Neutral lipid storage disease with myopathy

Description

Neutral lipid storage disease with myopathy is a condition in which fats (lipids) are stored abnormally in organs and tissues throughout the body. People with this condition have muscle weakness (myopathy) due to the accumulation of fats in muscle tissue. Other features of this condition may include a fatty liver, a weakened and enlarged heart (cardiomyopathy), inflammation of the pancreas (pancreatitis), reduced thyroid activity (hypothyroidism), and type 2 diabetes (the most common form of diabetes). Signs and symptoms of neutral lipid storage disease with myopathy vary greatly among affected individuals.

Frequency

Neutral lipid storage disease with myopathy is a rare condition; its incidence is unknown.

Causes

Mutations in the *PNPLA2* gene cause neutral lipid storage disease with myopathy. The *PNPLA2* gene provides instructions for making an enzyme called adipose triglyceride lipase (ATGL). The ATGL enzyme plays a role in breaking down fats called triglycerides. Triglycerides are an important source of stored energy in cells. These fats must be broken down into simpler molecules called fatty acids before they can be used for energy.

PNPLA2 gene mutations impair the ATGL enzyme's ability to break down triglycerides. These triglycerides then accumulate in muscle and tissues throughout the body, resulting in the signs and symptoms of neutral lipid storage disease with myopathy.

Learn more about the gene associated with Neutral lipid storage disease with myopathy

- PNPLA2

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Neutral lipid storage disease without ichthyosis
- NLSDM

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Neutral lipid storage myopathy (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853136/)

Genetic and Rare Diseases Information Center

- Neutral lipid storage myopathy (https://rarediseases.info.nih.gov/diseases/10288/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Neutral lipid storage disease with myopathy%22)

Catalog of Genes and Diseases from OMIM

- NEUTRAL LIPID STORAGE DISEASE WITH MYOPATHY; NLSDM (https://omim.org/entry/610717)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28neutral+lipid+storage+disease%29+AND+%28myopathy%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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