Surgical Management of Congenital Eyelid Coloboma

Michael Patipa, M.D.
Robert B. Wilkins, M.D.
Kurt W.L. Guelzow, M.D.

SUMMARY

Congenital eyelid colobomas are a partial or total absence of eyelid structures. The degree of severity determines the surgical techniques employed for repairing the eyelid. We feel that early surgical treatment reduces the risk of ocular scarring with satisfactory results. We present four cases of congenital upper eyelid colobomas of differing severity and discuss surgical approaches to these lid abnormalities.

INTRODUCTION

A coloboma of the eyelid is defined simply as an eyelid defect. This defect usually represents an absence of all the lid structures — skin, orbicularis, tarsus, conjunctiva, lashes, and glands. More rarely, the defect is partial or can be more extensive with the abnormality continued to the orbital margin. The surgical technique utilized to repair a coloboma depends upon the severity of the defect. We will discuss four surgical approaches to differing degrees of upper eyelid colobomas as we have utilized them in our clinical experience.

CASE I

This one-month-old female (M.H.) was evaluated for a coloboma of the right upper eyelid. She was the product of a normal pregnancy and delivery. The family history included two siblings with preauricular appendages, but with no other ocular or congenital stigmata.

Our initial examination revealed that the coloboma involved 30% of the right upper eyelid, with a small soft tissue mass at the superior edge of the defect (Figure 1). The rest of the ocular exam was normal. Bilateral preauricular appendages were present, but no other congenital abnormalities were noted. The patient was treated with a lubricating ointment, and surgery was planned for age four months. The cornea remained clear during this preoperative period.

The patient underwent a successful repair of the coloboma at the age of four months. The rectangular defect was converted to a pentagonal wedge. The soft tissue mass was in the resection. The lid margin defect was then closed in three layers. The mass was sent for pathologic evaluation and was found to be normal striated muscle. Evaluation over the next two years revealed an excellent cosmetic and functional surgical result with good lid mobility, normal lid margin configuration, and a clear cornea (Figure 2).

CASE II

This male child (R.C.A.) first was seen at age two weeks for evaluation of bilateral upper lid colobomas. He was the
product of an uncomplicated pregnancy with the exception of a premature birth. The family history was negative. Figure 3 illustrates the colobomas involving approximately 35% of both upper eyelids. There was no associated corneal exposure at that time and no other congenital defects. The baby was treated with a lubricating ointment three times daily and followed closely at six-week intervals.

At age 13 months, the child was noted to have bilateral exposure keratitis with corneal staining. We undertook surgical correction of the colobomas at that time to prevent jeopardizing the corneas.

The defects in both upper lids measured approximately 10 mm horizontally. The coloboma of the right upper eyelid was corrected by primary closure after conversion of the defect to a pentagonal wedge with a razor blade knife. A lateral canthotomy with cantholysis of the superior limb of the lateral canthal tendon was necessary to relieve tension on the lid margin. The defect was closed in three layers. Three 6-0 black silk sutures were used to reapproximate the lid margin.

Two weeks later, the left upper eyelid defect was corrected in a similar fashion, utilizing the lateral canthotomy with superior limb cantholysis for relaxation of the lid tissue and permitting closure of the defect.

The postoperative course was uneventful. Figure 4 demonstrates the result three years postoperatively. The child has no further difficulty with corneal exposure.

CASE III

This 10-week-old female child (C.R.) was seen for evaluation of a coloboma of the right upper eyelid. She was the product of a normal pregnancy and delivery. Her twin was normal. The family history was negative. A coloboma involving 40% of the right upper lid was present. She had been receiving lubricant ointment in the right eye since birth.

There was a small tag of lid tissue nasal to the defect. The superior fornix was obscured by a dense symblepharon which extended from the coloboma to the superior limbus and continued as a pannus over the superior cornea. The globe was hypertropic with mechanical restriction to downgaze. The right orbital rim was absent. The lower lid was normal. There was no corneal staining.

