Article • Deprivation Amblyopia in the Setting of Bilateral Congenital Cataracts: A Case Report

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ABSTRACT

Background: A cataract is an opacity of the crystalline lens that has the potential to cause vison loss or visual distortion. Etiologies of congenital cataracts include genetic mutation, hereditary, intrauterine infection during pregnancy, persistent fetal vasculature, association with metabolic disorders, and chromosomal syndromes. Congenital cataracts can present unilaterally or bilaterally and in a myriad of shapes, sizes, and colors. Depending on these characteristics, congenital cataracts may or may not affect vision. If they do, these cataracts warrant prompt surgical intervention to prevent deprivation amblyopia, which is reduced visual acuity due to a disruption to normal visual development. Optometrists play a crucial role in making the correct diagnosis and prompt referral. In addition, optometrists are responsible for managing these patients post-operatively for amblyopia treatment, correction of residual ametropia, and various post-operative complications.

Case Report: A four-year-old male with Down syndrome presented with bilateral dense cataracts, sensory nystagmus, and decreased visual acuity. Due to the cataracts, refractive error and retinal health were unable to be assessed clinically. The patient was referred for consultation for surgical management with a guarded prognosis. The patient was also referred for a visual evoked potential to determine visual potential.

Conclusion: If visually significant, one of the greatest concerns of congenital cataracts is deprivation amblyopia. Following prompt surgical intervention, optometrists must manage these patients closely, often over the course of their lives, to ensure optimal visual development and ocular health. Even if a patient presents outside the ideal treatment window, it is still important to recommend treatment, as there can be significant visual improvement recorded post-surgery.

Keywords: congenital cataracts, deprivation amblyopia, nystagmus, Down syndrome, cataract extraction

Introduction

Congenital cataracts are the leading cause of treatable childhood blindness.[1] The prevalence of congenital cataracts is 1 to 15 per 10,000 children globally and 1 to 3 per 10,000 children in developed countries.[2] One of the main concerns of congenital cataracts is irreversible vision loss from deprivation amblyopia. Amblyopia is the failure of cortical visual development in one or both eyes, which can cause decreased vision, reduced or absent stereopsis, and suppression of visual input.[3] Amblyopia can be caused by strabismus, refractive error, or obstruction of the visual pathway (deprivation).[4] Deprivation amblyopia most commonly results from congenital cataracts.[4]

Childhood cataracts can be congenital or acquired. Congenital etiologies include genetic mutation, hereditary, association with systemic and chromosomal disorders, intrauterine infection during pregnancy, and persistent fetal vasculature. Metabolic syndromes that are associated with congenital cataracts are Lowe syndrome, galactosemia, and Fabry disease.[5-7] The most common chromosomal disorder associated with congenital cataracts is Down syndrome.[8] A child with congenital cataracts may present with a positive family history of congenital cataracts, leukocoria, nystagmus, strabismus, photophobia, and delayed developmental milestones.[9] If the cataract is associated with a systemic condition, the examiner might also note a variety of non-ocular signs and symptoms, such as excess vomiting, lethargy, poor growth, or an overall failure to thrive.

Despite the ocular comorbidities of congenital cataracts, not all affect vision significantly. Therefore, not all congenital cataracts warrant a referral for surgical intervention. It is the role of the eye care provider to determine the level of visual disruption, if any, and to make prompt referrals as indicated. If a cataract is found to be a significant detriment to vision, ideally a cataract extraction is performed by six to eight weeks of life.[10] Once the cataract is removed, an intraocular lens may be placed, or the child may be left aphakic. If the latter is the case, the child will need to be fit with a high-plus contact lens to account for the lost dioptric power of the crystalline lens. In either case, optometrists play a vital role in the management of these patients due to their need for close follow-up, optical correction, and amblyopia treatment. This paper highlights the importance of early intervention in the setting of congenital cataract treatment and the importance of treatment even after the critical period has passed.

