

Disease

3-Methylcrotonyl-CoA carboxylase 2 deficiency¹

Gene

MCCC2

Inheritance	AR
Current Treatments	IV glucose during acute episodes,avoid fasting, carnitine, protein restricted diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine organic acids, plasma acylcarnitine profile, carnitine level ¹
Disease Frequency (per 100,000)	2.4 ²
Genes Included in Disease Frequency	1:36,000 MCCC1 and MCCC2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=mccc2&term=&cntry=&state=&city=&dist=

References

¹ <https://pubmed.ncbi.nlm.nih.gov/22642865/>

² 1;50,000