Optical Coherence Tomography in Rare Pediatric Cases

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Spectral-domain optical coherence tomography (SD-OCT) is becoming a common diagnostic tool and is known for aiding the diagnosis and treatment of several well-known pathologies, such as age-related macular degeneration, macular hole, and cystoid macular edema. I specialize in rare hereditary degenerative retinal pathologies and use a variety of methods, including psychophysical, visual field, and electrophysiology tests, to aid in diagnosis and treatment. However, SD-OCT has become a key part of my patient workup. Now I am able to couple functional information from other tests with high resolution, very detailed, highly informative data from OCT retinal scans. This capability has created a significant difference in the way I treat, examine, and diagnose my patients.

In addition to its value alongside other diagnostic equipment, OCT imaging proves invaluable in pediatric cases. A number of the typical ophthalmic tests used in adults are unusable with young patients who may be incapable of participating sufficiently in the test. Since our acquisition of the iVue SD-OCT (OptoVue) mounted on a specially designed cart and stabilizing arm (iStand, OptoVue), we can do retina, optic nerve and anterior segment scans while patients are recumbent under anesthesia.

CASE EXAMPLE 1

A boy aged 1 year was referred to us due to the presence of a leukocoria, a sign of retinoblastoma. An ocular oncologist ruled out retinoblastoma but suspected a retinal detachment. At this point, the retinal surgeon and I took the child into the OR to perform an exploratory investigation. We confirmed retinal detachment in 1 eye and, examining him with the iVue mounted on the iStand, we gathered important information from the fellow eye. Under ophthalmoscopic evaluation, his posterior pole appeared near normal. However, with SD-OCT, we discovered that it was diffusely delaminated, presenting with a carpet of cystic changes that were splitting the retina tangentially.

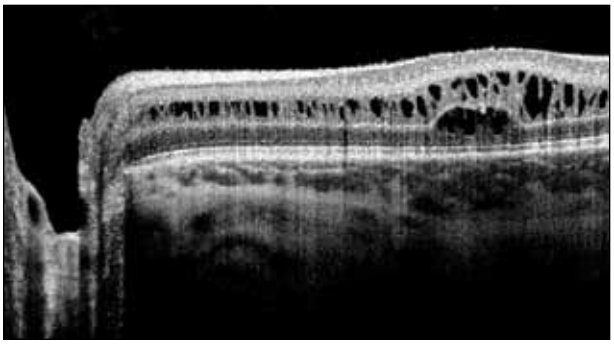

Figure 1. Macular SD-OCT scan intersecting the foveal region and extending nasally to the optic nerve head. The scan show clearly a layer of columnar polycystic spaces spanning across the middle of the neural retina, and additional cystic cavities within the photoreceptor layer at the foveal level. The perifoveal cystic changes blunt the severity of the foveal cystic changes. Such cystic changes were present wherever the iVue SD-OCT could reach. These findings, coupled with ERG findings and the presentation, strongly suggested the diagnosis of X-linked retinoschisis, which was confirmed by molecular genetic diagnostic testing.

Macular SD-OCT shows a cross-section of the retina, identifying changes in optical density and typically depicting the fovea as a downward pit. In this patient's OCT scans (Figure 1), the entire retina appeared similar to the cross-section of a sponge, showing no visible fovea. This aspect was not apparent on ophthalmoscopy because the entire macular region was delaminated, whereby the bulging of the fovea, obvious on SD-OCT, was blunted and unapparent on an en face view like that of ophthalmoscopy. Retinoschisis is uncommon, and this patient had a

particularly diffuse carpet of cystic changes across the retina. Using SD-OCT, however, we were able to identify easily the cystic changes across the posterior pole of the retina and even nasally to the disk. Almost everywhere we scanned, we found delamination of the retina.

We concluded that this patient experienced a retinal detachment secondary to X-linked juvenile retinoschisis (XLRS), which was subsequently confirmed by molecular genetic diagnostic testing for the gene causing XLRS. We also performed a flash electroretinogram (ERG) while the child was under anesthesia, which showed a cutoff of the b-wave in response to bright flashes, a typical finding in XLRS. Using only the ERG, we could have suspected the diagnosis of XLRS and then confirmed it with genetic testing. The SD-OCT, however, not only yielded a virtually immediate diagnosis, but, most important, made it possible to appreciate the sponge-like appearance of the entire back of the retina, which we could have not appreciated otherwise.

