

Disease

Primary systemic carnitine deficiency¹

Gene

SLC22A5

Inheritance	AR
Current Treatments	carnitine, frequent feeding and avoiding fasting
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma carnitine levels ¹
Disease Frequency (per 100,000)	0.7 ²
Genes Included in Disease Frequency	SLC22A5
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Carnitine+Deficiency&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK84551/>

² <https://www.ncbi.nlm.nih.gov/pubmed/27931018>