

Variant: NM_000314.7(PTEN):c.633C>G (p.Cys211Trp)

Version: 1.1

CA10603125 [↗](#)

279878 (ClinVar) [↗](#)

Gene: PTEN ([HGNC:5728](#))

Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: d2e370c6-311c-4c89-a383-c956681b1dfc

Approved on: 2025/10/3

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HGVS expressions

NM_000314.7:c.633C>G

NM_000314.7(PTEN):c.633C>G (p.Cys211Trp)

NC_000010.11:g.87952258C>G

CM000672.2:g.87952258C>G

NC_000010.10:g.89712015C>G

CM000672.1:g.89712015C>G

NC_000010.9:g.89701995C>G

NG_007466.2:g.93820C>G

ENST00000700029.2:c.633C>G

ENST00000710265.1:c.633C>G

ENST00000472832.3:c.633C>G

ENST00000688158.2:n.1368C>G

ENST00000688922.2:c.*463C>G

ENST00000700021.1:c.588C>G

ENST00000700022.1:c.493-5595C>G

ENST00000700023.1:n.1791C>G

ENST00000700024.1:n.2025C>G

ENST00000700025.1:n.1402C>G

ENST00000700029.1:c.467C>G

ENST00000706954.1:c.633C>G

ENST00000706955.1:c.*668C>G

ENST00000686459.1:c.*219C>G

ENST00000688158.1:c.*744C>G

ENST00000688308.1:c.633C>G

ENST00000688922.1:c.554C>G

ENST00000693560.1:c.1152C>G

ENST00000371953.8:c.633C>G

ENST00000371953.7:c.633C>G

ENST00000472832.2:c.60C>G

NM_000314.5:c.633C>G

NM_000314.6:c.633C>G

NM_001304717.2:c.1152C>G

NM_001304718.1:c.42C>G

NM_001304717.5:c.1152C>G

NM_001304718.2:c.42C>G

NM_000314.8:c.633C>G

Pathogenic

Met criteria codes 5

PP2PP3PS2_Very StrongPS4_ModeratePM2_Supporting

Not Met criteria codes 3

PM6PM1PS3

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specification: ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.1.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP			
PTEN c.633C>G (p.Cys211Trp) meets criteria to be classified as pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (PMID 30311380). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PS2_VS: Three pediatric patients (Internal lab contribution) confirmed de novo with WES trio. PP3: REVEL score >.7 (Score: 0.856). PS4_P: Vanderver et al (PMID 24375884) patient #21 with macrocephaly, developmental delay, and enlarged periventricular spaces. Phenotype score of +6, +1 proband point. Confirmed de novo pediatric patient with extreme macrocephaly (+4 SD), developmental delay, hemihypertrophy, autism and ID (Internal laboratory contribution). Phenotype score +5, +1 proband point. PM2_P: Absent in large sequenced populations (gnomad v4). PP2: PTEN is defined by the PTEN Expert Panel as a gene that has a low rate of benign missense variation and where missense variants are a common mechanism of disease.			
Met criteria codes			
PP2			PTEN gene has a low rate of benign missense variation and where missense variants are a common mechanism of disease.
PP3			REVEL score >.7 (Score: 0.856).
PS2_Very Strong			Three pediatric patients (Internal lab contribution) confirmed de novo with WES trio.
PS4_Moderate			Vanderver et al (PMID 24375884) patient #21 with macrocephaly, developmental delay, and enlarged periventricular spaces. Phenotype score of +6, +1 proband point. Confirmed de novo pediatric patient with extreme macrocephaly (+4 SD), developmental delay, hemihypertrophy, autism and ID (Internal laboratory contribution)
PM2_Supporting			Absent from gnomAD
Not Met criteria codes			
PM6			Internal lab contribution: one patient assumend de novo with macrocephaly and congenital heart defects. Pediatric phenotype score not high enough to receive weight.

PM1			not in critical domain
PS3			Cumulative functional score of -1.002 in Miguell et al. 2018 (PMID: 29706350), indicating WT-like phosphatase activity.

Curation History 

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