

Disease

galactose mutarotase deficiency¹

Gene

GALM

Inheritance	AR
Current Treatments	galactose/lactose-restricted diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma galactose level while on a galactose containing diet ²
Disease Frequency (per 100,000)	0.43 ³
Genes Included in Disease Frequency	GALM
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Galactosemias&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/30451973>

² <https://pubmed.ncbi.nlm.nih.gov/30451973/>

³ <https://www.ncbi.nlm.nih.gov/pubmed/30910422>