

Disease

Phosphoenolpyruvate carboxykinase deficiency, cytosolic

1, 2, 3

Gene

PCK1

Inheritance	AR
Current Treatments	IV glucose during acute episodes and avoid fasting
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	glucose, urine organic acids ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=%09Phosphoenolpyruvate%20carboxykinase%20deficiency&viewType

References

- ¹ <https://omim.org/entry/614168>
- ² <https://pubmed.ncbi.nlm.nih.gov/35868242/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34622459/>