Case Report

A four-year-old male presented with his mother to the University Eye Center for a second opinion regarding the patient's diagnosis of bilateral congenital cataracts. The patient's mother reported that the cataracts were present from birth. She also reported that the patient was holding objects and screens at a close working distance and had "shaking eye movements" and an inward eye turn of the right eye more so than the left eye. The patient was previously examined in Russia, and the mother reported that at one and a half months old, the patient's cataracts began "cracking," and some vision was gained. The mother stated that a doctor in Russia recommended to wait on pursuing cataract surgery until the patient reached five years of age. The patient was prescribed atropine eye drops to potentially allow more visual input into the eye. Unfortunately, the patient developed a skin rash, and the mother discontinued the drops after two days. They did not see an eyecare provider for a year. At the age of three years old, the patient saw an ophthalmologist in the United States who recommended cataract extraction. Again, the patient did not have the surgery and was lost to follow-up until his presentation at the University Eye Center.

The patient's medical history was positive for Down syndrome and bilateral congenital cataracts. Allergies to medicine included atropine per the patient's mother (however, the patient did receive a dilated fundus examination previously, with no adverse reactions). There was no known medication usage. Developmental history was positive for premature birth at twenty-eight weeks. The patient's walking milestone was delayed, and he was not speaking yet. The patient attended a special education school for children with Down syndrome, where he was receiving special services. The patient's mother reported that the patient was performing well in school and was able to complete matching activities. There was no previous ocular injury or surgery. Family history was unremarkable, including no family history of congenital cataracts.

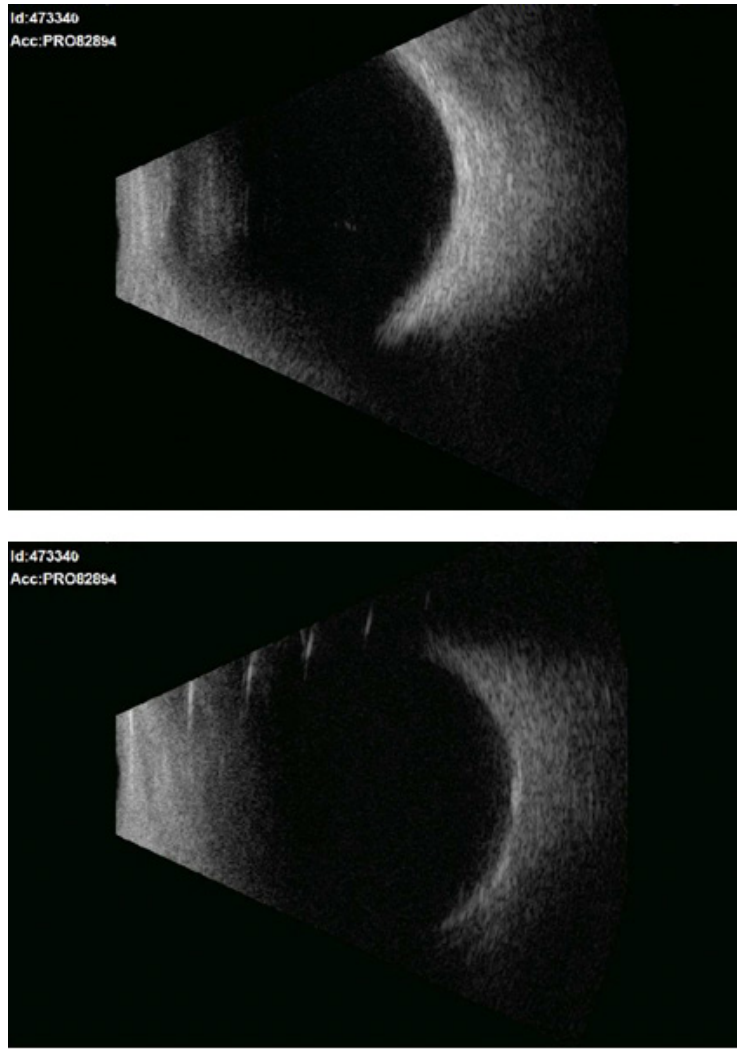

Figure 1. B-scan ultrasonography of the right eye (top image) and left eye (bottom image) of a male four-year-old patient prior to cataract surgery, showing a grossly flat and intact retina with no vitreous opacities

