

Variant: NM_000314.6(PTEN):c.1104T>C (p.Asp368=)

CA000293 [↗](#)

138835 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 044a4a6a-4939-4e73-8b9d-ff941b58e27d

Approved on: 2016-09-14

Published on: 2018-12-10

HGVS expressions

NM_000314.6(PTEN):c.1104T>C (p.Asp368=)

NM_000314.6:c.1104T>C

NM_000314.5:c.1104T>C

NM_001304717.2:c.1623T>C

NM_001304718.1:c.513T>C

NM_000314.7:c.1104T>C

NM_001304717.5:c.1623T>C

NM_001304718.2:c.513T>C

ENST00000371953.7:c.1104T>C

NC_000010.11:g.87965364T>C

CM000672.2:g.87965364T>C

NC_000010.10:g.89725121T>C

CM000672.1:g.89725121T>C

NC_000010.9:g.89715101T>C

NG_007466.2:g.106926T>C

Likely Benign

Met criteria codes **3**

BS1

BP7

BP4

Evidence Links **0**

Expert Panel

PTEN VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

PTEN VCEP

PTEN c.1104T>C (p.D368=) meets criteria to be classified as likely benign for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (Mester et al. 2018; manuscript in preparation). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). BS1: Allele frequency of 0.0057 (0.57%, 130/22,966 alleles) in the African subpopulation of the gnomAD cohort. (PMID 27535533) BP4: Synonymous variant where at least 2 out of 3 in silico models predict no splicing impact. BP7: Variant is synonymous (silent), nucleotide is not conserved, and no splicing impact is predicted.

Met criteria codes

BS1			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
BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



Showing 1 to 1 of 1 rows

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