

Variant: NM_000531.6:c.147C>T

Version: 1.0

CA10385792 [↗](#)

1166654 (ClinVar) [↗](#)

Gene: OTC ([HGNC:5009](#))

Condition: ornithine carbamoyltransferase deficiency ([MONDO:0010703](#))

Inheritance Mode: X-linked inheritance (recessive (HP:0001419))

UUID: e4b0d0d7-3c7f-41b8-aebf-49a2babb1955

Approved on: 2026/5/20

Published on: 2026/5/29

HGVS expressions

NM_000531.6:c.147C>T

NC_000023.11:g.38367360C>T

CM000685.2:g.38367360C>T

NC_000023.10:g.38226613C>T

CM000685.1:g.38226613C>T

NC_000023.9:g.38111557C>T

NG_008471.1:g.19878C>T

ENST00000039007.5:c.147C>T

ENST00000643344.1:c.147C>T

ENST00000039007.4:c.147C>T

ENST00000465127.1:c.172-298761C>T

ENST00000488812.1:n.239C>T

NM_000531.5:c.147C>T

Benign

Met criteria codes **3**

BA1

BP4

BP7

Not Met criteria codes **1**

PM1

Evidence Links **0**

Expert Panel

Urea Cycle Disorders VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Urea Cycle Disorders
Expert Panel Specifications to the ACMG/AMP Variant
Interpretation Guidelines for OTC Version 1.0.0

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Urea Cycle Disorders VCEP

The NM_000531.6:c.147C>T (p.Thr49=) variant in OTC is a synonymous variant in exon 2 of the OTC gene. There are 10 hemizygous males in gnomAD v.4.1.0. This variant being observed in ≥10 (female) homozygotes or (male) hemizygotes in the most current version of gnomAD available at the time of curation (v.4.1.0), The NM_000531.6:c.147C>T (p.Thr49=) meets BP4 (Splice AI predicts no impact of the variant on splicing) (BP4, BP7) In summary, this variant meets the criteria to be classified as a benign variant for OTC deficiency based on

the ACMG/AMP criteria applied, as specified by the ClinGen Urea Cycle Disorders Variant Curation Expert Panel (OTC specifications version 1.0.0): BA1, BP4, BP7

Met criteria codes

BA1			There are 10 hemizygous males in gnomAD v.4.1.0. This variant being observed in ≥ 10 (female) homozygotes or (male) hemizygotes in the most current version of gnomAD available at the time of curation (v.4.1.0) is an OTC-specific stand-alone criterion for benign variants. As a result, this variant meets this criterion (BA1).
BP4			The NM_000531.6:c.147C>T (p.Thr49=) meets BP4 (Splice AI predicts no impact of the variant on splicing) and the nucleotide is poorly conserved (Cactus 447 phyloP score -3.147).
BP7			SpliceAI predicts no impact on splicing (scores <0.2) and nucleotide is not highly conserved (conservation score in the Cactus alignment of 447 mammalian genomes is -3.147).

Not Met criteria codes

PM1			This variant is not located in a mutational hot spot and/or critical and well-established functional domain (e.g. active site of an enzyme) in OTC.
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Curation History



 

 

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