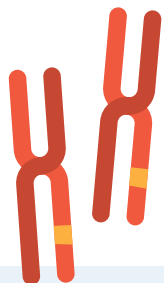


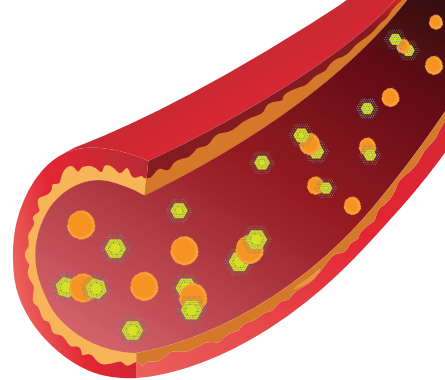
What is Sitosterolemia?

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 separated from the healthy nutrients of these foods and 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.

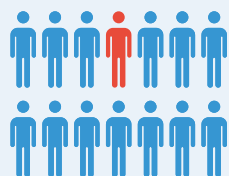


Cholesterol



Plant Sterol

How common is it?

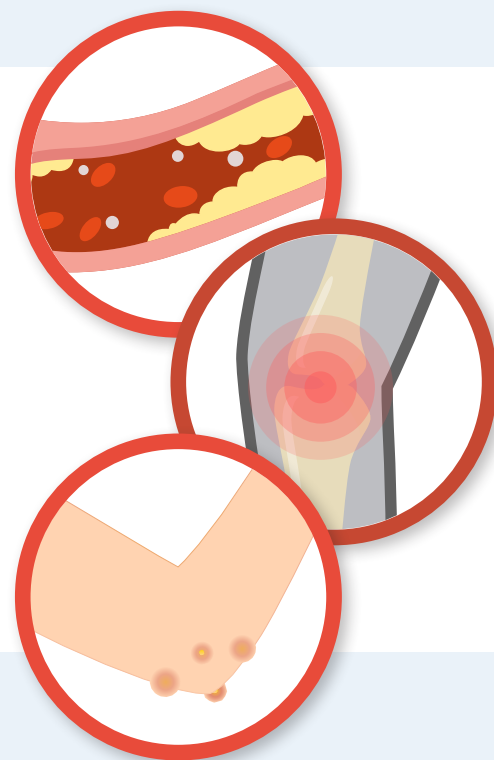


Only about 100 people with sitosterolemia have been reported. However, researchers believe that sitosterolemia may affect as many as 1 in 50,000 people that have not yet been diagnosed. If left undiagnosed and unmanaged, sitosterol can build up over time in arteries and cause a heart attack and/or stroke.

What are the symptoms?

The condition is usually first seen in childhood. People with the condition have sitosterol levels that are at least 10x greater than the usual blood level. In addition to sitosterol, cholesterol levels are often very high because the same “pump” which is supposed to eliminate plant sterols also gets rid of around 50% of the cholesterol in the diet. The good news is patients with sitosterolemia can dramatically lower their blood cholesterol by eating a low cholesterol diet.*

In addition to high blood cholesterol level, large lumps of cholesterol, called “xanthomas”, can appear on the elbows, knees and the Achilles tendon above the heel. Another symptom can be anemia, caused by plant sterols in the walls of red blood cells making them likely to break apart. Platelets – the cells in the blood which help prevent bleeding – can also be low in number and larger than usual. People with sitosterolemia may also experience joint stiffness and pain.



How is it treated?



While there is no cure for this disorder, symptoms CAN be controlled by reducing intake of plant sterols as well as by following a low-cholesterol diet. Foods high in plant sterols that should be avoided include some legumes (dried peas, beans, lentils, peanuts, almonds, walnuts, pecans, sunflower seeds, pumpkin seeds and sesame seeds) some fruits and vegetables (oranges, apples, berries, avocado, broccoli, cauliflower, brussels sprouts), vegetable oils, wheat germ oils, and shellfish.

However, diet alone is not enough. Several medications lower blood sitosterol levels by decreasing absorption. A drug called “ezetimibe” has been shown to lower blood sitosterol levels by 44% and medications called “bile acid sequestrants” give a 30% reduction.

*A dramatic drop in high blood cholesterol with a low cholesterol diet is a clue to the diagnosis of sitosterolemia. Other forms of genetic high blood cholesterol do not respond so dramatically to diet alone, without medication.