Cataracts and Glaucoma in Patients With Oculocerebrorenal Syndrome

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Background: Oculocerebrorenal syndrome is an X-linked recessive hereditary oculocerebrorenal disorder characterized by congenital cataract, mental retardation, and Fanconi syndrome of the proximal renal tubules. Other ocular findings include glaucoma, corneal opacity (keloid), enophthalmos, and hypotonia.

Objective: To describe the treatment of 7 patients (14 eyes) with bilateral cataracts associated with oculocerebrorenal syndrome.

Method: Retrospective review.

Results: Seven patients with oculocerebrorenal syndrome had visually significant bilateral cataracts detected on their first full ophthalmic examination. All underwent bilateral cataract surgery. The mean age (of 14 eyes) at cataract extraction was 1.25 (median, 1.1) months. Glaucoma diagnosis and treatment were more variable. The mean age (of 11 eyes) at glaucoma diagnosis was 24.1 (range, 0.2-70.0) months: the mean age was 0.2 month (of 4 eyes) when glaucoma was diagnosed before cataract extraction, and the mean age was 37.7 months (of 7 eyes) when glaucoma was diagnosed after cataract extraction. All eyes were followed up for a mean of 100.8 (range, 38-190) months.

Main Outcome Measure: Treatment of 7 patients (14 eyes) with bilateral cataracts associated with oculocerebrorenal syndrome.

Conclusions: Early identification and surgical removal of cataracts is recommended in patients with oculocerebrorenal syndrome. Despite this, visual acuity results will only rarely be better than 20/70, and nystagmus is likely. Patients should be monitored closely and regularly for changes in intraocular pressure, optic nerve cupping, and refractive error to rule out the development of glaucoma.

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Methods

We performed a retrospective review of the medical records of all patients with oculocerebrorenal syndrome at the Storm Eye Institute, Medical University of South Carolina. All patients identified with oculocerebrorenal syndrome had undergone bilateral cataract extraction (CE), and were included in this report. The diagnosis of oculocerebrorenal syndrome was made or confirmed based on the presence of Fanconi syndrome, mental retardation, and congenital cataract. Patient 2 had the diagnosis confirmed by genetic testing (a DNA probe for the OCRL1 gene).

In each of these eyes, an anterior limbal approach was used to perform a lensectomy with anterior vitrectomy. In brief, paracentesis openings were made at the 2- and 10-o'clock positions at the limbus. A vitrector handpiece and a 20-gauge blunt-tip irrigating cannula were inserted into the anterior cham-
A round 5-mm anterior vitrectorhexis was made, followed by lens aspiration, posterior capsulotomy, and anterior vitrectomy, without removing the instruments from the eye. A synthetic, absorbable suture was used to close each pars plana site. If an intraocular lens (IOL) was inserted, it was placed in the eye through a corneal or scleral tunnel after the lensectomy previously described.

Initial intraocular pressure (IOP) measurements were taken during cataract surgery. Patients with an IOP of lower than 22 mm Hg were followed up with serial examinations under anesthesia approximately every 3 to 4 months. Those who had an elevated IOP were given a trial of medical therapy, usually initiated with 0.25% timolol, and rechecked approximately 2 weeks after therapy was begun. In cases of failed medical therapy, surgical intervention was applied.

### RESULTS

Seven patients (14 eyes) diagnosed as having oculocerebrorenal syndrome had bilateral cataracts detected on their first full ophthalmic examination. The cataracts were dense nuclear cataracts (3-6 mm in size). Each was considered visually significant, and surgical removal was recommended. All patients underwent bilateral cataract surgery. Miotic pupils were also noted. Glaucoma diagnosis and treatment were more variable. The data for each patient are summarized in the Table. The mean age at CE was 1.25 (range, 0.25-4.00; median, 1.1) months, with a mean follow-up of 100.8 (range, 38-190; median, 75.0) months. It was difficult to assess the visual outcome in this group of patients, because many of them have severe developmental delay. However, only 3 patients (patients 4, 5, and 7) had a mental status that was amenable to checking Snellen visual acuity. Patient 4 had a visual acuity of 20/80 OU, with nystagmus and no fixation preference. Patient 5 had a visual acuity of 20/40 OD, but only light perception in the other eye, after uncontrolled glaucoma was treated with cyclocryotherapy. Patient 7 had visual acuities of 20/60 OD and 20/70 OS, despite the presence of nystagmus (Table).

