A potential target for peripheral neuropathy treatment discovered

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Whole exome sequencing has revealed a novel mechanism and potential target for treating peripheral neuropathy, a condition that afflicts millions of people in the United States alone.

Yale neurosurgeon Kristopher Kahle along with and colleagues at Vanderbilt University and the National Institutes of Health have discovered that a mutation resulting in a change of only one critical amino acid in a gene encoding a potassium/chloride transporter was sufficient to cause a progressive, early onset, and severe form of a predominantly motor neuropathy in humans. The mutation released an inhibitory "brake" on the transport of potassium and chloride into cells, which impaired cell volume regulation.

"Our study has shown the importance of dynamic cell volume regulation for the structure and function of the human nervous system," said Kahle, lead author of the research. "Modulating this pathway could be a potential novel approach to treating peripheral neuropathies, including those associated with diabetes."

The research was published Aug. 2 in the journal *Science Signaling*.

More information: K. T. Kahle et al. Peripheral motor neuropathy is associated with defective kinase regulation of the KCC3 cotransporter, *Science Signaling* (2016). DOI: 10.1126/scisignal.aae0546

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