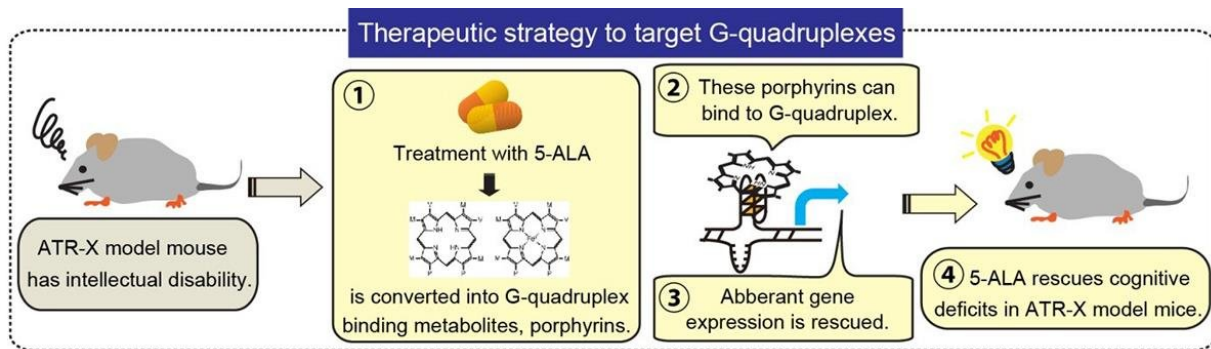


New advances in understanding and treating intellectual disorder

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Novel therapeutic strategy for intellectual disability. Credit: Norifumi Shioda

Researchers at Tohoku University in Japan have investigated an intellectual disorder (ATR-X) syndrome to reveal its cause, mechanism and a potential therapeutic strategy to decrease associated cognitive impairment.

"Disease related neurodevelopmental disorders are rare and there is still a lack of therapy to treat the various syndromes," said Professor Kohji Fukunaga at the Graduate School of Pharmaceutical Studies, who led the study.

Alpha-thalassemia X-linked intellectual disability (ATR-X) syndrome is a severe intellectual disability caused by ATRX gene mutations. The

researchers found that treatment with 5-aminolevulinic acid (5-ALA), which is converted into G-quadruplex-binding metabolites, rescues decreased synaptic plasticity and cognitive deficits seen in ATR-X model mice.

The findings suggest a potential therapeutic strategy to target G-quadruplexes and decrease [cognitive impairment](#) associated with ATR-X syndrome.

Among candidate ligands, 5-ALA has been applied clinically with minimal risk and approved for use following intracranial tumor resection in Europe, Canada, and Japan, where it has been used as a photosensitizer in photodynamic diagnostics applied in neurosurgery. These clinical applications potentially reduce the required approval time and cost of clinical trials, as pre-existing absorption, distribution, metabolism, excretion (ADME) and toxicity data are readily available.

The risk of failure is reduced as data relevant to 5-ALA safety and pharmacology is also available. The G-quadruplexes are involved in the pathology of other diseases and this discovery is expected to contribute to the possibility of new drug targets.

"5-aminolevulinic acid is proven to be safe and is already used in supplements in Japan, so we are ready to begin [clinical trials](#)," says Fukunaga. "There is also evidence that it may improve [autism spectrum disorders](#) which is a common disease in Japanese children."

More information: Norifumi Shioda et al. Targeting G-quadruplex DNA as cognitive function therapy for ATR-X syndrome, *Nature Medicine* (2018). [DOI: 10.1038/s41591-018-0018-6](https://doi.org/10.1038/s41591-018-0018-6)

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