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