

## **PROGRESS REPORT**

**Project title:** Probing protein mutagenesis and modulation in cystinosis

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Sep 2024– Feb 2025

### **Objectives:**

In this study, we aim to deepen our molecular understanding of mutations and explore the potential for modulating key proteins in cystinosis.

### **Executive overview of progress: Suitable for public disclosure**

Lysosomal membrane proteins that transport substrate small molecules across the membrane play a central role in both the cause and treatment of cystinosis. These proteins operate through transport cycle, transitioning among multiple conformational states. Mutations that prevent key membrane transport proteins from functioning properly can lead to cystinosis. Our research focuses on developing novel strategies to restore their function. Toward this goal, we have developed a screening platform, which can enable the potential identification of molecules that can modulate the functions of these proteins. Successful implementation of this project could set the stage to develop new treatments for cystinosis.