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## Scientists Discover Key Gene Associated with Glaucoma

Blinding diseases that affect children and young adults are mainly caused by defective genes, which are typically passed on from a parent to their children. One group of National Glaucoma Research–funded researchers has made it their mission to identify the culprit genes and find out how they act to cause blindness.

They recently discovered a gene called *GLIS1*, which is critical to maintaining the health of the tissue that regulates fluid drainage and pressure in the eye. Working with mouse models that are engineered to lack the *GLIS1* gene, researchers discovered the health of the trabecular meshwork is maintained in part by a protein product made from the *GLIS1* gene. Without the protein, the tissue degenerates.

This led researchers to propose that the *GLIS1* protein acts on a set of genes, rather than one particular gene, and may therefore affect the trabecular meshwork in numerous ways. Most importantly, they have detected a significant association between common genetic variants in *GLIS1* and glaucoma, thereby supporting the role of *GLIS1* as a glaucoma risk gene.

By further understanding the role this gene plays, scientists hope to be able to target ways to prevent or slow down the progression of glaucoma, and to find new insights into the origin of glaucoma inside the body.



**National Glaucoma Research–funded scientists have discovered a critical new glaucoma gene.**



## President's Corner

As we start the new year strong, I am excited by both the promising new research that will be conducted in 2022, and the results we are seeing grow out of prior science supported by National Glaucoma Research.

For instance, we are always excited to hear about additional research funding that builds upon the key, early grants that promising researchers received from us early in their careers. Recently, the NIH's National Eye Institute has announced the latest recipients of their Audacious Goals Initiative grants.

These grants are some of NIH's most prestigious vision research awards, and one of these is for a project led by a team of five scientists who are all current and past National Glaucoma Research grantees.

This is further evidence and strong affirmation for the high-quality science supported by National Glaucoma Research—and you. Your generosity is making a difference. Please enjoy this issue of your newsletter. Thank you.

Stacy Pagos Haller  
President

## The Importance of Your Eye's Drainage System

There are many different types of glaucoma (open- and closed-angle are the two most common forms), but they all share one thing—the importance of the trabecular meshwork for drainage.

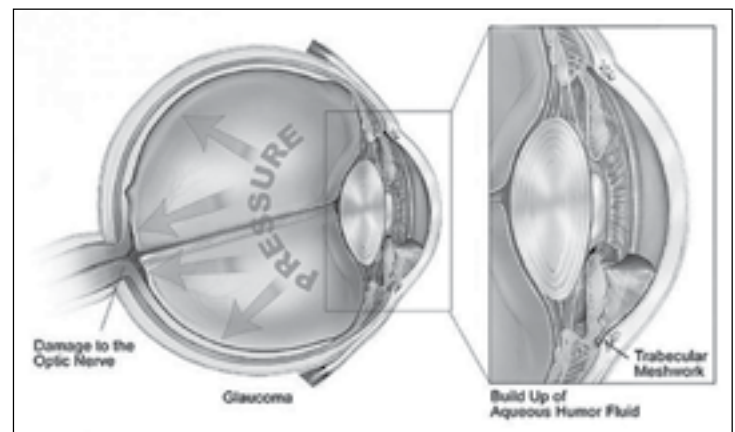
The majority of fluid (aqueous humor) draining out of the eye exits via the trabecular meshwork, through a structure called Schlemm's canal, into collector channels, then into the veins, and eventually back into the body's circulatory system.

When the trabecular meshwork functions improperly—whether due to increased age, congenital defect, or trauma—the outflow of fluid is decreased, raising pressure inside the eye and increasing the risk of developing glaucoma.

Most medications either decrease aqueous humor production or redirect the fluid away from the trabecular meshwork. Other, relatively newer medications can directly target the diseased trabecular meshwork to increase drainage.

In addition to medication, other treatments for glaucoma include laser trabeculoplasty (which vaporizes the pigment that may be affecting the cells in the drainage system to improve the drainage of fluid) and surgery.

Often considered minimally invasive, many glaucoma surgeries involve inserting stents into the eye to bypass the trabecular meshwork altogether, thereby decreasing eye pressure by routing fluid through Schlemm's canal.



**New drugs on the horizon may directly target the trabecular meshwork to decrease pressure inside the eye.**

## SPOTLIGHT ON ... Dr. Puya Gharahkhani, PhD

### Artificial Intelligence and Identifying the Risk for Glaucoma

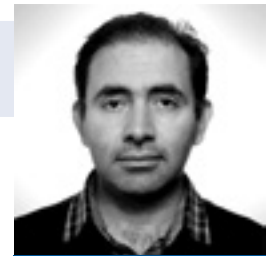
One of the first large-scale genetic studies to use artificial intelligence (AI) to accurately grade glaucoma-related damage of the optic nerve and retina is currently underway. Led by Puya Gharahkhani, PhD, at the QIMR Berghofer Medical Research Institute in Brisbane, Australia, and funded by National Glaucoma Research, this is the first study to investigate the genetic basis of eye nerve damage over time using AI approaches.

Current treatments for glaucoma are not capable of preventing blindness once the optic nerve is already injured. The study may uncover new potential candidate treatments for glaucoma, which are beneficial even after the nerve

is damaged. This new and time-efficient approach uses artificial intelligence to grade eye nerve damage, which will improve the accuracy of discovering a person's genetic risk for glaucoma.

