

Variant: NM_000252.3(MTM1):c.339T>C (p.Cys113=)

Version: 1.0

CA10539080 [↗](#)

285094 (ClinVar) [↗](#)

Gene: MTM1 ([HGNC:4534](#))

Condition: centronuclear myopathy ([MONDO:0018947](#))

Inheritance Mode: X-linked inheritance

UUID: 8fc253b7-3065-4ac1-a409-7f46aec90cde

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HGVS expressions

NM_000252.3:c.339T>C

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NC_000023.11:g.150614696T>C

CM000685.2:g.150614696T>C

NC_000023.10:g.149783169T>C

CM000685.1:g.149783169T>C

NC_000023.9:g.149533827T>C

NG_008199.1:g.51123T>C

ENST00000684910.1:c.136+18126T>C

ENST00000685439.1:c.-3-4342T>C

ENST00000685944.1:c.339T>C

ENST00000687215.1:c.54T>C

ENST00000687365.1:n.394T>C

ENST00000688152.1:c.339T>C

ENST00000688403.1:c.-300-24247T>C

ENST00000689314.1:c.384T>C

ENST00000689694.1:c.339T>C

ENST00000689810.1:c.339T>C

ENST00000690282.1:c.-300-24247T>C

ENST00000690351.1:c.137-4342T>C

ENST00000691232.1:c.-3-4342T>C

ENST00000691686.1:c.339T>C

ENST00000691851.1:c.339T>C

ENST00000692015.1:c.231+16010T>C

ENST00000692638.1:c.*88T>C

ENST00000692852.1:c.339T>C

ENST00000692915.1:c.*334T>C

ENST00000370396.7:c.339T>C

ENST00000306167.11:n.260-4392T>C

ENST00000370396.6:c.339T>C

ENST00000424519.1:c.231+16010T>C

ENST00000490530.1:n.278T>C

NM_000252.2:c.339T>C

NM_001376906.1:c.339T>C

NM_001376907.1:c.232-4342T>C

NM_001376908.1:c.339T>C

Benign

Met criteria codes 3

BA1

BP7

BP4

Not Met criteria codes 3

PP3

PM2

BS1

Evidence Links 0

Expert Panel

[Congenital Myopathies VCEP](#)

Criteria Specification Information

[Criteria Specification:](#) *ClinGen Congenital Myopathies Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MTM1 Version 1.0.0*

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Congenital Myopathies VCEP

The variant NM_000252.3:c.339T>C in MTM1 is a synonymous (silent) variant (p.Cys113=). The highest population filtering allele frequency in gnomAD v4.1.0 is 0.003242 (198/54107 alleles, 58 hemizygotes) in African/African American population, which is higher than the ClinGen Congenital Myopathies VCEP threshold (≥ 0.000016) for BA1, and therefore meets this criterion (BA1). In addition, SpliceAI does not predict impact to splicing, and the variant occurs at a nucleotide that is not conserved as shown by UCSC Genome Browser (BP4, BP7). In summary, this variant meets the criteria to be classified as benign for X-linked centronuclear myopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen Congenital Myopathies VCEP: BA1, BP4, BP7. (Congenital Myopathies VCEP specifications version 1; 8/7/2024)

Met criteria codes

BA1			The highest population filtering allele frequency in gnomAD v4.1.0 is 0.003242 (198/54107 alleles, 58 hemizygotes) in African/African American population, which is higher than the ClinGen Congenital Myopathies VCEP threshold (≥ 0.000016) for BA1, and therefore meets this criterion
BP7			The c.339T>C (p.Cys113=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by UCSC Genome Browser
BP4			The c.339T>C (p.Cys113=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by UCSC Genome Browser

Not Met criteria codes

PP3			The c.339T>C (p.Cys113=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by UCSC Genome Browser
PM2			The highest population filtering allele frequency in gnomAD v4.1.0 is 0.003242 (198/54107 alleles, 58 hemizygotes) in African/African American population
BS1			The highest population filtering allele frequency in gnomAD v4.1.0 is 0.003242 (198/54107 alleles, 58 hemizygotes) in African/African American population

Curation History 

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See Report	Preferred Variant Title	Classification ⓘ	Condition	Published Date	Version ⓘ	Criteria Specification	Gene
View	NM_000252.3(MTM1):c.339T>C (p.Cys...	Benign	Centronuclear Myopathy ↗	2024-10-01	1.0	ClinGen Congenital Myopathies Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MTM1 Version 1.0.0 ↗	MTM1 ↗

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