

# National Glaucoma Research Report

Better Health Through Research

Winter 2011

## Scientists Find Key Genetic Link to Glaucoma

### Risk levels vary among ethnicities

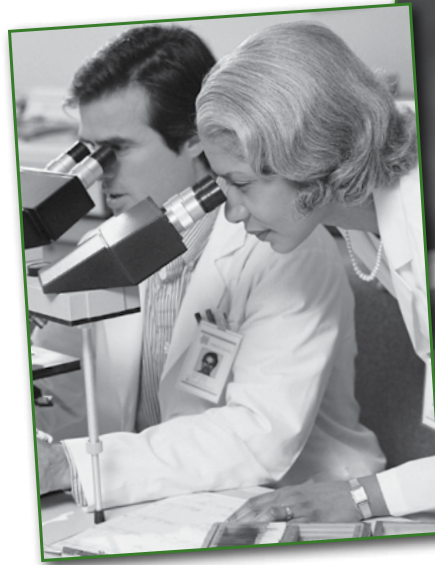
An international team of scientists has discovered perhaps the most important genetic risk factor for primary open-angle glaucoma, according to a study published in *Nature Genetics*.

Working with academic colleagues from Iceland, China, Sweden, the UK and Australia, researchers with deCODE genetics studied more than 40,000 human subjects and isolated a single-letter variation on chromosome 7 that appears to confer greater risk of developing glaucoma.

“The key to reducing the personal and public health impact of glaucoma is early diagnosis and treatment to slow the loss of sight,” says Kari Stefansson, deCODE’s executive chair and senior author of the study. “Discoveries such as today’s are important because we can fold them directly into tests to target screening and to detect and treat more disease earlier.”

Interestingly, the chromosome 7 variation appeared in approximately 6 percent of people with European ancestry but in less than 1 percent of study groups from Hong Kong and Shantou. However, in Chinese subjects, each copy of the variation conferred a much higher level of risk.

“This underscores the value of being able to systematically analyze the impact of genetic risk factors across continental ancestries,” says Stefansson. “Not only are these markers medically useful, they also tell us a bit about evolution and the spread of humanity across the globe.”



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# Detecting Glaucoma in Its Infancy

## New test could catch disease before it causes eye damage

Scientists are moving closer to developing a revolutionary new test that would save eyesight by diagnosing glaucoma years before it can now be detected, according to a study presented at the American Chemical Society.

Currently doctors use two techniques to detect the disease. One measures eye pressure by gently touching a special instrument to the eye's outer surface; the other uses an ophthalmoscope to look through the pupil of the eye at the optic nerve.

"All too often, these tests detect glaucoma after the disease has been silently causing damage to the optic nerve," explains study leader Chenxu Yu, Ph.D. "Years may pass between the first biological change associated with glaucoma inside the eye and diagnosis. We need ways of diagnosing glaucoma earlier, before permanent damage has occurred, so that patients can begin taking medication to control it."



Yu and his colleagues were able to use a common technique from chemistry labs — Raman spectroscopy — to shine a laser light through the eye's pupil. Optic nerve cells scatter the light, creating a rainbow-like spectrum that reveals the cells' chemical composition. Scientists can use this "snapshot" to identify the biochemical changes caused by glaucoma.

Yu and his colleagues have already used the technique on glaucomatous dogs with a 90 percent accuracy rate, and they hope to test it soon in clinical trials on humans. They estimate

the technique could be ready for use in eye doctor's offices within five years.

"We are confident that we're moving toward a breakthrough that will allow us to detect glaucoma at its earliest stage," says Yu. "We hope it will benefit millions of glaucoma patients and individuals at risk for this devastating eye disease worldwide."

## National Glaucoma Research Helping to Solve Mystery of Inherited Glaucoma

A scientist funded by National Glaucoma Research has received a prestigious Pew Scholarship in the Biomedical Sciences for her work in uncovering new treatments for inherited glaucoma.

Raquel Lieberman, Ph.D., of the Georgia Institute of Technology, has focused her efforts on a protein called myocilin, which forms part of the extracellular matrix that regulates eye pressure. When myocilin proteins mutate, eye pressure increases, leading to the retinal degeneration and vision loss associated with glaucoma.

"We hope to discover a drug molecule that will restore the secretion of non-mutant myocilin to the eye's matrix," says Lieberman. "This could prevent vision loss by preventing the buildup of mutant protein, controlling eye pressure and slowing retinal degeneration."

By better understanding the molecular structure of myocilin, Lieberman and her team hope to identify drugs that will successfully bind to the protein.

National Glaucoma Research has contributed \$100,000 toward Dr. Lieberman's work.