This sponginess had also prevented us from comprehending the extent of the cystic changes that occupied the foveal region. SD-OCT made the microanatomic retinal changes very visible and provided sufficient evidence to warrant prescription of carbonic anhydrase-inhibiting eye drops in an effort to manage the situation. Without this SD-OCT evidence, we would have decided to watch the patient instead. These drops have been shown to diminish significantly the macular cystic changes in XLRS patients. Therefore, the drops may make a huge difference in this child's visual acuity potential. It is possible that they may help reduce the overall retinal sponginess as well.

Showing the OCT images to the parents proved also very helpful. Seeing the sponge-like retina helped the mother to understand how weak the retina was and that it was capable of tearing fairly easily. It is rare for retinal detachments to occur in patients so young, and this patient is clearly at risk for a second retinal detachment in his only remaining eye.

CASE EXAMPLE 2

A girl aged 3 years was referred to me with suspicion of Leber's congenital amaurosis (LCA). She presented with severe monocular blindness, with essentially no light perception in her right eye. The left eye had visual acuity estimated at around 20/70. Under ophthalmoscopic examination, the fundus appeared discouraging in both eyes. My initial impression was that the eye with the better visual acuity had a retina less affected quantitatively. We then examined her under anesthesia with SD-OCT. The scans confirmed that her left eye was, in fact, much less affected (Figure 2). Unfortunately, the scans also confirmed that the macular region of her right eye was nearly devoid of photoreceptor inner and outer segments. There

