Manitoba Oculo-tricho-anal Syndrome: Complex Surgical Correction of an Extensive Upper Lid Coloboma with Congenital Symblepharon

Abstract

Manitoba Oculo-tricho-anal Syndrome (MOTA) is a rare autosomal recessive disorder associated with eyelid coloboma, cryptophthalmos, aberrant anterior hairline, bifid nose, and abdominal wall abnormalities. The name derives from the aboriginal Oji-Cree population of the Island Lake region of Manitoba, Canada, where it was first described. The genetic mutation involves FREM1, a protein involved in ensuring the integrity of basement membranes. The management of associated severe eye and eyelid malformations consists primarily of staged surgical interventions tailored to the patient's needs. We describe a unique surgical approach in a patient with this condition. The collection and evaluation of protected patient health information was HIPAA-compliant.

CASE REPORT

A 1-month old male was noted to have hypertelorism, an 18mm right upper lid coloboma with only a residual stump temporally associated with an extensive superomedial symblepharon obscuring the pupil in the context of MOTA. Surgical steps included reconstitution of the eyelid's anatomical landmarks with dissection of the symblepharon invading the opacified cornea, freeing the pupillary axis, creation of a superior fornix, advancement of myocutaneous/periosteal flaps, and insertion of an implant to recreate the absent eyelid. This resulted in significant improvement of ocular surface protection, quality of life, and allowed for amblyopia treatment.

KEYWORDS: Manitoba Syndrome; MOTA; FREM1 mutation; lid coloboma; congenital symblepharon; eyelid reconstruction.

INTRODUCTION

Manitoba Oculo-tricho-anal (MOTA) Syndrome is a rare autosomal recessive disorder associated with eyelid coloboma, cryptophthalmos, aberrant anterior hairline, bifid nose, and abdominal wall abnormalities. The name derives from the aboriginal Oji-Cree population of the Island Lake region of Manitoba, Canada, where it was first described. The genetic mutation involves FREM1, a protein involved in ensuring the integrity of basement membranes. The management of associated severe eye and eyelid malformations consists primarily of staged surgical interventions tailored to the patient's needs. We describe a unique surgical approach in a patient with this condition. The collection and evaluation of protected patient health information was HIPAA-compliant.

FIG. 1. Manitoba Syndrome: clinical features and initial approach.
A. Clinical photograph of the patient with extensive right upper lid coloboma with a minimal temporal stump, 6 hours of limbal invasion by the symblepharon, and an absent superior fornix. B. Initial conservative management with intensive ocular lubrication with ointment in conjunction with Vaseline on the skin and saran wrap cover to prevent further corneal dessication (acting as a humid camera).
anterior lamella (Figure 1C). Although this procedure was initially successful, the continuous adhesions of the superomedial fornix and gradual coverage of the pupil required further intervention nine months later.

Subsequently, the recurrent superior symblepharon was divided and an oral mucosa graft was positioned over the globe. The posterior lamella was reinforced with a tarsoconjunctival graft harvested from the contralateral side medially and a periosteal flap from the right lateral orbital rim. A bipedicled advancement myocutaneous flap just below the inexisten brow covered these grafts, and a full-thickness skin graft was inserted at the donor site. The patient was well and stable for fourteen months; however, adjusting polycarbonate glasses became quite challenging given his midfacial abnormalities and the child suffered a significant fall, which led to partial necrosis of the reconstructed eyelid.

The last procedure involved enlarging the superior fornix to allow for insertion of an oral mucosa graft and a conjunctival graft harvest from the ipsilateral inferior fornix (Figure 2A). Since we already had a strong anterior lamella, this was gently divided to allow the insertion of a thin porous spacer (Medpor®; Stryker, Oculoplastic Implants), which was trimmed to provide structural stability and a free pupil (Figure 2B). Currently, he has a free visual axis and continues using lubricating ointment only at night.

**DISCUSSION**

Congenital lid coloboma may occur in isolation or as part of a multitude of syndromes such as MOTA, BNAR (bifid nose, with/without anorectal and renal malformations), or Fraser. This occurs as a result of abnormal formation during development of lid folds and may cause severe disfiguration and visual impairment. Ophthalmologists are responsible for its diagnosis and management given the increased risk of ocular surface exposure, limited amount of limbal cells, and risk of amblyopia. Corrective measures such as protective glasses and corneal transplants are fundamental where it can aid in visual development. Although our patient had minimal regenerative corneal capability, and could not be refracted due to extensive scarring and opacity secondary to corneal exposure, vigilant monitoring of the cornea and pupil along with early surgical intervention is mandatory to optimize visual capacity. Our patient is currently a candidate for a corneal transplant.

We propose a staged methodical surgical approach during reconstruction of upper lid colobomas in patients with MOTA. Complex surgical repair has ranged from primary closure to a Mustarde-type switch flap with mucous membrane grafts to recreate a lid where there is insufficient conjunctival tissue. Although the type of reconstruction will depend on the severity of the coloboma and associated conditions, the combination of advanced myocutaneous flaps, periosteal flaps, oral mucosa and conjunctival grafts can offer favorable results while simultaneously improving cosmetic disfiguration, optimizing corneal protection, and offering a means to decrease the incidence of amblyopia. At the same time, it is vital to promptly reinforce the use of protective glasses. Had our patient not experienced a significant fall that resulted in necrosis of the reconstructed eyelid, further surgical intervention could have been spared.

**References**