Fucosidosis

Description

Fucosidosis is a condition that affects many areas of the body, especially the brain. The symptoms of fucosidosis can vary from person to person. Affected individuals have intellectual disabilities that worsen with age. Over time, people with this condition tend to lose skills they had previously learned, such as sitting, standing, walking, or talking. Additional signs and symptoms of fucosidosis can include slow growth, abnormal bone development (dysostosis multiplex), and rigid or stiff muscles (spasticity). People with fucosidosis may also have clusters of enlarged blood vessels that form small, dark red spots on the skin (angiokeratomas) and distinctive facial features that are often described as "coarse." Additional features of fucosidosis can include frequent respiratory infections, an enlarged liver and spleen (hepatosplenomegaly), and seizures.

In the past, fucosidosis has been divided into two types based on the symptoms and age of onset. Type 1 was used to describe the more severe form of the disorder, with symptoms typically appearing in infancy. Type 2 was used to describe cases with milder symptoms and a slower progression. Currently, many researchers consider the condition to be a spectrum with a wide range in severity.

Frequency

Fucosidosis is a rare condition. As of 2023, fewer than 200 cases have been reported in the literature. The highest incidence of fucosidosis has been described in Italy, Cuba, and in certain populations of the Southwestern United States.

Causes

Variants (also called mutations) in the *FUCA1* gene cause fucosidosis. The *FUCA1* gene provides instructions for making an enzyme called alpha-L-fucosidase. This enzyme plays a role in the breakdown of sugar molecules (oligosaccharides) that are attached to certain fats (glycolipids) and proteins (glycoproteins). Alpha-L-fucosidase is responsible for cutting off (cleaving) a sugar molecule called fucose toward the end of the breakdown process.

FUCA1 gene variants severely reduce or eliminate the activity of the alpha-L-fucosidase enzyme. A loss of enzyme activity results in an incomplete breakdown of glycolipids and

glycoproteins. These compounds gradually accumulate within various cells and tissues throughout the body. Brain cells are particularly sensitive to the buildup of glycolipids and glycoproteins. Damage to brain cells is thought to cause the neurological symptoms of fucosidosis. Glycolipids and glycoproteins also accumulate in other organs such as the liver, spleen, and skin.

Alpha-L-fucosidase is found in the lysosomes. Lysosomes are compartments in the cell that digest and recycle different types of molecules. Conditions like fucosidosis, that cause molecules to build up inside the lysosomes, are known as lysosomal storage disorders.

Learn more about the gene associated with Fucosidosis

- FUCA1

Inheritance

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

Other Names for This Condition

- Alpha-L-fucosidase deficiency
- Fucosidase deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Fucosidosis (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0016788/)

Genetic and Rare Diseases Information Center

- Fucosidosis (https://rarediseases.info.nih.gov/diseases/6473/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Fucosidosis%22)

Catalog of Genes and Diseases from OMIM

- FUCOSIDOSIS (https://omim.org/entry/230000)

Scientific Articles on PubMed

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

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