A 11-year-old male presented for a new patient comprehensive eye exam. He reported that the vision in his left eye was slightly reduced and hadn’t been “as good” as the right eye for a few years. The patient had broken his glasses 2 weeks before his visit.

His medical history was significant for chronic kidney disease (CKD) that was being managed by a nephrologist. The patient was taking vitamin D, sodium bicarbonate, and lisinopril. The family medical history was unremarkable.

The patient’s right eye was emmetropic, and the left eye had a low compound myopic refractive error. The patient’s mother, who accompanied the child, reported that the patient’s father had “chronic bilateral uveitis” for which he “received injections every few months.” No further information regarding the father’s diagnosis or treatment was known.

**A CLOSER LOOK**

Preliminary testing, including pupil testing, IOP, extraocular motility, confrontation visual fields, etc., was all normal. The patient’s best corrected distance visual acuity was 20/20-2 OD with plano sphere refraction and 20/25-2 OS with refraction of -1.00 -0.50 X 060.

The slit-lamp examination was unremarkable. A dilated fundus exam

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**AN INSIDE LOOK AT RENAL COLOBOMA SYNDROME**

A case report provides insights on this rare condition.

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**AT A GLANCE**

- Renal coloboma syndrome (RCS) is a multisystem genetic condition that predominantly affects the kidneys and eyes.
- Although RCS commonly results from PAX2 gene mutations, roughly half of people with RCS do not have an identifiable mutation in that gene.
- In the case presented here, an 11-year-old boy was diagnosed with this rare condition.
Figure 1. In the fundus of the right eye (A), optic disc excavation is apparent. In the fundus of the left eye (B), blood vessels emerge from the optic disc in a spoke-like manner. A slightly elevated yellow lesion is present nasal to the macula corresponding to bullous retinoschisis.

Figure 2. OCT through the macula in the right eye shows mild retinoschisis nasal to the macula.

Figure 3. OCT through the optic nerve in the right eye shows deep optic disc excavation with optic pit deep in the nerve canal. Retinoschisis and intraretinal fluid extend to the optic nerve canal.

Figure 4. OCT through the macula in the left eye shows more significant retinoschisis splitting the neurosensory retina.

Figure 5. OCT through the optic nerve in the left eye shows optic disc excavation with clear optic pit.

Because of the patient’s reduced visual acuity and retinal elevation, OCT scans of the maculas were obtained (Figures 2–5). The scans revealed bilateral optic disc pits along with bullous retinoschisis, worse in the left eye than the right.

A tentative diagnosis of renal coloboma syndrome (RCS) was made based on the ocular findings in tandem with the history of CKD. The patient was referred to a retina specialist for additional evaluation and to a geneticist for genetic testing.

After examination, the retina specialist agreed with the diagnosis of bilateral optic pit–associated bullous retinoschisis manifesting as a complication of RCS. Differential diagnoses included morning glory syndrome variant and X-linked retinoschisis (XLR). Genetic results revealed no mutations in the PAX2 gene.

The patient maintains kidney care and treatment with his nephrologist and rotates between the optometry and retina clinics every 6 months to be monitored for ocular changes. No progression of ocular involvement has been seen in the past 12 months.

DISCUSSION

RCS, also known as papillorenal syndrome, is a multisystem genetic condition that predominantly affects the kidneys and eyes. Ordinarily, patients with this ailment exhibit renal hypoplasia and optic nerve anomalies. However, genital abnormalities, hearing impairment, and central nervous system defects may also occur, among other irregularities.1

RCS is inherited in an autosomal dominant fashion and commonly results from PAX2 gene mutations.2 PAX2 gene transcription creates proteins (transcription factors) required for the development of the kidneys, eyes, central nervous system, and genital tract.3 Alterations in these proteins lead to malformations of the aforementioned body systems.