Entering unaided visual acuity was 20/150 in both eyes using Cardiff cards at fifty centimeters. Equal resistance to occlusion was noted. Extraocular motility testing was full in both eyes with poor fixation. A sensory nystagmus of both eyes was observed that was greater in lateral gazes. Confrontation visual fields were grossly full in both eyes. Pupils were equal, round,

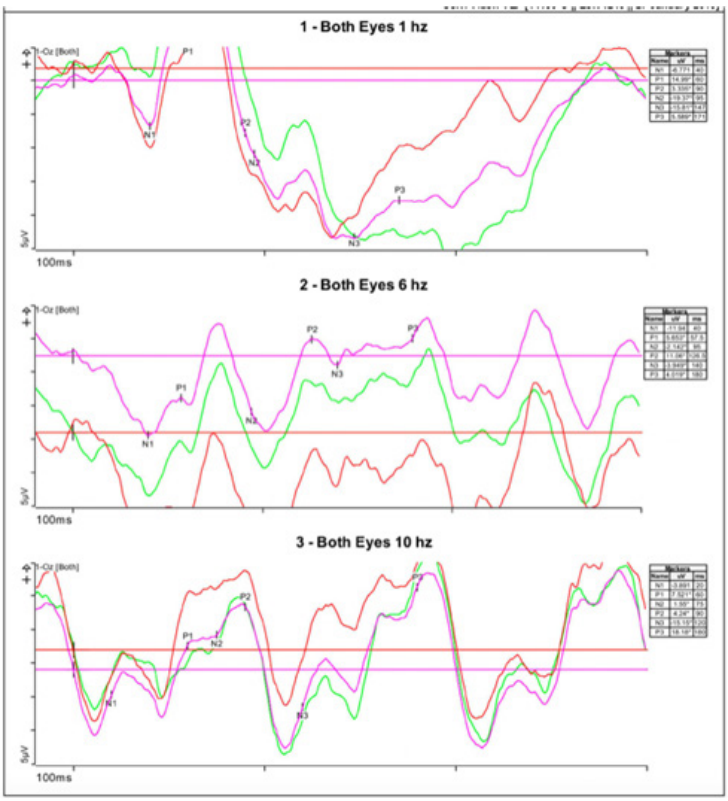

Figure 1. Flash visual evoked potential of both eyes of a male four-year-old patient, three months after cataract surgery including an intraocular lens implant, demonstrating adequate responses to all frequencies

and reactive to light with no afferent pupillary defect. Ocular alignment testing with Hirshberg revealed gross alignment at distance and near. No global or local stereopsis was appreciated. Retinoscopy was performed; however, no reflex was observed due to dense cataracts obstructing the retinal reflex, even with the use of high plus and high minus lenses. Anterior segment evaluation revealed prominent epicanthal folds of both eyes and dense bilateral lenticular opacities. Digital intraocular pressures were soft and equal to palpation. Upon dilated fundus examination (with tropicamide 0.5% and punctal occlusion), scattered sparse segments of peripheral retina were observed through breaks in the cataract. The cataract appeared to affect all layers of the lens, sparing sections of the far peripheral cortex of both lenses. The optic nerve, macula, and retinal vessels were unable to be assessed. B-scan was performed to rule out any gross retinal defects (Figure 1) and revealed a grossly flat and intact retina in both eyes with no vitreous opacities or masses. The patient was referred to pediatric ophthalmology for cataract extraction consultation with a guarded prognosis due to delayed intervention. The patient's mother was educated extensively on the need for follow-up after getting surgery and for appropriate optical correction.

