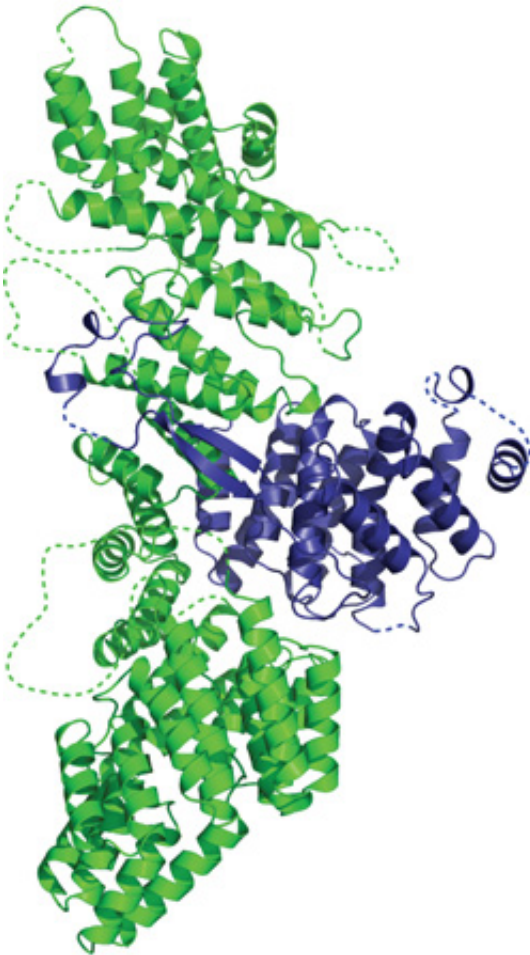


# How a mutated gene wreaks havoc on white matter

November 17 2015, by Bill Hathaway

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An inherited disease of myelin marked by slow, progressive neurological impairment is caused by mutations of a gene that controls lipid

metabolism, a finding that may shed insight into mechanisms to control the course of multiple sclerosis (MS), a Yale team has found.

Mutations in a single gene, called FAM126A, causes a panoply of pathologies, such as developmental delay, intellectual disability, [peripheral neuropathy](#), and muscle wasting, in addition to congenital cataracts. Until now the precise function of the gene was unknown.

The labs of Yale cell biologists Pietro De Camilli and Karin Reinisch found that the protein encoded by the gene, called hyccin, helps produce a lipid crucial to formation of the [myelin sheaths](#) that surround and protect the axons of neurons throughout the nervous system.

Their labs, working with other groups in the United States, Italy, and Germany, analyzed cells from patients suffering from the disease known as Hypomyelination and Congenital Cataract and found that FAM126A mutations results in the destabilization of an enzyme complex crucial to production of myelin.

In MS, the course of the disease is critically dependent upon the reformation of myelin sheaths after immune system attacks then destroys them, eventually leading to the death of the neurons. The researchers hypothesize that the lipid that hyccin helps generate may play a key role in creation of myelin sheaths in normal development as well as in recovering MS patients.

Postdoc Jeremy Baskin (now at Cornell) and graduate student Xudong Wu (now at Harvard) led the study in the De Camilli and Reinisch labs, respectively. The research was published Nov. 16 in the journal *Nature Cell Biology*.

**More information:** Jeremy M. Baskin et al. The leukodystrophy protein FAM126A (hyccin) regulates PtdIns(4)P synthesis at the plasma

membrane, *Nature Cell Biology* (2015). [DOI: 10.1038/ncb3271](https://doi.org/10.1038/ncb3271)

Provided by Yale University

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