

Variant: *NM_001258332.2:c.372C>A*

Version: 1.0

CA259481 [↗](#)

3639687 (ClinVar) [↗](#)

Gene: GALT ([HGNC:2592](#))

Condition: galactosemia ([MONDO:0018116](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 58857267-9ccc-410e-8413-e92fd4f15277

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HGVS expressions

NM_001258332.2:c.372C>A

NC_000009.12:g.34648773C>A

CM000671.2:g.34648773C>A

NC_000009.11:g.34648770C>A

CM000671.1:g.34648770C>A

NC_000009.10:g.34638770C>A

NG_009029.1:g.7136C>A

NG_028966.1:g.1589C>A

NG_009029.2:g.7185C>A

ENST00000691183.1:c.*287C>A

ENST00000378842.8:c.699C>A

ENST00000378842.7:c.699C>A

ENST00000450095.6:c.372C>A

ENST00000473506.6:c.*287C>A

ENST00000473529.5:n.858C>A

ENST00000487381.5:n.1389C>A

ENST00000489643.6:n.779C>A

ENST00000554085.5:c.*443C>A

ENST00000554550.5:c.*319C>A

ENST00000554638.5:n.1171C>A

ENST00000555020.5:n.1160C>A

ENST00000555086.5:n.703C>A

ENST00000555754.1:n.44C>A

ENST00000556244.1:c.686C>A

ENST00000556278.1:c.432+317C>A

ENST00000557706.5:n.1261C>A

NM_000155.3:c.699C>A

NM_001258332.1:c.372C>A

NM_000155.4:c.699C>A

Likely Benign

Met criteria codes **3**

BP7

BP4

PM2_Supporting

Evidence Links **0**

Expert Panel

Galactosemia VCEP [↗](#)

Criteria Specification Information

Evidence submitted by expert panel

Galactosemia VCEP

The c.699C>A (p.Val233=) variant (NM_000155.4) is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not predicted to be involved in splicing and the codon usage is not predicted to be significantly different (BP4, BP7). The c.699C>A variant in GALT is not found in gnomAD v4.1.0, which is lower than the ClinGen Galactosemia threshold (≤ 0.0005) for PM2_Supporting, and therefore meets this criterion (PM2_Supporting). To our knowledge, this variant is not reported in the clinical literature in an affected individual. The Galactosemia VCEP does not consider PM2_Supporting as conflicting data with BP4 and BP7. Therefore, this variant meets the criteria to be classified as likely benign for galactosemia. ACMG/AMP criteria applied, as specified by the ClinGen Galactosemia VCEP: PM2_Supporting, BP4, BP7 (GALT specifications v1.0.0).

Met criteria codes

BP7			The c.699C>A (p.Val233=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not predicted to be involved in splicing (BP4, BP7)
BP4			The computational splicing predictor SpliceAI gives a score of 0.06 for donor loss and 0.02 for donor gain suggesting that the variant has no impact on splicing (BP4)
PM2_Supporting			The c.699C>A variant in GALT is not found in gnomAD v4.1.0, which is lower than the ClinGen Galactosemia threshold (≤ 0.0005) for PM2_Supporting, and therefore meets this criterion (PM2_Supporting).

Curation History [↗](#)

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