

MACULAR DEGENERATION CENTER



INSIGHT

Genetic Testing for AMD? Promising but Premature, Say Experts

If your mother or brother suffers from age-related macular degeneration (AMD) can genetic testing reveal if you are likely to get it too? Can it also help determine what type of therapies you need to help prevent the disease, such as certain vitamin and mineral supplements?

Thanks to years of careful research coupled with recent technological advances, scientists are getting closer to answering these questions. Has the time come to rely on genetic testing to complete the picture?

Like other complex conditions, such as heart disease or cancer, AMD results from a combination of many factors, including your age, health habits such as smoking, and genetic makeup, says Michael Klein, M.D., director of Casey Eye Institute's Macular Degeneration Center. The disease, which normally affects people 50 and older, becomes more common as you age. Studies show that for people over age 65, about 3 percent have advanced (late) AMD, usually with some degree of visual impairment, while an additional 22 percent have changes in their retina. This means they are at increased risk of developing these late stages of the disease.

"In recent years, several genetic associations have been uncovered leading us to a better understanding of the disease," says Dr. Klein. For example, a group of genes strongly linked to late-stage AMD helps regulate the immune system, which influences inflammation in the human body. Scientists suspect there may be a con-

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Genetics of AMD *(Continued from page 1)*

nection between inflammation and development of AMD in patients with these gene mutations. Other gene defects associated with AMD involve lipid metabolism and changes in connective tissue.

As more is learned about AMD's underpinnings, medical researchers are using the information to help design new and more effective drug therapies and preventive measures. In addition, investigators are studying whether genetic information can help predict an individual's likelihood of developing the disease, as well as guide the use of current therapies.

Time for Genetic Testing?

What about the current role, if any, for genetic testing for AMD? As scientists continue to zero in on more AMD genes, questions arise about the value of routine genetic testing for at-risk patients, and to help decide the best treatment. In fact, various versions of these genetic tests are being recommended in some ophthalmology and optometry practices. Proponents of genetic testing claim that current tests provide enough information to justify the additional expense of the procedure, which is not covered by Medicare or Medicaid.

However, most experts in the field, including leading AMD clinicians, geneticists, and genetic statisticians, feel that routine genetic testing for AMD is not warranted at the present time. With regard to predicting risk of AMD, "genetic testing currently adds little to the known risk factors for progression to advanced (late) AMD," states Dr. Klein. "The most important of these factors is the appearance of the retina in an eye examination. Age, smoking history, and family history also contribute to the disease."

An ophthalmologist can estimate risk based on these facts, or use an online tool developed by Casey ophthalmologists in collaboration with the National Eye Institute (available on the Casey website www.ohsucasey.com). Although the tool can include genetic test results if available, the added information only minimally alters the risk estimate. Other proposed reasons for genetic testing, such as adjusting treatment and preventive measures—like the AREDS supplements—have yet to be proven accurate and reliable, according to most experts.

The View of the American Academy of Ophthalmology

The American Academy of Ophthalmology (AAO) agrees with this assessment of genetic testing for AMD. A recent review of the subject published by the AAO's Task Force on Genetic Testing recommends avoiding "routine genetic testing for genetically complex disorders like AMD" until it can be shown that patients with certain AMD genes can benefit from specific treatments or monitoring. In the meantime, the task force advises that genetic testing be confined to research.

"We agree with the current view of the AAO," says Dr. Klein. "All patients can benefit from a healthier lifestyle and regular eye exams," he says. "Although it is recommended that genetic testing be limited to research at this time, it has a very promising future in the clinical setting," says Dr. Klein. "We are confident that it will play a major role in determining who is at risk of advanced AMD and how they will respond to certain therapies." ●

Research Roundup

Current research studies at the Macular Degeneration Center include:

Gene Therapy for Wet AMD

Purpose: To evaluate the safety and dosing levels of a gene-based treatment, RetinoStat®, for wet AMD. In this study, two helpful genes are delivered directly to the retina, where they “turn on” proteins that block abnormal blood vessel growth in a sustained fashion. Enrollment is completed and study patients are being followed.

