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Published in the Russian Federation
European Journal of Medicine
Has been issued since 2013.
ISSN: 2308-6513
E-ISSN: 2310-3434
Vol. 9, Is. 3, pp. 150-154, 2015

DOI: 10.13187/ejm.2015.9.150
www.ejournal5.com



UDC 61

Upper Lid Coloboma – a Case Report

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Abstract

The upperlid colobomas are rare and usually associated with cryptophthalmos. The colobomas can either affect one or both eyes. They can be associated with life threatening syndromes which must always be looked out for. Our case however defied any of these associations.

Upper lid colobomas can lead to severe eyeball morbidities including blindness and must be attended to promptly either by conservative or surgical approaches depending on the size of the coloboma, the associated ocular morbidities and the age of the patient. The prognosis of the pathology is fairly good depending also on the surgeon's expertise and time of presentation of the patient to the surgeon.

Keywords: coloboma, repair, penthouse.

Introduction

Coloboma is a Greek word meaning removal of a part by mutilation [1, 2]. Colobomas could be congenital or acquired [3]. Typical eye colobomas are as a result of the failure of the choroidal fissure or fetal cleft to close during the developmental stage of the fetus. Colobomas can cause visual morbidity depending on the structures involved and how significant the defects are. Colobomas of the lids are rare and could affect either the upper or the lower lids or both lids, may be unilateral or bilateral and may be isolated or syndromic [3]. The management of the lid colobomas could be challenging depending on the extent of the defect, the visual affectation, associated eye diseases and the expertise of the surgeon. We therefore, present a 16-year-old female with a congenital, isolated upper eyelid coloboma that was managed with satisfactory aesthetic result from our hospital.

Case report

A 16-year-old female presented in our clinic with a complaint of inability to close her right eye from birth. Pregnancy history was not available however; patient said she was told she was delivered at term with the defect. General examination revealed a generally stable patient

systemically. Maxillofacial examination revealed a full thickness defect of about one-third of the right upper eyelid (Fig. 1a and b)



Figure 1a. Coloboma of the upper eye lid



Figure 1b: Coloboma of the upper lid pre-op (down gaze). (Straight gaze)

with mildly hyperemic conjunctiva in the right eye, clear cornea, normal anterior chamber, brisk pupil, normal iris, clear lens, pink disc with flat and complete retina in the right eye. No abnormality was detected in the left eye. However, the unaided visual acuities of the right and left eyes were 6/9 and 6/5 respectively. The IOPs were within normal limits (14mmHg and 12mmHg in the right and left eye respectively). The extra-ocular motilities were normal in both eyes; however, the left eye demonstrated lagophthalmos (Fig. 2).



Figure 2. Lagophthalmos with corneal sparing pre-op

Based on history and clinical examination an impression of congenital isolated upper eyelid coloboma was made.

Laboratory investigations including; full blood count, Urea and Electrolyte and Urinalysis were all within normal limits. The patient was subsequently slated for primary closure of the upper lid defect. After routine cleaning, draping and infiltration of the defect with 2% xylocaine local anaesthesia with 1 in 100,000 units of adrenaline, the defect was de-epithelized and fashioned into a pent-house shape. Closure of the resultant defect was done in layer with 6/0 vicryl suturing for the inner lamella and 5/0 black silk suture for the outer lid lamella. The upper lid margin was anchored to the cheek skin as frost suture with uneventful recovery; silk sutures were removed seven days post-operatively (Fig. 3).



Figure3. Direct repair of coloboma- 24 hours post-op

The eye was padded for 24 hours and removed while the frost suture was left in situ for 72 hours only. Three weeks post –operatively patient was able to close and open the eyes with no lagophthalmos, however there was a drooping of the eye lid (Fig. 4) probably from the resolving lid edema inducing mechanical ptosis.



Figure 4. Near normal (mildly drooping) upper lid -3 weeks post-op

Discussion

The first description colobomas was by Jacques Guillemeu in 1585, thereafter John Mustarde observed that upper lid coloboma may be an isolated condition, associated with other facial anomalies or associated with some syndromes [4]. Colobomas of the eye ball and lids are either congenital or acquired. The typical isolated colobomas of the eyes are rare. The incidence of the typical eye colobomas is between 0.5-0.7 in 10,000 births [2]. The lid colobomas are commoner in the congenital colobomas. The mutation in the PAX2 gene seems more implicated in congenital eye colobomas. Colobomas of the eye could involve virtually all eye tissues including the lids, the iris, the choroid, the retina and the optic nerve. Acquired colobomas can be secondary to either surgical or non-surgical traumas.

Upper lid colobomas occur as a result of the failure of the lateral and nasal frontonasal mesenchymal processes of the optic cups of the optic vesicles to fuse together during the developmental stage of the embryo resulting in loss of the tarsal plate and muscularis layer of the upper lid. The lower lid colobomas are in turn as a result of the failure of the nasal and the lateral processes of the maxillary mesenchymal process to fuse together with the resultant loss of the lower lid tarsal plate and the muscular layer [5].

