Fucosidosis

Subjects: Others
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Fucosidosis is a neurodegenerative disorder which progresses inexorably. Clinical features include coarse facial features, growth retardation, recurrent upper respiratory infections, dysostosis multiplex, and angiokeratoma corporis diffusum.

Keywords: Fucosidosis ; neurodegenerative disorder ; FUCA1 gene

1. Introduction

Fucosidosis is caused by mutations in the *FUCA1* gene resulting in α-L-fucosidase deficiency. Only 36 pathogenic variants in the *FUCA1* gene are related to fucosidosis. Most of them are missense/nonsense substitutions; six missense and 11 nonsense mutations. Among deletions there were eight small and five gross changes. So far, only three splice site variants have been described—one small deletion, one complete deletion and one stop-loss mutation. The disease has a significant clinical variability, the cause of which is not well understood. The genotype–phenotype correlation has not been well defined.

2. 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.

3. 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.

3.1. The gene associated with Fucosidosis

- FUCA1

4. 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.

5. Other Names for This Condition

- Alpha-fucosidase deficiency
- Fucosidase deficiency

- Fucosidase Deficiency Disease

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