European Journal of Ophthalmology / Vol. 13 no. 8, 2003 / pp. 714-717

SHORT COMMUNICATION

Case report

Unilateral giant coloboma of the upper eyelid associated with other congenital anomalies (33 years follow-up of surgical repair)

G. SCASSELLATI-SFORZOLINI[1], B. SCASSELLATI-SFORZOLINI[2]

[1]University Eye Clinic of Bologna and Casa di Cura Villa Verde, Bologna
[2]Department of Ophthalmology, University of Ancona - Italy

PURPOSE. *To describe a case of congenital unilateral giant coloboma and its successful surgical repair with 33 years of follow-up.*
CASE REPORT. *A 6-year-old boy presented with a congenital unilateral giant coloboma of the right upper eyelid associated with madarosis of the eyebrows, microphthalmos, dystopia of the hair, and coloboma of the apex of the nose. The patient underwent surgical repair of the multiple anomalies in different steps.*
DISCUSSION. *A multiple-step, two-layer technique for the reconstruction of the right upper eyelid was performed in a 6-year-old boy with congenital unilateral giant coloboma associated with multiple ocular and facial anomalies. After 33 years of follow-up, the cosmetic results are excellent, although it has not been possible to preserve the visual function of the right eye, which had to be enucleated.* (Eur J Ophthalmol 2003; 13: 714-7)

KEY WORDS. *Congenital dystopia of the hair, Congenital madarosis of the eyebrows, Giant congenital coloboma of the eyelid, Microphthalmos*

Accepted: xxxxxxx

INTRODUCTION

Anomalies of development of the eyelids occur between the seventh and ninth week of gestation (1). The extreme form of unilateral coloboma (giant coloboma or ablepharia) of the upper eyelid is rare. The coloboma has a quadrangular shape, the tarsus is absent, and a remnant is usually present in the lateral portion of the eyelid. This anomaly can be isolated or associated with epibulbar dermoids, microphthalmos, cryptophthalmos, orbitofacial clefts, other facial anomalies, syndactylism, genital abnormalities, or renal aplasia, or can be a part of complex syndromes such as Franceschetti and Goldenhar syndromes (1-4). At birth, a dermatocele may cover the upper half of the cornea with a cutaneous epithelium, the superior rectus muscle is often absent, and Bell phenomenon does not occur. Therefore, the cornea inevitably ulcerates and the eye becomes blind if this condition is not treated soon after birth (2, 3).

We report an unusual case of giant unilateral coloboma of the upper eyelid associated with madarosis of the eyebrow and nasal coloboma. The patient underwent multiple surgeries and was followed for 33 years.

1120-6721/714-04$15.00/0

Case report

A 6-year-old boy was referred to one of us (G.S.-S.) in 1966 for the correction of a coloboma of the right upper eyelid (Fig. 1). Visual acuity was light perception in the right eye and 20/20 with spectacle correction in the left eye. The patient had a history of frequent and painful corneal ulcers. No family history of eye disease was present. Examination of the right periorbital region showed a 1-cm-long thin line of hair corresponding to the medial eyebrow. A giant quadrangular coloboma of the right upper eyelid with insertion of the skin of the forehead on the lower third of the cornea was evident. Only two remnants of the eyelid could be seen laterally (7 mm long) and medially (smaller) to the coloboma. Several lines of irregular eyelashes were present and the upper punctum and canaliculus were patent. A slight entropion of the inferior eyelid with a wedge of hair reaching the external third of the superior orbital arcade could be observed. A small coloboma with vascularized tip of the nose and an ogival palate were also present.

Slit-lamp examination of the right eye revealed a microphthalmic eye with xerosis and marked neovascularization of the inferior third of the cornea, modest conjunctival hyperemia, and iris heterochromia. Ocular tension was 14 mm Hg in the left eye and digitally normal in the right. The fundus of the left eye was normal, whereas the fundus of the right eye could not be explored. Plain X-rays of the head revealed no abnormalities.

