NOVEL TREATMENT FOR GALACTOSEMIA

THERAPEUTICS
Repurposing known endoplasmic reticulum stress inhibitors to treat GALT-deficiency.

TECHNOLOGY SUMMARY
Galactosemia is a rare, inherited condition where people cannot metabolize galactose due to mutations in GALT, GALE, and GALK1. The disorder appears in 1 in 40,000 live births and can be fatal if left untreated, but no long-term treatment exists. Galactosemia patients also experience intellectual and speech deficits, motor function loss, ataxia, and infertility.

The inventors have discovered that accumulation of Gal1-p results in a unique form of endoplasmic reticulum (ER) stress in fibroblasts of patients with Galactosemia. The proposed invention repurposes known ER stress inhibitors, such as Salubrinal, which have been shown to reduce phosphorylation of several intracellular proteins involved in ER stress for the treatment of Galactosemia.

FEATURES AND BENEFITS
- Novel mechanism of action.
- ER stress inhibitors can also be combined with novel small molecule GALK inhibitors.
- Salubrinal has an established safety profile and has been approved by the FDA.
- Restoration of P13K/Akt signaling pathway alleviates all disease phenotypes of Galactosemia.
- Mouse model of Galactosemia available for drug screening and development.

RECENT PUBLICATIONS

INVENTOR PROFILE
Kent Lai, Ph.D., Professor - Pediatrics
Bijina Balakrishnan, Ph.D., Post-Doctoral Fellow - Pediatric Genetics