

Treacher Collins Syndrome, Intellectual Disability, and Self-Inflicted Bilateral Traumatic Cataracts: A Case Report

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ABSTRACT

Introduction: Treacher Collins syndrome is a congenital craniofacial disorder with unique ocular signs and an increased risk of a variety of visual abnormalities, including strabismus and amblyopia. Although most patients with Treacher Collins syndrome are of average intelligence, it can also be present in patients with intellectual disabilities. Patients with autism spectrum disorder and/or intellectual disability are at risk for developing self-injurious behavior, which is often directed at the head and face. Traumatic cataracts are

one of the more serious injuries that can occur secondary to this behavior. When cataracts become visually significant, extraction is required. After cataract extraction, aphakia can be corrected with intraocular lenses, glasses, or contact lenses.

Case Report: A 6-year-old African American male with Treacher Collins syndrome, intellectual disability, hearing impairment, and autism spectrum disorder presented with bilateral traumatic cataracts secondary to self-injurious behavior. His vision declined rapidly as the cataracts progressed. His mobility became extremely limited and he was no longer able to fixate on faces. After cataract extraction, the patient was left aphakic due to an increased risk of complications from ongoing self-injurious behavior directed at the eyes and face. He was prescribed aphakic sports goggles due to these behaviors and his external ear malformation. He will be considered for secondary intraocular lens implantation in the future.

Conclusion: Patients with Treacher Collins syndrome and those who exhibit self-injurious behavior are at an increased risk for developing ocular abnormalities and should be monitored closely. For pediatric patients with cataracts, the decision whether or not to proceed with an intraocular lens insertion should be based on the patient's behaviors, visual demands, and age. When choosing a treatment approach, it is essential to evaluate the patient's lifestyle and visual demands, not just the diagnosis.

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INTRODUCTION

Treacher Collins syndrome is a congenital craniofacial disorder that affects the bones and soft tissue of the head and face. It has a prevalence of 1 in 50,000 live births and is due to genetic mutations in the *TCOF1*, *POLR1C*, and *POLR1D* genes.¹ There are numerous characteristic facial abnormalities associated

with this condition, including downward-sloping palpebral fissures, hypertelorism, and occasionally eyelid coloboma or madarosis of the lower lid. These patients are more likely to experience other visual abnormalities, such as strabismus and amblyopia, than are patients without the condition.² The majority of patients with Treacher Collins syndrome are of average intelligence, but the condition can also be present in patients with intellectual disabilities, as seen in this case.

Individuals with intellectual disabilities and autism often develop self-injurious behavior (SIB), which is repetitive, deliberate, and self-abusive. Between 25% and 35% of patients with autism and/or intellectual disability exhibit SIB at any given time.³ The self-abuse is often directed at the head and face, which puts the eyes at significant risk of trauma.³ While self-injurious behavior usually results in minor soft tissue damage, more serious injuries, such as traumatic cataracts, retinal detachment, and ocular perforation can occur.⁴

Visually-impairing cataracts in children require surgical excision due to the risk of visual deprivation amblyopia. After the cataract has been extracted, an intraocular lens (IOL) can be immediately implanted or the patient can remain aphakic and use either glasses or contact lenses. There are risks and benefits to each option, but numerous studies have shown that there is no significant difference in visual function when vision is corrected with an intraocular lens or with lenses. However, there is a higher rate of complications in those undergoing primary IOL implantation.⁵⁻⁹

As the prevalence of autism spectrum disorders continues to rise, it is essential that providers be aware of the potential ocular complications of self-injurious behavior. There are limited reports of traumatic cataracts secondary to SIB in the literature, and none of them involve a patient with a craniofacial disorder and the corresponding ocular characteristics. Patients with special needs and atypical craniofacial anatomy often have

unique demands that should be taken into account when choosing a treatment modality.

CASE REPORT

Chief Complaint

A 6-year-old African American male presented to the urgent care clinic with a chief complaint of sudden onset white pupil in the right eye. He was an established patient at the Illinois Eye Institute and had been seen for annual eye exams since infancy without any previous finding of leukocoria. Per the patient's mother, he had visited the emergency room one week prior for a subconjunctival hemorrhage in the right eye secondary to self-injurious behavior. She first noticed the leukocoria the day before the exam.

