

In This Issue

- P.1 New Genetic Variants Found in People With Glaucoma
- P.2 President's Corner
Pioneering Research Reverses Glaucoma Induced Vision Loss
- P.3 Spotlight On...
Dr. Karen Curtin
Gene Therapy and Glaucoma
- P.4 Tips for Caring for Someone With Glaucoma
- P.5 New Recipe:
Mango Salsa Pizza
Supporting Your Family and Glaucoma Research

New Genetic Variants Found in People With Glaucoma

Your support helped make this breakthrough possible

A major international science team at Massachusetts Eye and Ear Infirmary that included several National Glaucoma Research-funded scientists has identified dozens of new glaucoma genes and genetic variants in people who have glaucoma. This major study, the largest of its kind, compared the genes of people with glaucoma to people without it.

“Glaucoma is one of the most strongly genetic human diseases, which is why we are looking at the genetic architecture of the disease to find clues on how to prevent and treat it,” Professor Stuart MacGregor, a co-senior researcher on the study, said in a recent press release.* “We’re hopeful that understanding the biological processes and knowing which genes control them could help scientists develop new drugs in the future.”

By contrasting the genetic codes of all participants, researchers also identified 44 new gene loci and confirmed the placement of 83 previously reported loci with connections to glaucoma.

Loci are a fixed spot on a chromosome where specific genes are located. By finding a particular gene’s “address” through loci, researchers hope to improve screening, diagnosis, and treatment of glaucoma.

Most prior genetic studies focused on people of European descent, despite African and Asian ancestry groups having the highest rates of glaucoma. This study was notable for being the first to compare people of European, African, and Asian descent. The findings of this large study set the stage for reexamining the therapeutic options for glaucoma.

**Massachusetts Eye and Ear Infirmary, News Release, 02/24/21*



Researchers hope to develop new glaucoma drugs, treatments, and screenings from this discovery.



President's Corner

Imagine a world where eye doctors could treat people at risk for glaucoma . . . *years before their vision was affected.*

It might sound impossible, but as you'll read in this issue, exciting new research is advancing vision science quicker than ever before. New discoveries are broadening our understanding of how glaucoma changes the eye and even helping us understand who is more likely to develop it.

The progress being made right now was scarcely imaginable even five years ago. Advances like the ones listed inside offer hope to anyone who has glaucoma or has a loved one affected by it. We still have a long way to go to reach a cure, but we're getting closer every day.

Thank you for standing with us, and for making this life-changing research possible.

Stacy Pagos Haller
President

Pioneering Research Reverses Glaucoma- Induced Vision Loss



Promising treatment may hold the key to restoring sight.

A collaboration by Harvard scientists has successfully reprogrammed cells in mice to reverse vision loss from glaucoma, as well as normal (not disease-related) vision loss associated with aging. Their pioneering work is the first demonstration that it may be possible to safely reprogram complex tissues, such as the nerve cells of the eye, to an earlier age.

Harvard researchers Bruce Ksander, PhD, and Meredith Gregory-Ksander, PhD, along with their colleagues, used a gene editing technique that can reverse chemical modifications that naturally happen to key genes over time. This treatment is shown to promote optic nerve regeneration and reverse vision loss due to injury as well as natural aging.

Drs. Gregory-Ksander and Ksander attribute their success in this discovery, in part, to the early support they received from National Glaucoma Research and donors like you.

“Regaining visual function after the injury occurred has rarely been demonstrated by scientists,” Dr. Gregory-Ksander said. “This new approach, which successfully reverses multiple causes of vision loss in mice without the need for a retinal transplant, represents a new treatment modality in regenerative medicine.”

SPOTLIGHT ON ... Dr. Karen Curtin

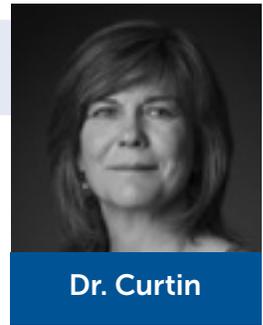
Preventing Vision Loss by Predicting and Treating Exfoliation Syndrome

Exfoliation syndrome (XFS) is marked by the accumulation of abnormal threadlike white fibers throughout the body over time. The buildup of these fibers in the front of the eye is a common cause of glaucoma. However, it is not yet known why certain individuals with XFS have rapid progression to vision loss, while others never develop glaucoma.

Karen Curtin, PhD, with the University of Utah, believes that by studying the records of exfoliation patients, her team can develop a targeted way to accurately predict which patients' eyes will rapidly develop glaucoma. Their hope is to eventually create an easy-to-administer

prediction tool for clinicians that can be widely used for patient assessment and referral, as well as timing for intervention, such as medication or surgery, to lower intraocular pressure.

"It is my belief that using resources available to us, we can and will make a difference in the quality of the lives of patients with exfoliation of the eye who are at risk for developing debilitating glaucoma," Dr. Curtin said. "By identifying factors that can predict progression and developing a simple tool to assess these conditions early on, clinicians will be able to intervene much earlier to ameliorate future vision loss in the care of their patients."



Dr. Curtin

"We are extremely grateful to have the opportunity that a National Glaucoma Research award affords to assemble a top-notch research team and undertake the rigorous scientific investigation we propose."

