

Disease

Pyruvate dehydrogenase phosphatase deficiency¹

Gene

PDP1

Inheritance	AR
Current Treatments	ketogenic diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	lactate, pyruvate, urine organic acids ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/study/NCT05257005?cond=pdp1&viewType=Table&rank=1

References

¹ <https://omim.org/entry/608782>

² <https://pubmed.ncbi.nlm.nih.gov/31392110/>