These genes will subsequently be used to develop a genetic test to identify people at high risk of developing eye nerve damage and glaucoma, and may allow for treatment of the damaged eye nerve, which currently is not possible.

“Thank you very much for your support of National Glaucoma Research and of my work,” Dr. Gharahkhani says. “Together, we can make a difference for the future by preventing blindness.”



Dr. Gharahkhani

Dr. Gharahkhani's work will help us better understand how glaucoma damages the eye and how it might be prevented.

### Scoring Glaucoma Risk Based on Genetic Factors

New research leads to understanding glaucoma risk

People with a close relative who has glaucoma have a greater risk of developing the disease themselves. However, no single genetic mutation is responsible; instead, subtle variations in the “coding” (or chemical makeup) of genes may be contributing factors. In research jargon, these variations are known as single-nucleotide polymorphisms (SNPs) and have been identified through large collaborative studies analyzing enormous numbers of genetic samples. After years of discovery, National Glaucoma Research-funded scientists have zeroed in on the most common area of genetic risk for glaucoma, which involves variants in the myocilin (MYOC) gene. They have developed and tested a polygenic risk score (PRS) based on the presence of those variants. Highest-scoring individuals are at risk for glaucoma 10 years earlier than the lowest-scoring ones, and

they are at a 15-fold increased risk of developing advanced glaucoma requiring surgical intervention. Researchers hope their glaucoma PRS will soon help eye doctors personalize treatments, including earlier intervention for high-risk individuals and possibly pursuing less intensive monitoring and treatment in lower-risk groups.



## Turkey Chili

*Stay warm with this healthy carb and high-fiber meal.*

### Ingredients

1/2 pound chopped cooked turkey

2 cups canned kidney beans, rinsed  
and drained

1 cup chopped zucchini

1 cup chopped celery

1 cup diced tomatoes

1 cup low-sodium vegetable broth

1/2 cup chopped onion

1/2 cup chopped bell peppers

1 teaspoon chopped fresh garlic

3/4 tablespoons chili powder

1/2 teaspoon olive oil

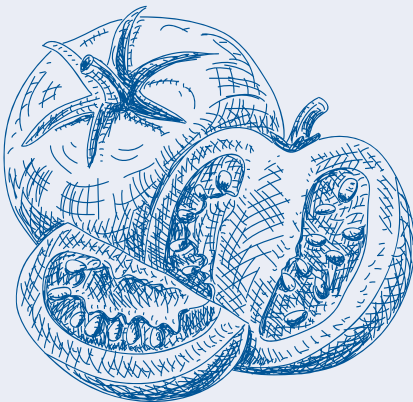
1/2 teaspoon cumin seed

1/2 teaspoon brown sugar

### Instructions

1. Preheat your oven to 475<sup>0</sup> F and coat a glass baking dish with cooking spray.
2. Place zucchini in a single layer in the dish, and roast for 8 to 10 minutes or until tender and lightly browned.
3. While the zucchini roasts, place oil and onions in a soup pot. Sauté the onions until brown, then add celery and peppers, and continue to sauté.
4. Add turkey, garlic, cumin seed, and chili powder to the soup pot. Cover and simmer for 5 minutes.
5. Stir in the tomatoes, brown sugar, beans, broth, and roasted zucchini. Cover and simmer for 15 minutes.
6. Serve immediately and enjoy.

*Makes four servings.*





# Eye Health for the New Year

For people with glaucoma or at risk of developing it, a healthy lifestyle that includes regular exercise and a nutritious diet is especially important. In addition to benefiting one's overall physical and mental well-being, it promotes eye health. Maintaining mental and emotional health is also important. Here are some of our tips for healthy living:

## Eat a Varied, Healthy Diet

- Carotenoids, antioxidants, vitamins A and D, zinc, and omega-3 fatty acids may all contribute to better vision.

## Limit Caffeine

- Evidence suggests that high amounts of caffeine may increase eye pressure.

## Exercise

- Studies indicate that exercise can lower eye pressure.

## Maintain a Healthy Weight

- It keeps blood pressure at a normal level and helps control other medical conditions.

## Stop Smoking

- Smoking increases blood pressure and inflammation in the eye, which leads to an increased chance of cataracts and diabetes.

## Protect Eyes from Sunlight

- Prevent overexposure to sunlight by wearing wide-brimmed hats and high-quality sunglasses.

## Visit Your Eye Doctor Regularly

- Comprehensive eye exams should be conducted as often as your eye doctor recommends.

## Take Medications as Prescribed

- Take any prescribed medications consistently and exactly as directed by your eye doctor.

## Stop Glaucoma and Receive Income for Life

If you are tired of the fluctuating stock market and want to receive fixed payments, a charitable remainder annuity trust may provide you with the stability you desire. It pays a fixed amount to you or your designated beneficiaries each year, based on the value of the property at the time the trust is funded. The funds remaining in the trust after all payments have been made will be used to help end glaucoma.

If you have already designated National Glaucoma Research as a beneficiary of a trust, please let us know. Contact Charlie Thomas at 301-556-9362 or [plannedgiving@brightfocus.org](mailto:plannedgiving@brightfocus.org) so we can thank you for your foresight, welcome you into our de la Cuesta Legacy 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](http://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](http://brightfocus.org).

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