Understanding Usher Syndrome: New Findings

Sometimes, research results are not only gratifying professionally, but personally as well. Such is the case for Jesse D. Sengillo, an RPB Medical Student Fellowship recipient based at Columbia University Medical Center. He, his mentor Stephen H. Tsang, MD, PhD (a former RPB Medical Student Fellowship recipient) and colleagues from around the world recently collaborated on a study to better characterize the genetic and physiologic characteristics of Usher syndrome—the most common cause of deaf-blindness, and the disease that affects Sengillo’s mother.

People with Usher syndrome have both retinitis pigmentosa (RP)—an inherited, irreversible and progressive form of blindness—as well as hearing loss.

The researchers identified a pattern between certain types of genetic mutations and the amount of residual cone photoreceptor function, which is needed for vision. They also reported that there may be a threshold of sorts for the severity of genetic mutations that, if exceeded, explains the presence of hearing loss.

This work will jump-start further study on Usher syndrome and help to target enrollment for future clinical trials. It is also critical information to help physicians to counsel their patients about expected clinical outcomes and disease management.

“Usher syndrome and other forms of RP have an immense impact on the person who is affected,” said Sengillo. “It’s a special feeling to know that our work may someday benefit [my mom] and many others.”

Predicting Glaucoma Damage Before Vision Loss

Most glaucoma tests measure cell death to indicate disease severity. While accurate, testing for cell death is, in a sense, too late. Once cells die, they cannot be brought back to life. In an exciting new development, RPB-supported researchers at Washington University School of Medicine in St. Louis published a study identifying a biomarker that could help to predict glaucoma damage before vision loss.

“There hasn’t been a reliable way to predict which patients with glaucoma have a high risk of rapid vision loss,” said principal investigator Rajendra S. Apte, MD, PhD, the Paul A. Cibis Distinguished Professor of Ophthalmology and Visual Sciences.

“But we’ve identified a biomarker that seems to correlate with disease severity in patients, and what that marker is measuring is stress to the cells rather than cell death.”

“If we can identify when cells are under stress, then there’s the potential to save those cells to preserve vision,” said Dr. Apte.

Next steps for the researchers include sampling fluid from human eyes with glaucoma to correlate glaucoma progression with levels of the biomarker, with the ultimate goal of creating therapies that more effectively preserve vision in people with glaucoma.

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Healthy Vision Tips

Zeaxanthin, lipoic acid and curcumin (found in turmeric) may prevent or inhibit the progression of diabetic retinopathy.

Vitamin D (from milk, fish and exposure to the sun) may protect against early age-related macular degeneration (AMD).

Sulforaphane, found in broccoli, may protect against retinal degenerative diseases.

The antioxidants lutein and zeaxanthin (from dark leafy greens, colorful fruits and vegetables, egg yolks or supplements) may protect against AMD.

For more vision-preserving tips, download RPB Fact Sheets at http://bit.ly/FactSheets17

Research to Prevent Blindness

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RPB-supported researchers at the University of Southern California have developed a way to diagnosis retinoblastoma (a sometimes-fatal cancer of the eye that is most common in children) without removing the eye. The innovative approach uses a liquid biopsy that takes fluid from the front of the eye. The new, sight-preserving procedure was developed when researchers discovered that the fluid called the aqueous humor contains tumor-derived genetic material.

Researchers at Massachusetts Eye and Ear have identified key compounds related to blood vessel growth (angiogenesis) in wet age-related macular degeneration (AMD), using RPB support. Their recently published study suggests that it may be possible to prevent the vision loss observed in wet AMD by increasing the expression of specific bioactive lipid metabolites (which regulate inflammatory immune cells) in the retina. These metabolites show promising therapeutic potential not only for AMD, but also for other major conditions that involve angiogenesis and inflammation, such as cardiovascular disease and cancer.

RPB-supported researchers at the University of Illinois at Chicago have identified an enzyme (heparanase) present in the cornea that triggers inflammation. The study looked at human corneal cells during and after a herpes simplex virus-1 infection, which causes persistent inflammation, but heparanase may be a key factor in other inflammatory disorders, including dry eye disease. A heparanase-blocking drug could represent a novel treatment for such conditions.

A 3-D rendering of the enzyme heparanase.

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Choroidal neovascular lesion with associated blood vessels (red) and immune cells (green).

Pediatric ocular oncologists at Children’s Hospital Los Angeles and the USC Roski Eye Institute.

Sulforaphane, found in broccoli, may protect against retinal degenerative diseases.

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