A visual evoked potential was ordered to further assess visual potential. A flash visual evoked potential was performed (Figure 2) rather than pattern due to poor patient fixation and cooperation. The flash visual evoked potential demonstrated adequate response to all frequencies, implying normal visual cortex response to a flash stimulus. The patient and mother were advised to follow up with the pediatric ophthalmologist for surgical intervention and to return for assessment of optical correction, amblyopia therapy, and assessment of visual potential. Genetic testing was recommended to potentially link the cataract with a specific gene mutation, as well as to perform a pattern visual evoked potential if patient cooperation allows. The patient was scheduled for cataract extraction consultation. The ophthalmologist planned to implant an intraocular lens rather than leaving the patient aphakic according to the patient's mother. The patient has yet to return for his follow-up examination, so the results of the cataract extraction, if performed, are unknown.

Discussion

Etiologies of Congenital Cataracts

Etiologies of congenital cataracts include genetic mutation/hereditary, intrauterine infection during pregnancy, persistent fetal vasculature, and association with chromosomal syndromes. About fifty percent of bilateral cases have a genetic basis,[11] making it a likely etiology in the case of this patient. Of course, this cannot be confirmed until genetic testing is performed. Inheritance pattern tends to be autosomal dominant;[11] therefore, asking about family history of childhood cataracts is paramount if hereditary cataracts are suspected.

Intrauterine infections are responsible for numerous congenital anomalies, congenital cataracts being one of them. Intrauterine infections include toxoplasma, others (syphilis, parvovirus, varicella zoster), rubella, cytomegalovirus, and herpes, otherwise known as "TORCH." Rubella is the most common TORCH infection associated with congenital cataracts.[12] Cataracts secondary to rubella tend to be dense, bilateral, pearly white opacities. Other ocular complications of intrauterine infections include pigmentary retinopathy, microphthalmos, glaucoma, iris dystrophy, or chorioretinitis.[12] If suspicious for intrauterine infection, it is important to inquire about any fever, rash, or flu-like symptoms during pregnancy.

Persistent fetal vasculature is a failure of regression of the fetal hyaloid vasculature. It is unilateral in 90% of cases,[13] but bilateral cases have been reported.

The type of cataract associated with persistent fetal vasculature is a capsular cataract, but it can also be associated with other ocular complications such as glaucoma, corneal opacification, coloboma, retinal detachments, and spontaneous vitreous hemorrhage.[14] Diagnosis of persistent fetal vasculature is made by B-scan ultrasonography, where the remnant hyaloid vasculature would be apparent. This diagnosis was ruled out in this patient due to the unremarkable B-scan and clear vitreous.

There are many chromosomal syndromes that are associated with congenital cataracts. Down syndrome is the most common chromosomal syndrome, with a frequency of 1 case in every 700 to 1,000 live births.[15] Multiple cataract types have been associated with Down syndrome; however, cerulean blue dot cataracts are the most prevalent.[16] In addition to cataracts, children with Down syndrome are disproportionately affected by a host of ocular complications, such as blepharitis, Brushfield spots, strabismus, nystagmus, and accommodative disorders.[17] The patient discussed in this report had a positive history for Down syndrome, and it is unknown whether his cataracts were from a genetic mutation (as genetic testing was not performed) or whether the cataracts were associated with his diagnosis of Down syndrome.

Surgical Intervention: Indications and Considerations

If surgery is indicated, unilateral cataracts should be removed within the first 4 to 6 weeks of life and bilateral cataracts within the first 6 to 8 weeks of life for optimal visual prognosis.[18] As we know from the work of Hubel and Wiesel, deprivation of typical visual input and experiences during a critical period can irreversibly alter pathways in the visual cortex, leading to decreased visual performance.[19] Indications for surgery include significantly reduced visual acuity, cataract greater than 3 millimeters in size, cataract on the visual axis, and cataract associated with strabismus or nystagmus.[20] The association with nystagmus and/or strabismus indicates a disruption to binocularity, as was the case for this patient. As a rule of thumb, if the cataract is large enough to obstruct the examiner's view of the posterior ocular structures, it is likely that the cataract is also obstructing the patient's view of the world.

