

Variant: *NM_000531.6(OTC):c.137A>G (p.Lys46Arg)*

Version: 1.1

CA121291 [↗](#)

10994 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 49573b1f-663f-4820-99c1-cd9eeebbc98e

Approved on: 2026/6/11

Published on: 2026/9/11

HGVS expressions

NM_000531.6(OTC):c.137A>G (p.Lys46Arg)

NM_000531.6:c.137A>G

NC_000023.11:g.38367350A>G

CM000685.2:g.38367350A>G

NC_000023.10:g.38226603A>G

CM000685.1:g.38226603A>G

NC_000023.9:g.38111547A>G

NG_008471.1:g.19868A>G

ENST00000039007.5:c.137A>G

ENST000000643344.1:c.137A>G

ENST00000039007.4:c.137A>G

ENST00000465127.1:c.172-298771A>G

ENST00000488812.1:n.229A>G

NM_000531.5:c.137A>G

Benign

Met criteria codes **1**

BA1

Not Met criteria codes **3**

BP4

PP3

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*

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

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

Evidence submitted by expert panel

Urea Cycle Disorders VCEP

The NM_000531.6:c.137A>G (p.Lys46Arg) is a missense OTC variant in exon 2. The MAF >5% in all populations except in East Asians with more than one hundred thousand combined homo- and hemizygotes (BA1). In summary, this variant meets the criteria to be classified as benign 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.

Met criteria codes

BA1

i

✓

The frequency of the G allele is >1% in all populations except in East Asians, and has combined homo-/hemizygotes over one hundred thousand. BA1 applies per OTC-specific criteria established by the UCD VCEP.

Not Met criteria codes

BP4

i

✗

REVEL score of 0.407 is lower than the 0.644 threshold for PP3 but higher than the OTC gene-specific 0.290 cut-off for BP4.

PP3

i

✗

REVEL score of 0.407 is lower than the 0.644 threshold for PP3 but higher than the OTC gene-specific 0.290 cut-off for BP4.

PM1

i

✗

Not in any of the designated hotspots per the OTC specifications

Curation History [↗](#)

⛶

☰

▼

⬇

▼

Showing 1 to 2 of 2 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.