Only one patient (patient 3) underwent a bilateral IOL implantation at CE, at the age of 5 weeks. His best visual acuity is deemed central, unsteady, and maintained in both eyes, and he has had no adverse sequelae from the IOL in either eye. Age at surgery, glaucoma propensity in these eyes, and poor pupil dilation were recorded as reasons the other patients did not undergo IOL implantation at surgery. Patient 5 required a subsequent operation to aspirate cortical lens material from each eye, 3 months after the original cataract surgery. This repalpitation of lens cortex escaped the capsular bag remnant (Soemmering ring cataract) and appeared within the pupillary space. The material aspirated easily. Patient 1 underwent removal of a pupillary membrane and pearls at the age of 6 years after being unavailable for follow-up for 2 years. None of the patients have undergone secondary IOL implantation.

Of the 14 eyes, 9 were diagnosed as having glaucoma based on repeated IOP measurements higher than 21 mm Hg. Recorded gonioscopic results for 4 eyes 2 years following CE found open angles for 360°, except for one clock hour of peripheral anterior synechiae located superiorly. Intraocular pressure control was achieved without surgery in 6 of 9 eyes. One patient (patient 6) received a Baerveldt implant in each eye after trabeculotomy failed to control the IOP.

The age at glaucoma diagnosis varied. Many of the eyes were diagnosed as having glaucoma at cataract surgery when they underwent examination under anesthesia within the first week of life (patients 5 and 6). Patient 5 underwent 180° cyclocryotherapy 3 times to both eyes between the ages of 17 and 24 months. The left eye...
became phthisical. Patient 6 underwent bilateral 180° trabeculotomies 1 month after CE; they failed, necessitating a Baerveldt implant in both eyes 1 month later. He has done well, with medical treatment for the 4 years of follow-up since then. In 2 patients (patients 2 and 4), the pressure became elevated within months to a few years after the cataract diagnosis. These patients have been successfully treated medically. Patient 4 required pupilloplasty for miosis and corectopia exacerbated by echothiophate iodide (Phospholine Iodide). Of our 7 patients, 4 are African American, 2 are white, and 1 is of mixed ancestry. Patients with oculocerebrorenal syndrome have bilateral cataracts that are usually deemed visually significant at or near birth. An increased IOP and global systemic developmental delay can be expected. Mechanized vitrector instrumentation is essential in the management of the congenital cataracts in these patients. Complete removal of all lens material and a primary posterior capsulotomy and an anterior vitrectomy will reduce the chances of secondary membrane formation. Cortex reproliferation required a subsequent operation in one patient (patient 5) (2 eyes), but the material aspirated easily and did not recur. Patient 1 had a secondary membrane removed in one eye at the age of 6 years, following a CE in 1985 using techniques that have since been improved.

We have treated all but one of the patients described herein by leaving the eyes aphakic after surgery, and using spectacles for optical rehabilitation. In one patient (patient 3), primary IOL implantation was performed at the age of 5 weeks, with spectacles used for residual hyperopia. This patient developed nystagmus despite this early optical correction. A longer follow-up will be needed to determine if IOL implantation is useful in patients with oculocerebrorenal syndrome. This patient has recently developed IOPs in the mid 20s and is being treated medically. Glaucoma, common in these patients, can cause marked axial length changes, which would make primary IOL power selection difficult.

Despite early identification and surgical treatment, 5 patients (patients 1-4 and 7) developed nystagmus. In a previous study, researchers stated that the best visual acuity that can be expected is in the range of 20/100. However, the means by which this number was formulated are not stated. In our series, only 3 patients had a mental status that was amenable to obtaining the Snellen letter visual acuity. One patient (patient 5) had a visual acuity of 20/40 OD (no nystagmus) and light perception in the fellow eye, while another patient (patient 7) had visual acuities of 20/60 OD and 20/70 OS (with nystagmus). Patient 4 had a visual acuity of 20/80 OU, and demonstrated no fixation preference.

It seems that even with early surgery and early optical replacement of the crystalline lens, the visual acuity is not likely to be normal in these patients, and nystagmus usually develops. The nystagmus was not noted at birth in these patients. It was recorded as a sensory nystagmus from poor visual development. After repeated review of the medical records, no predictive factor for poor visual development or acuity could be rec-
The lack of eye growth predictability in these infants with lens implantation at CE can be performed safely, but the cataracts and prompt optical correction. Intraocular tagmus is likely, even with early surgical removal of acuity is not expected to be better than 20/70 and nystagmus is likely, even with early surgical removal of the cataracts in patients with oculocerebrorenal syndrome to Xq24-q26 by use of restriction fragment length polymorphisms. J Clin Invest. 1987;79:282-285.


REFERENCES