

Disease

Biotinidase deficiency¹

Gene

BTD

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

References

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