

Disease

Galactokinase deficiency^{1, 2}

Gene

GALK1

Inheritance	AR
Current Treatments	galactose/lactose-restricted diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	erythrocyte galactokinase enzyme activity ³
Disease Frequency (per 100,000)	1.0225 ⁴
Genes Included in Disease Frequency	GALK1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Galactosemias&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/29580649>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/22632133>
- ³ <https://pubmed.ncbi.nlm.nih.gov/22632133/>
- ⁴ <https://www.ncbi.nlm.nih.gov/pubmed/21290184>