OCT Trials

Purpose: To test the capabilities of high-speed optical coherence tomography (OCT) in patients with dry or wet AMD. OCT uses light waves to produce extremely detailed cross-sectional images of eye structures. Investigators are studying whether this new version of OCT can visualize and measure blood vessel growth as well as fluorescein angiography, which involves the injection of a contrasting agent to highlight the problem vessels. The team is also comparing how retinal anatomy and blood flow differ among study patients in early, intermediate and advanced dry AMD.

Genetics of Age-Related Macular Degeneration Studies

Researchers at the Macular Degeneration Center are using advanced whole genome sequencing technology to uncover gene variations in large families and other populations affected by AMD. Finding these genes casts light on the mechanisms of the disease and is the key to more accurate early detection and precise treatments. The study, supported by a major grant from the National Eye Institute, is being carried out in partnership with genetic statisticians at the Texas Biomedical Research Institute in San Antonio, Texas, as well as other collaborators in the U.S. and elsewhere.

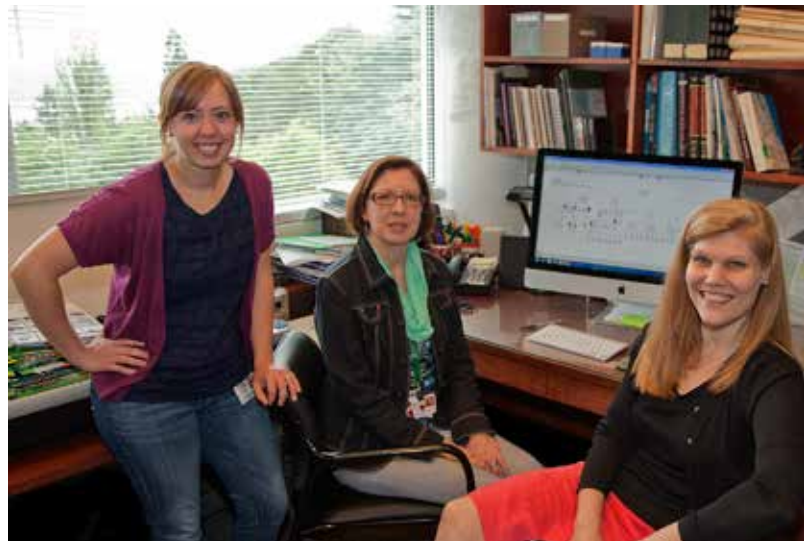
SEATTLE Study (For Dry AMD)

Purpose: To learn if an oral study medication, emixustat hydrochloride, slows progression to advanced dry AMD (geographic atrophy) compared to placebo in people with dry AMD. Enrollment is completed and study patients are being followed.

On the Horizon

The Macular Degeneration Center expects to launch several new clinical trials of experimental medications for both wet and advanced dry AMD. More details will be available in the coming months.

For more information about our research activities, please contact the program coordinator at 503 494-3537 or visit our website at www.caseyamd.com



Members of the Genetics of AMD research team include l to r: Laura Quinn, Tammy Martin, Ph.D., and Jennifer Maykoski.

Ask an Expert: Christina Flaxel, M.D.

Nutritional Supplements for Age-Related Macular Degeneration

What kind of dietary supplements are recommended for age-related macular degeneration (AMD)?

We recommend the formula based on the Age-Related Eye Disease Studies (AREDS and AREDS 2). These were large-scale clinical trials sponsored by the National Eye Institute that were conducted over many years. The supplements, a high-dose combination of antioxidants and zinc, were shown to reduce the risk of advanced (late) AMD and vision loss in people at high risk.

I am in the early stages of AMD. Should I be taking supplements?

The studies showed that the supplements were helpful for people with moderate macular degeneration or who have advanced macular degeneration in one eye. It did not benefit individuals in the early stages of the disease or whose eyes are healthy. Your eye doctor can tell you what stage you are in.

Are there side effects?

