

Disease

Lysinuric protein intolerance¹

Gene

SLC7A7

Inheritance	AR
Current Treatments	protein restriction, carnitine, citrulline, lysine supplementation, sodium benzoate
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	24-hour urinary excretion of cationic amino acids ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Lysinuric+protein+intolerance&term=&cntry=&state=&city=&dis

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1361/#lpi.Summary>

² <https://www.ncbi.nlm.nih.gov/books/NBK1361/#lpi.Diagnosis>