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Variant: NM_000155.4(GALT):c.855G>T (p.Lys285Asn)

Version: 1.0

CA340107

3621 (ClinVar)

Gene: GALT (HGNC:2592)
Condition: galactosemia (MONDO:0018116)
Inheritance Mode: Autosomal recessive inheritance
UUID: b256f847-fbb4-4bb7-842f-cb45be7cc8d3
Approved on: 2026/6/23
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HGVS expressions

NM_000155.4:c.855G>T
NM_000155.4(GALT):c.855G>T (p.Lys285Asn)
NC_000009.12:g.34649032G>T
CM000671.2:g.34649032G>T
NC_000009.11:g.34649029G>T
CM000671.1:g.34649029G>T
NC_000009.10:g.34639029G>T
NG_009029.1:g.7395G>T
NG_028966.1:g.1848G>T
NG_009029.2:g.7444G>T
ENST00000691183.1:c.*443G>T
ENST00000378842.8:c.855G>T
ENST00000378842.7:c.855G>T
ENST00000450095.6:c.528G>T
ENST00000488412.2:n.111G>T
ENST00000489643.6:n.935G>T
ENST00000554550.5:c.*475G>T
ENST00000554638.5:n.1327G>T
ENST00000555020.5:n.1316G>T
ENST00000555086.5:n.962G>T
ENST00000555754.1:n.303G>T
ENST00000556278.1:c.432+576G>T
ENST00000557706.5:n.1430G>T
NM_000155.3:c.855G>T
NM_001258332.1:c.528G>T
NM_001258332.2:c.528G>T

Pathogenic

Met criteria codes 5

PM3_Very Strong | PP3 | PM2_Supporting | PS3_Supporting | PP4_Moderate

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.855G>T variant in GALT is a missense variant predicted to cause substitution of lysine to asparagine at amino acid 285 (p.Lys285Asn). This variant has been detected in multiple individuals with galactosemia in the compound heterozygous and homozygous states (e.g., p.Q188R, c.329-2A>C, p.L195P, PM3_Very Strong, PMIDs: 31954591, 18210213, 25592817, 34030713). Multiple patients with this variant displayed RBC GALT enzyme activity <10%, which is highly specific for galactosemia (PP4_Moderate; PMIDs: 31954591, 25592817). GALT enzyme activity in purified recombinant human protein studies and when expressed in yeast showed absence of GALT activity indicating that this variant impacts protein function (PS3_Supporting; PMIDs: 31954591, 18210213, 25592817, 34030713). The filtering allele frequency of the c.855G>T variant in GALT is 0.00025 for Latino 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 0.883, which is equal to or above the threshold of 0.7, evidence that correlates with impact to GALT function (PP3). In summary, this variant meets the criteria to be classified as pathogenic for galactosemia based on the ACMG/AMP criteria applied, as specified by the ClinGen Galactosemia VCEP: PM3_Very Strong, PP4_Moderate, PS3_Supporting, PM2_Supporting, PP3 (GALT specifications v1.0.0).

Met criteria codes

PM3_Very Strong	✔	This variant has been detected in at least 24 individuals with galactosemia. Of those individuals, 22 were compound heterozygous for the variant and a pathogenic or likely pathogenic variant and 9 of those were confirmed in trans by family testing. 2 affected individuals were homozygous for the variant. PMIDs: 31954591, 18210213, 25592817, 34030713 (PM3_Very Strong).
PP3	✔	The computational predictor REVEL gives a SCORE of 0.883, 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 of the c.855G>T variant in GALT is 0.00025 for Latino 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).
PS3_Supporting	✔	GALT enzyme activity in purified recombinant human protein studies and when expressed in yeast showed absence of GALT activity indicating that this variant impacts protein function (PMIDs: 31954591, 18210213, 25592817, 34030713)(PS3_Supporting).
PP4_Moderate	✔	At least one patient with this variant displayed [DESCRIBE PHENOTYPE], which is highly specific for galactosemia (PP4_Moderate), PMIDs, ClinVar SCVs, Internal lab contributors)

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.855G>T (p.Lys2…	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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