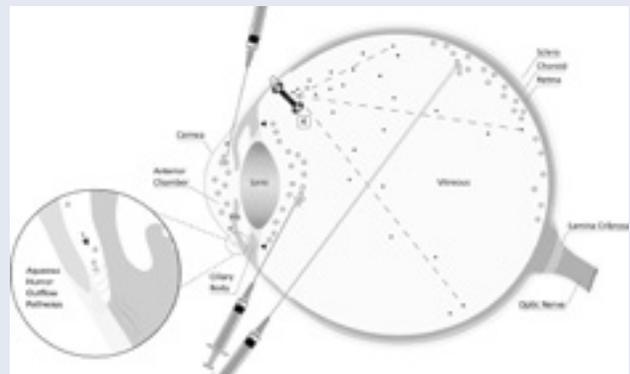
Gene Therapy and Glaucoma

New therapies show much promise for new treatments

With ocular gene therapy, genetic material such as DNA or RNA is injected into the eye with the purpose of achieving a therapeutic effect. One common and effective way to administer gene therapy is by using benign virus particles, or vectors, that carry the genetic material into the cells of interest.

The advantages of gene therapy include the ability to specifically target known disease mechanisms and cells, and the potential that a one-time treatment will result in a long-term therapeutic effect and possibly a cure. By injecting the gene therapy vector directly into the eye, the treatment reaches the desired target more effectively than by eye drops or oral medications. The eye is well suited for gene therapy because it forms a separate compartment where inflammation is relatively muted compared to other organs; this is referred to as "immune privilege."

Gene therapy can be used to address genetic risk factors by either replacing or silencing defective genes that contribute to the development and progression of glaucoma. Gene therapy can also be applied to enable cells within the eye to produce drugs continuously, thereby replacing the need for daily eye drop medication. The first ocular gene therapy was approved for treatment of one form of retinal childhood glaucoma just three years ago.



Tips for Caring for Someone With Glaucoma

If you have a friend or family member with glaucoma, you can provide meaningful, caring, and practical support. Here are some ways to help your loved one with glaucoma:

Low-Vision Aids

Help your loved one acquire optical devices, called low-vision aids, which can help people better use their remaining vision and keep doing tasks and the things they enjoy.

Many low-vision aids are covered by health insurance. Some examples include:

- Customized magnifiers for reading, knitting, and other near tasks
- Computerized text-to-speech devices
- Handheld or spectacle-mounted telescopes for seeing objects in the distance

Home Safety

You can help your loved one make adjustments at home that can improve visibility and reduce the risk of a fall. Suggestions include:

- **Lighting:** Make sure the home is well lit with high-wattage light bulbs and additional lamps.
- **Organization:** Remove unnecessary household clutter. Offer to help organize and label important items.



- **Contact information:** Create a list of important phone numbers in large print on bold-lined paper. Include doctors, transportation, and emergency contacts, and put the list in a convenient place.
- **Mobility:** Use brightly colored tape to mark stairs or slopes. Eye-catching colors that contrast with the flooring work best.

Although it may be difficult for your loved one to ask for help, it's important to communicate openly and clearly. Encourage him or her to be specific about what you can do to help. Caring for a loved one with glaucoma can be rewarding but also challenging—be sure you meet your own needs as well.

For other resources for people living with glaucoma, visit brightfocus.org/NGRresources.

Please clip & keep.

Mango Salsa Pizza

A tasty and simple citrus twist on a classic dish.

Ingredients

- 1/2 cup chopped red bell peppers
- 1/2 cup chopped green bell peppers
- 1/2 cup chopped mango
- 1/2 cup pineapple tidbits
- 1/2 cup minced onion
- 1 Tablespoon lime juice
- 1/2 cup chopped fresh cilantro
- 12-inch premade whole-grain pizza crust



Instructions

1. Preheat the oven to 425°F. In a small bowl, combine the peppers, mango, pineapple, onion, lime juice, and cilantro; set aside.
2. Coat a pizza pan with cooking spray and roll the pizza crust over it. Place it in the oven and bake for 15 minutes.
3. Take the pizza crust out of the oven and cover it with the mango salsa mixture. Place the pizza back in the oven and bake until the toppings are hot, about 5-10 minutes or until the crust is browned. Slice and serve immediately.

Makes four servings.

Supporting Your Family and Glaucoma Research

If you are looking for a way to pass on some of your assets to your family while reducing or eliminating gift or estate taxes, a charitable lead trust is an excellent option. By making a contribution of your property to fund a trust, you can support National Glaucoma Research while receiving a gift or estate tax deduction at the time of the gift. Then, after a set period, your family receives the trust asset, plus any additional growth in value.

If you have already designated us as a beneficiary of a charitable lead trust, please let us know. Contact Charlie Thomas at 301-556-9362 or plannedgiving@brightfocus.org so we can thank you for your foresight, welcome you into our Heritage Society, and ensure that your gift is used as you intend. For more information regarding all the ways you can leave a legacy of support, go to brightfocus.org/plannedgiving.

Thank you for supporting National Glaucoma Research!

Please share this newsletter with someone who might be interested in learning more about some of the latest advancements in research to diagnose, prevent, treat, and cure glaucoma. This newsletter is published by National Glaucoma Research, a program of BrightFocus Foundation®, a nonprofit organization located at 22512 Gateway Center Drive, Clarksburg, Maryland 20871, 301-948-3244, brightfocus.org.

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