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GENE THERAPY RESEARCH SHOWS PROMISE IN EYE DISEASE THAT CAUSES BLINDNESS

DALLAS --Using a form of gene therapy, researchers in Dallas and Los Angeles have succeeded in correcting an inherited eye disease of mice called *retinal degeneration slow (rds)*, involving the same gene that causes retinitis pigmentosa in humans.

Currently there is no cure for retinitis pigmentosa, which has blinded an estimated 100,000 Americans and 1.5 million people worldwide. Studies conducted by Dr. Gabriel H. Travis, a neurobiologist at The University of Texas Southwestern Medical Center at Dallas, and Dr. Dean Bok of the Jules Stein Eye Institute at UCLA suggest that gene therapy eventually may offer an effective treatment for some forms of the disease.

In research reported in this month's issue of the journal *Neuron*, Travis and Bok introduced a DNA "cassette" containing a normal copy of the *rds* gene into fertilized eggs of *rds* mutant mice, which are named for their disease. The eggs were re-implanted into their mothers and allowed to develop into adult mice. When the retinal tissues of the transgenic offspring were analyzed, the mice were found to have entirely normal retinas, despite the fact that they still had a copy of the mutated *rds* gene that typically would predestine them to lose their vision.

"This study suggests that somatic gene therapy for some forms of retinitis pigmentosa may be possible," said Travis. "We're still far away from human trials, though."

(More)

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The form of gene therapy the researchers conducted is known as germ cell therapy. Since the transplanted gene is present in all tissues and is passed on indefinitely to subsequent generations, germ cell therapy is unsuitable as a treatment for humans, said Travis. He is working with collaborators in San Francisco, however, to insert the normal *rds* gene into a modified herpes simplex virus, which can be injected directly into the retinas of affected adult mice. If that technique is effective in mice, it could lead to safe gene therapy for humans afflicted with retinitis pigmentosa.

"This study is exciting because it is the first naturally occurring animal mutation for inherited retinal degeneration that has direct relevance to human retinitis pigmentosa," said Bok, the Dolly Green Professor of Ophthalmology at UCLA's Jules Stein Eye Institute.

Retinitis pigmentosa is an inherited eye disease that begins with night blindness and loss of peripheral vision. It is relentlessly progressive, resulting in total blindness, usually by middle age.

When images enter the normal eye, they are focused by the cornea onto the retina at the back of the eye, where light-sensitive nerve cells known as rods and cones convert images into nerve impulses. The rods allow vision in dim light. The cones detect color and detail. Retinitis pigmentosa causes the rods, and sometimes the cones, to degenerate over time.

Travis, the lead author of the paper, is an assistant professor of psychiatry at UT Southwestern and a John Merck Fund Scholar. The research was funded by the National Eye Institute and the National Retinitis Pigmentosa Foundation Fighting Blindness.

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