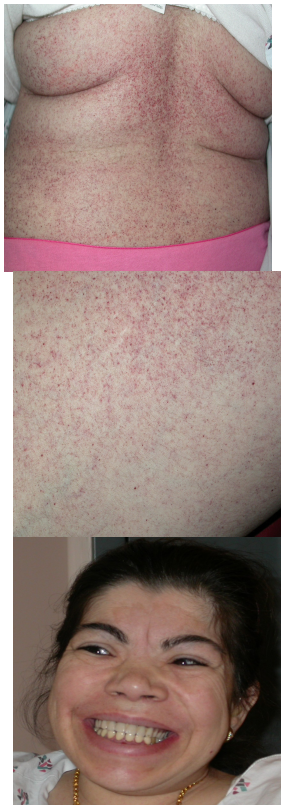

fucosidosis

In fucosidosis, an enzyme (alpha-fucosidase) is missing in the body. Without it, the body can't finish breaking down certain sugar, fat, and protein molecules into their useful parts. The partially-broken-down molecules build up in cells throughout the body and damage them. This causes many types of symptoms which often become worse over time.

Not much is known about how often fucosidosis occurs. Less than 100 individuals with fucosidosis have been written about in the medical literature worldwide. A gene for the disorder has been found on chromosome 1, and a similar gene sequence has been found on chromosome 2. An individual must inherit two copies of the defective gene (one from each parent) in order to develop fucosidosis.

Types and symptoms of fucosidosis

Some researchers believe there are two types of fucosidosis, while others classify it into three types. Still others believe there are not different types but rather different ways that fucosidosis may develop. If types are used, the symptoms are usually classified as: **Type I** (also called severe)

- begins in the first 3-18 months of life
- coarse facial features
- enlarged liver (hepatomegaly), spleen (splenomegaly), and/or heart (cardiomegaly)
- abnormal bone formation of many bones of the body (dysostosis multiplex)
- mental retardation
- seizures
- progressive deterioration of the brain and spinal cord
- increased or decreased perspiration

Type II (also called moderate)

- begins between 1 and 2 years of age
- symptoms similar to Type I but milder, and progress more slowly
- horny or warty growths over blood vessels on the skin (angiokeratomas)

Those who classify fucosidosis into three types suggest that Type I starts at 10 months, Type II at 18 months, and Type III (juvenile form) at 1-2 years old. Type I and Type II are considered severe and Type III mild.

Diagnosis

If a diagnosis of fucosidosis is suspected based on the child's physical appearance and symptoms, a special urine test will check for partially-broken-down sugars (oligosaccharides). If they are present, a blood sample or skin sample (biopsy) will be taken. In fucosidosis the blood or skin will have below-normal amounts of alpha-fucosidase in it.

Treatment

There is as yet no way to stop or reverse fucosidosis, so treatment focuses on the symptoms an individual has, such as medications for seizure control.

It is possible that if an individual with fucosidosis received a bone marrow transplant (BMT), he or she would receive healthy bone marrow cells which would produce normal amounts of alpha-fucosidase. However, only a few individuals with fucosidosis have experimentally received BMT, and not enough information is available to know if it is an effective treatment for fucosidosis.