

Variant: NM_177438.3(DICER1):c.1089C>T (p.Phe363=)

Version: 1.0

CA10583223 [↗](#)

242030 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: c13d3d4a-1321-4983-b5b8-da45714e3656

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HGVS expressions

NM_177438.3(DICER1):c.1089C>T (p.Phe363=)

NM_177438.3:c.1089C>T

NC_000014.9:g.95124483G>A

CM000676.2:g.95124483G>A

NC_000014.8:g.95590820G>A

CM000676.1:g.95590820G>A

NC_000014.7:g.94660573G>A

NG_016311.1:g.37940C>T

ENST00000529720.2:c.1089C>T

ENST00000531162.7:c.1089C>T

ENST00000674628.2:c.1089C>T

ENST00000675540.2:c.1089C>T

ENST00000696733.1:c.1089C>T

ENST00000696734.1:c.1089C>T

ENST00000696736.1:c.1089C>T

ENST00000696737.1:c.1089C>T

ENST00000696921.1:n.2195C>T

ENST00000696922.1:n.1498C>T

ENST00000696923.1:c.1089C>T

ENST00000696924.1:c.1089C>T

ENST00000696925.1:n.1498C>T

ENST00000696927.1:n.692C>T

ENST00000696928.1:n.1286C>T

ENST00000343455.8:c.1089C>T

ENST00000393063.6:c.1089C>T

ENST00000526495.6:c.1089C>T

ENST00000532939.3:c.1089C>T

ENST00000556045.6:c.1089C>T

ENST00000674628.1:c.1089C>T

ENST00000675995.1:c.1089C>T

ENST00000343455.7:c.1089C>T

ENST00000393063.5:c.1089C>T

ENST00000526495.5:c.1089C>T

ENST00000527414.5:c.1089C>T

ENST00000541352.5:c.1089C>T

NM_001195573.1:c.1089C>T

NM_001271282.2:c.1089C>T

NM_001291628.1:c.1089C>T
NM_030621.4:c.1089C>T
NM_177438.2:c.1089C>T
NM_001271282.3:c.1089C>T
NM_001291628.2:c.1089C>T
NM_001395677.1:c.1089C>T
NM_001395678.1:c.1089C>T
NM_001395679.1:c.1089C>T
NM_001395680.1:c.1089C>T
NM_001395682.1:c.1089C>T
NM_001395683.1:c.1089C>T
NM_001395684.1:c.1089C>T
NM_001395685.1:c.1089C>T
NM_001395686.1:c.807C>T
NM_001395687.1:c.684C>T
NM_001395688.1:c.684C>T
NM_001395689.1:c.684C>T
NM_001395690.1:c.684C>T
NM_001395691.1:c.522C>T
NM_001395692.1:c.1089C>T
NM_001395693.1:c.1089C>T
NM_001395694.1:c.1089C>T
NM_001395695.1:c.1089C>T
NM_001395696.1:c.684C>T
NM_001395697.1:c.-480C>T
NM_001395698.1:c.684C>T
NM_001395699.1:c.1089C>T
NM_001395700.1:c.1089C>T
NR_172715.1:n.1303C>T
NR_172716.1:n.1434C>T
NR_172717.1:n.1601C>T
NR_172718.1:n.1601C>T
NR_172719.1:n.1434C>T
NR_172720.1:n.1434C>T

Likely Benign

Met criteria codes 2

BP7 BP4

Not Met criteria codes 11

PS1 PS2 BP2 PP1 PM6
PM2 PM1 PM5 BA1 BS1
BS4

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

Evidence submitted by expert panel

DICER1 and miRNA-Processing Gene VCEP

The NM_177438.3:c.1089C>T (p.Phe363=) variant is a synonymous (silent) variant that is not predicted to impact splicing by MaxEntScan or SpliceAI (BP4, BP7). The total allele frequency in gnomAD v4.1.0 is 0.000001368 (2/1461842 alleles) with a highest population minor allele frequency of 0.00003312 (2/60394 alleles) in a population of undisclosed ancestry (PM2_Supporting, BS1, and BA1 are not met). In summary, this variant meets the criteria to be classified as Likely Benign for DICER1-related tumor predisposition based on the ACMG/AMP criteria applied, as specified by the ClinGen DICER1 VCEP: BP4, BP7. (Bayesian Points: -2; VCEP specifications version 1.3.0; 04/23/2024)

Met criteria codes

BP7			The NM_177438.3:c.1089C>T (p.Phe363=) variant is a synonymous (silent) variant that is not predicted to impact splicing by MaxEntScan or SpliceAI (BP4, BP7).
BP4			The NM_177438.3:c.1089C>T (p.Phe363=) variant is a synonymous (silent) variant that is not predicted to impact splicing by MaxEntScan or SpliceAI (BP4, BP7).

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
BP2			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
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			The total allele frequency in gnomAD v4.1.0 is 0.000001368 (2/1461842 alleles) with a highest population minor allele frequency of 0.00003312 (2/60394 alleles) in a population of undisclosed ancestry (PM2_Supporting, BS1, and BA1 are not met).
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			The total allele frequency in gnomAD v4.1.0 is 0.000001368 (2/1461842 alleles) with a highest population minor allele frequency of 0.00003312 (2/60394 alleles) in a population of undisclosed ancestry (PM2_Supporting, BS1, and

BA1 are not met).

BS1



The total allele frequency in gnomAD v4.1.0 is 0.000001368 (2/1461842 alleles) with a highest population minor allele frequency of 0.00003312 (2/60394 alleles) in a population of undisclosed ancestry (PM2_Supporting, BS1, and BA1 are not met).

BS4



No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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