# RESEARCH ROUNDUP

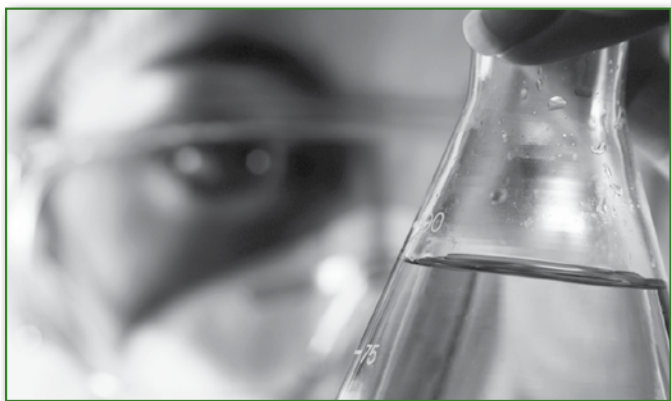
## Vitamin E could help deliver glaucoma medicine



Contact lenses loaded with Vitamin E can keep glaucoma medicine in contact with the eye for nearly 100 times longer than regular commercial lenses, according to a team of scientists at the University of Florida in Gainesville.

Currently, only about 1 to 5 percent of the drugs in eye drops actually reach the cornea of the eye. “The problem,” says lead researcher Anuj Chauhan, Ph.D., “is within about two to five minutes of putting drops in the eye, tears carry the drug away and it doesn’t reach the targeted tissue. Much of the medicine gets absorbed into the bloodstream, which carries it throughout the body where it could cause side effects.”

Chauhan’s team found that clusters of Vitamin E molecules slowed the escape of glaucoma medication from the lens into the eye, increasing the duration of the drug’s release and enhancing its therapeutic impact.



In addition to relieving glaucoma, Chauhan adds, contact lenses treated with Vitamin E could ameliorate other eye diseases, including cataracts and dry eye, while protecting eyes from UV radiation. Clinical trials of the new lenses could begin within a year or two.

## “Fuzzy fibers” shown to enhance eye drainage

Newly designed carbon drainage tubes could give glaucoma patients a better, more lasting alternative to silicone tubes, say researchers with Mobius Therapeutics in St. Louis.

Eye surgeons who operate on glaucoma patients normally use silicone shunts to drain fluid from the affected eye. However, the body often responds to these shunts by producing fibroblasts, a kind of scar tissue that blocks tubes and prevents drainage, necessitating frequent tube replacement.

In response, Mobius owner Ed Timm and his research team, including inventor Khalid Lafdi, distinguished research engineer at UDRI, have produced a special “fuzzy fiber” drain. This carbon scaffold is coated in chemically modified nanotubes that, Timm says, prevent the formation and buildup of fibroblasts.

“Not only is the material completely biocompatible, eliminating the risk for rejection by the body,” says Timm, “it will also serve to preserve the longevity of the implant by keeping it from becoming blocked with tissue. I believe this will completely change the thought process in the design of future ophthalmic devices.”

## Chairman’s Corner

In the absence of a cure for glaucoma, our next best bet is early diagnosis.

That’s because the sooner we catch this disease, the sooner we can begin treating it — and the more successful we will be in arresting vision loss.

Unfortunately, by the time glaucoma presents its symptoms, vision loss has already begun. For that reason, I was particularly excited to hear about a new test — described in this issue of *National Glaucoma Research Report* — that uses

## Catching it early

spectroscopes to track glaucoma from its earliest onset. While the use of this test in doctors’ offices is still years away, it does hold out hope of giving us greater leverage against this terrible disease.

In the meantime, please encourage your loved ones to get regular eye examinations from a qualified professional. We don’t have a cure yet. But if we can catch this disease in its earliest stages, we can give ourselves a fighting chance against it.

# Feel Good About Your Gift



## Planned giving can help fight glaucoma

Through thoughtful planned giving, you can help us search for a glaucoma cure, while also passing along the values that have guided your life.

When you include National Glaucoma Research in your will, your gifts of cash or stock will put you front and center in the fight to discover

treatments that will benefit millions. You may also be able to reduce your estate taxes and leave a larger inheritance for your loved ones.

For additional information, or if you want to discuss planned giving options, please contact Barbara Spitzer, Gift Planning Officer, at 1-800-437-2423 or via email at [bspitzer@ahaf.org](mailto:bspitzer@ahaf.org).

## Thank you for thinking of National Glaucoma Research!



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then simply click on the National Glaucoma Research link to learn more about what's new in the world of research and information about risk factors for glaucoma. You can also follow us on Twitter ([@glaucoma\\_](https://twitter.com/glaucoma_)) or become a fan on Facebook.

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