Fuchs’ Dystrophy

This issue is focused on Fuchs’ endothelial corneal dystrophy—its definition, symptoms, what it’s like to have this disease and what we are doing about it.

What is it?

Fuchs’ endothelial corneal dystrophy (Fuchs) is a hereditary condition of the inner cell layer of the cornea known as the endothelium (see above). The endothelium provides the appropriate balance of fluid in the cornea, keeping it thin and crystal clear.

In Fuchs patients, the cells in the endothelium begin depositing abnormal material on the membrane to which they are attached (Descemet’s membrane—the blue line in the figure above). These deposits look like mounds and are called guttae. As the guttae enlarge they distort and diffract light causing glare and poor vision. With further growth, they allow fluid into the cornea, causing it to swell and become hazy. One of the first signs of the disease maybe that you begin to notice glare with headlights at night or in bright sunlight.

As the disease progresses, your vision may become foggy or blurry in the morning, and clear as the day progresses. This occurs because evaporation of fluid from the cornea is decreased while your eyes are closed, and accumulation of fluid in the cornea during the night results in corneal swelling.

While there are steps you can take to stave off the disease, once vision is diminished sufficiently to hurt your quality of life, a cornea transplant is needed.

From a Patient’s Perspective

We met Miriam Zimmerman in the fall of 2011. She had come to Indianapolis from San Francisco for her DMEK cornea transplant and met with us to discuss her experience. After speaking with her, it was obvious she had done her research and her story could benefit others. At our request, Miriam agreed to document her story, a journey dating back to 2001, through diagnosis of Fuchs, choosing a surgeon, and her personal reflections, so we may share it with others.

After being diagnosed with Fuchs in 2001, Miriam received her annual checkups without much progression. It wasn’t until she could no longer drive at night to get to the night class she taught that she began actively seeking a solution.

Surgical Outcome and Reflections

At her one-month checkup with her San Francisco corneal specialist, she received excellent test results. His first words were, “beautiful!” followed by “excellent work.” The graft was clear and fully attached. After her two-month checkup, she ordered glasses and now enjoys perfect vision, symptom-free.

As Miriam shares her personal reflections, we learn “...what has made this process so much more manageable for me was my original research into treatment options for Fuchs that enabled me to make the best choices in procedure and physician. Having knowledge about what to expect demystified the dread and equipped me with optimism and confidence. Reading about others’ experiences was integral to gathering this knowledge.”

A Desire to Give Back

Miriam shares, “An unexpected outcome of the experience is that I feel highly motivated to give back in some manner for the incredible gift of vision that has been bestowed on me by someone.” You can find Miriam’s entire DMEK Diary on our homepage at www.cornea.org. If you would like to share your story, please email Jessica@cornea.org.
Our Mission: To Give People Back the Use of Their Eyes

Welcome New Fellows!

The Foundation is pleased to announce the addition of two new Cornea Fellows! Zach Burkhart, MD (left) and Matthew Feng, MD (right) are pictured with their families.

Dr. Burkhart received his undergraduate degree from the University of Notre Dame and his medical degree from the University of Texas Southwestern Medical School at Dallas. After an internship at UT Medical Branch in Austin, he trained as an ophthalmology resident at the University of Texas at Houston. Dr. Burkhart is interested in all anterior segment surgery, particularly cornea, cataract, and refractive surgery. He enjoys sports, painting, playing/writing music and spending time with his wife and two children.

Dr. Feng graduated magna cum laude from Harvard College and was a Dean's Merit Scholar at the University of Pittsburgh School of Medicine. As a resident at the University of Arizona, he became interested in corneal surgery and has published research on corneal imaging for pre-LASIK screening. His hobbies include nature photography, travel, and in-line skating. Once upon a time, he played the piano at Carnegie Hall. Tragically, he is a rabid Pittsburgh Steelers fan.

Happenings

September 28, 2012
Mark your calendar for the annual Cornea Research Luncheon and Auction! Reserve your seat early—single tickets are $40, sponsorships begin at $250.

This year, we’re taking a new direction and featuring a fabulous silent auction filled with works of art from artists, patients and the visually impaired. Please join us to learn more about the Foundation and support its valuable work!

November 17, 2012
The Focus on Education seminar will be held at the Ritz Charles in Carmel, Indiana. Offering 7 hours of CE to attending optometrists while hearing from contemporary experts on today’s topics.

June 4, 2013
Cornea Classic Golf Outing—Please mark your calendar! Call us now at 317-814-2993 to ensure you receive the brochure when it’s printed.