Discordantly, roughly half of people with RCS do not have an identifiable PAX2 gene mutation.2 Evidently, PAX2 gene expression is involved in a multitude of disease processes and has even been linked to ovarian cancer.4

Kidney Findings

Kidney findings associated with RCS include hypoplasia, vesicoureteral reflux, renal hypertension, and
proteinuria.5 Research illustrates that renal hypoplasia, vesicoureteral reflux, and CKD are correlated with an increased risk of end-stage renal disease.6,7 Approximately 33% of patients with RCS will develop end-stage renal disease.8

Although the management of children with CKD has improved, the mortality rate for them remains higher compared to their healthy peers. Nowadays, children with CKD are more likely to die from heart complications or infection than from kidney failure.9

Treatment for CKD mainly consists of hemodialysis, peritoneal dialysis, and kidney transplant.9 In addition, vitamin D, sodium bicarbonate, and lisinopril—all three of which were prescribed for this patient—have been shown to benefit patients with CKD.10-12

Symptoms of CKD can vary and can be nonspecific; they can include fatigue, drowsiness, nausea, and sleep disturbance, among others.5,13,14 A diagnosis of CKD is often arrived at through a blood and urine test, regularly adjoined with ultrasound imaging and tissue biopsy.

Ocular Findings

Ocular findings may include optic disc excavation, optic disc pits, retinal colobomas, and varying degrees of visual impairment.8 Ocular diagnosis requires a detailed fundus examination. OCT scans of the retina, macula, and/or optic nerve can be helpful aids in diagnosis.

Although there is no consensus on a gold standard treatment for macular retinoschisis, a number of management options are available. Surveillance is an acceptable approach if a mild case of retinoschisis presents with minimal symptoms. A topical carbonic anhydrase inhibitor may promote resorption of intraretinal fluid in retinoschisis.15

Vitrectomy has been shown to aid in retinoschisis resolution and is especially useful if there is concomitant vitreoretinal traction.16 Laser photocoagulation is rarely used in maculopathies due to the risk of permanent vision defects. Naturally, if macular retinoschisis progresses to retinal detachment, a retinal detachment repair would be needed. Lastly, although RCS-associated bullous retinoschisis is unique to XLRs, it is encouraging that gene therapy for XLRs has shown promise in improving visual acuity.16

For this patient, because the bullous retinoschisis and visual impairment were relatively mild in each eye, management consisted of close observation without intervention.

As noted in the presentation above, differential diagnoses for RCS include a variant of morning glory syndrome and XLRs. Detailed descriptions of the differential diagnoses, along with the classification stages of CKD, are beyond the scope of this article.

It is important to note that, although genetic testing revealed no mutations in the PAX2 gene, approximately half of patients with RCS do not have PAX2 gene anomalies.2

**IT PAY TO INVESTIGATE YOUR PATIENTS’ CONCERNS**

RCS is a rare condition with the potential for significant systemic and ocular consequences. Although RCS can affect a variety of the body’s organs, its impact is usually limited to the kidneys and eyes.

Optometrists are privileged to have the opportunity to peer into the eyes of patients and acquire a snapshot into their systemic health. We can develop an understanding of their wellness and discover findings that may reveal meaningful concerns. The eyes may or may not be the windows to the soul, but they can serve as a window to overall health.

Through astute clinical investigation, our patient’s inquiry into his reduced vision was answered. Furthermore, he and his mother were made aware of the likely connection between his eyes and long-standing kidney disease. They will now continue close observation with frequent eye examinations. If any ocular changes occur, they can be addressed promptly.

This case reminds us of a couple of key points in our field of optometry. First, further investigation is warranted in patients exhibiting less-than-expected vision. Second, optometrists are well-suited to embrace medical eye care. I’m sure we can all agree that we have the ability to provide more than just refractive care via glasses and contact lenses. As our nation’s health care networks become increasingly integrated, optometrists can assume a pivotal role in the system to elevate ourselves professionally, enhance our practices, and provide the best care to our patients.

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