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Variant: NM_000155.4(GALT):c.203A>C (p.His68Pro)

Version: 1.0

CA260400

36143 (ClinVar)

Gene: GALT (HGNC:2592)
Condition: galactosemia (MONDO:0018116)
Inheritance Mode: Autosomal recessive inheritance
UUID: d1b2f8ee-3d9b-49cf-8981-4907896a1a98
Approved on: 2026/6/15
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HGVS expressions

NM_000155.4:c.203A>C
NM_000155.4(GALT):c.203A>C (p.His68Pro)
NC_000009.12:g.34647209A>C
CM000671.2:g.34647209A>C
NC_000009.11:g.34647206A>C
CM000671.1:g.34647206A>C
NC_000009.10:g.34637206A>C
NG_009029.1:g.5572A>C
NG_028966.1:g.25A>C
NG_009029.2:g.5621A>C
ENST00000691183.1:c.203A>C
ENST00000378842.8:c.203A>C
ENST00000378842.7:c.203A>C
ENST00000450095.6:c.1A>C
ENST00000465543.6:n.542A>C
ENST00000468099.2:n.243A>C
ENST00000472111.5:n.244A>C
ENST00000473506.6:c.203A>C
ENST00000473529.5:n.250A>C
ENST00000485531.1:n.196A>C
ENST00000487381.5:n.229A>C
ENST00000489643.6:n.233A>C
ENST00000554085.5:c.203A>C
ENST00000554139.5:n.256A>C
ENST00000554330.5:n.200A>C
ENST00000554550.5:c.203A>C
ENST00000554638.5:n.227A>C
ENST00000554897.5:c.203A>C
ENST00000554944.5:n.233A>C
ENST00000555020.5:n.233A>C
ENST00000555086.5:n.207A>C
ENST00000555214.5:n.212A>C
ENST00000556157.1:n.310A>C
ENST00000556244.1:c.87A>C
ENST00000556278.1:c.203A>C
ENST00000556403.5:n.216A>C
ENST00000556494.5:n.235A>C

ENST00000557541.5:n.396A>C
ENST00000557706.5:n.317A>C
NM_000155.3:c.203A>C
NM_001258332.1:c.1A>C
NM_001258332.2:c.1A>C

Likely Pathogenic

Met criteria codes 4

PP3 PM3 PP4_Moderate 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.203A>C variant in GALT is a missense variant predicted to cause substitution of histidine to proline at amino acid 68 (p.His68Pro). The filtering allele frequency (the upper threshold of the 95% CI of 4/1461862) of the c.203A>C variant in GALT is 0.0000027 for South Asian 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 0.861, 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 1 individual with galactosemia; This individual was compound heterozygous for the variant and a pathogenic variant (p.Gln188Arg) and were confirmed in trans by Sanger sequencing (PM3; PMID: 38469090). The patient with this combination of variants displayed severe GALT deficiency with 0% GALT activity, which is highly specific for galactosemia (PP4_moderate; PMID: 38469090). 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: PP4_Moderate, PM3, PP3, PM2_Supporting (GALT specifications v1.0.0).

Met criteria codes

PP3	✔	The computational predictor REVEL gives a score of 0.861, which is equal to or above the threshold of 0.7, evidence that correlates with impact to GALT function (PP3).
PM3	✔	This variant has been detected in at least 1 individual with galactosemia; This individual was compound heterozygous for the variant and a pathogenic variant (p.Gln188Arg) and was confirmed in trans by Sanger sequencing (PM3; PMID: 38469090)
PP4_Moderate	✔	A patient with p.His68Pro and p.Gln188Arg in the compound heterozygous state displayed severe GALT deficiency with 0% GALT activity, which is highly specific for galactosemia (PP4_moderate; PMID: 38469090)
PM2_Supporting	✔	The filtering allele frequency (the upper threshold of the 95% CI of 4/1461862) of the c.203A>C variant in GALT is 0.0000027 for South Asian 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).

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.203A>C (p.His6…	Likely Pathogenic	Galactosemia	2026-06-15	1.0	ClinGen Galactosemia Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for GALT Version 1.0.0	GALT

Showing 1 to 1 of 1 rows

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