

Variant: NM_000155.4(GALT):c.270T>C (p.Asp90=)

Version: 1.0

CA464595620 [↗](#)

913656 (ClinVar) [↗](#)

Gene: GALT ([HGNC:2592](#))
Condition: galactosemia ([MONDO:0018116](#))
Inheritance Mode: Autosomal recessive inheritance
UUID: 718cdaa6-7244-410b-add6-1faec06cfaaa
Approved on: 2026/7/20
Published on: 2026/7/20

HGVS expressions

NM_000155.4(GALT):c.270T>C (p.Asp90=)
NM_000155.4:c.270T>C
NC_000009.12:g.34647509T>C
CM000671.2:g.34647509T>C
NC_000009.11:g.34647506T>C
CM000671.1:g.34647506T>C
NC_000009.10:g.34637506T>C
NG_009029.1:g.5872T>C
NG_028966.1:g.325T>C
NG_009029.2:g.5921T>C
ENST00000691183.1:c.270T>C
ENST00000378842.8:c.270T>C
ENST00000378842.7:c.270T>C
ENST00000450095.6:c.50+251T>C
ENST00000465543.6:n.609T>C
ENST00000468099.2:n.543T>C
ENST00000472111.5:n.311T>C
ENST00000473506.6:c.253-32T>C
ENST00000473529.5:n.317T>C
ENST00000485531.1:n.496T>C
ENST00000487381.5:n.529T>C
ENST00000489643.6:n.282+251T>C
ENST00000554085.5:c.*14T>C
ENST00000554139.5:n.323T>C
ENST00000554330.5:n.250-32T>C
ENST00000554550.5:c.252+251T>C
ENST00000554638.5:n.527T>C
ENST00000554897.5:c.252+251T>C
ENST00000554944.5:n.283-32T>C
ENST00000555020.5:n.300T>C
ENST00000555086.5:n.274T>C
ENST00000555214.5:n.261+251T>C
ENST00000556157.1:n.394T>C
ENST00000556244.1:c.257T>C
ENST00000556278.1:c.252+251T>C
ENST00000556403.5:n.283T>C
ENST00000556494.5:n.302T>C

ENST00000557541.5:n.446-32T>C
ENST00000557706.5:n.617T>C
NM_000155.3:c.270T>C
NM_001258332.1:c.50+251T>C
NM_001258332.2:c.50+251T>C

Uncertain Significance

Met criteria codes **2**

PM2_Supporting PP3

Not Met criteria codes **1**

BP4

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*

 **Criteria Specification Approval History**

 **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Galactosemia VCEP

The c.291C>T variant is a synonymous (silent) variant that the computational splicing predictor SpliceAI gives a delta score of 0.2 for acceptor gain; however, the acceptor gain prediction is the canonical acceptor (PP3). The filtering allele frequency of the c.270T>C variant in GALT is 0.00006614 for Admixed American chromosomes by gnomAD v4.1.1, which is lower than the ClinGen Galactosemia threshold (≤ 0.0005) for PM2_Supporting, and therefore meets this criterion (PM2_Supporting). In summary, this variant meets the criteria to be classified as variant of uncertain significance for galactosemia based on the ACMG/AMP criteria applied, as specified by the ClinGen Galactosemia VCEP: PM2_Supporting, PP3

Met criteria codes

PM2_Supporting			The filtering allele frequency of the c.270T>C variant in GALT is 0.00006614 for Admixed American chromosomes by gnomAD v4.1.1, which is lower than the ClinGen Galactosemia threshold (≤ 0.0005) for PM2_Supporting, and therefore meets this criterion (PM2_Supporting).
PP3			The computational predictor SpliceAI gives a delta score of 0.2 for acceptor gain, but the acceptor gain prediction is the canonical acceptor.

Not Met criteria codes

BP4			The computational predictor SpliceAI gives a delta score of 0.2 for acceptor gain, but the acceptor gain prediction is the canonical acceptor.
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