

Variant: *NM_000314.8(PTEN):c.64G>C (p.Asp22His)*

Version: 1.0

CA10578904 [↗](#)

233456 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: f16fe142-883f-4de7-9f82-815a073aa806

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HGVS expressions

NM_000314.8(PTEN):c.64G>C (p.Asp22His)

NM_000314.8:c.64G>C

NC_000010.11:g.87864533G>C

CM000672.2:g.87864533G>C

NC_000010.10:g.89624290G>C

CM000672.1:g.89624290G>C

NC_000010.9:g.89614270G>C

NG_007466.2:g.6095G>C

NG_033079.1:g.3905C>G

ENST00000700029.2:c.64G>C

ENST00000710265.1:c.64G>C

ENST00000472832.3:c.64G>C

ENST00000688922.2:c.64G>C

ENST00000700021.1:c.64G>C

ENST00000700022.1:c.64G>C

ENST00000706954.1:c.64G>C

ENST00000706955.1:c.64G>C

ENST00000686459.1:c.64G>C

ENST00000688158.1:c.64G>C

ENST00000688308.1:c.64G>C

ENST00000693560.1:c.583G>C

ENST00000371953.8:c.64G>C

ENST00000371953.7:c.64G>C

ENST00000462694.1:n.66G>C

ENST00000487939.1:n.85G>C

ENST00000610634.1:c.-39G>C

ENST00000618586.1:n.33G>C

NM_000314.5:c.64G>C

NM_000314.6:c.64G>C

NM_001304717.2:c.583G>C

NM_001304718.1:c.-642G>C

NM_000314.7:c.64G>C

NM_001304717.5:c.583G>C

NM_001304718.2:c.-642G>C

Likely Pathogenic

Met criteria codes 4

PP2PP3PM6_StrongPM2_Supporting

Not Met criteria codes 3

PS1PS3PM5

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 3.0.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel			
<div><div>PTEN VCEP</div><div>PTEN c.64G>C (p.Asp22His) meets criteria to be classified as likely 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). PM6_Strong: De novo (parentage not confirmed) in two patients with the disease and no family history (Internal laboratory contributor(s) SCV000577329.3). PM2_Supporting: Absent in large sequenced populations (PMID 27535533). 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. PP3: REVEL score = 0.965 (>0.7).</div></div>			
Met criteria codes			
PP2	<div><div></div><div></div></div>	<div><div></div><div></div></div>	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.
PP3	<div><div></div><div></div></div>	<div><div></div><div></div></div>	REVEL score = 0.965 (> 0.75)
PM6_Strong	<div><div></div><div></div></div>	<div><div></div><div></div></div>	GeneDx: Two cases, both proven de novo via targeted testing only (parentage not confirmed). Both children with macrocephaly NOS, autism, and developmental delays.
PM2_Supporting	<div><div></div><div></div></div>	<div><div></div><div></div></div>	absent in gnomAD
Not Met criteria codes			
PS1	<div><div></div><div></div></div>	<div><div></div><div></div></div>	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	<div><div></div><div></div></div>	<div><div></div><div></div></div>	Mighell et al (2018): High confidence variant, cum_score = -0.8245 (> -1.11)
PM5	<div><div></div><div></div></div>	<div><div></div><div></div></div>	3 close matches in ClinVar p.Asp22Tyr: N/A (BLOSUM = -3) p.Asp22Gly: VUS - Ambry (BLOSUM = -1) p.Asp22Glu: VUS - 3billion (BLOSUM = 2)

Curation History 		
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