Rx-Genes
Treatment Guidance at the Point of Care

Disease

Maple syrup urine disease, type Ib[1]

Gene

BCKDHB

Inheritance	AR
Current Treatments	dietary management, thiamine
Evidence for Treatment	guideline[2, 3]
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, urine organic acids[1]
Disease Frequency (per 100,000)	0.71[4]
Genes Included in Disease Frequency	DBT, BCKDHA, BCKDHB
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Maple+Syrup+Urine+Disease&term=&cntry=&state=&city=&dist

References

[1] https://www.ncbi.nlm.nih.gov/books/NBK1319/

[2] https://southeastgenetics.org/ngp/guidelines.php/105/MSUD%20Nutrition%20Guidelines/Version%201.54

[3] http://www.bimdg.org.uk/store/guidelines/ER-MSUD-v4_684428_09092016.pdf

[4] https://www.ncbi.nlm.nih.gov/pubmed/25022224