

Disease

Primary coenzyme Q10 deficiency 2¹

Gene

PDSS1

Inheritance	AR
Current Treatments	CoQ10 supplementation
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Disease Frequency (per 100,000)	1 ¹
Genes Included in Disease Frequency	COQ6, COQ4, COQ2, COQ7, COQ8A, COQ8B, COQ9, PDSS1, PDSS2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=coenzyme+Q10+deficiency&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK410087>