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Lauren Gunderson
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Golf Outing Success

On June 21, 2012, the Foundation hosted the 15th annual Cornea Classic Golf Outing at Wood Wind Golf Club. The event raised more than $40,000! We are thankful for a great planning committee and all those who helped us reach our goal. Mark your calendar for June 4, 2013!

Save the date! This fall, please join us for the Cornea Research Luncheon and Auction featuring art from professional artists, grateful patients and the visually impaired. Marianne Glick of GlickArt, Inc. will serve as honorary chair.

Are you an artist? Let us know! We would appreciate a donation of your artwork. Artworks of all media are being accepted! If you would like to receive an invitation or volunteer on the committee please call our office.

Fall Luncheon

When: September 28, 2012
Where: Renaissance Hotel
Carmel, Indiana
Feature: Silent Art Auction
All proceeds benefit CRFA!

Pictured (left to right): Mike Dickerson (board member), Dr. Price Sr., and Dr. Price Jr.
I am pleased to report that we are continuing to develop improved treatments for Fuchs’ endothelial corneal dystrophy.

Ten years ago Fuchs was usually treated by cutting out the entire center of the cornea and sewing in a donor cornea with sutures. With this technique, called penetrating keratoplasty (PK, Figure 1), it took a year or longer to get the sutures out, and the visual results were unpredictable. In addition, the cornea never heals completely, so PK patients have to be careful never to bump the eye and break open the incision.

Since 2001, we have helped pioneer a series of less invasive, small incision techniques, known as endothelial keratoplasty (EK). At first DLEK, then DSEK and now DMEK. EK is now the preferred surgical treatment for Fuchs. In contrast to penetrating keratoplasty, EK selectively replaces the diseased endothelial cell layer (back layer) while leaving 95% of a patient’s cornea intact (Figure 2).

DMEK is now an amazing option for patients with Fuchs. We implant just a single healthy cell layer from the donor cornea through an extremely small incision that heals rapidly, restoring the cornea to a normal thickness. We have found that DMEK provides the best vision and greatly reduces the risk of transplant rejection compared with earlier techniques. For Fuchs patients who have a full thickness graft or DSEK, if the original graft ever needs to be replaced, we can now replace it with a DMEK.

Steroid Study

The risk of rejection is so low now with DMEK, that we are conducting a study to see if we can safely cut back on the corticosteroid eye drops that are used to prevent cornea transplant rejection. The steroid eye drops have a number of undesirable side effects and increase the pressure in the eye in about one third of patients. This is dangerous because it can damage the optic nerve that sends visual information to the brain. So far the study results look very promising!

Visit www.youtube.com/user/cornearesearch to view a DMEK procedure and more!

Continued Improvement

We use air, instead of sutures, to hold a DSEK or DMEK graft in place. Sometimes we have to add more air to make sure the graft sticks. Recently we successfully improved the DMEK technique to reduce the chance that more air will be needed.

Genetic Study

We also made great progress in figuring out what causes Fuchs. In collaboration with other eye centers around the country, we collected blood samples from hundreds of Fuchs and control patients then analyzed the samples to find out how the DNA (the body’s instruction manual) differs in Fuchs patients.

Differences in a gene called TCF4 are strongly associated with Fuchs. This gene is interesting because it contains a repetitive DNA sequence (called a ‘tri-nucleotide repeat’ that sometimes gets copied too many times when cells divide. As many as 20 additional DNA regions are associated with Fuchs, although none as strongly as TCF4.

The differences we see in the age of onset and the severity of Fuchs between different people, even between family members, may be explained by the specific combination of these genes that each person inherits. This new information will help in developing tests for early Fuchs diagnosis and may provide clues for new treatments to halt progression.

Visit www.youtube.com/user/cornearesearch to view a DMEK procedure and more!
Ask Your Doctor
By Dr. Francis Price, Jr.

Q: I have Fuchs's dystrophy and have been researching my different treatment options online. I was excited to learn about DMEK and the results you've experienced. However, I've also been reading about the possibility of using an eye drop to help regrow cells instead of having a cornea transplant. Do you see this as a potential treatment available in the foreseeable future?

A: I'm happy to hear you are taking the initiative in researching your treatment options. Currently, the Japanese are evaluating use of an eye drop, called a RROCK inhibitor, as a potential treatment for corneal endothelial problems, such as with Fuchs's dystrophy. Unlike the U.S., Japan has a severe shortage of donor corneas. For this reason, they have also experimented with growing endothelial cells in a laboratory and injecting them into the eye to see how the results compare with DSEK or DMEK. While it sounds exciting, it will probably take between 5 and 10 years before we know whether these new treatments are safe and effective for use in our patients.

Thank you for your continued interest and support! All donations to the Foundation are tax-deductible and support our valuable research.