A Yeast Model for Screening Mutations in Cystinosis

Breakthroughs in research can potentially aid in the understanding of rare genetic disorders such as Cystinosis. Here, the identification of a yeast homologue and design of a complimentary assay makes it possible to isolate and identify carriers of Cystinosis.

Background:
Cystinosis is a rare autosomal recessive genetic disease affecting approximately 500 children in the United States, with only about 2000 cases worldwide. It is caused by a mutation in the gene cystinosin (CTNS), causing a buildup of the amino acid cystine in cells. This further leads to cell crystallization and early cell death. Ultimately, the disease destroys all the organs of the body. Unfortunately, there is no cure available for this disease, but it a genetic screening method may prove useful.

Technology Description:
Dr. Neta Dean from the Department of Biochemistry and Cell Biology and Dr. X. Gao have discovered that a structural and functional homologue of CTNS can be obtained using the yeast ERS1 gene. They have thus developed a novel yeast complementary assay that can be used to identify heterozygous carriers of Cystinosis.

Advantages
- Swift and economical screening technique to determine carriers of Cystinosis

Applications
- Yeast biology
- Genetic screening
- Research tool
- Potential Cystinosis treatment

Patents and Publications:
- Patent Pending