Once it is determined that a cataract extraction is indicated, the surgeon may elect to implant an intraocular lens or to leave the child aphakic. The aphakic child will need to be fit with a contact lens to make up for the lenticular power lost. This is dependent on the surgeon and the patient's age and is determined on a case-by-case basis. Generally, most surgeons leave the child aphakic if less than six months old and will recommend waiting until the child is at least two years old before implanting an intraocular lens.[10] In the case of this patient, the ophthalmologist recommended an intraocular lens implant due to his older age.

The Infant Aphakia Treatment Study compared intraocular lens and contact lens correction in monocular aphakia patients less than six months old. According to the Infant Aphakia Treatment Study, there was no significant difference in visual acuity outcome when comparing between the two groups at five years.[21] While visual acuity was similar, the number of adverse reactions in the intraocular lens group was higher than the contact lens group. The intraocular lens group also required more surgeries.[21] Taking cost into consideration, the patient cost was greater in the contact lens group. With that being said, contact lenses require many follow-up visits and a commitment from the entire family, as they are the ones doing the insertion, removal, and maintenance of the contacts.

Post-Surgical Complications

Following cataract extraction, complications can include glaucoma, infection, and visual axis opacification. The Infant Aphakia Treatment Study also identified the percentage of subjects who developed glaucoma following cataract extraction. At one year following surgery, 12% of subjects developed glaucoma.[22] At 10 years following surgery, that number rose to 22%.[23] This percentage was similar in both the intraocular lens group and the contact lens group. Increased risk of developing glaucoma was associated with a younger age of surgery and persistent fetal vasculature.[22] The increased risk of glaucoma in this population highlights the importance of measuring intraocular pressures at follow-up examinations, even if years have passed since the surgery.

Infection or endophthalmitis is another complication of cataract extraction. It occurs in less than 5% of pediatric cataract surgeries and may present days to weeks after surgery.[24] Risk factors include nasolacrimal duct obstruction, periorbital eczema, and upper respiratory illness at the time of surgery.

Visual axis opacification is a complication that occurs in virtually 100% of cases if the posterior capsule is left intact. To combat this, some surgeons will remove the posterior capsule and perform an

anterior vitrectomy.[25] This is because the treatment of visual axis opacification requires an Nd:YAG laser capsulotomy, for which younger patients will not be able to fixate accurately.

Optometric Post-Surgical Management

Optometrists have a vital role in the post-surgical management of pediatric cataract patients. Whether the child is aphakic or has an intraocular lens, they will need to be fit with contact lenses and/or glasses, as well as started on a patching schedule for amblyopia treatment.

If aphakic, contact lenses are the refractive correction of choice as they will minimize aniseikonia, especially in the case of unilateral aphakic patients. Glasses would result in poor image quality due to the required high-plus powers, discomfort due to the weight of the lenses, and poor cosmesis due to the magnification effect of plus lenses. When choosing contact lens parameters, optometrists must take into consideration the child's steeper corneas, small aperture size, and their need for high plus power and high oxygen permeability. In terms of prescribing, children less than two years old should be corrected for near and therefore over-plussed by about two to three diopters since their entire world is up close.[9] When the child is older and ambulating, they need to be fully corrected for both distance and near and will require a bifocal over the contact lenses. In patients with an intraocular lens, a bifocal is still recommended, with the distance portion being either plano or any residual correction from the implant.