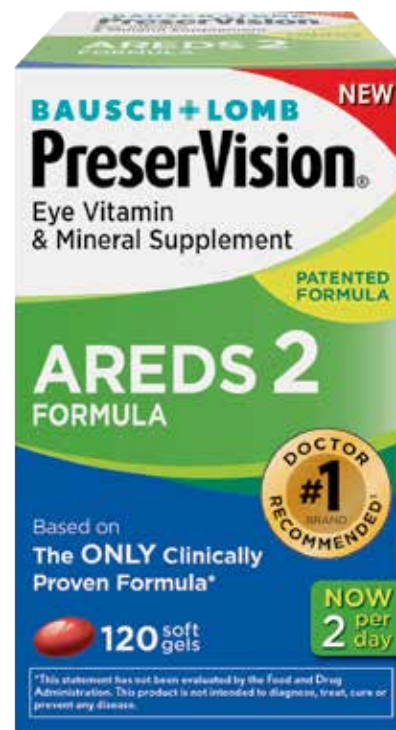
The first AREDS study found few possible side effects. However, beta carotene, which was included in the original formula, may raise the risk of lung cancer in smokers and people who have recently quit. We recommend smokers and non-smokers stick with an AREDS 2 formula that replaces beta carotene with lutein and zeaxanthin.

Because these products contain high levels of nutrients, be sure to check with your primary care doctor or eye doctor before starting the regimen, especially if you are being treated for a chronic disease and taking several medications.

Where can I find AREDS supplements?

They are available without a prescription in stores that sell dietary supplements. Although there are several formats and types, we suggest patients take the formula with lutein and zeaxanthin instead of beta carotene.

Below is an example of an eye vitamin and mineral supplement containing the currently recommended formula based on AREDS 2.



PreserVision® Eye Vitamin AREDS 2 Formula

	Per Serving	Per Day
Vitamin C	250 mg	500 mg
Vitamin E	200 IU	400 IU
Zinc	40 mg	80 mg
Copper	1 mg	2 mg
Lutein	5 mg	10 mg
Zeaxanthin	1 mg	2 mg

New Co-Director of the Macular Degeneration Center



Christina Flaxel, M.D., Professor of Ophthalmology at OHSU Casey Eye Institute, has been named the new Co-Director of the Macular Degeneration Center, joining Director Michael Klein, M.D., in this leadership

role. A faculty physician in the Retina Division for more than seven years, Dr. Flaxel has a special interest in the treatment and research of macular degeneration and has been an investigator in several important studies of the disease.

"I am delighted to help lead the Macular Degeneration Center during such an exciting period of discovery in the lab and clinic," says Dr. Flaxel. "Not only is the Center involved in supporting this crucial work, but we are committed to raising awareness about the disease and recent advances in prevention and treatment," she says, noting that "our public education efforts will become more urgent" as our population grows older and more people are diagnosed with AMD. ●

Oregon Program Loans Special Telephone Equipment for Sight Impaired

Do you or does someone you know have difficulty using a telephone because of vision loss? Oregon's Telecommunications Devices Access Program (TDAP) loans adaptive telephone equipment at no charge to eligible Oregon residents who have a hearing, vision, speech, thinking or mobility impairment. The devices include big button phones and separate caller I.D. units using voice or large print. The phones also have other helpful features, such as talking key pads, amplified sound and a built-in loud ringer.

For more information about the program, call 800 848-4442 or 503 373-7171 or visit www.rspf.org



Free Audio Recordings Offer Tips for Managing Vision Loss

Hadley, a nonprofit organization that supports people with vision loss, is offering a series of free audio recordings on CD that provide tips and techniques to handle everyday tasks. Topics range from marking your stove or oven and managing medications to indoor mobility and using adaptive devices and technology.

The recordings encourage independent living and teach individuals how they can continue their favorite activities with diminished vision. To get started, call the Low Vision Focus@Hadley program at 1 855 830-5355 or visit www.LowVisionFocus.org to register. You do not need to be legally blind to participate in the program.

Simple Lighting Tips Can Brighten Your Life

If you're one of the millions of seniors with age-related macular degeneration — or AMD — reading a newspaper or book may be a frustrating experience. In some cases, gradually impaired central vision may hamper your ability to see fine details, such as newsprint, the instructions on a prescription bottle or the stitches in your needlework project.

While your first instinct may be to shop for a magnifier, using the right kind of lighting for close-up tasks can make all the difference in the world, says John Boyer, O.D., clinical director of OHSU Casey Eye Institute's Vision Rehabilitation Center. One of the troubling symptoms of AMD is a decline in contrast sensitivity, which can make reading particularly challenging. "Patients may think a magnifier can make up for the loss, but what you really may need is better illumination. Contrast improves dramatically with increased lighting positioned close to the reading material," he explains.

