Fucosidosis

Fucosidosis is a condition that affects many areas of the body, especially the brain. Affected individuals have intellectual disability that worsens with age, and many develop dementia later in life. People with this condition often have delayed development of motor skills such as walking; the skills they do acquire deteriorate over time. Additional signs and symptoms of fucosidosis include impaired growth; abnormal bone development (dysostosis multiplex); seizures; abnormal muscle stiffness (spasticity); clusters of enlarged blood vessels forming small, dark red spots on the skin (angiokeratomas); distinctive facial features that are often described as "coarse"; recurrent respiratory infections; and abnormally large abdominal organs (visceromegaly).

In severe cases, symptoms typically appear in infancy, and affected individuals usually live into late childhood. In milder cases, symptoms begin at age 1 or 2, and affected individuals tend to survive into mid-adulthood.

In the past, researchers described two types of this condition based on symptoms and age of onset, but current opinion is that the two types are actually a single disorder with signs and symptoms that range in severity.

Frequency

Fucosidosis is a rare condition; approximately 100 cases have been reported worldwide. This condition appears to be most prevalent in Italy, Cuba, and the southwestern United States.

Causes

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 complexes of sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins) and fats (glycolipids). Alpha-L-fucosidase is responsible for cutting (cleaving) off a sugar molecule called fucose toward the end of the breakdown process.

*FUCA1* gene mutations severely reduce or eliminate the activity of the alpha-L-fucosidase enzyme. A lack of enzyme activity results in an incomplete breakdown of glycolipids and glycoproteins. These partially broken down compounds gradually accumulate within various cells and tissues throughout the body and cause cells to malfunction. Brain cells are particularly sensitive to the buildup of glycolipids and glycoproteins, which can result in cell death. Loss of brain cells is thought to cause the neurological symptoms of fucosidosis. Accumulation of glycolipids and glycoproteins...
also occurs in other organs such as the liver, spleen, skin, heart, pancreas, and kidneys, contributing to the additional symptoms of fucosidosis.

Inheritance Pattern

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

• Alpha-fucosidase deficiency
• Fucosidase deficiency
• Fucosidase Deficiency Disease

Diagnosis & Management

Genetic Testing Information

• What is genetic testing?
  /primer/testing/genetictesting
• Genetic Testing Registry: Fucosidosis

Research Studies from ClinicalTrials.gov

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Fucosidosis%22+OR+%22fucosidosis%22

Additional Information & Resources

Health Information from MedlinePlus

• Health Topic: Carbohydrate Metabolism Disorders
  https://medlineplus.gov/carbohydratemetabolismdisorders.html
• Health Topic: Genetic Brain Disorders
  https://medlineplus.gov/geneticbraindisorders.html
• Health Topic: Neurologic Diseases
  https://medlineplus.gov/neurologicdiseases.html
• Health Topic: Seizures
  https://medlineplus.gov/seizures.html
Genetic and Rare Diseases Information Center

- Fucosidosis
  https://rarediseases.info.nih.gov/diseases/6473/fucosidosis

Educational Resources

- MalaCards: fucosidosis
  https://www.malacards.org/card/fucosidosis
- Orphanet: Fucosidosis
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=349

Patient Support and Advocacy Resources

- ISMRD: The International Advocate for Glycoprotein Storage Diseases
  http://www.ismrd.org/
- Metabolic Support UK
  https://www.metabolic.supportuk.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/fucosidosis/
- The MAGIC Foundation
  https://www.magicfoundation.org/
- The MPS Society (UK)
  http://www.mpssociety.org.uk/diseases/related-diseases/fucosidosis/

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- FUCOSIDOSIS
  http://omim.org/entry/230000

Sources for This Summary


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Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services