Medical History

The patient had been previously diagnosed with Treacher Collins syndrome (see Figure 1), autism spectrum disorder, intellectual disability, hearing deficiency, and self-abuse directed at the face. Surgical history included cleft palate repair, tracheostomy, and implantation of a gastrostomy tube. The patient was severely developmentally delayed and non-verbal. He was ambulatory, but almost completely uncommunicative. He did not speak, sign, or understand most spoken language or gestures. He was not taking any medications and did not have any allergies.

Per the patient's mother, the patient developed self-injurious behavior at the age of 3 years old. He would forcefully strike himself in the eyes and head, so he often wore arm restraints. When his arm mobility was limited, the patient would then resort to striking his head against nearby objects. He required frequent hospital visits because of this behavior. To decrease the severity and frequency of these injuries, the patient often



Figure 1: Facial characteristics of Treacher Collins syndrome

wore either a hockey helmet with a mask or arm restraints.

Ocular History

The patient's ocular history was remarkable for a longstanding intermittent right exotropia and moderate hyperopia and astigmatism. Glasses had previously been prescribed, but compliance was very poor as the patient constantly removed the glasses. The patient had no significant visual demands as he did not participate in any visual activities such as coloring, watching TV, pointing at objects, etc. Spectacle correction was discontinued the previous year and was to be reconsidered at a future date.

Family History

The patient's parental family history was unknown, as the patient was adopted. However, the patient had a twin brother who had been adopted by another family. Per the patient's mother, his brother did not display any of the medical conditions found in the patient.

Examination

Clinical examination was restricted due to limited communication, so objective testing was performed. The patient was unable to perform preferential-looking visual acuity testing, but he was able to fixate and follow with both the right and left eyes. He exhibited a 30 prism diopter intermittent right exotropia, but all other entrance testing was normal. Anterior segment evaluation revealed dense cortical cataracts worse in the right eye than the left eye, hypertelorism, and downward-sloping palpebral fissures (see Figure 2). Intraocular pressures were soft and equal via tactile evaluation. The posterior pole could not be viewed due to the cataracts and limited patient cooperation, so a B-scan was performed (see Figures 3a and 3b). The scan revealed intact retinas with vitreous opacities in both eyes.



