LIFE-ALTERING MOMENTS IN OPTOMETRY

Two patients, two lives helped, thanks to the keen observation skills of this OD.

BY MOLLY ANN CLYMER, OD, FAAO

Optometrists serve as gatekeepers of health care in the ophthalmic setting. We often see patients when they are young and healthy, and we are in a position to detect both ocular and systemic disease during the course of our examinations.

Some of the diseases we may be the first to recognize can be life-threatening or life-changing, so it's important for us always to perform at the top of our game when seeing patients. You never know what you're going to find, as shown in the two cases presented below.

PATIENT NO. 1

When a 61-year-old woman scheduled an emergency appointment because she noticed new flashes and floaters, I assumed I would find a posterior vitreous detachment (PVD) and my role would be to rule out any potential retinal complications. Not surprisingly, that's exactly what I found—at first.

Initial Findings

The patient presented with a medical history including multiple sclerosis and hypercholesterolemia and an unremarkable ocular history. She was taking 40 mg atorvastatin (Lipitor, Pfizer) daily, and her entering VA was 20/30 in each eye (OU). Her pupils were unremarkable, and IOP was 11 mm Hg in the right eye (OD) and 12 mm Hg in the left (OS). Anterior slit-lamp examination revealed moderate cataract OU, and her initial dilated fundus examination showed a PVD with an unremarkable retinal exam OD.

When I finished my initial examination, the patient mentioned that she had been losing "the top half" of the vision in her right eye at times for the past 2 months. She said she knew she had a new floater, but that she perceived the top half of the vision in her right eye going completely black for about 3 to 5 minutes at a time and then returning to normal. "It always comes back, so I wasn't too worried, but I thought I should tell you." I added transient vision loss OD to her assessment and decided to take a second look at her fundus.

Digging Deeper

Within seconds of positioning her back at the slit lamp and focusing on her optic nerve, I was surprised to see a large, thick, mobile plaque exiting the central retinal artery and passing through the inferior bifurcations of the vessels. This was only in my view for a few seconds and had not been visible to me during my first fundus examination.

I quickly explained to the patient what I had just seen. She denied experiencing any other stroke-like symptoms and insisted that she was compliant with her atorvastatin. She reported a strong family history of carotid artery disease, and I explained the urgency of the clinical findings in the setting of transient vision loss.

I called her primary care practitioner and explained the emergent clinical findings. Our mutual decision was to send

FACTS TO KNOW ABOUT MYASTHENIA GRAVIS

- **What it is:** Myasthenia gravis is a chronic autoimmune neuromuscular disease that causes weakness in the skeletal muscles. The hallmark of the disease is muscle weakness that worsens after periods of activity and improves after periods of rest.
- **Who is affected:** Both men and women across all racial and ethnic groups can be affected, but it most commonly affects women younger than 40 years and men older than 60 years. It is neither inherited nor contagious.
- **How the eyes are involved:** Certain muscles, such as those that control eye and eyelid movement, are often (but not always) involved in the disorder.
- **What are the symptoms?** Symptoms may include ptosis; blurred or double vision; a change in facial expression; difficulty swallowing; shortness of breath; impaired speech; and weakness in the arms, hands, fingers, legs, and neck.
- **How is it diagnosed?** Several tests may be ordered to confirm a diagnosis, including a physical and neurological examination; an edrophonium test; antibody blood tests; electrodiagnostic testing; diagnostic imaging (to look for thymoma commonly found in patients with myasthenia gravis); and pulmonary function testing.
- **Is there a cure?** Although there is no known cure, current therapies (eg, immunosuppressive drugs, anticholinesterase medications, thymectomy) can control symptoms and often allow people to have a relatively good quality of life.

Source: Myasthenia gravis fact sheet. National Institute of Neurological Disoders and Stroke. www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Myasthenia-Gravis-Fact-Sheet. Accessed August 23, 2019.

her to the emergency room (ER) right away. I spoke with the attending ER physician and recommended a full stroke workup, including a carotid Doppler test, echocardiogram, electrocardiogram, MRI of the brain, and any additional tests he felt were necessary. I asked the patient to follow up with me in 1 month.

Fast Forward

The ER physician ordered an MRI of the patient's brain and magnetic resonance angiography of the head and neck, which revealed a segment of high-grade stenosis in the proximal cervical segment of the right internal carotid artery and focal high-grade stenosis at the origin of the right external carotid artery.

At her 1-month follow-up visit, the patient reported that all symptoms of transient vision loss had resolved after she had emergent right-sided carotid endarterectomy surgery. She was discharged with instructions to take aspirin daily. This patient let me know how appreciative she was of my examination and immediate referral.

