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NOVEL TREATMENT FOR GALACTOSEMIA

THERAPEUTICS

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

TECHNOLOGY TYPE

Drug Repurposing
Galactosemia
Cerebellar Ataxia
Rare Metabolic Disease
Combination Theory
Galactose

STAGE OF DEVELOPMENT

- Proof of concept demonstrated in mouse model of Galactosemia.

- Preliminary data shows increase in ovarian follicles and effect of pro-survival signals in mice.

- Additional studies required.

IP PROTECTION

PCT filed.

LEARN MORE

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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 alldisease phenotypes of Galactosemia.
- Mouse model of Galactosemia available for drug screening and development.

RECENT PUBLICATIONS

Balakrishnan, B., Nicholas, c., Siddiqi, A., Chen, W., Bales, E., Feng, M., Johnson, J., Lai, K. (2017). Reversal of aberrant P13K/Akt signaling by Salubrinal in a GalT-deficient mouse model. *Biochimica et Biophysical Acta*. 1863(12):3286-3293. doi: 10.1016/j.bbadis.2017.08.023

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