Figure 2: Traumatic cataract OD

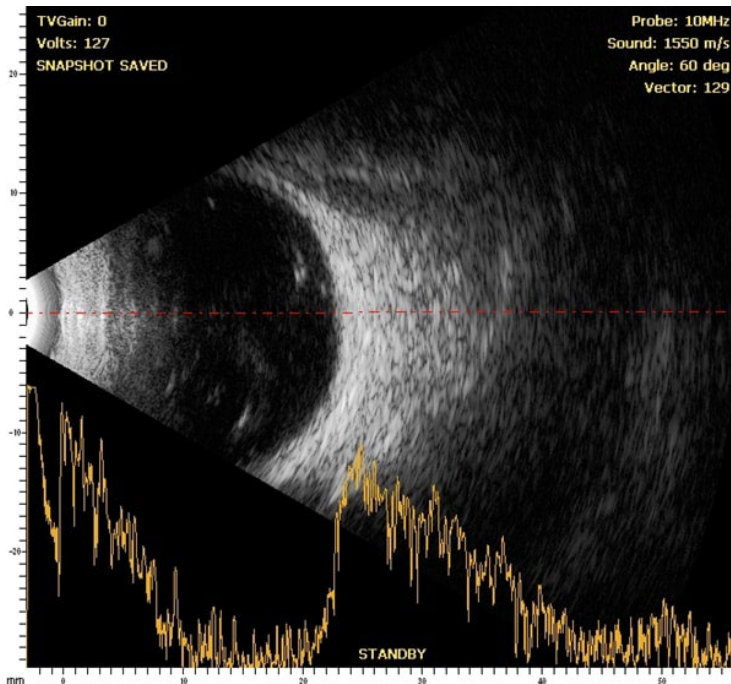


Figure 3a: B scan OD

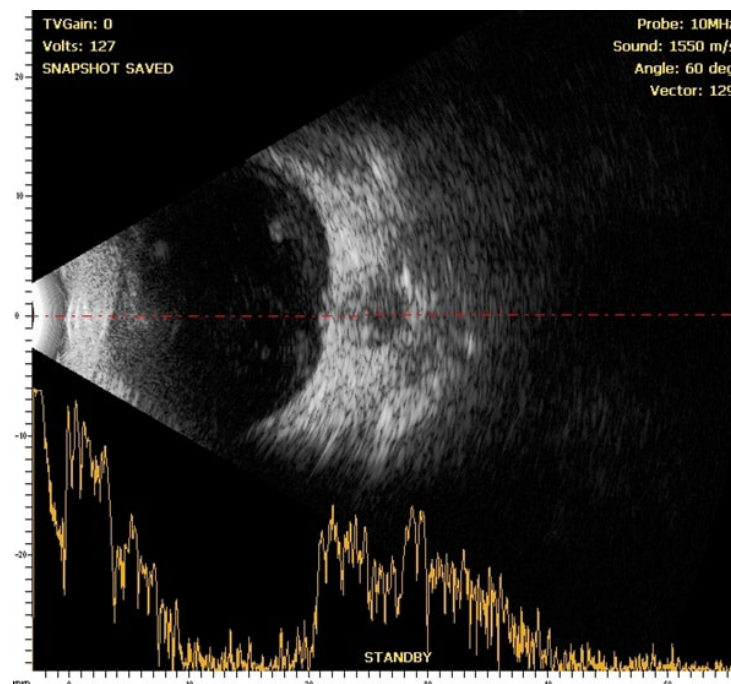


Figure 3b: B scan OS

The patient was referred to a pediatric ophthalmologist for further evaluation and he returned four days later for his consultation. Exam findings were largely the same, with no significant differences noted by the ophthalmologist. The patient was diagnosed with suspected traumatic cataracts in both eyes and cataract extraction without intraocular lens implantation was scheduled in six weeks.

However, the patient and his mother returned to the clinic five days later. The patient's mother reported a significant decline in the patient's vision and quality of life. His teacher expressed concern that he was bumping into chairs and desks at school and had difficulty navigating his environment. At home, the patient had ceased to make eye contact and had fallen down the stairs. These were all new concerns that had developed over the last few days.

Upon exam, the patient was unable to fixate and follow a target with either eye, but pupils were round and reactive. Examination revealed progression of the cataracts in both eyes, with a 4+ cataract in the right eye and a 3+ cataract in the left eye. The pediatric ophthalmologist was consulted, and surgery was rescheduled for the following week due to the patient's precipitous decline in functional vision.

The cataract in the left eye was extracted first, followed by the cataract in the right eye three weeks later. Intraocular lenses were not implanted due to concerns about perforation and dislocation secondary to ongoing head trauma. The patient was prescribed aphakic sports goggles with the potential for secondary IOL implantation or contact lenses at a later date. Unlike previous attempts at correction, he successfully adapted to the sports goggles and wore them nearly full-time. Visual acuity could not be determined using preferential looking cards, but he achieved fixate and follow vision in both eyes following the surgery. The patient's mother reported a significant improvement in mobility and functional vision.

He was once again able to safely navigate his environment and make eye contact.

DISCUSSION

Treacher Collins Syndrome

Treacher Collins syndrome is a craniofacial disorder also known as mandibulofacial dysostosis. It is a congenital genetic condition that affects the bone and soft tissue of the head and face.