All's Well That Ends Well

Sometimes an optometrist is handed the perfect opportunity to make a huge impact on someone's quality of life. I honestly think luck was on my side that day, as I just happened to be looking at that vessel at the exact right time. Even without seeing it clinically, I probably still would have recommended a workup, but the visualization of the plaque certainly caused me to expedite the process.

This case reminds me never to rush a patient when he or she is explaining symptoms, even if a brand new complaint arises when you're totally finished with your exam and ready to see the next patient. Never ignore a symptom reported by a patient, even if it seems insignificant, and never miss that second opportunity to prevent a potentially catastrophic event. Listen carefully and look again—it could save someone's life.

PATIENT NO. 2

I saw an 83-year-old woman the same day that she called our office asking to see an eye doctor as soon as possible. She explained that she had just been discharged from the hospital and was struggling to focus her eyes.

Initial Findings

A detailed and complex history of the present illness was obtained when the patient arrived. She had recently visited the ER three times with complaints of "inflamed eyelids, dizziness, blurred vision, diplopia, weakness, and fatigue." She reported having multiple tests, including a brain MRI with a normal result.

The patient was admitted to the hospital after multiple ER visits, diagnosed with sinus and urinary tract infections. She continued to struggle with blurred vision and diplopia that made her feel intoxicated. She reported difficulty holding her eyelids open. The patient was placed on intravenous antibiotics for her diagnosed infections and was then discharged to a rehabilitation facility.

According to the patient's daughter, her doctors felt that her diplopia was secondary to a potassium deficiency. She was adamant that prior

to the onset of all these symptoms she was active, attending the YMCA nearly every day for aquatic fitness. She was frustrated with her new inability to perform basic activities of daily life secondary to her symptoms.

Digging Deeper

The patient's ocular history included cataract surgery OU, Nd:YAG capsulotomy OU, and macular hole repair OS. Her medical history included hypertension, renal disease, anxiety, and osteoporosis. Her entering best corrected distance acuity was 20/25 OD and 20/30 OS. Her confrontation visual fields were restricted superiorly secondary to bilateral ptosis, and her pupils were reactive to light OU with no evidence of afferent pupillary defect.

Distance cover test showed no tropia or phoria, and she reported no diplopia. The patient's extraocular muscle movements presented as an external ophthalmoplegia showing restrictions in abduction, adduction, elevation, and depression. Her lids showed ptosis OU with palpebral fissure measurements of 10 mm OU. IOP was 12 mm Hg OD and 14 mm Hg OS. Dilated fundus examination was unremarkable.

Déjà Vu

Before meeting this patient I had seen a few patients with myasthenia gravis, and this was at the top of my differential diagnosis once I examined her. Although I was accustomed to seeing milder disease, I highly suspected myasthenia gravis based on her complaints and clinical presentation.

I asked the patient to maintain upgaze for 2 minutes and then remeasured her palpebral fissures at 7 mm OD and 8 mm OS. For this patient, looking up was challenging due to her external ophthalmoplegia presentation, which confirmed worsening ptosis with fatigue.

I had the patient perform the ice test for 2 minutes, which showed a significant improvement in her ptosis, and I documented this with photos. The ice test is highly sensitive and specific for myasthenia gravis. The patient commented on her improved superior visual field following the ice test.

Confirming Suspicions

I ordered myasthenia antibody blood tests and referred the patient to a neuro-ophthalmologist. During the examination I called him to make him aware of the significance of her symptoms, and he recommended that I prescribe oral prednisone for her to take until he was able to evaluate her (I saw her on a Friday and he saw her the following Monday). When he evaluated the patient, he confirmed the diagnosis and referred her to a neuromuscular specialist, who admitted her to the hospital and continued her oral prednisone in addition to oral pyridostigmine.

During her appointment with me, I explained that I highly suspected an autoimmune condition called myasthenia gravis (see *Facts to Know About Myasthenia Gravis*). I told her that this diagnosis would explain all of her previously unexplained symptoms, and I reassured her that treatment was available and that she had the potential to regain her previous quality of life.

Make a Myasthenia Mental Note

This patient's case highlights that it is important for members of our profession to be familiar with the symptoms of myasthenia gravis and confident in identifying them. ERs are designed to manage life-threatening conditions, and emergency physicians may not recognize the symptoms of myasthenia gravis on a regular basis. We must make a conscious effort to consider myasthenia as a differential diagnosis when we are confronted with the symptoms mentioned in this case.

I recommend finding a neuro-optometrist or neuro-ophthalmologist with whom to establish a good working relationship. Further, it's good practice to ask all patients who present with ptosis if they are experiencing any systemic symptoms such as shortness of breath, generalized weakness, or fatigue.

Helping this patient get properly diagnosed and treated significantly improved her quality of life.

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Questioning clinical findings and a past clinician's advice put this patient—and her mother—on the right track with the proper treatment.

