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Variant: NM_000155.4(GALT):c.772C>T (p.Arg258Cys)

Version: 1.0

CA266764

92522 (ClinVar)

Gene: GALT (HGNC:2592)
Condition: galactosemia (MONDO:0018116)
Inheritance Mode: Autosomal recessive inheritance
UUID: 489d5ea6-a834-44c0-b755-49b67288ec95
Approved on: 2026/6/23
Published on: 2026/6/23

HGVS expressions

NM_000155.4(GALT):c.772C>T (p.Arg258Cys)
NM_000155.4:c.772C>T
NC_000009.12:g.34648846C>T
CM000671.2:g.34648846C>T
NC_000009.11:g.34648843C>T
CM000671.1:g.34648843C>T
NC_000009.10:g.34638843C>T
NG_028966.1:g.1662C>T
NG_009029.2:g.7258C>T
ENST00000691183.1:c.*360C>T
ENST00000378842.8:c.772C>T
ENST00000378842.7:c.772C>T
ENST00000450095.6:c.445C>T
ENST00000473506.6:c.*360C>T
ENST00000489643.6:n.852C>T
ENST00000554085.5:c.*516C>T
ENST00000554550.5:c.*392C>T
ENST00000554638.5:n.1244C>T
ENST00000555020.5:n.1233C>T
ENST00000555086.5:n.776C>T
ENST00000555754.1:n.117C>T
ENST00000556244.1:c.759C>T
ENST00000556278.1:c.432+390C>T
ENST00000557706.5:n.1334C>T
NM_000155.3:c.772C>T
NM_001258332.1:c.445C>T
NM_001258332.2:c.445C>T

Likely Pathogenic

Met criteria codes 4

PP3 PM2_Supporting PP4_Moderate PM3_Strong

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.772C>T variant in GALT is a missense variant predicted to cause substitution of arginine by cysteine at amino acid 258 (p.Arg258Cys). The filtering allele frequency (the upper threshold of the 95% CI of 71/1180028) of the c.772C>T variant in GALT is 0.00004863 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 0.952, which is equal to or above the threshold of 0.7, evidence that correlates with impact to GALT function (PP3). This variant has been detected in at least 4 individuals with galactosemia. Of those individuals, all were compound heterozygous for this variant and a pathogenic or likely pathogenic variant (PM3_strong, Internal lab contributors, PMID: 34485021). At least two patients with this variant displayed RBC GALT enzyme activity <10%, which is highly specific for galactosemia (PP4_Moderate, Internal lab contributors). 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: PM3_Strong; PP4_Moderate; PM2_Supporting; PP3 (GALT specifications v1.0.0).

Met criteria codes

PP3	✔	The computational predictor REVEL gives a SCORE of 0.952, which is equal to or above the threshold of 0.7, evidence that correlates with impact to GALT function (PP3).
PM2_Supporting	✔	The filtering allele frequency (the upper threshold of the 95% CI of 71/1180028) of the c.772C>T variant in GALT is 0.00004863 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 RBC GALT enzyme activity <10%, which is highly specific for galactosemia (PP4_Moderate, Internal lab contributors).
PM3_Strong	✔	This variant has been detected in at least 4 individuals with galactosemia. Of those individuals, all were compound heterozygous for this variant and a pathogenic or likely pathogenic variant (PM3_strong, Internal lab contributors, PMID: 34485021).

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.772C>T (p.Arg2...	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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