On June 10, 1966, a rectangular cutaneous flap was delineated (Fig. 2a) and released from its deep muscular connections with the levator muscle up to its insertion on the cornea; the levator muscle was then grasped with three catgut (6/0) stitches (Fig. 2b). No tarsus or fibers of the orbicularis muscle were found. The thin cutaneous flap was passed underneath the levator muscle to cover the bulb (Fig. 2c). The lateral palpebral remnant was split and the incision extended temporally. The two flaps were then sutured together superiorly (to re-establish the upper fornix) and to the lateral edge of the medial remnant of the upper eyelid using catgut (6/0) (Fig. 2d). The levator muscle was passed between the two layers and sutured with a central and two lateral stitches with catgut (6/0). The two lateral stitches were also used to suture a thin cartilaginous flap removed from the postauricular cartilage and inserted underneath the levator muscle (Fig. 2e). The extensive loss of substance was covered with a thin dermo-epidermal flap removed from the auricle and the postauricular skin. A wet and squeezed cotton cylinder was placed over the virtual palpebral sulcus and the operating field was then covered with a second moist cotton cylinder (Fig. 2f).

On September 21, 1966, reconstruction of the right eyebrow with a hair-bearing rotate peduncle flap using a dental paste mould and removal of a full thickness triangle of dystopic hair was performed, followed by a skin graft.

On November 6, 1967, deepening of the superior fornix with a flap of labial mucosa was performed.

On September 11, 1968, a section of a small symblepharon bridle at the superior fornix. Transposition of a fascicle of the marginal orbicularis muscle from the lower to the upper eyelid to improve the closure of the upper eyelid, increase the thickness of the palpebral margin, and reduce the inferior entropion was done.

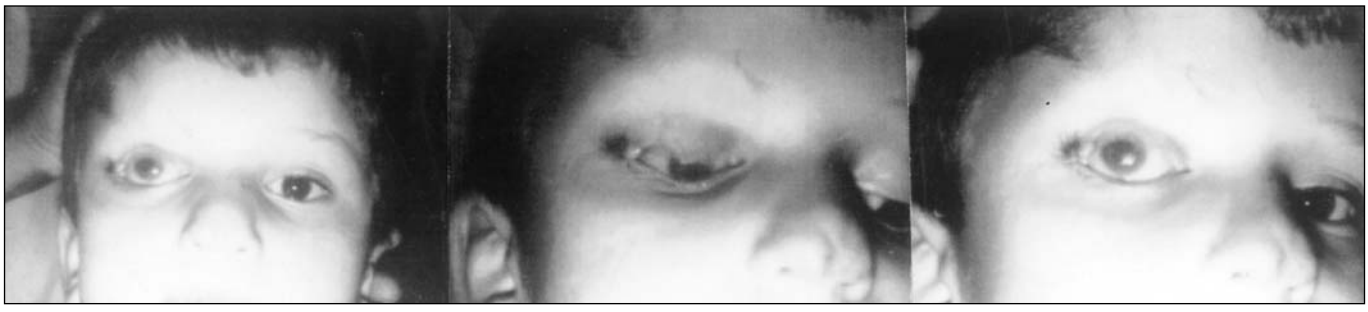

Fig. 1 - *Six-year-old boy with congenital unilateral coloboma of the upper eyelid associated with madarosis of the eyebrows, microphthalmos, dystopia of the hair, and coloboma of the apex of the nose.*

