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Rare genetic condition may provide insights on Parkinson's and other late-onset diseases

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A new article suggests that an enzyme deficiency seen in the lysosomal storage disorder Krabbe's disease may point to new mechanisms underlying certain late-onset neurodegenerative diseases such as Parkinson's disease.

Patients with Krabbe's disease lack galactosylceramidase, which is needed to make the protective myelin coating around nerve cells. Unfortunately, there is currently no cure, and most infants with the degenerative disorder die at a very young age.

"It's been established that mutations to lysosomal enzymes, as present in Gaucher disease, can be a strong genetic risk factor for Parkinson's disease. In this commentary, we highlight recent evidence that suggests pathophysiological changes due to galactosylceramidase mutations in Krabbe's disease may also be related to the pathology of late-onset alpha-synucleinopathies," said Dr. Michael. Marshall, co-author of the *Journal of Neuroscience Research* article. "While this association requires further investigation, we hope that future larger cohort studies may be able to confirm either a genetic link or common pathological mechanisms between the diseases."

"If a connection with Parkinson's disease is demonstrated, this would prove invaluable for developing novel therapeutic targets based on our current understanding of Krabbe's disease and for establishing new biomarkers for identifying at-risk patients," added co-author Dr. Ernesto Bongarzone.

The study is part of a special issue to mark the 100th anniversary of Dr. Knud Krabbe's discovery of the disease. The issue highlights advances in the field and discoveries of new treatments using stem cell therapies that can help to prolong life.

More information: Michael S. Marshall et al. Beyond Krabbe's disease: The potential contribution of galactosylceramidase deficiency to neuronal vulnerability in late-onset synucleinopathies, *Journal of Neuroscience Research* (2016). DOI: 10.1002/jnr.23751