Treacher Collins syndrome has an annual incidence of 1 in 50,000 live births and is due to mutations in the *TCOF1*, *POLR1C*, and *POLR1D* genes. Approximately 80-90% of cases are due to mutations in *TCOF1*.¹ Although it is a genetic disorder, only 40% of cases are inherited while 60% of cases are due to new mutations. Treacher Collins has an autosomal dominant inheritance pattern when it is transmitted through the *TCOF1* gene. The *POLR1C* gene transmits in an autosomal recessive pattern, and the inheritance of the *POLR1D* gene is unknown.¹

Signs of Treacher Collins syndrome are highly variable, ranging from severe to barely noticeable. Characteristic signs include underdeveloped facial bones, micrognathia, cleft palate, hearing loss due to underdeveloped ears, and difficulty breathing or swallowing due to irregularities of the larynx, pharynx, and neck muscles.² Patients with Treacher Collins syndrome are usually of average intelligence, with no intellectual disabilities or impairments.

Ocular signs of this condition include downward-sloping palpebral fissures, hypertelorism, and occasionally eyelid coloboma or madarosis of the lower lid. Patients with Treacher Collins syndrome are also at an increased risk of developing other visual abnormalities. Hertle, et al. found that 37% have strabismus, 27% have vision loss, and 58% of these patients exhibit significant refractive errors.¹⁰ Although it is a rare condition, providers must be aware of the presentation of Treacher Collins syndrome and its associated ocular findings.

Self-Injurious Behavior

Self-injurious behavior is defined as repetitive, deliberate, self-abusive behavior that is commonly seen in patients with intellectual disabilities (ID) and autism. Self-injurious behavior is seen more frequently in males with intellectual disabilities than in females.¹¹ These behaviors can include banging, hitting, and kicking of the head and body as well as biting, choking, pinching, poking, scratching, and self-restraint. The most common types of self-injurious behaviors are hitting and banging of the head, as seen in this patient.³

Numerous studies have shown a positive correlation between SIB, autism and/or intellectual disability, and communication deficits. One study found that 25% of children with ID exhibit self-injurious behavior.³ Another study of 152 adolescents on the autism spectrum found that over 35% were regularly self-abusive.¹² Self-injurious behavior is transient and is not diagnostic of either autism or intellectual disability. Since these behaviors can come and go throughout a patient's lifetime, not all patients with self-injurious tendencies exhibit these behaviors at any given time. One study estimated that up to 50% of patients with autism spectrum disorder are self-injurious at some point in their lives.¹³ Delays with expressive and receptive language skills are also associated with an increased risk of developing self-injurious behavior. For these patients, functional communication training may be beneficial and should be considered as a potential referral.¹⁴

Self-injurious behavior is also directly correlated with the severity of a patient's intellectual disability and autism symptoms. Of patients with profound and severe intellectual disabilities, between 25% and 15.5% exhibit SIB. Meanwhile, only 7% of those with moderate and 4% of those with mild intellectual disabilities are self-abusive.¹¹ For patients with autism spectrum disorder, the main risk factor for SIB during adolescence is

the severity of autism symptoms. Adolescents with better communication skills are less likely to exhibit self-injurious behavior, and cognitive development is considered protective against SIB in children.¹²

The majority of injuries sustained from self-injurious behavior are mild, soft tissue injuries such as excoriations and bruises. However, more severe injuries can occur, especially when force is directed at delicate tissues such as the eyes. The most common ocular injury from self-injurious behavior is corneal abrasion, but a study of children with intellectual disabilities and SIB found that 2.1% develop traumatic cataracts.⁴

Cataracts and Treatment Considerations

Traumatic cataracts can be caused by blunt trauma, penetrating injury, ionizing radiation, infrared radiation, or electric shock. Cataracts secondary to blunt trauma are the result of coup and contrecoup forces, which are generated by the initial blunt impact and the radiating shockwaves that follow. The exact pathophysiology is unknown, but it is believed that these forces result in either dysfunction of the anterior lens epithelium or rupture of the anterior lens capsule. Either of these injuries can result in lenticular edema and ultimately, opacification and development of a traumatic cataract.¹⁵

Progression of traumatic cataracts is highly variable. In a study of 137 children with a history of trauma, the average time to cataract development was six months. However, presentation ranged from one day to nine years. The density and severity of the traumatic cataracts also varied greatly.¹⁶

