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Refractive lens exchange with intraocular lens implantation in hyperopic eyes of a patient with Angelman syndrome

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Abstract

We describe a patient with Angelman syndrome with severe developmental delay who was visually impaired by uncorrected high hyperopia and poor control of accommodation. Refractive lens exchange with intraocular lens implantation was performed in both eyes when the patient was 22 years of age. Satisfactory anatomical and functional outcomes were achieved and maintained during 3 years of follow-up. Refractive lens exchange can be useful in patients with severe neurobehavioral disorders in the presence of high refractive error and poor accommodative control.

Refractive surgery is now performed more frequently at a younger age in patients with severe anisometropia or bilateral high ametropia resistant to conventional therapy using spectacles or contact lenses.^{1–4} Although corneal refractive surgery is less invasive than intraocular lens (IOL) implantation, it is not suitable for correcting extreme refractive errors.⁴ In eyes with these errors, phakic IOL (pIOL) implantation or refractive lens exchange (RLE) with IOL implantation can be useful alternatives. In the United States, there are no Food and Drug Administration–approved pIOLs for hyperopic correction. We report the 3-year postoperative outcome of RLE with IOL implantation in both eyes of a patient with Angelman syndrome with severe developmental delay and uncorrected high hyperopia with poor accommodative control.

CASE REPORT

In 1992, a patient presented to us with a complaint about deviation of his eyes from his mother. The diagnosis was Angelman syndrome (Figure 1) with developmental delay, spontaneous laughter, excitability, hand flapping, and a short attention span. The vision was recorded as central and steady fixation in each eye. Refraction at the time of strabismus surgery for exotropia revealed marked hyperopia of +7.5 diopters (D) in both eyes. During a visit in 2006, the patient's mother stated that use of spectacles was becoming more and more difficult. Each new pair of glasses was broken by the patient despite attention from her and the caregivers. During the previous year, the mother had spent approximately \$2500 on

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glasses. Poor accommodative control was also noted by dynamic retinoscopy. Because of the uncorrected hyperopia and poor accommodative control, the patient's functional behavior was that of a severely visually impaired person.

The options of lens surgery and laser refractive surgery were discussed with the mother. Refractive lens exchange with IOL implantation was performed in both eyes by one of us (M.E.W.) when the patient was 22 years of age. Preoperative biometry measurements were done under anesthesia at the time of surgery. The axial length was 21.36 mm in the right eye and 21.37 mm in the left eye. The anterior chamber depth was 3.45 mm and 3.55 mm, respectively. Refractive lens exchange with a 32.0 D SN60AT IOL (Alcon, Inc.) was performed in the left eye, with a refractive aim of -1.50 D. After 4 months, RLE with a 30.0 D SN60WF IOL (Alcon, Inc.) was performed in the right eye, with a refractive aim of -1.00 D. The posterior capsule was left intact in both eyes at the time of surgery. At the 3-year follow-up visit, the patient had central and steady fixation. Visually, the patient could manage his day-to-day activities. The mother noted a marked improvement in his quality of life after surgery. An examination under anesthesia was performed since a thorough examination in the clinic was not possible. The intraocular pressure was 15 mm Hg in the right eye and 12 mm Hg in the left eye. Retinoscopy revealed a refractive error of $-0.5 +1.5 \times 45$ in the right eye and -2.5 in the left eye. The axial length was 21.46 mm and 21.50 mm, respectively. Mild posterior capsule opacification (PCO) was observed in both eyes (Figure 2). This was thought to be visually insignificant.

DISCUSSION

In 1965, Angelman⁵ described “puppet” children, named after the characteristic signs that are associated with Angelman syndrome. A few reports describe the ocular manifestations of this syndrome.^{6–8} Consistent systemic findings in Angelman syndrome patients are mental retardation, speech impairment, ataxia, frequent spontaneous laughter with happy demeanor, excitability, hand flapping, hypermotoric behavior, short attention span, and seizures.⁸ Characteristic facial features include prognathism, protruding tongue, macrostomia, widely spaced teeth, and microcephaly.⁸ Other associated characteristics include excessive chewing behavior and sleep fragmentation. Common ocular findings include iris hypopigmentation (79%), choroidal hypopigmentation (70%), esotropia or exotropia (44%), and refractive errors, usually hyperopia.^{7,8} Our patient had more severe hyperopia than that reported in Angelman syndrome patients.⁶ Dickinson et al.⁶ report 2 children who had 4.00 D of hyperopia.

Our patient had functionally severe visual impairment before the RLE. The uncorrected hyperopia exacerbated issues related to the neurobehavioral disorder. Bilateral RLE with IOL implantation was performed because the refractive error was not within the normal range of corneal refractive surgery. However, RLE brings with it concerns about endothelial cell damage, PCO, and risks associated with intraocular surgery (eg, infection and retinal detachment). Lens removal abolishes the ability to accommodate. Thus, this surgery is generally not offered during the prepresbyopic period when pIOL implantation is the treatment of choice. However, our patient had poor accommodation even at his prepresbyopic age. In addition, no pIOLs for hyperopic powers are available in the U.S. Given this background, despite an anterior chamber depth of 3.5 mm, we chose to perform RLE. The procedures were deemed to be medically necessary and were funded through Medicaid. Multifocal IOL implantation was discussed but not chosen for this patient.

The most challenging aspect of RLE in young individuals is maintenance of a clear visual axis. Posterior capsule opacification is one of the most challenging aspects of lens surgery in these patients. Lessons from pediatric cataract surgery are a useful source of information

when performing RLE, even in young adults. Standard age-appropriate surgical strategies should be used. These include corneal tunnel, manual anterior capsulorhexis (vitrectorhexis for infants), bimanual irrigation/aspiration, hydrophobic IOL implantation, and consideration of primary management of the posterior capsule. In young children who have lens removal surgery, PCO develops rapidly and frequently if the posterior capsule is left intact. To prevent PCO, posterior capsulectomy with or without vitrectomy is considered an essential surgical step. In patients who are likely to cooperate for an office-based neodymium:YAG (Nd:YAG) laser procedure 18 to 24 months after lens removal surgery, the posterior capsule is often left intact.⁹ Refractive lens exchange is generally offered to children who are not cooperative, so cooperation for office-based Nd:YAG laser capsulotomy is questionable even in older children in this category. Neodymium:YAG laser capsulotomy in children often requires additional laser energy and has a higher chance of reopacification or reclosure. If the PCO in our patient becomes visually significant, it may require surgical removal. Our decision to leave the posterior capsule intact was related to our patient's tendency to self-manipulate and traumatize his eyes. Simplifying the surgery and retaining the full barrier between the anterior and posterior aspects of the eyes was thought to be important in anticipation of the possible trauma to the eyes. However, patients such as ours may benefit from primary posterior continuous curvilinear capsulorhexis even at 22 years of age.

Refractive lens exchange has certain advantages for the visual rehabilitation of patients. However, we caution that the RLE technique should be reserved for extreme situations until there are more data available to prove the long-term safety of this procedure.

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Figure 1.
Typical facial appearance of patient with Angelman syndrome.

Figure 2.
View of eye after RLE with IOL implantation.