

Disease

Sorbitol dehydrogenase deficiency with peripheral neuropathy^{1, 2}

Gene

SORD

Inheritance	AR
Current Treatments	epalrestat and ranirestat
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	mass spectrometry analysis of serum sorbitol levels ³
Disease Frequency (per 100,000)	1 ⁴
Genes Included in Disease Frequency	SORD
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Sorbitol+dehydrogenase&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/32367058>

² <https://omim.org/entry/618912>

³ <https://www.ncbi.nlm.nih.gov/pubmed/32367057>

⁴ <https://pubmed.ncbi.nlm.nih.gov/32367058/>