

Disease

# Galactosemia<sup>1</sup>

Gene

## GALT

|                                                 |                                                                                                                                                                                                                                           |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                        |
| <b>Current Treatments</b>                       | Galactose free diet                                                                                                                                                                                                                       |
| <b>Evidence for Treatment</b>                   | guideline <sup>2</sup>                                                                                                                                                                                                                    |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                       |
| <b>Non-Molecular Test</b>                       | erythrocyte galactose-1-phosphate uridylyltransferase (GALT) enzyme activity <sup>1</sup>                                                                                                                                                 |
| <b>Disease Frequency (per 100,000)</b>          | 2.27 <sup>3</sup>                                                                                                                                                                                                                         |
| <b>Genes Included in Disease Frequency</b>      | GALT                                                                                                                                                                                                                                      |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Galactosemias&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Galactosemias&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1518/>
- <sup>2</sup> <https://www.ncbi.nlm.nih.gov/pubmed/27858262>
- <sup>3</sup> [https://legacyscreening.phe.org.uk/policydb\\_download.php?doc=518](https://legacyscreening.phe.org.uk/policydb_download.php?doc=518)