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# *Variant: NM_000155.4(GALT):c.379A>G (p.Lys127Glu)*

Version: 1.0

*CA259377*

*25167 (ClinVar)*

**Gene**: GALT (HGNC:2592)
**Condition**: galactosemia (MONDO:0018116)
**Inheritance Mode**: Autosomal recessive inheritance
**UUID:** d76e8033-3166-4164-a062-1c6945d96704
**Approved on**: Tue, 08 Sep 2026 00:00:00 +0000
**Published on**: Fri, 23 Oct 2026 00:00:00 +0000

## *HGVS expressions*

**NM_000155.4(GALT):c.379A>G (p.Lys127Glu)**
**NM_000155.4:c.379A>G**
NC_000009.12:g.34647833A>G
CM000671.2:g.34647833A>G
NC_000009.11:g.34647830A>G
CM000671.1:g.34647830A>G
NC_000009.10:g.34637830A>G
NG_009029.1:g.6196A>G
NG_028966.1:g.649A>G
NG_009029.2:g.6245A>G
ENST00000691183.1:c.330A>G
ENST00000378842.8:c.379A>G
ENST00000378842.7:c.379A>G
ENST00000450095.6:c.52A>G
ENST00000465543.6:n.718A>G
ENST00000472111.5:n.635A>G
ENST00000473506.6:c.330A>G
ENST00000473529.5:n.515A>G
ENST00000485531.1:n.820A>G
ENST00000487381.5:n.764A>G
ENST00000489643.6:n.283-282A>G
ENST00000554085.5:c.*123A>G
ENST00000554139.5:n.558A>G
ENST00000554330.5:n.542A>G
ENST00000554550.5:c.254A>G
ENST00000554638.5:n.851A>G
ENST00000554897.5:c.254A>G
ENST00000554944.5:n.575A>G
ENST00000555020.5:n.535A>G
ENST00000555086.5:n.383A>G
ENST00000555214.5:n.262-215A>G
ENST00000556244.1:c.366A>G
ENST00000556278.1:c.253-282A>G
ENST00000556494.5:n.500A>G
ENST00000557541.5:n.523A>G
ENST00000557706.5:n.941A>G
NM_000155.3:c.379A>G

NM_001258332.1:c.52A>G
NM_001258332.2:c.52A>G

**Likely Pathogenic**

**Met criteria codes** 4

PP3 PP4 PM2_Supporting PM3_Strong

**Not Met criteria codes** 1

PM5

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 1.1*

**Criteria Specification Approval History**

**Criteria Specifications for this VCEP**

Evidence submitted by expert panel

***Galactosemia VCEP***

**The c.379A>G variant in GALT is a missense variant predicted to cause substitution of lysine by glutamic acid at amino acid 127 (p.Lys127Glu). The filtering allele frequency (the upper threshold of the 95% CI of 9/1614030) of the c.379A>G variant in GALT is 0.0000036 for Non-Finnish European 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). The computational predictor REVEL gives a score of 0.816, which is equal to or above the threshold of 0.7, evidence that correlates with impact to GALT function (PP3). At least one patient with this variant displayed GALT erythrocyte activity of <3.3 %, which is highly specific for galactosemia (PP4, PMID: 38444572). This variant has been detected in at least 7 individuals with galactosemia. All of those individuals were compound heterozygous for this variant and a pathogenic variant (PM3_Strong; PMID: 10384398, 11397328, 22870861, 31954591, 38444572). 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: PM2_Supporting, PP3, PP4, PM3_Strong (GALT specifications v1.1)**

**Met criteria codes**

| | | |
|---|---|---|
| **PP3** | ✔ | The computational predictor REVEL gives a score of 0.816, which is equal to or above the threshold of 0.7, evidence that correlates with impact to GALT function (PP3). |
| **PP4** | ✔ | At least one patient with this variant displayed GALT erythrocyte activity of <3.3 %, which is highly specific for galactosemia (PP4, PMID: 38444572). |
| **PM2_Supporting** | ✔ | The filtering allele frequency (the upper threshold of the 95% CI of 9/1614030) of the c.379A>G variant in GALT is 0.0000036 for Non-Finnish European 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). |
| **PM3_Strong** | ✔ | This variant has been detected in at least 7 individuals with galactosemia. All of those individuals were compound heterozygous for the variant and a pathogenic variant (PM3_Strong; PMID: 10384398, 11397328, 22870861, 31954591, 38444572). |

**Not Met criteria codes**

| | | | |
|---|---|---|---|
| **PM5** | | ✖ | GALT c.380A>G (p.Lys127Arg) is a VUS |

## Curation History

Showing 1 to 1 of 1 rows

| See Report | Preferred Variant Title | Classification | Condition | Published Date | Version | Criteria Specification | Gene |
|---|---|---|---|---|---|---|---|
| View | NM_000155.4(GALT):c.379A>G (p.Lys1... | Likely Pathogenic | Galactosemia | 2026-10-23 | 1.0 | ClinGen Galactosemia Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for GALT Version 1.1 | GALT |

Showing 1 to 1 of 1 rows

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