Gaucher's Disease

Introduction

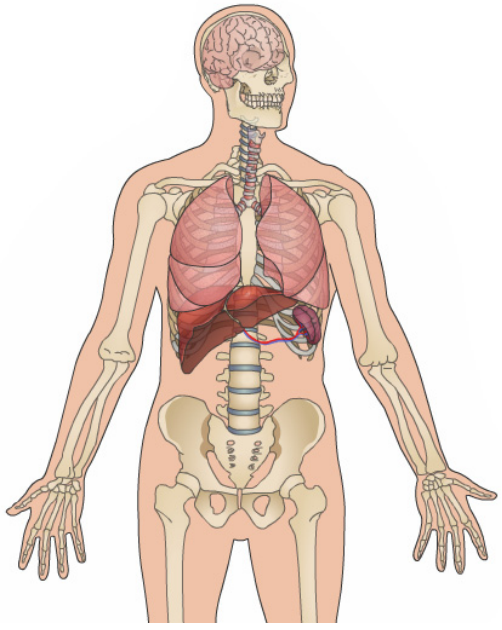
Gaucher's disease is a rare disorder that causes too much of a fatty substance to build up in your organs. These organs can include the spleen, liver, lungs, bones and, sometimes, your brain. The fatty substance prevents these organs from working properly.

Gaucher's disease has no cure. Treatment options depend on which type of Gaucher's disease you have. It may include medicine and enzyme replacement therapy.

This reference summary explains Gaucher's disease. It talks about causes and symptoms of Gaucher's disease, as well as diagnosis and treatment options.

Gaucher's Disease

Gaucher's disease is a type of lipid storage disease. Lipids are fatty materials that include oils, fatty acids, waxes and steroids, such as cholesterol and estrogen. Gaucher's disease happens when you do not have enough of an enzyme called glucocerebrosidase. This enzyme helps the body process a fatty substance called glucocerebroside.



Not having enough of the enzyme causes too much of the fatty substance to build up in certain organs. Organs that can be affected include the:

- Bones.
- Brain.
- Liver.
- Lungs.
- Spleen.

When the fatty substance builds up in the organs, they cannot work properly. This can cause a variety of symptoms.

Last reviewed: 04/23/2018

There are three types of Gaucher's disease. Type 1 is the most common form. It can cause:

- Liver and spleen enlargement.
- Bone pain and broken bones.
- Lung and kidney problems.

Type 1 does not affect the brain. It can happen at any age. Type 2 causes severe brain damage. It happens only in infants. Most children who have type 2 Gaucher's disease die by age 2. Type 3 usually starts in childhood or adolescence. There may be liver and spleen enlargement. The brain is gradually affected.

Symptoms

Symptoms of Gaucher's disease vary widely. Even siblings with the disease can have very different symptoms.

Symptoms of type 1 can include:

- An enlarged liver and/or spleen.
- Anemia.
- Bone problems, including weakening of the bones, bone pain and broken bones.
- Feeling extremely tired.

Anemia is a condition in which your blood has a lower than normal number of red blood cells.

Other symptoms of type 1 can include:

- Delayed puberty.
- Easy bruising.
- Nosebleeds.
- Yellow spots in your eyes.

Puberty is the time in life when a boy or girl becomes sexually mature. It is a physical change that usually happens between ages 10 and 14 for girls and ages 12 and 16 for boys.

Last reviewed: 04/23/2018

Type 2 can cause the same symptoms as type 1, as well as:

- Brain problems, such as learning disabilities.
- Rigidity and stiffness.
- Seizures.

Type 2 can also cause symptoms of dementia, including:

- Behavioral problems.
- Memory loss.
- Personality changes.

Type 3 may cause symptoms seen in both type 1 and type 2, as well as:

- Abnormal eye movements.
- Loss of muscle coordination.

Type 1 is the mildest form, while type 2 is often the most severe. Type 3 can vary in intensity. If you or your child has any of the above symptoms or other changes, contact a health care provider.

Causes

Gaucher's disease is inherited, which means children get the gene from their parents. This section explains the genetic cause of Gaucher's disease.

Genes tell the body to make certain substances. Every person has thousands of genes. Our genes make us look the way we do. They also have a lot to do with our health. Each person has 23 pairs of chromosomes. Genes are found on the chromosomes. The gene that controls how much glucocerebrosidase enzyme is made in the body is called GBA.

A mutation, or change, in the GBA gene causes the body to not make enough glucocerebrosidase enzyme. A person who has Gaucher's disease inherits one mutated copy of the GBA gene from each parent. Sometimes a person will only be a carrier of the disease, meaning that he or she can pass on a defective gene but has no health problems. This happens if a person only inherits one mutated GBA gene instead of two.

Last reviewed: 04/23/2018

To learn more about the possible combinations of the GBA gene and its effects on your health or the health of future children, talk to your health care provider. Some medical facilities offer genetic counseling. Genetic counseling provides information and support to people who have, or may be at risk for, genetic disorders. A genetics professional meets with you to discuss genetic risks. You may follow up with genetic testing.

Diagnosis

Your health care provider will first ask about your symptoms and medical history. You will also be asked about your family's medical history. A physical exam will be performed. If your health care provider thinks you may have Gaucher's disease, blood tests may be done. These tests can measure the level of glucocerebrosidase enzyme or see if you have two mutated GBA genes.

The sample of blood is sent to the lab for testing. People with Gaucher's disease have low levels of glucocerebrosidase enzyme in their blood. A genetic test looks for the mutated gene related to Gaucher's disease. This can help determine which type of Gaucher's disease you or your child has.

Treatment

Gaucher's disease has no cure. Treatment options vary depending on the type. Some people with type 1 may have such mild symptoms that they do not need treatment.

Treatment for types 1 and 3 may include medicine and enzyme replacement therapy. These are usually very effective. There is no good treatment for the brain damage caused by types 2 and 3. Enzyme replacement therapy replaces the deficient enzyme with artificial enzymes. These replacement enzymes are administered in an outpatient procedure through a vein. Other medicines may also be given. These include the oral medicine miglustat (Zavesca®). This medicine can help with the production of glucocerebrosidase enzyme.

Last reviewed: 04/23/2018

A bone marrow transplantation may be used to treat severe cases of Gaucher's disease. For this treatment, blood-forming cells that have been damaged by Gaucher's are removed and replaced. This may reverse many of Gaucher's signs and symptoms.

Summary

Gaucher's disease is a rare disorder that causes too much of a fatty substance to build up in your organs. These organs can include the spleen, liver, lungs, bones and, sometimes, your brain. The fatty substance prevents these organs from working properly.

There are three types of Gaucher's disease:

- Type 1 is the most common form and can cause liver and spleen enlargement, bone pain, broken bones and lung and kidney problems.
- Type 2 causes severe brain damage.
- Type 3 may cause liver and spleen enlargement. The brain is gradually affected.

A person who has Gaucher's disease inherits one mutated copy of the GBA gene from each parent. The gene that controls how much glucocerebrosidase enzyme is made in the body is called GBA. A mutation, or change, in the GBA gene causes the body not to make enough of the enzyme.

Gaucher's disease has no cure. Treatment options depend on which type of Gaucher's disease you have. Treatment may include medicine and enzyme replacement therapy.

Last reviewed: 04/23/2018