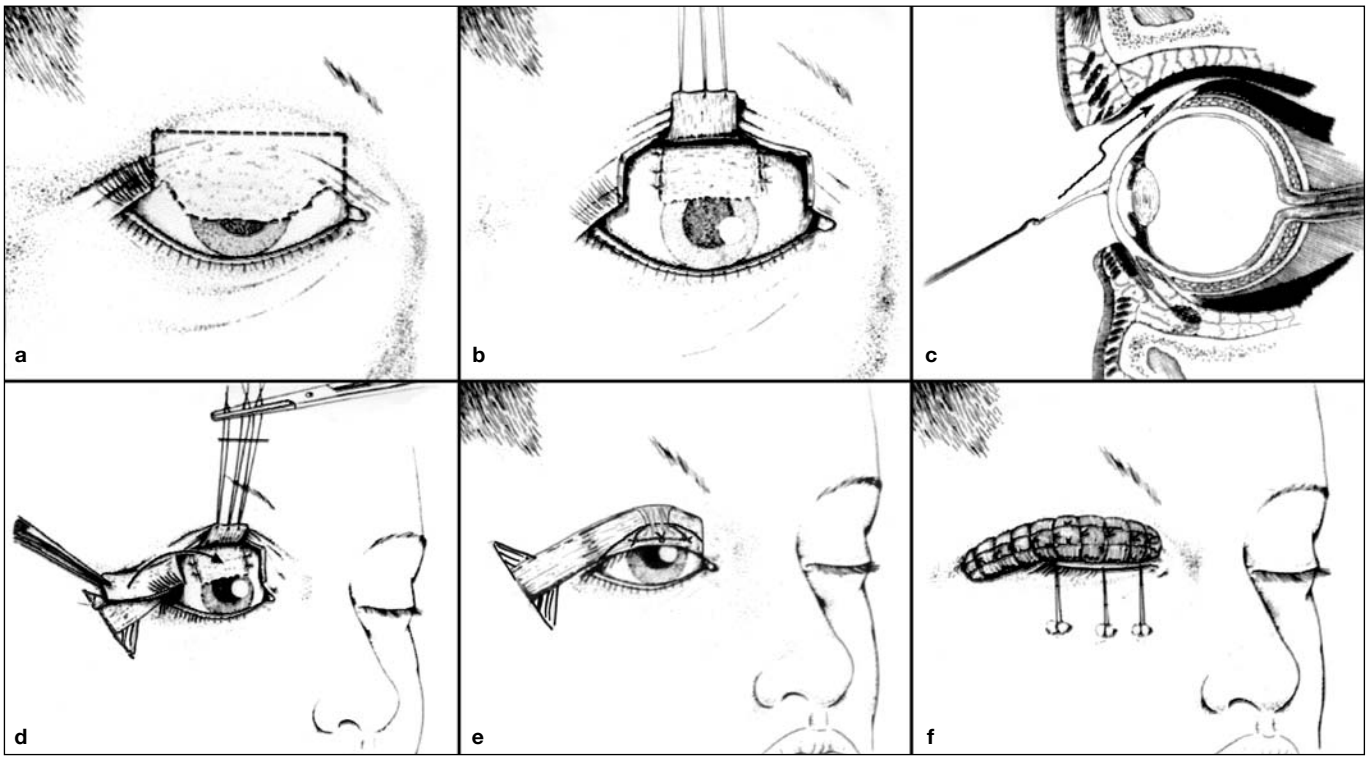

Fig. 2 - *Surgical technique of reconstruction of colobomatous upper eyelid and madarosis.* **a)** *A rectangular cutaneous flap is delineated.* **b)** *The flap is released from its deep muscular connections with the levator muscle up to its insertion on the cornea; the levator muscle is grasped with three catgut (6/0) stitches.* **c)** *The thin cutaneous flap is passed underneath the levator muscle to cover the bulb.* **d)** *The lateral palpebral remnant is split and the incision extended temporally. The two flaps are then sutured together superiorly and to the lateral edge of the medial remnant of the upper eyelid.* **e)** *The levator muscle is passed between the two layers and sutured with a central and two lateral stitches. A thin cartilaginous flap, removed from the postauricular cartilage, is inserted underneath the levator muscle. The loss of substance is covered with a thin dermo-epidermal flap removed from the auricle and the postauricular skin.* **f)** *A wet and squeezed cotton cylinder is placed over the virtual palpebral sulcus and the operating field is covered with a second moist cotton cylinder.*

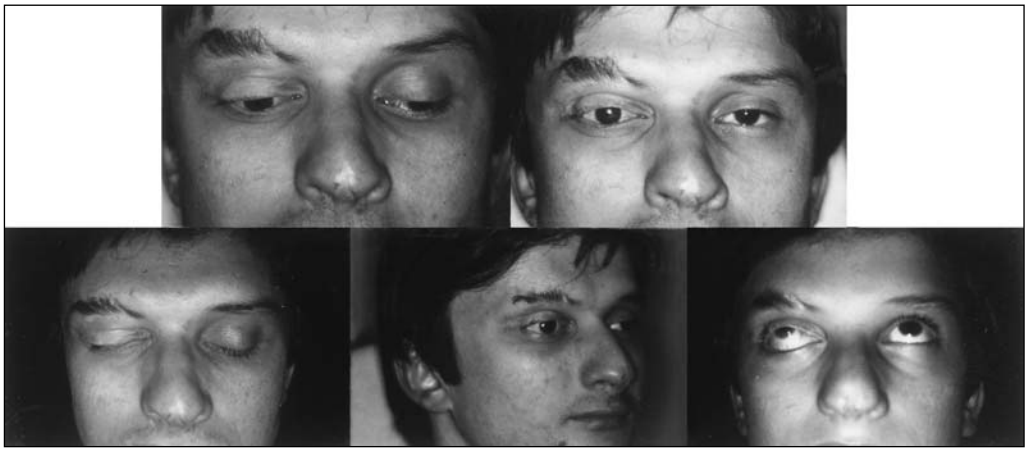

Fig. 3 - *Same patient as Figure 1 at age 39 years.*

On October 23, 1975, correction of the coloboma of the apex of the nose was performed (Prof. Cavina, head of the Plastic Surgery Department of the S. Orsola Hospital, Bologna).

