

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

Version: 2.1

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

Met criteria codes 2

PVS1 PM2_Supporting

Not Met criteria codes 19

PM1 PM5 PM4 BA1 BS2
BS1 BS4 BS3 BP4 BP3 BP2
BP7 PS1 PS2 PS3 PS4 PP1
PP3 PP4

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 1.4
- Criteria Specification Approval History
- Criteria Specifications for this VCEP

DICER1 and miRNA-Processing Gene VCEP

The NM_177438.3: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). Although this variant has been observed in an individual with thyroid nodules undergoing genetic sequencing, to our knowledge, this variant has not been reported in individuals with phenotypes highly specific to DICER1-related tumor predisposition (PS4 not met; Internal lab contributor). This variant is absent from gnomAD v4.1.1 (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. (VCEP specifications version 1.4.0) Based on Tavtigian et al 2020 (PMID:32720330), the total points applied = 9, which equates to a classification of LP.

Met criteria codes

PVS1			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).
PM2_Supporting			This variant is absent from gnomAD v4.1.1 (PM2_Supporting).

Not Met criteria codes

PM1			Not missense
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
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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.
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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			Although this variant has been observed in an individual with thyroid nodules undergoing genetic sequencing, to our knowledge, this variant has not been reported in individuals with phenotypes highly specific to DICER1-related tumor predisposition (PS4 not met; Internal lab contributor).
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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

Curation History 

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