Cataract extraction is especially important in pediatric patients due to the risk of amblyopia. Generally, traumatic cataracts have a better prognosis than unilateral congenital cataracts because in most instances of pediatric traumatic cataracts, the patient has already developed accurate binocular fixation prior to the development of the cataract. Expected

visual outcome of traumatic cataracts varies greatly and depends on the onset, duration, and severity of the opacification as well as any associated comorbidities from trauma.¹⁶

Intraocular lens implantation is not always performed immediately following cataract extraction. Implantation of an intraocular lens at the time of cataract extraction is known as primary IOL insertion, while secondary IOL insertion is performed months or years after removal of the cataract. In these instances, the patient is left aphakic and vision is corrected with glasses or contact lenses until IOL insertion is performed.

In pediatric patients, there has been extensive research into the outcomes of primary IOL insertion and aphakia corrected with a contact lens or glasses. The Infant Aphakia Treatment Study compared contact lenses and intraocular lenses for correcting aphakia secondary to cataract extraction in 114 infants with unilateral congenital cataract. The median age at the time of surgery was 1.8 months.¹⁷ After one year, there was no difference in visual acuity between the groups, but there was a higher rate of post-surgical complications, adverse events, and additional surgical procedures in the patients who had received primary IOL implantation.⁵ Visual acuity was also tested at 4.5 years with HOTV optotypes. Again, there was no difference in visual acuity between the children whose vision was corrected with an IOL and those whose vision was corrected with a contact lens.⁶ Based on these findings, it was recommended that patients under the age of 6 months be left aphakic and corrected with a contact lens instead of an intraocular lens due to the similar visual acuity outcomes and reduced number of adverse events of contact lenses versus IOLs.^{6,18}

While the Infant Aphakia Treatment Study provides extensive data on the treatment of congenital cataracts, it does not address other forms of cataract that develop later in childhood. Another study evaluated the

outcomes of cataract surgery performed on 400 patients between the ages of 2 and 16 years, of whom 57% had traumatic cataracts. This study also found no difference in visual acuity between primary and secondary IOL insertion in this patient population.⁷

While secondary IOL insertion is often used in the pediatric population, it can also be performed in adults. Studies of senile cataracts and military veterans with traumatic cataracts found no significant difference in visual outcome between primary and secondary IOL insertion.^{8,9}

Extensive research has shown no significant difference in visual outcomes between primary IOL insertion and aphakia in infants, children, and adults with congenital, senile, or traumatic cataracts.⁵⁻⁹ Both options were considered in this patient, who had the complicating factors of ongoing self-abuse to the head and eyes and atypical facial features that could cause difficulty with spectacle correction. While compliance with correction would have been guaranteed with an intraocular lens, primary IOL implantation would have put the patient at significant risk of dislocation of the IOL, re-opening of the surgical wound, or other adverse events. For this reason, the patient was left aphakic after surgery and corrected with sports goggles.

Although contact lenses are often the treatment of choice in aphakia, this patient was prescribed sports goggles for three reasons. First, sports goggles would provide protection to his eyes and reduce the risk of additional injuries from blunt trauma. Second, a contact lens would very likely be dislodged in this patient's case and proper care and replacement would be difficult due to the patient's extreme dislike of having his eyes and face touched. Finally, sports goggles would be better suited for this patient than traditional spectacles due to his unique facial anatomy and absence of fully formed external ears. Secondary IOL insertion will be considered in the future if the patient's self-injurious behavior changes or abates.

CONCLUSION

Patients with Treacher Collins syndrome, intellectual disabilities, and autism spectrum disorder are at an increased risk of developing visual abnormalities. While patients with Treacher Collins syndrome are at risk of developing strabismus and vision loss due to structural abnormalities of the eye and socket, patients with ID and/or autism spectrum disorder are at risk of developing self-injurious behavior. These patients should be monitored closely due to the potential for ocular injury and treated as needed. Traumatic cataracts can be surgically removed, but there are risks and benefits of both primary IOL insertion and aphakia. Treatment should take into account the patient's lifestyle and visual demands, as these are often unique in the special needs population.

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