

CASE STUDY





Figure 1: Optos photos of the right and left eyes showing extensive damage to the RPE and choroid with scalloped edges





Figure 2: OCT scans of the right and left eyes showing damage to the outer retina, RPE, and choroid adjacent to the foveal center.



PHYSICIAN SPOTLIGHT

BELINDA SHIRKEY, MD

Dr. Shirkey is a seasoned vitreoretinal surgeon. As a partner and the Medical Director for Retina Associates of Kentucky, she specializes in both surgical and medical retinal disease. Dr. Shirkey treats all surgical vitreoretinal conditions including floaters, macular holes and puckers, vitreous hemorrhage and all varieties of retinal detachment.

As many patients who suffer from macular disease can attest, the macula is critical to central, or "fine vision." This is the vision that allows us to see well enough to read and drive. Fortunately, many conditions that affect the macula, such as puckers and holes, are surgically reparable. Groundbreaking advancements in surgical instrumentation and technique have resulted not only in improved outcomes, but shorter recovery for those undergoing these procedures. These sight-saving surgeries allow patients to live more independently and return to enjoying the day to day activities of their lives.

A 38 year old male presented to our office for a second opinion on his retinal condition. He had been told for years that he had retinitis pigmentosa (RP), a degenerative disease of the rod photoreceptors in the retina. He had developed significant peripheral visual field loss and, as a result, was no longer able to drive. He also suffered from a severe decrease in low light sensitivity. His central visual acuity, however, remained fairly good at 20/30 + 2 in the right eye and 20/25 in the left.

Clinical photographs of his eyes can be seen in Figure 1. There is substantial peripheral obliteration of not only the retinal pigment epithelium (RPE), but also the underlying choroidal vasculature. The pattern of this damage is scalloped at its edges. The classic "bone spicule" pigmentation of RP is not present. The optical coherence tomography (OCT) scans shown in Figure 2 highlight the damage to the outer retina, RPE, and choroidal layers adjacent to the foveal center. These findings are more consistent with another degenerative retinal disease known as choroideremia. Genetic testing was recommended, which confirmed this diagnosis.

Choroideremia is an X-linked recessive mutation in the CHM gene. Because of this, males are predominantly affected, as they only require one copy of the mutation whereas females require two. This gene is involved in the production of the Rab escort protein-1 (REP-1), which is essential in assisting transport of proteins within cells in the retina. The absence of REP-1 leads to premature cell death in the retina and associated tissues. The damage begins in the peripheral retina but eventually advances centrally resulting in blindness, usually in late adulthood.

While there is currently no available treatment for choroideremia, there are exciting clinical trials active in the US and elsewhere evaluating gene therapy as a potential means of slowing, stopping, or potentially reversing some of the damage caused by this disease. This research came about due to the FDA approval of Luxturna (Spark Therapeutics, Inc.) in December of 2017 for treatment of RPE65-associated inherited retinal diseases (IRD), such as Leber's Congenital Amaurosis and certain forms of RP. Luxturna is actually the first FDA-approved gene therapy for IRD. It utilizes a viral vector approach to introduce a functional RPE65 gene into cells to replace the defective gene. That

same approach is now being applied to numerous other forms of IRD. We were able to connect this particular patient with the clinical trial head for an active choroideremia gene therapy trial in the US, and he is trying to enter this trial at one of its three sites.

This is an amazing time for those affected by IRD! We at Retina Associates of Kentucky have partnered with the Foundation For Fighting Blindness as well as Spark Therapeutics to provide free genetic testing to patients with IRD in order to identify their mutations and potentially connect them with active clinical trials or perhaps even an FDAapproved treatment, if their mutation is in the RPE65 gene. These are blinding diseases for which there have traditionally been no effective treatments. If you know someone with an IRD, please let them know that there may now be a way to treat them, so it is important they identify their specific mutation. Retina Associates of Kentucky is happy to assist them!



