

Disease

Propionic acidemia 1^{1, 2}

Gene

PCCA

Inheritance	AR
Current Treatments	diet, carnitine, biotin, metronidazole, liver transplantation, N-carbamylglutamate
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, urine organic acids ⁴
Disease Frequency (per 100,000)	0.33 ⁵
Genes Included in Disease Frequency	PCCA and PCCB
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Propionic+acidemia&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK92946/>
- ² <https://pubmed.ncbi.nlm.nih.gov/33552909/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/33595124/>
- ⁴ <https://www.ncbi.nlm.nih.gov/books/NBK92946/#propionic-a.Diagnosis>
- ⁵ <https://www.ncbi.nlm.nih.gov/pubmed/30760309>