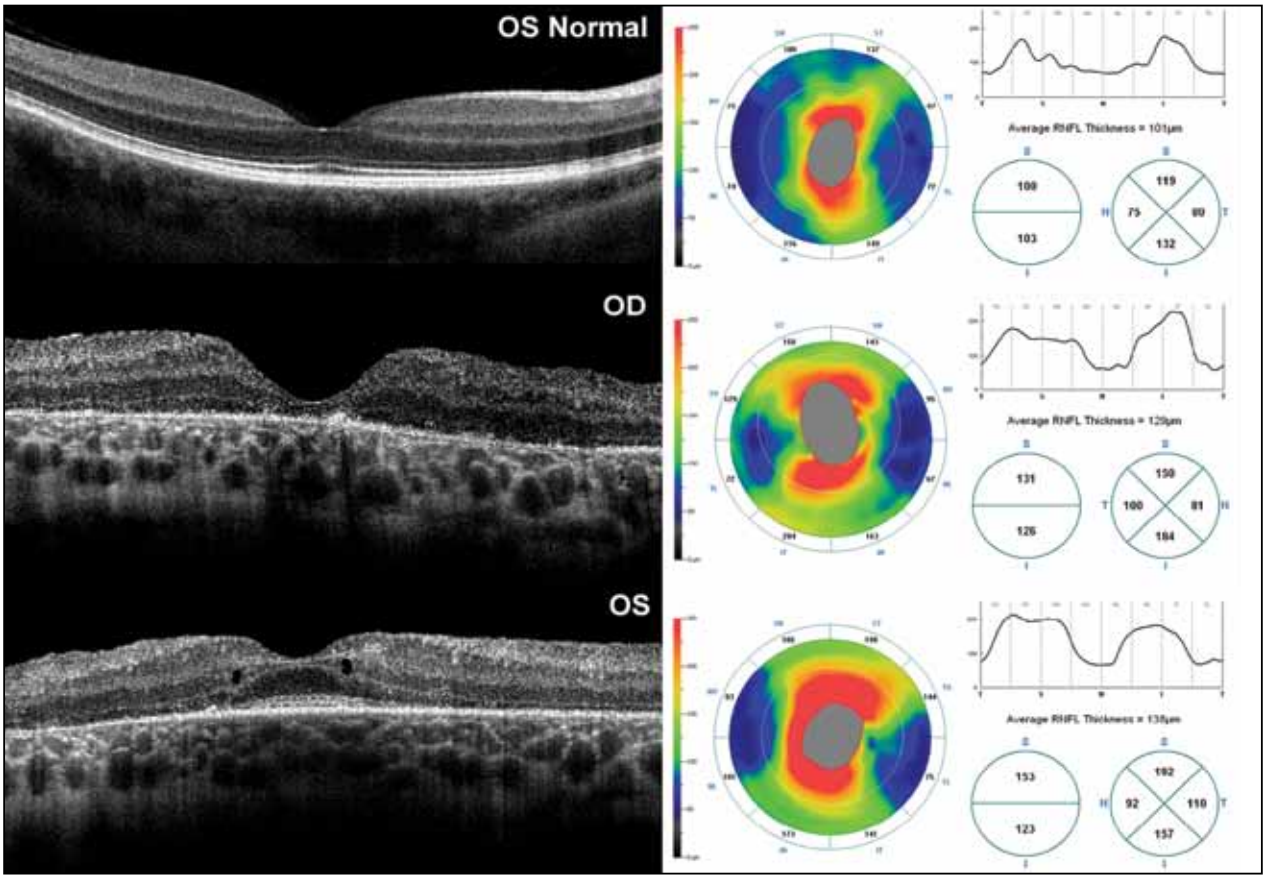

Figure 2. Comparison of retinal macular and disc retinal nerve fiber layer (RNFL) SD-OCT scans in a normal subject (top rows) and in a patient with autoimmune neuroretinopathy (AINR). In the right eye of the AINR patient, retinal lamination is markedly deranged, and there is severe widespread loss of both the photoreceptor outer-inner segment boundary and of the retinal pigment epithelium (RPE). In the left eye, retinal lamination is far better preserved, and so are photoreceptors in the foveal area, and the RPE is also much less affected. There is also evidence for mild perifoveal cystoid macular edema. The RNFL is markedly increased in thickness in both eyes. These findings go hand in hand with the serologically demonstrated presence of autoantibodies recognizing both retinal and optic nerve antigens, and the robust response to periocular depot steroid therapy in this patient.

was some residue of photoreceptor cells, but the outer segments had already been lost through most of the macula. The left eye also suffered significant damage to the macula and there was mild cystoid macular edema, but the fovea was largely intact, allowing 20/70 visual acuity.

As I was conducting the fundus exam, the asymmetry between the 2 eyes captured my attention, as it is very unusual in hereditary diseases. The nerves also appeared hyperemic, which is atypical of degenerative diseases. An SD-OCT scan of the optic nerve confirmed that the retinal nerve fiber layer was significantly thicker than expected in a normal eye, suggesting inflammation (Figure 2).

At this point, I began to suspect primary or secondary autoimmune disorder of the retina and optic nerves. This is typically thought of as an adult disease, as no previous case of autoimmune disorder had surfaced in a child this young. However, the SD-OCT scans matched those of adults with autoimmune disorders of the retina and optic nerve. Blood tests for autoantibodies against the retina and optic nerve returned strongly positive. Although we were uncertain if the trigger for the autoimmune reaction was some form of congenital predisposition or hereditary disease, we could at least confirm the suspected role of autoantibodies in worsening vision, and, from there, identify a treatment target. Treatment options for autoimmune diseases include heavy doses of oral cortisone or other immunosuppressant, or injections of periocular depot steroids, which would be ongoing and, given the patient's age, would require anesthesia.

The information gained from the SD-OCT scans was

instrumental in correctly diagnosing the disease. It also provided evidence to persuade me and the family of the necessity of obtaining the autoantibody tests and, thereafter, of giving the patient steroid injections under anesthesia. She has been receiving injections every 3-4 months for a period of 18 months and has regained visual acuity. Her right eye has improved from no light perception to perception of hand motion, and her left eye improved from 20/70 to 20/30. Initially, she was nonfunctional, and now she functions near normally.

So far, she has not manifested high intraocular pressure or developed a cataract, the 2 greatest risks of periocular steroid injections. However, these possibilities represent secondary concerns considering the possibility of blindness in patients with autoimmune disorder of the retina. Without periocular steroids, this patient would likely have lost light perception in her left eye as well. Without the information that we gained with SD-OCT imaging in this patient, we would have not been able to make the correct diagnosis or decided to start such treatment.

SUMMARY

SD-OCT imaging is expanding our knowledge of the posterior segment of the eye and aiding in the diagnosis and treatment of a number of pathologies. Its flexibility and noninvasive nature make it particularly useful with pediatric patients, who are unable to undergo many other ocular diagnostic exams. ■

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