Inheritest®
CARRIER SCREEN

Fucosidosis

What is fucosidosis?

Fucosidosis is an inherited metabolic disease characterized by intellectual disability, delayed motor skill development, progressive neurological deterioration, and distinctive facial features.[1,3] Individuals with fucosidosis have a deficiency of the alpha-L-fucosidase enzyme, which helps to break down fucose-containing compounds within lysosomes in cells.[2,3] Without this enzyme, fucose-containing compounds accumulate in various tissues and lead to a spectrum of associated symptoms. Fucosidosis is also known as alpha-L-fucosidase deficiency and belongs to a group of diseases called lysosomal storage disorders.[3]

What are the symptoms of fucosidosis and what treatment is available?

The severity and age of onset of fucosidosis are variable. In severe cases, symptoms typically appear in infancy, with rapid psychomotor regression and severe neurologic deterioration. In milder cases, symptoms usually begin at one to two years of age and survival is longer.[1]

Symptoms of fucosidosis may include[1,3]:

- Angiokeratomas (benign skin lesions)
- Progressive psychomotor retardation
- Neurologic deterioration
- Distinctive facial features
- Dysostosis multiplex (skeletal abnormalities seen by X-ray)
- Abnormal muscle stiffness
- Impaired growth
- Hypotonia (poor muscle tone)
- Progressive intellectual disability

There is no cure for fucosidosis. Treatment includes supportive care for symptoms.[3]

How is fucosidosis inherited?

Fucosidosis is an autosomal recessive disease caused by mutations in the *FUCA1* gene.[1] An individual who inherits one copy of a *FUCA1* gene mutation is a carrier. Carriers are not affected with fucosidosis. An individual who inherits two *FUCA1* mutations, one from each parent, is expected to be affected with fucosidosis.

If both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for fucosidosis?

Fucosidosis is a rare condition that can occur in individuals of all races and ethnicities. It may occur more commonly in certain populations in the southwest U.S., Cuba, and Italy.[4]

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/fucosidosis

National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/fucosidosis/

Children Living with Inherited Metabolic Diseases (CLIMB): http://www.climb.org.uk/

The International Advocate for Glycoprotein Storage Diseases: http://www.ismrd.org/glycoprotein_diseases/fucosidosis

References

1. Fucosidosis. *Genetics Home Reference.* Available at http://ghr.nlm.nih.gov/condition/fucosidosis. Accessed February 2, 2016.
2. *FUCA1. Genetics Home Reference.* Available at: http://ghr.nlm.nih.gov/gene/FUCA1. Accessed February 2, 2016.
3. Fucosidosis. *National Organization for Rare Disorders.* Available at http://rarediseases.org/rare-diseases/fucosidosis/. Accessed February 2,2016.
4. Willems PJ, *et al*. Spectrum of mutations in fucosidosis.*Eur J Hum Genet.* 1999;7:60-67