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Cells from skin create model of blinding eye disease

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For the first time, Wisconsin researchers have taken skin from patients and, using induced pluripotent stem cell (iPSC) technology, turned them into a laboratory model for an inherited type of macular degeneration.

Dr. David Gamm, director of the UW's McPherson Eye Research Institute, said that while Best disease is relatively rare, having a patient-specific model of the eye disease, which destroys the macula of the retina, could lead to a greater understanding of more common eye disorders such as age-related macular degeneration.

"This model gives us a chance to understand the biological effects of human gene mutations in a relatively expeditious manner," says Gamm, associate professor of ophthalmology and visual sciences and pediatrics. "Ultimately, we hope the model will help us craft treatments to slow or reverse the course of Best Disease."

Gamm and lead researchers Dr. Ruchira Singh and Dr. Wei Shen, all members of the UW's Waisman Center, took skin samples from members of two Chicago-area families with Best disease.

Children in those families have a 50-percent chance of inheriting the gene that causes the disease, which begins destroying the macula as early as age three. Using samples of affected and unaffected siblings, they turned the skin into stem cells, then into retinal pigment epithelium, the cells of the eye that are affected by the disease.

In the laboratory dish, they were able to track the changes that underlie a lesion on the retina that resembles "egg yolk," and progresses to a stage called "scrambled egg," which destroys the central vision.

The UW model revealed some of the cellular processes causing the disease. The models of the Best disease patients showed a buildup of fluid and old photoreceptor cells, indicating something gone wrong with the ability to degrade and remove debris such as dead cells. On a molecular level, the Best cells were slow to degrade rhodopsin, a biological pigment in photoreceptor cells, and had differences in calcium signaling and oxidative stress.

"These results give us some ideas where to look for therapies that would allow us to interfere with the disease process," says Gamm. "And the stem cell model gives us a chance to test those therapies before trying them on patients."

Even more important, on a human level, is how excited some of the family members were to participate in understanding and eventually treating a disease that has plagued generations of their families.

"These family members know they're not getting treated directly as a result of this study, but they're doing it out of concern for the next generation," Gamm said. "That brings peace to them, to know that they're not passive victims of this disease, but instead, active players in the discovery process."

The chief research officer of the Foundation Fighting Blindness, which helped fund the Best disease project, says the method holds promise for a number of retinal conditions.

"We are delighted by the highly innovative research of Dr. Gamm and his lab in harnessing stem cells to better understand complex retinal

diseases and move us closer to vision-saving treatments and cures," says Dr. Stephen Rose. "His techniques can be used to help characterize and overcome the entire spectrum of inherited retinal conditions."

The study on a model for Best disease is being published online today in the journal *Human Molecular Genetics*.

Provided by University of Wisconsin-Madison

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