The risk of corneal exposure and the restricted motility predisposing the eye to develop an amblyopia prompted early surgery. In order to prevent extended occlusion of the eye, a lateral canthotomy with cantholysis of the superior limb of the lateral canthal tendon together with a semicircular flap temporally were used to approximate the coloboma margins. The symblepharon was excised from the superior limbus, and full motility was restored on forced ductions. The defect margins were freshened with a razor blade knife and then closed in three layers with 6-0 silk sutures.

The one-year postoperative course has been uneventful. A residual hypertropia is being treated with patching. The palpebral fissure width is approximately 9 mm and appears symmetric to the other eye. The levator function appears normal.

CASE IV

This seven-month-old female patient (I.R.R.) presented with an almost total absence of her left upper eyelid. She was the product of a normal pregnancy and term delivery. Family history was noncontributory. Three previous opera-
tions had been performed on the left upper eyelid prior to our
evaluation in attempts to protect the exposed cornea.

Ophthalmologic examination revealed a normal right eye.
The left upper eyelid consisted of a 2-mm tag of tissue at the
lateral canthus. The eyebrow was absent. The superior
fornix was obliterated by tissue of mucous membrane
consistency which extended from the coloboma to the
superior aspect of the globe. A dense epithelial vascular
pannus involved the upper two thirds of the cornea. That
portion of the cornea beneath the lower lid was clear. The
lower eyelid appeared normal. Gross motility examination
revealed a 35 prism diopter left esotropia and a slight left
hypertropia (Figures 5 & 6).

An examination under anesthesia revealed a normal
superior punctum. There was no upper eyelid tissue
between the punctum and the tiny lateral 2-mm tag of
tissue. The fundus could not be evaluated in detail through
the hazy cornea.

It was felt that a Cutler-Beard bridge flap from the left
lower lid would adequately close the large eyelid defect.

The procedure was performed in the classic fashion. The
tissue beneath the superior orbital rim was divided into
three layers and the edges were freshened. The tissue on
the posterior margin of the coloboma was separated
carefully from the globe at the limbus. No underlying sclera
defect was seen. This mucous membrane was then sutured
to the conjunctiva from the lower lid utilizing a running 6-0
plain catgut suture. The lower lid orbicularis muscle layer
was sutured to the layer felt to represent remnants of the
levator muscle with 5-0 gut sutures. The skin was closed
routinely with 6-0 black silk sutures. All tissues appeared
well vascularized at the end of the procedure.

The patient was followed at six-week intervals, and after
six months the second stage of the Cutler-Beard procedure
was performed. The bridge flap was released after determin-
ing the point of incision to obtain a symmetrical upper eyelid.
The postoperative course was uneventful and the patient
now has adequate corneal protection. At the last examina-
tion, four and a half years postoperative, the upper eyelid
has excellent color and configuration. There is a 4-mm
palpebral fissure with poor levator function (Figure 7).
Further procedures are contemplated to improve the
residual ptosis.

DISCUSSION

Etiology: Numerous theories for the development of
eyelid colobomas have been proposed through the years.
The various hypotheses have been divided into basically
two categories: hereditary and external or mechanical
origins.

Clinical Findings: Although there are many locations for
eyelid colobomas, the most common position is at the
junction of the medial and middle thirds of the upper lid. No
lid appendages or accessory structures usually are seen
within the colobomas.9,10 Associated orbital and ocular
anomalies include eyebrow defects, dermoid tumors or
dermolipomas of the lid or cornea,9,10 cutaneous bridges from
the lid to the cornea, obstruction of the nasolacrimal drainage
system, malformation of the canule, sym-
blepharon from the defect to the globe,9 corneal opacity, iris
abnormalities (corectopia and coloboma), anterior polar
cataract, and dislocation of the lenses and coloboma of the
choroid.11
Case I displayed a small mass of striated muscle tissue at the superior edge of the coloboma. Case II exhibited no associated ocular or orbital abnormalities. Case III exhibited adhesions from the defect to the globe with restricted motility and also absence of an upper orbital rim. Case IV represented a much more severe lid coloboma with numerous associated findings including absence of eyebrow, extensive synblepharon, and a large corneal opacity felt to be scarring secondary to long term exposure. Intraocular abnormalities could not be determined due to the condition of the cornea.

