

Disease

Branched-chain ketoacid dehydrogenase kinase deficiency^{1, 2}

Gene

BCKDK

Inheritance	AR
Current Treatments	dietary management, branch chain aminoacid supplementation
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, urine organic acids ³
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=BCKDK&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/24449431>
- ² <https://pubmed.ncbi.nlm.nih.gov/35216372/>
- ³ <https://www.ncbi.nlm.nih.gov/books/NBK1319/>