On December 4, 1986, the patient underwent evisceration of the right eye and implant of an acrylic prosthesis according to Fox. A few months later, an esoprosthesis was applied.

On March 17, 1999, the latest examination (Fig. 3), the cutaneous grafts were isochromic. The palpebral sulcus and fold of the right upper eyelid were symmetric with the contralateral ones. Alopecia of the eyelashes of the upper eyelid could be observed. Both the closure and the elevation of the upper eyelid were satisfactory. The anophthalmic socket was wide and the prosthesis presented a good motility. Two thin linear scars could be seen temporally and at the apex of the nose.

DISCUSSION

We report a case of giant coloboma of the upper eyelid of the right eye, madarosis of the eyebrow, dystopia of the hair, and microphthalmos associated with a vascularized coloboma of the tip of the nose and ogival palate.

Two surgical techniques are usually employed to repair a giant coloboma of the eyelid: the full thickness operation of Cutler-Beard (5) (when the aponeurosis of the levator muscle is preserved) and that of Mustardé (6) (when the coloboma is subtotal). Both procedures utilize a full thickness segment of the lower eyelid, which is sutured into the defect in the upper eyelid. Complications are not rare with these techniques. In order to try to avoid some of the complications, the original Cutler-Beard method has been modified by several authors (7-10).

In our case, a multiple-step, two-layer technique for the reconstruction of the upper eyelid was used. The purpose of this approach was to give support and motility to the upper eyelid, without weakening the inferior one. The superficial half of the lateral palpebral stump and adjacent temple were utilized to restore the deep layer of the upper eyelid, while a small autoplastic cartilaginous flap and the levator muscle were superimposed to it. These layers were covered with a thin dermo-epidermal flap removed from the auricle and the postauricular skin (superficial layer). The eyebrow was reconstructed with a pedunculated hair-bearing flap and a no stitch technique. The appearance of the patient was further improved with enucleation of the right eye and acrylic implant. After more than 30 years from the first operation, the cosmetic results are remarkable. Unfortunately, the visual function of the right eye could not be preserved, as the cornea was already completely opaque and vascularized when the patient was referred to us.

Reprint requests to:
Prof. Guidobaldo Scassellati-Sforzolini
Viale Oriani 31
40137 Bologna, Italy
e-mail: bscassellati@hotmail.com

REFERENCES

1. Chaine G, Aufier JL. Affections génétiques des paupieres. In: Encyclopédie Médico-Chirurgicale-Ophthalomologie. Vol II. Paris: Ed. Techniques; 1999: 21 100 A10.
2. Rougier Y, Tessier P, Hervouët, et al. Chirurgie plastique orbito-palpébrale. Paris: Masson; 1977.
3. Duke Elder S. System of ophthalmology. London: Kimpton; 1966: Vol III, part 1, 231-4, 298-300; Vol III, part 2, 835-40, 878.
4. Nouby G. Congenital upper eyelid coloboma and cryptophthalmos. Ophthal Plast Reconstr Surg 2002; 18: 373-7.
5. Cutler NL, Beard C. Method of partial and total upper lid reconstruction. Am J Ophthalmol 1955; 39: 1-7.
6. Moustardé JC. Repair and reconstruction of the orbital region. A practical guide. Baltimore: Williams & Wilkins; 1966.
7. Hübner H. Totalersatz des Oberlides. Klin Monatsbl Augenheilkd 1976; 169: 207-11.
8. Putterman AM. Viable composite grafting in eyelid reconstruction. Am J Ophthalmol 1978; 85: 237-41.
9. Wesley RE, Mc Cord CD Jr. Transplantation of eyebank sclera in the Cutler-Beard method of upper eyelid reconstruction. Ophthalmology 1980; 87: 1022-8.
10. Kadoi C, Hayasaka S, Kato T, et al. The Cutler-Beard bridge flap technique with use of donor sclera for upper eyelid reconstruction. Ophthalmologica 2000; 214: 140-2.