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Genetically Precise Human Cellular Models for Lowe Syndrome and Ocular Disease Research

Stanford researchers have developed a genome engineering platform for generating genetically precise human cellular models of Lowe syndrome, a rare inherited disorder frequently associated with severe ocular complications, including early-onset glaucoma.

Current Lowe syndrome disease models rely largely on animal systems, gene knockouts, or non-ocular cell types that fail to accurately reproduce patient-specific OCRL gene mutations and human disease biology. As a result, mechanistic studies and therapeutic development for OCRL-associated ocular disease remain significantly limited.

This technology enables the generation of isogenic, mutation-specific human cellular models that closely replicate clinically relevant Lowe syndrome mutations in human trabecular meshwork cells, a key ocular cell type involved in regulating intraocular pressure. The platform combines CRISPR-based base editing and prime editing with induced pluripotent stem cell (iPSC) engineering to introduce or correct disease-causing mutations in the OCRL gene, followed by differentiation into trabecular meshwork-like cells.

This genetically precise platform enables the study of mutation-specific disease mechanisms and the evaluation of emerging therapeutic approaches, including gene editing and gene therapy strategies. By generating disease-relevant trabecular meshwork models, it supports therapeutic discovery and preclinical development for Lowe syndrome and other rare genetic ocular disorders.

Stage of Development
Proof of concept

Applications

- Lowe syndrome disease modeling
- Glaucoma and ocular disease research
- Patient-specific iPSC model development
- Rare disease therapeutic screening

Advantages

- Genetically precise human cell models
- Disease-relevant ocular cell type
- Enables mutation-specific therapeutic testing
- Alternative to animal and knockout models
- Replicates clinically relevant OCRL mutations

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