

Disease

Carnitine palmitoyltransferase I deficiency^{1, 2}

Gene

CPT1A

Inheritance	AR
Current Treatments	infants should eat frequently during the day and have cornstarch continuously at night; fasting should not last more than 12 hours during illness, surgery, or medical procedures; adults need a high-carbohydrate, low-fat diet to provide a constant supply of carbohydrate energy and medium-chain triglycerides to provide approximately one third of total calories. Triheptanoin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma acylcarnitine profile, total and free carnitine levels and urine organic acids ³
Disease Frequency (per 100,000)	0.15 ³
Genes Included in Disease Frequency	CPT1A
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/NBK1527/#cpt1a.Summary>
- ² <https://pubmed.ncbi.nlm.nih.gov/32885845/>
- ³ <https://www.ncbi.nlm.nih.gov/books/NBK1527/>