i.e. Treacher Collins Syndrome characteristically involve iris and optic nerve coloboma and Cat eye syndrome usually contain associated with coloboma of iris, choroid and optic nerve. [3, 4, 7, 9]

Eyelid protects the globe from trauma, excessive light, and moves the tears toward the lacrimal drainage system and maintains integrity of tear films. [5] Eyelid defect increases susceptibility for proneness to environmental hazard in temperate climate with abundance of dust and dry air promoting conjunctival and corneal xerosis and ulceration. The eyelid defects are encountered in congenital anomalies, trauma, and post excision for neoplasm. [7. 8. 9] Further, eyelid defect can be categorized into small, medium and large respectively, on basis of sizes of defective eye lid area with comparison to total eyelid surface area, have respectively 20-25%, 30 to 50%, and 50% deficient. [5] Current case had smaller variety of upper lid coloboma.

Depending on presence of associated defects of retina, cornea, iris or upper lid defect, patient can present with a range of clinical symptoms including proptosis, loss of vision, increased intraocular pressure, and visual impairment and even blindness in potentially missed cases. Clinically the patient presents with orbital pain, hypoesthesia, and restriction of ocular movement, diplopia or proptosis and vision loss. Oththalmolocial evaluation along with systemic evaluation by appropriate imaging like ultrasonography, or CT scan can detect and aids in identifying the presence of defect and degree of ocular disorder requiring care. [1, 5, 7, 9, 10]

The aim of management is to put a constant vigilance to look for any potentially vision impairing pathology, raised intraorbital pressure. There is no cure for coloboma; but regular, periodic monitoring is advised according to the complexity of the eye involvement. In the present case, ophthalmologist advised regular use of artificial tear, use of eye protecting goggle and eye ointment application along with regular and periodic follow-up to detect early abnormality. [3, 5, 7, 8, 10] As our cases had small coloboma, she is still awaiting surgical correction by reconstruction by direct approximation using semicircular flap (Tenzel's) surgery. [5] These cases need regular follow-up and care under multidisciplinary team.

4. Conclusion

Awareness of occurrence of congenital ocular coloboma, which may have associated other systemic developmental anomaly of cardiac, gastrointestinal, renal or central nervous system. Further ocular coloboma has potential for threatening of vision or causing permanent blindness, if defects are not detected early and prompt remedial measures for corneal defect, glaucoma are not made early. As such abnormality is responsible for about 4-11% childhood blindness, which is potentially preventable, if detected early and prompt therapeutic intervention can lead to good functional outcome.

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