Regional defects also occur in association with colobomas. Goldenhar's syndrome exhibits eyelid colobomas in association with other aspects of oculoauriculovertebral dysplasia including preauricular appendages.12 Treacher-Collins syndrome or mandibulofacial dysostosis consists of lateral lower lid colobomas, macrostomia, hypoplasia of the facial bones and other associated anomalies.11 Congenital colobomas also have been reported to occur in association with cleft lip and palate.12,14 Case I had preauricular appendages. Case II was noted to have an umbilical hernia. Cases III and IV displayed no distinct defects.

Clinical Approach: The clinical approach to eyelid coloboma repair is dictated by the severity of the individual situation; this in turn is generally a reflection of the location and size of the deformity. Lower lid colobomas do not require emergency measures as often as those of the upper lid, for they will rarely jeopardize the corneal integrity.

We no longer wait beyond six months of age to correct colobomas as was done in the early Case II. The possibility for the development of corneal exposure and scarring moves one to action more quickly. The potential for this problem is obviously greater when close follow-up cannot be guaranteed or when adequate examination of the cornea is difficult in the uncooperative child. One may see corneal involvement only after it becomes extensive. The psychological trauma to the patient and family during an extended period of delay also must be considered.

In our experience, eyelid colobomas are generally not as large as they appear clinically when the margins of the defect are placed under normal tension. Therefore, a less difficult repair can be anticipated than might be expected initially.14

Surgery was performed on Cases I and III at age four months without waiting for exposure keratitis to develop. Therefore, these cases reflect more accurately our present surgical approach. The procedures were tolerated well, and satisfactory cosmetic and functional results were attained. Case II was followed for 13 months, until exposure keratitis was no longer controlled by conservative methods, at which time surgery was done. Case IV was corrected surgically at age seven months, immediately after our initial evaluation, in order to prevent further corneal damage and possible loss of the eye.

After the decision is made to operate, one must decide on a mode of treatment which will fit the given case. For small defects involving less than 30% of the lid, a primary closure usually can be performed with good results. The edges are freshened, the defect is converted into a pentagonal wedge, and a three-layer closure is completed with three 6-0 silk sutures.10-12

Excess tension on the wound margins can result in a wound dehiscence or in ptosis secondary to a tethering effect on the taut lid. This becomes a problem in closing defects involving between 30% and 50% of the lid. Many times, as in Case II, in which 35% of the lid margin was involved, a lateral canthotomy with appropriate cantholysis must be performed to obtain adequate relaxation of the lid margin.12 Additional relaxation can be obtained with a semicircular flap as in Case III.2

If the defect involves greater than 50% of the lid margin, as in Case IV, one must be prepared to perform a major lid reconstruction utilizing one of a number of techniques described in the literature.11 Bridge flaps after Cutler and Beard8 or Hughes10 can be employed. The Cutler-Beard technique obtained very satisfactory results in a difficult situation in Case IV. The attendant risk of extended occlusion and development of amblyopia must be recognized with major lid reconstructions in a pediatric population. Therefore, a decision regarding the visual potential of the eye should be made prior to employing these techniques for lid reconstruction.

Congenital eyelid colobomas vary in severity. Early surgical intervention is advocated if quality of pediatric anesthesia and familiarity with lid anatomy are excellent. Satisfactory cosmetic and functional results can be obtained if careful preoperative and intraoperative planning are employed. This approach avoids common complications, be they ocular or psychological.

REFERENCES
CONGENITAL EYELID COLOBOMA


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