CASE REPORT

Isolated Bilateral Upper Lid Coloboma - A Case Report

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ABSTRACT

Background: *Congenital upper lid colobomas may be associated with ocular and systemic anomalies. This paper reports an isolated bilateral upper lid coloboma.*

Method: *A report of a case of bilateral upper lid coloboma with discussion of relevant literature.*

Results: *A 5-month old healthy baby girl presented with isolated bilateral upper lid coloboma. The coloboma was as large as two thirds of each upper lid with symblepharon. She had no other associated congenital anomalies reported by various workers. The upper lid defect in each eye was repaired in two stages. She had a flap from the lower lid using the lid switch technique and flap separation 2 weeks after the first surgery, in a combined surgery by a plastic surgeon and ophthalmologists. A good functional and cosmetic result was achieved from the treatment. The patient was discharged five days after the second surgery.*

Conclusion: *This case shows that lid coloboma could be an isolated problem in a child. Early and appropriate surgical intervention is necessary for good visual and functional outcome.*

KEY WORDS: *Lid Coloboma; Fraser Syndrome; Delleman and Goldenhar's Syndrome.*

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INTRODUCTION

Congenital upper lid colobomas may be associated with ocular and systemic anomalies[1]. They constitute a potential threat to vision at an early age and a significant cosmetic blemish later in life.

The role the ophthalmologist involves both the diagnosis and management of the various ocular structure defects and management of the visual development. Early surgical intervention is usually required when the defect is larger than one third of the eyelid margin. Close monitoring of the visual development of patients with congenital upper eyelid colobomas is also essential since the risk of amblyopia in these patients is high.

Many reports of upper eyelid defects described various ocular and systemic anomalies which may often require multiple anaesthetics for examination of the eye, excision of skin tags and repair of cleft palate as in Delleman syndrome[2,3].

This case report illustrates the fact that isolated upper eye lid coloboma may be a serious problem. Early diagnosis and treatment is required for a significant positive visual and cosmetic outcome.

CASE REPORT

In December 1999, ophthalmic treatment was sought for a 5-month old infant after the parents had noticed that the patient always had both eyes opened all the time and that she rarely closed her eyes even when sleeping.

The patient was a product of a full term pregnancy and normal spontaneous vaginal delivery in a private hospital in Lagos, Nigeria. Birth weight was 2.8kg. The patient was the product of nonconsanguinous marriage and the neonatal period was unremarkable. She had normal developmental milestones and a healthy sister. This patient was reviewed by a team of paediatricians, neurologists and plastic surgeons who managed the patient together with the ophthalmologists. Her vision was very good as she could hold the biro and other smaller toys from the examiner's hand. Systemic and neurological examinations were normal. The haematological studies, the electrolyte and urea were all within the normal limit. The main findings were on ocular examination.

The patient had examination under general anaesthesia (EUA) and repair of upper lid

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defect in both eyes done in January 2000. EUA disclosed the following findings. Bilateral upper lid coloboma measured 10mm in the right upper lid; symblepharon of the upper lid to the upper limbus was noticed. The vertical and horizontal corneal diameters were 12mm and 11.5mm respectively in either eye. The intraocular pressures with Perkins tonometer were 10mmHg in the right eye and 11mmHg in the left eye. The corneas were slightly hazy, anterior chambers were well formed; the lenses were normal but there was diffuse mottling of the retinal pigment epithelium.

After a full ophthalmologic examination under anaesthesia the operation to repair the defects followed. This involved the separation of symblepharon. The upper lid defect in each eye was freshened by excision of the edges, repaired with a flap from the lower lid using the Lid switch technique in a combined surgery by the plastic surgeon and the ophthalmologists.

Revision of the wound with flap separation and insetting was done on the 14th day after the initial surgery. She had an uneventful postoperative period. Patient was discharged five days after the second surgery.

At 3 weeks post operative review, the patient had good movement of both upper lids when blinking. She had good vision in both eyes as demonstrated by localization of objects and following a moving toy.

DISCUSSION

There have been few reports of upper lid coloboma in association with malformation of midfacial structures; however isolated occurrence of lid coloboma is a rare feature[1, 2]. Our patient had no other oculoauriculovertebral dysplasia or congenital heart disease as reported in other patients[2-4].

There was a reported case of a 4-year-old male child who presented with meningitis in Delhi who was found to be a case of oculoauriculovertebral dysplasia with tetralogy of Fallot. He had epibulbar dermoid in addition to having upper lid coloboma, double elevator palsy and grade 1 Duane's retraction syndrome in his right eye and dermolipoma in the left eye[2]. The symptom complex described Goldenhar's syndrome which is a disease complex of structures developed from the 1st and 2nd brachial arch.

Another case of an infant with oculocerebrocutaneous dysplasia (Delleman) syndrome has been reported[3]. This patient presented with total alopecia in the scalp, periorbital skin appendages, hypertrophy of the skin, left side orbital cyst, lid coloboma, left palate, neonatal seizures, cerebral hemiatrophy, multiple intracranial cystic spaces and enlarged lateral ventricles. The anomalies often predispose the patient to multiple anaesthetic exposure[3].

In Los Angeles an infant with Fraser syndrome with complete left cryptopthalmos has been reported[4]. The patient had a favourable long-term visual outcome after ocular reconstruction.

In our own reported case there was bilateral incomplete cryptopthalmos. The reconstruction was accomplished in a stepwise manner. The upper eyelids were dissected from the upper corneal surface, the conjunctiva fornices were well formed and so there was no need for reconstruction, while the upper lid coloboma was repaired with a flap from the lower eyelid. `The lid flap gives the best tissue match in texture, colour and functional adaptation. The sizes of the lid defects preclude mobilization and direct closure. There was no need for repeated general anaesthesia to correct the various anomalies that normally accompany lid coloboma like in Delleman, Fraser's and Goldenhar's syndrome[5-7]. Not much of oculoplastic surgery is being done by ophthalmologists in this country[8]. However, plastic lid surgery could be one of the most challenging but satisfying areas of ophthalmic practice. There was significant preservation of eyelid function and good visual prognosis due to the isolated nature of the defect.

CONCLUSION

Upper lid coloboma which is usually a part of anomalies in various syndromes like Delleman syndrome, Fraser and Goldenhar's syndrome can present as an isolated problem in rare cases. The result of the treatment with

oculoplastic and corneal surgery is very encouraging. This patient had useful vision and good eyelid movement when blinking.

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