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Variant: NM_000155.4(GALT):c.607G>A (p.Glu203Lys)

Version: 1.0

CA340108

3622 (ClinVar)

Gene: GALT (HGNC:2592)
Condition: galactosemia (MONDO:0018116)
Inheritance Mode: Autosomal recessive inheritance
UUID: 2af77308-4074-4045-8e13-0949f2a9e0a4
Approved on: 2026/6/23
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HGVS expressions

NM_000155.4(GALT):c.607G>A (p.Glu203Lys)
NM_000155.4:c.607G>A
NC_000009.12:g.34648376G>A
CM000671.2:g.34648376G>A
NC_000009.11:g.34648373G>A
CM000671.1:g.34648373G>A
NC_000009.10:g.34638373G>A
NG_009029.1:g.6739G>A
NG_028966.1:g.1192G>A
NG_009029.2:g.6788G>A
ENST00000691183.1:c.*195G>A
ENST00000378842.8:c.607G>A
ENST00000378842.7:c.607G>A
ENST00000450095.6:c.280G>A
ENST00000472111.5:n.863G>A
ENST00000473506.6:c.*195G>A
ENST00000473529.5:n.766G>A
ENST00000485531.1:n.1201G>A
ENST00000487381.5:n.992G>A
ENST00000489643.6:n.382G>A
ENST00000554085.5:c.*351G>A
ENST00000554139.5:n.853G>A
ENST00000554550.5:c.*227G>A
ENST00000554638.5:n.1079G>A
ENST00000554944.5:n.956G>A
ENST00000555020.5:n.763G>A
ENST00000555086.5:n.611G>A
ENST00000555214.5:n.428G>A
ENST00000556244.1:c.594G>A
ENST00000556278.1:c.352G>A
ENST00000556494.5:n.728G>A
ENST00000557706.5:n.1169G>A
NM_000155.3:c.607G>A
NM_001258332.1:c.280G>A
NM_001258332.2:c.280G>A

Likely Pathogenic

Met criteria codes 4

PP3 PM3 PM2_Supporting PP4_Moderate

Not Met criteria codes 1

PM5

Evidence Links 0

Expert Panel

Galactosemia VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen Galactosemia Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for GALT Version 1.0.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Galactosemia VCEP

The c.607G>A variant in GALT is a missense variant predicted to cause substitution of glutamic acid by lysine at amino acid 203 (p.Glu203Lys). This variant has been detected in a proband with classical galactosemia. The patient carried the variant with the Duarte variant (N314D) in cis and was compound heterozygous with a pathogenic variant (c.329-2A>C, also known as IVSC), which was confirmed in trans by family testing (PM3, PMID: 7887416). The patient with this variant displayed severe GALT deficiency with 0.6umol/g Hb/h (normal range = 17-45), which is highly specific for galactosemia (PP4_Moderate, PMID: 7887416). The filtering allele frequency of the c.607G>A variant in GALT is 0.000009530 for European (non-Finnish) chromosomes by gnomAD v4.1.0, which is lower than the ClinGen Galactosemia threshold (≤0.0005) for PM2_Supporting, and therefore meets this criterion (PM2_Supporting). The computational predictor REVEL gives a score of 0.767, which is above the threshold of 0.7, predicting a damaging impact on GALT function (PP3). In summary, this variant meets the criteria to be classified as likely pathogenic for galactosemia based on the ACMG/AMP criteria applied, as specified by the ClinGen Galactosemia VCEP: PP4_Moderate, PM3, PP3, PM2_Supporting (GALT specifications v1.0.0).

Met criteria codes

PP3	✔	REVEL=0.767
PM3	✔	This variant has been detected in a proband with classical galactosemia. The patient carried the variant with the Duarte variant (N314D) in cis and was compound heterozygous with a pathogenic variant (c.329-2A>C, also known as IVSC), which was confirmed in trans by family testing (PM3, PMID: 7887416).
PM2_Supporting	✔	The filtering allele frequency of the c.607G>A variant in GALT is 0.000009530 for European (non-Finnish) chromosomes by gnomAD v4.1.0, which is lower than the ClinGen Galactosemia threshold (≤ 0.0005) for PM2_Supporting, and therefore meets this criterion (PM2_Supporting).
PP4_Moderate	✔	At least one patient with this variant displayed severe GALT deficiency with 0.6umol/g Hb/h (normal range = 17-45), which is highly specific for galactosemia (PP4_Moderate, PMID:7887416).

Not Met criteria codes

PM5	✖	Not met. No case data for p.Glu203Gly.

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000155.4(GALT):c.607G>A (p.Glu2...	Likely Pathogenic	Galactosemia	2026-06-23	1.0	ClinGen Galactosemia Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for GALT Version 1.0.0	GALT

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