

Disease

Galactose epimerase deficiency^{1, 2}

Gene

GALE

Inheritance	AR
Current Treatments	galactose/lactose-restricted diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	erythrocyte uridine diphosphate galactose-4-epimerase enzyme activity ³
Disease Frequency (per 100,000)	1.4 ⁴
Genes Included in Disease Frequency	GALE
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Galactosemias&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK51671/#gale-def.Summary>

² <https://www.ncbi.nlm.nih.gov/pubmed/20075179>

³ <https://www.ncbi.nlm.nih.gov/books/NBK51671/>

⁴ https://www.ncbi.nlm.nih.gov/books/NBK51671/#gale-def.Genetically_Related_Allelic_Dis