Patients visiting the rehabilitation center at Casey often are pleasantly surprised to find how much easier it is to make out letters on the eye chart or to read a brochure



John Boyer, O.D., demonstrates proper positioning of lighting

when a light is aimed directly on the reading material, says Dr. Boyer. "The key is to bring a light to you, not the task to the light," he says.

In the dark about proper lighting for your vision problems? Give these five easy strategies a try.

1. For best reading, use lights that can be placed 4 to 7 inches from the print. This distance is needed whether or not you are reading with special glasses or a magnifier.

2. Try different kinds of light bulbs. Options are compact fluorescent, halogen or LEDs. (Traditional incandescent bulbs are being phased out and may no longer be available at your local store). Whichever type of light you use, be sure to place the bulb closer to the page than you are.

3. If seated at a table or desk, place a lamp right next to your reading. Sitting in your living room chair or bed? You will need a lamp with a long arm that will reach far enough so that the light bulb can be placed in front of your shoulder, not over your shoulder.

4. Before heading out to a restaurant or theater, tuck a small LED light into your pocket or handbag. While the darker mood lighting may be romantic, it may make viewing the menu or program nearly impossible. You can also use the light from your smart phone or a flashlight app.

5. Browse your local home improvement store and online to find a variety of lighting options. Also consider retail outlets that specialize in office products or art and craft supplies. ●

Save the Date!

Macular Degeneration and Low Vision Expo

September 27, 2014
Doubletree Hotel – Lloyd Center
1000 NE Multnomah – Portland, Oregon
Open to the Public
9 AM to 4 PM

Exhibits of visual aids and community resources

Presentations by Casey faculty physicians

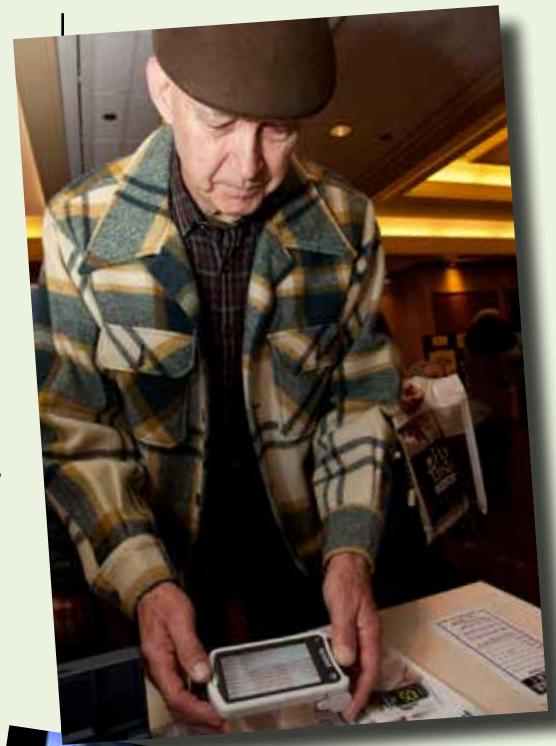
- Diagnosis, treatment and research advances for age-related macular degeneration
- Managing vision loss — the tried, the true and what's new

Small group sessions led by instructors from Oregon Commission for the Blind

- Using Apple products (iPhone, iPad & iTouch) to read, communicate, navigate & more
- Android devices and low tech products for everyday life

Details about registration, exhibitors and event schedule will be mailed to subscribers of this newsletter later this summer and posted on our website, www.caseyamd.com

For more information or to be added to the Insight newsletter mailing list, call the Macular Degeneration Center at 503 494-3537 or email kahnj@ohsu.edu





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Connect with the Macular Degeneration Center!

Education and outreach is a top priority of Casey Eye Institute's Macular Degeneration Center, a national leader in research and patient care for age-related macular degeneration (AMD). If you'd like to be on our mailing list to receive the *Insight* newsletter and other information — or have a speaker for your group, please call the center at 503 494-3537 or email us at kahnj@ohsu.edu.

Learn more about AMD and the work of the Macular Degeneration Center at www.caseyamd.com