

Variant: *NM_177438.3(DICER1):c.238G>T (p.Glu80Ter)*

Version: 2.0

[CA390889868](#) 

[649946 \(ClinVar\)](#) 

Gene: DICER1 ([HGNC:23405](#))

Condition: DICER1-related tumor predisposition ([MONDO:0100216](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 23ee9f03-2243-4d18-b1eb-aec9c811e84f

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HGVS expressions

NM_177438.3:c.238G>T

NM_177438.3(DICER1):c.238G>T (p.Glu80Ter)

NC_000014.9:g.95132584C>A

CM000676.2:g.95132584C>A

NC_000014.8:g.95598921C>A

CM000676.1:g.95598921C>A

NC_000014.7:g.94668674C>A

NG_016311.1:g.29839G>T

ENST00000529720.2:c.238G>T

ENST00000531162.7:c.238G>T

ENST00000674628.2:c.238G>T

ENST00000675540.2:c.238G>T

ENST00000696733.1:c.238G>T

ENST00000696734.1:c.238G>T

ENST00000696736.1:c.238G>T

ENST00000696737.1:c.238G>T

ENST00000696739.1:n.686G>T

ENST00000696740.1:c.238G>T

ENST00000696921.1:n.469G>T

ENST00000696922.1:n.647G>T

ENST00000696923.1:c.238G>T

ENST00000696924.1:c.238G>T

ENST00000696925.1:n.647G>T

ENST00000696928.1:n.435G>T

ENST00000343455.8:c.238G>T

ENST00000393063.6:c.238G>T

ENST00000526495.6:c.238G>T

ENST00000531162.6:c.238G>T

ENST00000532939.3:c.238G>T

ENST00000556045.6:c.238G>T

ENST00000674628.1:c.238G>T

ENST00000675995.1:c.238G>T

ENST00000343455.7:c.238G>T

ENST00000393063.5:c.238G>T

ENST00000526495.5:c.238G>T

ENST00000527414.5:c.238G>T

ENST00000529206.1:n.379G>T

ENST00000541352.5:c.238G>T
NM_001195573.1:c.238G>T
NM_001271282.2:c.238G>T
NM_001291628.1:c.238G>T
NM_030621.4:c.238G>T
NM_177438.2:c.238G>T
NM_001271282.3:c.238G>T
NM_001291628.2:c.238G>T
NM_001395677.1:c.238G>T
NM_001395678.1:c.238G>T
NM_001395679.1:c.238G>T
NM_001395680.1:c.238G>T
NM_001395682.1:c.238G>T
NM_001395683.1:c.238G>T
NM_001395684.1:c.238G>T
NM_001395685.1:c.238G>T
NM_001395686.1:c.-37G>T
NM_001395687.1:c.-37G>T
NM_001395688.1:c.-37G>T
NM_001395689.1:c.-37G>T
NM_001395690.1:c.-37G>T
NM_001395691.1:c.-221G>T
NM_001395692.1:c.238G>T
NM_001395693.1:c.238G>T
NM_001395694.1:c.238G>T
NM_001395695.1:c.238G>T
NM_001395696.1:c.-37G>T
NM_001395697.1:c.-1331G>T
NM_001395698.1:c.-37G>T
NM_001395699.1:c.238G>T
NM_001395700.1:c.238G>T
NR_172715.1:n.583G>T
NR_172716.1:n.583G>T
NR_172717.1:n.750G>T
NR_172718.1:n.750G>T
NR_172719.1:n.583G>T
NR_172720.1:n.583G>T

Likely Pathogenic

Met criteria codes **2**

PM2_Supporting PVS1

Not Met criteria codes **24**

PS1 PS2 PS3 PS4 PP1 PP2
PP3 PP4 BA1 PM6 PM1
PM3 PM5 PM4 BS2 BS1
BS4 BS3 BP4 BP3 BP1 BP2
BP5 BP7

Evidence Links **0**

Expert Panel

[DICER1 and miRNA-Processing Gene VCEP](#)

Criteria Specification Information

[Criteria Specification:](#) *ClinGen DICER1 and miRNA-Processing Gene Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for DICER1 Version 1.3.0*





[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)
















DICER1 and miRNA-Processing Gene VCEP




























The NM_177438.2:c.238G>T (p.Glu80Ter) variant in DICER1 is a nonsense variant predicted to cause a premature stop codon in biologically-relevant exon 3/27 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant is absent from gnomAD v4.1.0 (PM2_Supporting). In summary, this variant meets the criteria to be classified as Likely Pathogenic for DICER1-related tumor predisposition based on the ACMG/AMP criteria applied, as specified by the ClinGen DICER1 VCEP: PVS1, PM2_supporting. (Bayesian Points: 9; VCEP specifications version 1.3.0; 08/27/2024)

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PVS1			Predicted to undergo NMD: Stop codon 5' of p.Pro1850; UniProt: Helicase ATP-binding domain from p.51-227; This variant in exon 3. Start codon in exon 2. Second methionine is at p.11. The NM_177438.2:c.238G>T (p.Glu80Ter) variant in DICER1 is a nonsense variant predicted to cause a premature stop codon in biologically-relevant exon 3/27 leading to nonsense-mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).

Not Met criteria codes

PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			This evidence code is not currently applicable for DICER1 VCEP curations.
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			This evidence code is not currently applicable for DICER1 VCEP curations.
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			This evidence code is not currently applicable for DICER1 VCEP curations.
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			N/A
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3			This evidence code is not currently applicable for DICER1 VCEP curations.
BP1			This evidence code is not currently applicable for DICER1 VCEP curations.
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			This evidence code is not currently applicable for DICER1 VCEP curations.
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline



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