

## Disease

# Biotinidase deficiency<sup>1</sup>

## Gene

### BTD

|                                                 |                                                                                                                                                                                                                                                             |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                                          |
| <b>Current Treatments</b>                       | biotin                                                                                                                                                                                                                                                      |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                                              |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                         |
| <b>Non-Molecular Test</b>                       | biotinidase enzyme activity in serum or plasma <sup>2</sup>                                                                                                                                                                                                 |
| <b>ClinGen</b>                                  | <a href="https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC098">https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC098</a>                                                                                   |
| <b>Disease Frequency (per 100,000)</b>          | 1.67 <sup>3</sup>                                                                                                                                                                                                                                           |
| <b>Genes Included in Disease Frequency</b>      | BTD                                                                                                                                                                                                                                                         |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Biotinidase+deficiency&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Biotinidase+deficiency&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

## References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1322/>
- <sup>2</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1322/#biotin.Diagnosis>
- <sup>3</sup> <https://ghr.nlm.nih.gov/condition/biotinidase-deficiency#statistics>