Research to Prevent Blindness

NEWS

FALL 2015

EYE RESEARCH MATTERS

Lifestyle Choices Can Drive AMD Disease Risk

Everything you already (should) know about reducing your risk for developing age-related macular degeneration (AMD) is even more important if you have a genetic predisposition for the disease. That finding comes from a new study co-funded by RPB and the National Eye Institute, which indicates that there is an interaction between genes and risk-influencing behavior.

First of all, unhealthy dietary choices, smoking and lack of exercise increase your AMD risk whether or not you carry genes associated with greater AMD risk. But, if you have inherited a version of the complement factor H (CFH) gene that is known to be associated with greater AMD risk, then a history of heavy smoking, consistently not exercising or eating enough fruits and vegetables significantly raises your risk of developing AMD.

Earlier studies have shown that: eating a healthy diet (with plenty of dark, leafy greens, colorful fruits and vegetables, steamed or broiled cold-water, fatty fish, and nuts) can slow the progression of AMD; and getting exercise (at least 10 hours a week of light exercise or at least eight hours of moderate activity such as brisk walking) and stopping or never smoking significantly reduce your AMD risk.

So, risky CFH gene or not, by making healthy lifestyle choices you may be able to lower your risk of AMD or slow its progression.

Eye Drops Treat Cataracts in Dogs, Humans Next?

RPB-supported researchers have partially reversed cataracts in dogs using eye drops and injections of a naturally occurring steroid in the human body. The discovery is pointing to an eye drop treatment for cataracts for people, and the impact will be global.

Cataracts are the leading cause of blindness worldwide, accounting for more than half of all cases. They most commonly occur over time, with age, when the transparent, tight protein structure of the human lens is disrupted, usually by protein aggregation caused by oxidative stress.

The National Eye Institute estimates that by age 80, more than half of all Americans either have a cataract or have had surgery to remove one.

The only treatment now available for cataracts is surgical removal of the natural lens and replacement with an artificial lens. While the surgery has very high success rates, in developed countries it contributes to higher healthcare costs. As populations age, estimates indicate that cataract surgeries will double in the next 20 years. Because cataract surgery emerges as an option later in life, delaying the development of the disease by as little as ten years might eliminate the need for nearly half of those surgeries.

Enter lanosterol, which the human body produces as a precursor to the creation of cholesterol. RPB-supported scientists identified a genetic mutation that interfered with the production of lanosterol in the cases of three children who had a severe, inherited cataract condition. In lab tests, lanosterol was shown to dissolve aggregated lens proteins.

"While we have yet to determine precisely how lanosterol is working, we are gratified that the treatment is working," said to RPB resesarcher Dr. Kang Zhang, professor of ophthalmology and chief of ophthalmic genetics at UC San Diego.

Before testing can begin in humans, researchers will check the toxicity of lanosterol, then create an eye drop version of the drug, hopefully within a year.

EYE RESEARCH MATTERS

Don't Forget Your Vitamin D

In another RPB/NEI co-funded study, deficient vitamin D levels were also found to raise AMD risk. "These findings support the notion of biologic synergy. That is, that one's genes, lifestyle factors and nutrition all come together in a synergistic way to mediate inflammation, which is a key mechanism involved in AMD," said Julie A. Mares, PhD, of the University of Wisconsin-Madison. "There's a large body of evidence that unhealthy lifestyle habits are associated with inflammation and that CFH risk alleles augment inflammatory responses. Vitamin D is believed to suppress inflammation, which is thought to enhance the AMD disease processes both directly and indirectly."

Ebola Lingers In the Eye

Ebola may remain in a surviving patient's eye, causing severe ocular complications. That's the finding in an RPB-supported study conducted at Emory University, where Dr. Ian Crozier—who contracted the virus while working in an Ebola treatment center in Sierra Leone—returned two months after his near-death-but-successful treatment for the disease. He was experiencing intense eye pain, soaring eye pressure and vision loss. Without immediate grant support to explore the dramatic development, the Emory department of ophthalmology was able to apply RPB's Unrestricted Grant to conduct this timely research. A combination of anti-inflammatory and experimental anti-viral drugs eventually led to the restoration of his sight. The discovery has wide implications for thousands of Ebola virus disease survivors and health care providers in West Africa.

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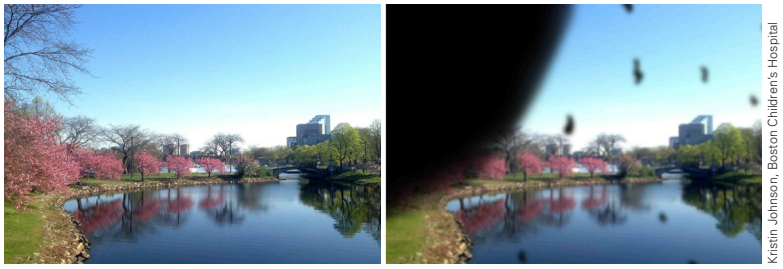

Kristin Johnson, Boston Children's Hospital

Retinal detachment can cause visual field loss, floaters and blurring. If untreated, the detachment can progress, leading to loss of all vision.

Preserving Sight Following Retinal Detachment

RPB-supported vision researchers have identified targets for treatments that would prevent vision loss in patients following retinal detachment. The discovery holds promise for the many patients who, even though they have corrective surgery soon after experiencing a retinal detachment, continue to lose sight. The research shows, for the first time, that a component of the body's immune system significantly contributes to photoreceptor cell death and that inhibiting the pathway is protective.

"It is our hope that future studies will allow us to develop specific therapeutics that harness this knowledge," says RPB Special Scholar Award recipient Kip M. Connor, PhD, Harvard Medical School. Ultimately, the findings may have much broader impact, because degeneration of photoreceptors also occurs as a result of blunt trauma or as a side effect of a variety of diseases including diabetic retinopathy, ocular tumors, retinopathy of prematurity, and age-related macular degeneration.

Renewed Hope for Gene Therapy for RP

A few years ago, in a major milestone, RPB-supported researchers used gene therapy to restore partial sight in patients with Lebers congenital amaurosis, a form of retinitis pigmentosa (RP). For a while, the patients' sight improved. But earlier this year, reports indicated that the treatments were wearing off. The patients' sight was again degenerating. One proposed explanation was that the disease had already reached a point of no return when the initial gene therapy had been applied; the photoreceptors were already beyond permanent rescue.

But new findings are rekindling hope for the use of that gene therapy and others, and there are indications that it can be effective at any stage in the progression of the disease. "We have demonstrated a stable, prolonged halting of a retinal degeneration," says RPB researcher Stephen H. Tsang, MD, PhD, Columbia University College of Physicians & Surgeons. The Tsang team found that, if the gene therapy was optimally delivered, it could be effective at any time during the course of the disease.

Meanwhile, another team of RPB-funded collaborators at the Universities of Pennsylvania and Florida used gene therapy to restore sight in dogs with naturally occurring, late-stage RP. Before the therapy can be tested in people with a more commonly found form of RP, additional research is needed to assess its safety. Both sets of findings suggest that RP can be treated despite being diagnosed after the disease has compromised many cells.

A GIFT TO RPB CAN SAVE SIGHT

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