Typically eye ball colobomas hardly cause significant visual impairment except where there is significant colobomas involving a large portion of the retina and the optic nerve. The lid colobomas pose more challenges to the eyes. There is more tendency of losing the entire eye ball from exposure keratopathy.

The severity of eye morbidity due to upper lid colobomas is dependent on the grade of the coloboma. Nouby classified upper lid colobomas into 5 grades [6, 7] as follows:

- Grade 1- Coloboma without cryptophthalmos
- Grade 2- Coloboma with abortive cryptophthalmos
- Grade 3- Coloboma with complete cryptophthalmos
- Grade 4- Classical cryptophthalmos with absence of all eye lid structures and complete coverage of the eye by skin

- Grade 5- Severe cryptophthalmos with severe deformity of the nose and ectropion of the lip

Nouby suggested that upper lid colobomas, cryptophthalmos and facial deformities could be one anomaly [6]. Our case report did not have any associated facial deformities.

Lid colobomas ideally are not sight threatening except where the cornea is compromised as in very severe colobomas. Our patient only exhibited a mild visual drop of 6/12 unaided perhaps due to amblyopia, as compared with the normal (left) eye that had a visual acuity of 6/5 unaided. Visual loss in colobomas is difficult to resolve depending on the extent of damage.

Of all colobomas of the eye the lid colobomas are the commonest. The lid colobomas could either be acquired or congenital. A study conducted in the United States sometime revealed that of the 26 cases reviewed with congenital colobomas the upper lid colobomas were the commonest type of congenital colobomas [2]. Most upper lid colobomas are associated with cryptophthalmos and thus syndromic. The case presented here was isolated with no associated eye or systemic findings on gross examinations.

Syndromes associated with upper eye lid colobomas include; Fetal alcohol syndrome[3], a rare autosomal recessive condition characterized by eyelid colobomas, cryptophthalmos, anophthalmia or microphthalmia, an aberrant hairline, a bifid or broad nasal tip, gastro-intestinal anomalies or omphalocele and anal stenosis, the Charge syndrome[8] consisting of coloboma, heart disease, atresia choanae, retarded growth and development, genital hypoplasia, ear abnormality with deafness, the Fraser Syndrome, an autosomal recessive disorder with life expectancy of less than one year with features of cryptophthalmos, ear and nose defects, skeletal defect like syndactyly, urogenital and central nervous system defects, laryngeal stenosis, lung and liver defects, the renal coloboma syndrome [4] also known as the papillorenal syndrome, an autosomal dominant syndrome due to mutation of the PAX2 gene characterized by optic nerve dysplasia and renal hypodysplasia with eye findings of microcornea, retinal coloboma, scleral staphyloma, optic nerve cyst and pigmentary macular dysplasia, wide excavated dysplastic optic disc with radial retinal vessels emerging from the disc margins directly, referred often to as the morning glory anomaly or the optic nerve coloboma and the Manitoba oculotrichoanal syndrome [9] which is a bilateral eyelid coloboma syndrome with bifid nasal tip hydrometrocolpos and vaginal atresia.

Treatment of colobomas may be conservative or surgical. The position, severity of the coloboma, associated morbidities and syndromic involvement will determine the time and the type of intervention. Conservative or medical intervention include the use of lubricants, artificial tears, humid chambers and nocturnal padding [4, 7, 10] all in a bid to avoid dry eye associated problems. The surgical modalities [4, 9, 10] depend on the size and the type of the coloboma. Defects of about 1/3 or less can easily be closed by direct closure using the 2-layered approximation of the tarsus and the skin, after de-epithelising the edges of the defects[4, 9, 10]. This was the mode of treatment in the presented case here since the defect was about a third of the whole lid. Lateral cantholysis may be done in difficult cases [4, 10, 11].

For defects of 40% and 50% a 2-staged surgery is advocated [7, 12, 13]. The modified Cutler-Beard [7, 12, 13]. procedure in which the tarso-conjunctiva of the lower lid is fashioned to fill the upper lid defect while a free retroauricularis or the sliding skin grafts are used to cover the raw tarso-conjunctival surface for defects that are between 40% and 50%, while defects greater than 50%, the Tenzel or the modified Tenzel semi-circular lateral canthal flaps are used [7, 12, 13]. Other modalities that are applied in such huge defects include the Lateral Modified Hughes procedure and the full-thickness Lid rotation Flaps [7, 12, 13]

Conclusion

The upperlid colobomas are rare and usually associated with cryptophthalmos. The colobomas can either affect one or both eyes. They can be associated with life threatening syndromes which must always be looked out for. Our case however defied any of these associations.

Upper lid colobomas can lead to severe eyeball morbidities including blindness and must be attended to promptly either by conservative or surgical approaches depending on the size of the coloboma, the associated ocular morbidities and the age of the patient. The prognosis of the pathology is fairly good depending also on the surgeon's expertise and time of presentation of the patient to the surgeon.

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