

What is Sitosterolemia (Phytosterolemia)?

Sitosterolemia is a rare inherited plant sterol storage disease. It can lead to premature thickening of artery walls due to plaque (fat build up), as well as other abnormalities. Plant sterols are similar in structure to cholesterol and occur naturally in foods in our diets.

Common Symptoms Include:

- Xanthomas (Yellowish cholesterol rich fat deposits) on the elbow, back of the heel or under the eyes)
- Blood platelet abnormalities
- Premature coronary atherosclerosis
- High blood cholesterol in a child (without a family history of heart disease or parents with high cholesterol)



What causes Sitosterolemia?

Normally we consume plant sterols in our diet. These plant sterols enter the intestinal cells, but then are pumped back out into the gut for disposal. This keeps plant sterols out of the body.

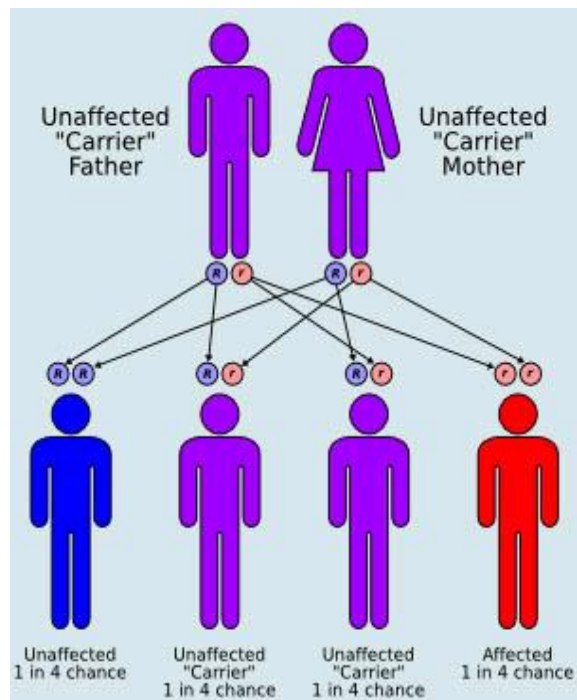
Sitosterolemia is caused by a mutation (or change) in the **ABCG8** and **ABCG5** genes. This mutation leads to abnormal proteins that are unable to pump plant sterols out of the intestine and liver cells. Therefore, high concentrations of plant sterols such as Sitosterol, Campesterol, Stigmasterol, Desmosterol and Lathosterol accumulate throughout the body.

Genetics

Sitosterolemia is an inherited autosomal recessive genetic condition. A person needs to inherit two copies of the mutated (or changed) ABCG8 or ABCG5 genes, one from each parent.

The parents are "carriers" of Sitosterolemia, meaning that they have one normal functioning copy and one non-functioning copy of the genes.

With each pregnancy, carriers of Sitosterolemia have a 1 in 4 or 25% chance of having a child with the disease.



There have been approximately 100 patients worldwide diagnosed with Sitosterolemia. Some people have no signs or symptoms and may go undetected.

How is Sitosterolemia diagnosed?

Sitosterolemia is diagnosed by measurement of elevated plant sterols in the blood.

What is the treatment for Sitosterolemia?

Medication and a diet low in plant sterols and cholesterol are the keys to treating Sitosterolemia.

Take **Ezetrol** as prescribed _____

<u>Avoid</u> foods high in Plant Sterols	Comments
All vegetable oils – this includes canola, olive, corn oil, etc. All margarines Shortening This includes foods made with the above such as potato chips and baked goods.	OK to use Lard (an animal fat) or coconut oil in small amounts at the table or in cooking
Olives and avocados	Pickles are OK
All types of Nuts and Peanuts	
Peanut butter, nut butters	
Seeds, such as sunflower seeds	
Chocolate	Hard candies, LifeSavers, peppermints, licorice are OK
Shell fish, such as Shrimp, Lobster and Crab	Fish such as salmon, tuna and pickerel are OK
<u>Limit</u> foods high in cholesterol	
Eggs – limit to 2 per week	Egg whites are OK
Liver (beef or chicken)	
Beef	Choose chicken, turkey, fish, lean pork more often than beef or lamb
Butter	Use lard or coconut oil instead
Whole milk and cream	Use skimmed milk – it provides the same amount of calcium and protein