Submitted by Todd J. Purkiss, MD, PhD "With the development of finer microsurgical equipment over the last several years, I can delicately remove harmful tissue from the eye, while preserving the patient's sensitive macula. As a surgeon, it is incredibly gratifying to restore a patient's best vision through the surgical repair of vitreoretinal disease."

Belinda Shirkey, MI

Dr. Shirkey also treats chronic retinal conditions such as macular degeneration, diabetic eye disease and vascular occlusion. Fortunately, patients suffering from these conditions can receive convenient treatment right in our office.

While Dr. Shirkey primarily sees patients in Lexington, she also sees patients in RAK's Richmond, Danville, Frankfort and Somerset satellite locations.

WHAT'S HAPPENING



Federation of the Blind Symposium Hilton Garden Hotel, Louisville



Meeting Hilton Hotel, Downtown

Lexington



Museum

Ashland New Office Provider Open House



Lexington Academy of Eye Surgeons & Physcians Meeting







(800) 627-2020

THE RETINA TIMES

Fall 2019 · Issue #14

RETINA ASSOCIATES OF KENTUCKY RECEIVES A PERFECT SCORE FROM MEDICARE

July 16, 2019, Lexington, Kentucky

Every physician at Retina Associates of Kentucky received a perfect score from Medicare in the quality and cost effectiveness of its medical care. Medicare set up a quality and cost scoring system after passage of the Medicare Access and CHIP Reauthorization Act of 2015 (MACRA) which was signed into law on April 16, 2015.

Medicare's scoring system was designed to measure the quality and efficiency of medical care, drive improvement in care processes and health outcomes, increase the use of healthcare information, and reduce the cost of care. Thomas Stone, MD, Chairman and President of Retina Associates of Kentucky stated that the physicians at Retina Associates of Kentucky have a long history of providing the highest quality retina care to patients with a sensitivity to the high cost of medical care and continuously work to improve both the quality and cost of the care they provide. Retina Associates of Kentucky provides retina care in eleven locations in Kentucky from the I-65 corridor east to West Virginia, conducts research on new retina treatments, and provides a fellowship training program for the next generation of retina surgeons.

For more information contact Kristin Willard at kwillard@retinaky.com.

NEW ASHLAND OFFICE





We at Retina Associates of Kentucky are excited to announce the opening of our brand new office at 1700 Winchester Avenue in downtown Ashland.

We are proud to have served the patients of Ashland and the surrounding area for over 20 years, and are grateful that your continued support has allowed us to grow.

AWARDS & RECOGNITION

Retina Associates of Kentucky is proud to highlight stellar team members with our Awards and Recognition program!











JOIN US

The surgeons at Retina Associates of Kentucky are the first in Kentucky to have 3D Digital Assisted Vitreoretinal surgery, powered by TruVision. We are committed to education and showing our colleagues in eye care how we take care of patients. If you are an eye doctor, we welcome you to join us in surgery or clinic for observing. If you have interest please feel free to contact Kristin Willard at (502) 649-3681 or by



email: kwillard@retinaky.com.

RESEARCH

If you are interested in information regarding past clinical trials or participation criteria in our current clinical trials, please contact our research department: **Diana Holcomb** - Clinical Research Manager **PH (859) 264-2905** | **dholcomb**@**retinaky.com**



MAIN OFFICES

Lexington

120 N. Eagle Creek Drive, Suite 500 Lexington, KY 40509

Louisville

6420 Dutchmans Parkway, Suite 70 Louisville, KY 40205

Ashland

1700 Winchester Avenue Ashland, KY 41101

OUR OTHER LOCATIONS

Bardstown Danville Frankfort Lexington - West London Prestonsburg Richmond Shelbyville Somerset

OUR PHYSICIANS

William J. Wood, MD Rick D. Isernhagen, MD Thomas W. Stone, MD John W. Kitchens, MD Todd J. Purkiss, MD, PhD Belinda L. Shirkey, MD Sheila Garcia Santana, MD Blake A. Isernhagen, MD Jack L. Hollins, MD



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