

Repurposed ALS drug shows promise in mouse model of rare childhood genetic disorder

November 19 2021



Credit: Unsplash/CC0 Public Domain

Riluzole, a drug approved to treat amyotrophic lateral sclerosis (ALS), a disease affecting nerve cells controlling movement, could slow the

gradual loss of a particular brain cell that occurs in Niemann-Pick disease type C1 (NPC1), a rare genetic disorder affecting children and adolescents, suggests a study in mice by scientists at the National Institutes of Health.

The study was conducted by Forbes D. Porter, M.D., Ph.D., of NIH's *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), and colleagues in the National Human Genome Research Institute and National Institute of Arthritis and Musculoskeletal and Skin Disease. It appears in *Molecular Genetics and Metabolism*. The study was supported in part by a grant from the Ara Parseghian Medical Research Foundation.

NPC1 results from an impaired ability to move cholesterol through cells, leading to difficulty controlling movements, liver and lung [disease](#), impaired swallowing, intellectual decline and death. Much of the movement difficulties in NPC1 result from gradual loss of brain cells known as Purkinje neurons. The researchers found that mice with a form of NPC1 have a diminished ability to lower levels of glutamate—a brain chemical that stimulates neurons—after it has bound to a neuron's surface. High levels of glutamate can be toxic to cells. The researchers believe the buildup of glutamate contributes to the brain cell loss seen in the disease. Riluzole blocks the release of glutamate and hence delays the progression of ALS in people.

In the current [study](#), mice with NPC1 survived 12% longer when treated with riluzole, compared to untreated mice. The researchers believe that riluzole or similar drugs may provide a way to slow disease progression in patients with NPC1.

More information: Antony Cougnoux et al, Reduction of glutamate neurotoxicity: A novel therapeutic approach for Niemann-Pick disease, type C1, *Molecular Genetics and Metabolism* (2021). [DOI:](#)

[10.1016/j.ymgme.2021.11.008](https://doi.org/10.1016/j.ymgme.2021.11.008)

Provided by NIH/Eunice Kennedy Shriver National Institute of Child Health and Human Development

Citation: Repurposed ALS drug shows promise in mouse model of rare childhood genetic disorder (2021, November 19) retrieved 22 July 2024 from <https://medicalxpress.com/news/2021-11-repurposed-als-drug-mouse-rare.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.