

Disease

Carnitine-acylcarnitine translocase deficiency^{1, 2, 3}

Gene

SLC25A20

Inheritance	AR
Current Treatments	high-carbohydrate, low-fat diet, medium chain triglycerides, frequent feeds, carnitine, avoid fasting, triheptanoin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Disease Frequency (per 100,000)	0.4 ⁴
Genes Included in Disease Frequency	SLC25A20
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC25A20&term=&cntry=&state=&city=&dist=

References

- ¹ <https://pubmed.ncbi.nlm.nih.gov/35862567/>
- ² <https://pubmed.ncbi.nlm.nih.gov/33085788/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32885845/>
- ⁴ <https://www.medicalhomeportal.org/newborn/cact-deficiency>