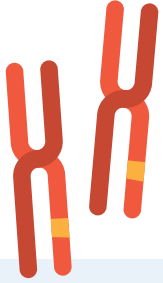
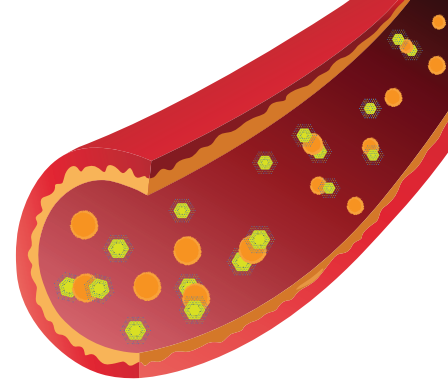


# What is Sitosterolemia?

Sitosterolemia is an inherited disorder in which waxy substances from plants are absorbed into the body and build up in the blood and arteries. These waxy substances are called "plant sterols". The principle one is sitosterol.

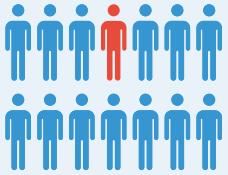


Usually, plant sterols are cleared from the intestine before ever entering the body. This is done by a protein "pump" called ABCG5/ABCG8. Sitosterolemia develops when the gene responsible for the creation of this pump is broken.

The condition is "recessive" which means that a broken copy of the gene must be inherited from each parent. The parents, who only have one broken copy of the gene, do not have the disorder.



## How common is it?

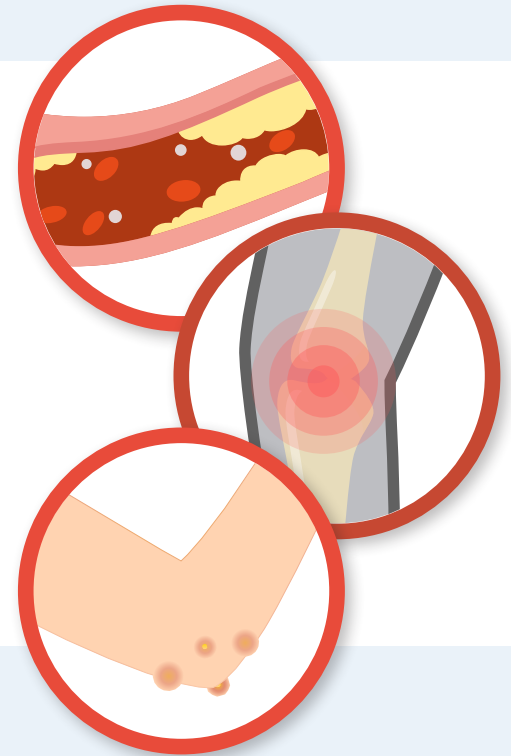


Sitosterolemia is thought to be a rare disease, however it may be more common than we think. Recent studies estimate that it may affect as many as 1 in 200,000 individuals in the general population. If left untreated, sitosterolemia can lead to premature hardening of the arteries (atherosclerosis), so it is important to diagnose it and start treatment as soon as possible.

## What are the symptoms?

The buildup of plant sterols in different parts of the body leads to a variety of symptoms, which may vary from person to person. Plant sterols can deposit in the arteries of the heart, causing a heart attack at a very young age. Plant sterols may also build up in the joints, causing joint stiffness and pain. It is common to see fatty deposits under the skin called "xanthomas" on hands, elbows, knees, and the Achilles tendon above the heel.

Patients with sitosterolemia often, but not always, have very high blood cholesterol levels. Routine laboratory tests do not measure plant sterols. When sitosterolemia is suspected, plant sterol levels in the blood should be measured using special tests. Sitosterol levels may be 10-25 times higher than normal. Genetic testing of mutations in ABCG5 and ABCG8 confirms the diagnosis.



## How is it treated?



*\*A dramatic drop in cholesterol with a diet low in plant sterols is a clue to the diagnosis. Other forms of genetic high blood cholesterol do not respond so dramatically to diet.*

Dietary changes are very effective in people with sitosterolemia and should focus on restriction of plant sterols. Foods high in plant sterols that should be avoided include vegetable oils, margarine, wheat germ, nuts, seeds, legumes, avocados, chocolate, and shellfish.

### Medication Options

- Ezetimibe (Zetia) is the first-line treatment for people with sitosterolemia. It works by reducing plant sterol and cholesterol absorption from food in the gut to the bloodstream. Ezetimibe may lower blood levels of plant sterols by up to 50%.
- Bile acid sequestrants are considered in those with an incomplete response to ezetimibe. These medications work by increasing the elimination of plant sterols and cholesterol in the stool and typically reduce plant sterol levels by about 30%.

To find a lipid specialist in your area, use the "find a clinician" tool on [learnyourlipids.com](http://learnyourlipids.com).