Sitosterolemia

Other names: Plant Sterol Storage Disease **Gene name:** ABCG5, ABCG8

What is Sitosterolemia?

Sitosterolemia is a rare genetic condition. It causes the body to store up excess fats that are found in both animals and plants. The condition is an autosomal recessive disorder that is caused by changes in the ABCG5 or ABCG8 gene.

Most plant fats are absorbed and digested from the foods we eat. People with sitosterolemia are unable to properly digest the fats. These plant fats tend to build up in the blood and arteries. Children are born with sitosterolemia and will not grow out of it.

What are the early signs and symptoms?

Signs and symptoms can vary from person to person, but common symptoms of the condition include:

- High cholesterol that is not controlled with medication
- Xanthomas or visible fatty deposits that can be seen under the skin
- Joint stiffness and pain
- Blood abnormalities such as: low platelet count, abnormally large platelets or abnormally shaped red blood cells
- All patients will have elevated levels of fats in their blood

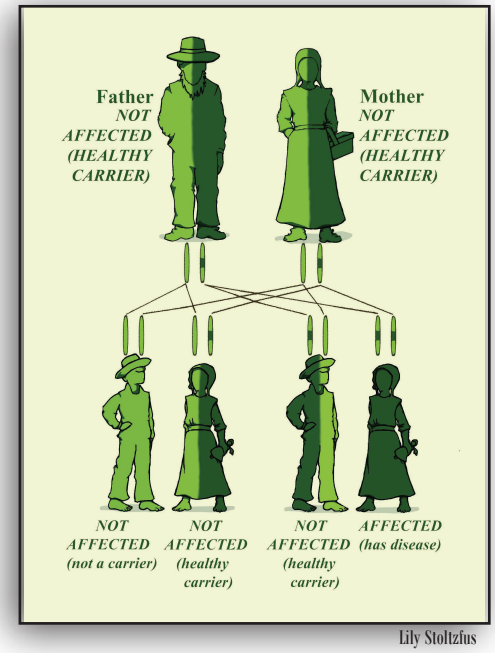
Management

The amount of plant fats and cholesterol levels should be monitored. The size, number and areas of the fatty deposits should be checked at least 1 or 2 times a year. Treatment for sitosterolemia can vary but mainly focuses on reducing the levels of fats and cholesterol, and to prevent the growth of the fatty deposits. The treatments or therapies used to do this are a diet low in vegetable oil, margarine, nuts, seeds, avocados, chocolate and shellfish along with prescription medications.

A blood test can reveal increased levels of these fats. Sitosterolemia is found when patients have higher amounts of those fats in their blood and tissues. It is recommended that treatment be started at the time of diagnosis.

Diagnosis

A gene test is required to make the diagnosis. To arrange this, please see the contact information below.



Autosomal Recessive Inheritance

Genes are instructions for growth and development. We have two copies of each gene. One copy is inherited from our mother, and one copy is inherited from our father. Children with Sitosterolemia have a spelling change in BOTH copies of their *ABCG5 or ABCG8* genes. This means they have no working copy of the *ABCG5 or ABCG8* instruction.

People with one normal copy of the *ABCG5 or ABCG8* gene and one misspelled copy are called "healthy carriers".

When two healthy carrier parents have a child, there is a:

- 1 in 4 chance the child will have Sitosterolemia, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *ABCG5 or ABCG8* gene and be neither affected nor be a carrier

This informational pamphlet is a project of New Leaf Center - a member of the Plain Community Health Consortium (PCHC)

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Center for Special Children (La Farge, WI) **608-625-4039**
Central Pennsylvania Clinic (Belleville, PA) **717-935-2065**
Clinic for Special Children (Strasburg, PA) **717-687-9407**
DDC Clinic (Middlefield, OH) **440-632-1668**
Midwest Genetics Network (Okemos, MI) **517-324-8300**
The Community Health Clinic (Topeka, IN) **260-593-0108**
UPMC Children's Hospital of Pittsburgh (Pittsburgh, PA) **412-692-5070**
WeCare Clinic - Medical Care for Special Needs (Pembroke, KY) **270-962-7383**