

Disease

Sitosterolemia 2¹

Gene

ABCG8

Inheritance	AR
Current Treatments	diet low in shellfish sterols and plant sterols, ezetimibe, cholestyramine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma plant sterol concentrations ¹
Disease Frequency (per 100,000)	2 ²
Genes Included in Disease Frequency	ABCG8, ABCG5
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Sitosterolemia&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK131810/>

² <https://ghr.nlm.nih.gov/condition/sitosterolemia#statistics>