

Variant: *NM_000531.6(OTC):c.264AAG[1] (p.Arg89del)*

Version: 1.1

CA2424871675 [↗](#)

957857 (ClinVar) [↗](#)

Gene: [OTC](#)

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

Inheritance Mode: X-linked inheritance

UUID: 1013949b-d83b-492b-adf2-b808a41e801c

Approved on: 2026/6/11

Published on: 2026/9/11

HGVS expressions

NM_000531.6:c.264AAG[1]

NM_000531.6(OTC):c.264AAG[1] (p.Arg89del)

NC_000023.11:g.38369846_38369848del

CM000685.2:g.38369846_38369848del

NC_000023.10:g.38229099_38229101del

CM000685.1:g.38229099_38229101del

NC_000023.9:g.38114043_38114045del

NG_008471.1:g.22364_22366del

ENST00000039007.5:c.267_269del

ENST000000643344.1:c.267_269del

ENST00000039007.4:c.267_269del

ENST000000465127.1:c.172-296275_172-296273del

ENST000000488812.1:n.353+6_353+8del

NM_000531.5:c.267_269del

NM_000531.6:c.267_269del

Uncertain Significance

Met criteria codes **2**

PM4

PM2_Supporting

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*

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

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

Evidence submitted by expert panel

Urea Cycle Disorders VCEP

The NM_000156.6:c.264AAG[1] (p.Arg89del) variant in OTC is an in-frame deletion of the Arg89 residue, which is located in the loop between the B3 beta-sheet and the H2 alpha-helix and is immediately adjacent to the STRT motif which is involved in carbamylphosphate binding and catalysis (PM4). This variant is absent in gnomAD v4.1.0 (PM2_Supporting). In summary, this variant meets the criteria to be

classified as a variant of uncertain significance 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): PM4, PM2_Supporting.

Met criteria codes

PM4			The Arg89 residue is located in the loop between the B3 beta-sheet and the H2 alpha-helix. The Arg89 residue is immediately adjacent to the STRT motif which is involved in carbamylphosphate binding and catalysis.
PM2_Supporting			Absent from gnomAD v4.1.0

Curation History



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