BY LESLIE O'DELL, OD, FAAO

The day was like most every other day of patient care. I saw one patient after another, reviewing test findings and changing treatments accordingly. Then I walked into an exam room to find a 13-year-old female patient waiting for me. This fact alone was out of place for me because, at the time, my mean patient age was 70. I assumed it would be an easy vision exam and an opportunity to make up some lost time from my slow morning. That changed once I began my slit-lamp evaluation.

PLAYING DETECTIVE

I noted cornea verticillata, also known as vortex keratopathy, bilaterally on the patient's corneas—out of place for sure. I asked the patient's mother to change places with her daughter, and I examined her corneas, finding the same vortex patterns and bilateral presentation.

WHY I'LL NEVER FORGET THIS PATIENT ENCOUNTER

- It was the first and only time I've seen a patient with Fabry disease. I knew nothing about this potentially life-threatening genetic disease and had a clue only from attending continuing education taught by fellow ODs.
- The diagnosis helped to improve the quality of life for this patient and her mother.
- It taught me not to settle with a previous benign diagnosis and to stay proactive in my patients' care.

I asked the mother if her previous eye care practitioner had discussed these findings with her. She said that they had discussed the issue previously, but her clinician thought them to be benign and left it at that. I excused myself from the examination room and went to my office to dig through course materials I had saved until I found notes from a course given by Joseph P. Shovlin, OD, FAAO, about this exact patient presentation.

Armed with a bit more information, I returned to the examination room to ask additional questions. The patient's mother reported having some gastrointestinal issues and chronic pain that she thought to be fibromyalgia. She and her daughter both described experiencing a burning sensation in their hands and feet (acroparesthesias) whenever they were febrile, but they assumed it was normal.

We collected blood samples from the patient and her mother and sent the samples out to a specialty lab for confirmation of Fabry disease.

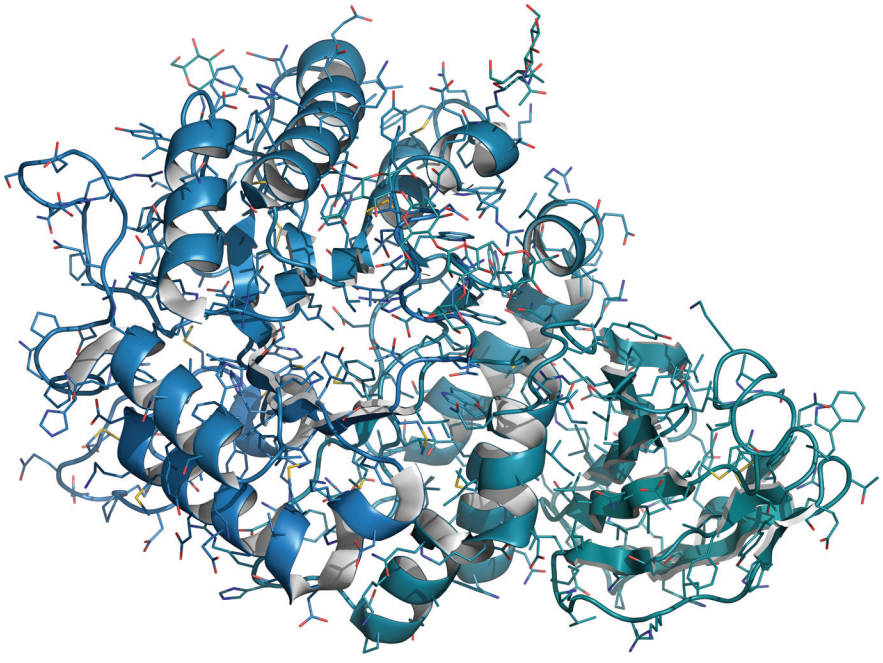

Figure. Fabry disease is caused by a deficiency of the enzyme alpha-galactosidase A.

REACHING A DIAGNOSIS

This is where my learning about Fabry disease began. It is a rare genetic disease caused by a deficiency of the enzyme alpha-galactosidase A (Figure), which causes a buildup of a type of fat called globotriaosylceramide (Gb3, or GL-3) in the body. Cornea verticillata can be a presenting feature in asymptomatic patients with Fabry disease.

For years, this mother was told not to worry, that women can be carriers and not be affected, when, in fact, carriers do have symptoms and are at risk for life-threatening complications of Fabry disease, including ischemic or hemorrhagic strokes and interstitial lung disease.

After confirming the diagnoses of Fabry disease in this young patient and her mother and properly referring them for treatment, I can rest assured that they will receive lifelong treatment with the enzyme replacement therapy agalsidase beta (Fabrazyme, SanofiGenzyme), which will help normalize their kidney and heart function and the blood supply to their brains.

That young patient is now a married adult pursuing a career in nursing. Read *Why I'll Never Forget This Patient Encounter* for a list of lessons learned. ■

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