



## ABCG8 gene

ATP binding cassette subfamily G member 8

### Normal Function

The *ABCG8* gene provides instructions for making sterolin-2, which makes up half of a protein called sterolin. The other half of the sterolin protein, sterolin-1, is produced from a gene called *ABCG5*. Sterolin is involved in eliminating plant sterols, which are fatty components of plant-based foods that cannot be used by human cells.

Sterolin is a transporter protein, which is a type of protein that moves substances across cell membranes. It is found mostly in cells of the intestines and liver and transports plant sterols. After plant sterols are absorbed from food into intestinal cells, the sterolin transporters in these cells pump them back into the intestinal tract. Sterolin transporters in liver cells pump the plant sterols into a fluid called bile that is released into the intestine. From the intestine, the plant sterols are eliminated with the feces. This process removes most of the dietary plant sterols, and allows only about 5 percent of these substances to get into the bloodstream. Sterolin also helps regulate levels of cholesterol, another fatty substance found in animal products, in a similar fashion; normally about 50 percent of cholesterol in the diet is absorbed by the body.

### Health Conditions Related to Genetic Changes

#### Sitosterolemia

At least 28 *ABCG8* gene mutations have been identified in people with sitosterolemia, which is a condition caused by accumulation of plant sterols. The mutations result in a defective sterolin transporter and impair the elimination of plant sterols and, to a lesser degree, cholesterol from the body. These fatty substances build up in the arteries, skin, and other tissues, resulting in clogged blood vessels that can impair blood flow (atherosclerosis), fatty skin growths (xanthomas), and the additional signs and symptoms of sitosterolemia. Excess plant sterols in red blood cells likely make their cell membranes stiff and prone to rupture, leading to a reduced number of red blood cells (anemia). Changes in the lipid composition of the membranes of red blood cells and platelets may account for the other blood abnormalities that sometimes occur in sitosterolemia.

#### Other disorders

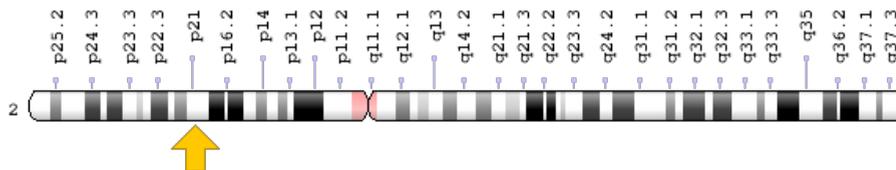
Certain normal variations (polymorphisms) in the *ABCG8* gene are associated with an increased risk of gallstones, which are small pebble-like deposits in the gallbladder or the bile ducts. Bile ducts carry bile (a fluid that helps digest fats) from the liver, where bile is produced, to the gallbladder, where it is stored, and to the small intestine, where it aids in digestion. Researchers suggest that the *ABCG8*

gene changes that increase the risk of gallstones may result in a sterolin transporter protein that pumps more cholesterol than usual into bile. This leads to the presence of more cholesterol than can be dissolved in the bile fluid in the bile ducts and gallbladder, resulting in the formation of gallstones.

## Chromosomal Location

Cytogenetic Location: 2p21, which is the short (p) arm of chromosome 2 at position 21

Molecular Location: base pairs 43,831,942 to 43,882,988 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ABCG8\_HUMAN
- ATP-binding cassette sub-family G member 8
- ATP-binding cassette, sub-family G (WHITE), member 8
- ATP-binding cassette, subfamily G, member 8
- GBD4
- sterolin-2
- sterolin 2
- STSL

## Additional Information & Resources

### Educational Resources

- Madame Curie Bioscience Database: Fat Absorption and Lipid Metabolism in Cholestasis  
<https://www.ncbi.nlm.nih.gov/books/NBK6420/>
- The Human ATP-Binding Cassette (ABC) Transporter Superfamily: ABCG Genes  
<https://www.ncbi.nlm.nih.gov/books/NBK3/#A215>

### Clinical Information from GeneReviews

- Sitosterolemia  
<https://www.ncbi.nlm.nih.gov/books/NBK131810>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ABCG8%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY G, MEMBER 8  
<http://omim.org/entry/605460>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ABCG8.html](http://atlasgeneticsoncology.org/Genes/GC_ABCG8.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ABCG8%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:13887](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:13887)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:64241>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/64241>
- UniProt  
<https://www.uniprot.org/uniprot/Q9H221>

### **Sources for This Summary**

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