The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000155.4(GALT):c.267C>G (p.Tyr89Ter)

Version: 1.0

CA373279118

2675829 (ClinVar)

Gene: GALT (HGNC:2592)
Condition: galactosemia (MONDO:0018116)
Inheritance Mode: Autosomal recessive inheritance
UUID: baccb028-f48c-4069-badf-19548f4aaffa
Approved on: 2026/6/23
Published on: 2026/6/23

HGVS expressions

NM_000155.4(GALT):c.267C>G (p.Tyr89Ter)
NM_000155.4:c.267C>G
NC_000009.12:g.34647506C>G
CM000671.2:g.34647506C>G
NC_000009.11:g.34647503C>G
CM000671.1:g.34647503C>G
NC_000009.10:g.34637503C>G
NG_009029.1:g.5869C>G
NG_028966.1:g.322C>G
NG_009029.2:g.5918C>G
ENST00000691183.1:c.267C>G
ENST00000378842.8:c.267C>G
ENST00000378842.7:c.267C>G
ENST00000450095.6:c.50+248C>G
ENST00000465543.6:n.606C>G
ENST00000468099.2:n.540C>G
ENST00000472111.5:n.308C>G
ENST00000473506.6:c.253-35C>G
ENST00000473529.5:n.314C>G
ENST00000485531.1:n.493C>G
ENST00000487381.5:n.526C>G
ENST00000489643.6:n.282+248C>G
ENST00000554085.5:c.*11C>G
ENST00000554139.5:n.320C>G
ENST00000554330.5:n.250-35C>G
ENST00000554550.5:c.252+248C>G
ENST00000554638.5:n.524C>G
ENST00000554897.5:c.252+248C>G
ENST00000554944.5:n.283-35C>G
ENST00000555020.5:n.297C>G
ENST00000555086.5:n.271C>G
ENST00000555214.5:n.261+248C>G
ENST00000556157.1:n.391C>G
ENST00000556244.1:c.254C>G
ENST00000556278.1:c.252+248C>G
ENST00000556403.5:n.280C>G
ENST00000556494.5:n.299C>G

ENST00000557541.5:n.446-35C>G
ENST00000557706.5:n.614C>G
NM_000155.3:c.267C>G
NM_001258332.1:c.50+248C>G
NM_001258332.2:c.50+248C>G

Likely Pathogenic

Met criteria codes 2

PVS1 PM2_Supporting

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.267C>G (p.Tyr89Ter) (NM_000155.4) variant in GALT is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 3/11 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1; PMIDs: 2940989, 22944367). Additionally, this truncation would eliminate the active site. This variant was only detected on one chromosome 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). In summary, the ClinGen Galactosemia VCEP classified the variant as likely pathogenic based on PVS1 + PM2_supporting (GALT specifications v1.0.0).

Met criteria codes

PVS1	✔	The c.267C>G (p.Tyr89Ter) (NM_000155.4) variant in GALT is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 3/11 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1; PMIDs:2940989, 22944367).
PM2_Supporting	✔	This variant was only detected on one chromosome 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).

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.267C>G (p.Tyr8...	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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