When prescribing glasses or bifocals, polycarbonate is a must. For pediatric patients in general, polycarbonate lenses are recommended due to their durability and protective qualities. Special considerations for children in bifocals include segment height and type of bifocal. Since these children have no accommodation, it is vital that they have near correction for optimal vision at all distances, whether aphakic or with an intraocular lens implant. Segment height should be higher than what would be prescribed for an adult to ensure that the child is looking through the bifocal and thus receiving the clearest image possible. A flat top 28 or executive bifocal style is also preferred for the same reasons. Add power can be +2.50 to +3.00 diopters, with the segment height ranging from mid-pupil to lower iris, depending on age and maturity level. In this patient, additional consideration should be given regarding frame type (i.e., shorter temple lengths, lower nose pads, lower nose bridge design) to accommodate the facial features of a child with Down syndrome.

Amblyopia in this population can be severe depending on the timing in which the cataract was removed, whther it was unilateral or bilateral, and the age of the child. To make matters more difficult, there is no universal consensus on patching schedules. Most clinicians follow the guidelines from the Infant Aphakia Treatment Study, which entail patching one hour per day per month of life until about eight months old. After eight months old, the patching schedule shifts to half of all waking hours.[26] Patching can be a challenge, as the child will likely be resistant, especially in the case of unilateral aphakia. Encouragement with activities that the child enjoys can be helpful, and there are a variety of patches (adhesive and cloth) that can be used to aid in compliance. After initiating patching, follow-up examinations should be scheduled every four to six weeks and thereafter every three months once improvement in acuity has been noted.[9] Patching therapy is performed generally until the child is six years old, but some studies have recommended patching until nine years old.[27,28] If a plateau is reached and it is unclear whether patching is still indicated, a pattern visual evoked potential can be useful in determining an appropriate end point. A pattern visual evoked potential can provide an objective, quantitative assessment of visual acuity;[28] however, it requires patient cooperation, which is why a flash was performed in lieu of a pattern visual evoked potential in this patient. Bilateral congenital cataracts tend to have a more favorable visual prognosis compared to unilateral; however, the presence of nystagmus and/or strabismus leads itself to a more guarded prognosis,[20,30] as is the case in this patient. It is of importance to note that just because a child presents with cataracts outside the recommended window for removal or outside the critical period, that does not mean that cataract extraction should be deferred. Although each case is unique, having cataract extraction is recommended even in delayed presentations, as there have been significant visual improvements recorded post-surgery.[31,32]

Visual Perceptual Effects of Late Treatment of Visual Deprivation

The effect of visual deprivation on visual acuity has been vastly studied. However, it is also of importance to note other consequences that visual deprivation may have on a child, vision-related and beyond. One study

found that in children ages 3 to 6 years old, deprivation amblyopia was associated with lower self-perception of peer acceptance and physical competence.[33] This lower self-perception may discourage children from partaking in social interactions and physical activities compared to their normally sighted peers, which can in turn negatively impact their quality of life.

McKyton et al. looked at the effects of delayed cataract surgery on shape recognition in Ethiopian children with early-onset complete bilateral cataracts. It was found that after delayed cataract extraction, only low-level cues for shape recognition, such as color, size, and shape, were identifiable by the newly sighted children.[34] Mid-level cues for shape recognition, such as occlusion and shading, remained deficient in these same children even after two years following cataract extraction.[34] Vision is an amalgamation of many intricate neuronal pathways, including those for visual perception. Difficulty navigating busy visual environments, difficulty with object recognition, and difficulty with face recognition[35] are all examples of visual perceptual deficits that go beyond a measurement of visual acuity but have just as much of an impact on quality of life.

Conclusion

There are many factors that must be considered when managing the pediatric cataract population. Pediatric patients who have had cataract extraction(s) require frequent follow-up examinations for a multitude of reasons. They are at increased risk of decreased visual acuity, ocular complications, visual perceptual deficits, and lower self-perception. They require optical correction in the form of glasses and/or contacts, as well as a rigorous patching schedule. The risk of complications such as developing glaucoma is also higher in this population; however, the benefit of removing the cataract outweighs the risk of glaucoma. The road to maximal visual potential is often a long one, requiring support from family and eye care providers alike.

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