

Disease

Carnitine palmitoyltransferase II deficiency^{1, 2, 3}

Gene

CPT2

Inheritance	AR
Current Treatments	bezafibrate, High-carbohydrate (70%) and low-fat (<20%) diet to provide fuel for glycolysis, carnitine, one third of the calories as even-chain medium chain triglycerides (MCT), triheptanoin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC1022
Disease Frequency (per 100,000)	0.21 ⁴
Genes Included in Disease Frequency	CPT2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Carnitine+Palmitoyl+Transferase&term=&cntry=&state=&city=&

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1253/>
- ² <https://omim.org/entry/255110>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32885845/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/33495527/>