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# Researchers discover mutation involved in neurodegeneration

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A mutation that increases the level of a special class of sphingolipids—molecules important to cell structure and signaling—can lead to neurodegeneration due to problems with neuronal membranes, reports a research team led by Jackson Laboratory Research Scientist Lihong Zhao, Ph.D. and Professor Patsy Nishina, Ph.D.

Sphingolipids are a family of biological molecules structurally related to fats. They add to the structure of the membrane, and they are often involved in sending signals between cells. They are particularly important in the brain, and many common neural disorders (including Niemann-Pick disease, Gaucher disease and Tay-Sachs disease) are related to problems with sphingolipid metabolism.

The precursors of sphingolipids, called long-chain bases, from which all sphingolipids are derived, are of a certain length, 18 carbons, but they can also be found at 16- and 20-carbon lengths, with the length being controlled by a multi-protein enzyme called serine palmitoyltransferase (SPT). The authors studied a mouse model with a mutation called Stellar, which exhibits symptoms of neurodegeneration. They determined that the Stellar mutation, which occurs in one of the proteins that make up the SPT complex, causes an increase in the number of "long" 20-carbon sphingolipids.

"It's interesting to note," Nishina says, "that having a certain amount of 20-carbon sphingolipids actually appears to be important in healthy animals. But what's important are the levels of these 20-carbon

molecules compared with those of the 18-carbon variety."

**More information:** Zhao et al.: Elevation of 20-carbon long chain bases due to a mutation in serine palmitoyltransferase small subunit b results in neurodegeneration. *Proceedings of the National Academy of Sciences*, published online before print October 5, 2015, DOI: 10.1073/pnas.1516733112

Provided by Jackson Laboratory

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