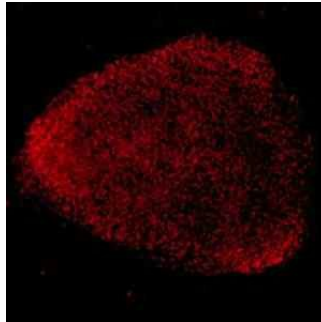

Precision Medicine Hope for Blindness



University of Iowa researchers and Columbia University Medical Center ophthalmologists have together used a new technology for repairing disease-genes and have been able to correct a blindness-causing gene mutation in stem cells derived from a patient. The research is published in *Scientific Reports*.

The new technique is called CRISPR and can repair the genetic mutation that is primarily responsible for retinitis pigmentosa (RP). RP degrades the retina and leads to blindness. The researchers targeted the most common variant of RP which is caused by a single mistake in the gene called RPGR. This is the first time that researchers have been able to replace a defective gene associated with a sensory disease in stem cells that were derived from a patient's tissue.

Alexander Bassuk, MD, PhD, and Vinit Mahajan, MD, PhD and his team generated stem cells from patient skin cells and repaired the damaged gene by using an editing technique that is so precise that it can correct a single DNA change that had damaged the RPGR gene. Since the corrected tissue was derived from the patient's own stem cells, it could be transplanted without any harmful drugs to prevent tissue rejection.

"With CRISPR gene editing of human stem cells, we can theoretically transplant healthy new cells that come from the patient after having fixed their specific gene mutation," says Mahajan, clinical assistant professor of ophthalmology and visual sciences in the UI Carver College of Medicine. "And retinal diseases are a perfect model for stem cell therapy, because we have the advanced surgical techniques to implant cells exactly where they are needed."

Stephen Tsang, MD, PhD, the László Z. Bitó Associate Professor of Ophthalmology and associate professor of Pathology & Cell Biology at CUMC explains that the X-linked form of RP is the ideal candidate for a precision medicine approach because a common mutation is responsible for 90 percent of the cases. Using CRISPR, the researchers were able to do fast and accurate editing and could retransplant the cells with a much lesser risk of rejection by the immune system.

The success that has been seen with RPGR is quite promising and could potentially lead to the treatment of other forms of conditions that are caused by mutations in other genes. Other gene therapies for RP are undergoing clinical trials but unlike the CRISPR-based technique, these therapies supplement the activity of the defective gene but do not correct the original mutation. This new technique offers an improvement over current therapies and could restore a patient's vision due to its accuracy.

The technique has not yet been tested in humans so there are still some concerns regarding its safety. However, it does hold potential with respect to fixing diseased genes.

Source: [University of Iowa Healthcare](#)

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