

Variant: *NM_000314.8(PTEN):c.131G>A (p.Gly44Asp)*

Version: 1.0

CA377784538 [↗](#)

427582 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 0cca48d0-e46c-4b58-b93d-f7a1f6a45afa

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HGVS expressions

NM_000314.8(PTEN):c.131G>A (p.Gly44Asp)

NM_000314.8:c.131G>A

NC_000010.11:g.87894076G>A

CM000672.2:g.87894076G>A

NC_000010.10:g.89653833G>A

CM000672.1:g.89653833G>A

NC_000010.9:g.89643813G>A

NG_007466.2:g.35638G>A

ENST00000700029.2:c.131G>A

ENST00000710265.1:c.131G>A

ENST00000472832.3:c.131G>A

ENST00000688158.2:n.899+13638G>A

ENST00000688922.2:c.131G>A

ENST00000700021.1:c.131G>A

ENST00000700022.1:c.131G>A

ENST00000706954.1:c.131G>A

ENST00000706955.1:c.*166G>A

ENST00000686459.1:c.131G>A

ENST00000688158.1:c.*275+13638G>A

ENST00000688308.1:c.131G>A

ENST00000693560.1:c.650G>A

ENST00000371953.8:c.131G>A

ENST00000371953.7:c.131G>A

ENST00000462694.1:n.133G>A

ENST00000610634.1:c.29G>A

NM_000314.5:c.131G>A

NM_000314.6:c.131G>A

NM_001304717.2:c.650G>A

NM_001304718.1:c.-575G>A

NM_000314.7:c.131G>A

NM_001304717.5:c.650G>A

NM_001304718.2:c.-575G>A

Pathogenic

Met criteria codes 7

PS3_Supporting

PS4_Moderate

PP2

PP3

PM2_Supporting

PM6

PS2

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 3.2.0

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

PTEN VCEP			
<p>PTEN c.131G>A (p.Gly44Asp) meets criteria to be classified as Pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.1.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PS2: De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (PMID 28475857). PM6: Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. (internal laboratory contributor(s)). PS4_M: Probands with phenotype specificity score of 2-3.5. (PMID 19265751, 24375884, 28526761). 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 > 0.7 (score of this variant =0.96). PM2_Supporting: Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort. (PMID 27535533). PS3_Supporting: Abnormal in vitro cellular assay or transgenic model with phenotype different from wild type that does not meet PS3. (PMID 21828076).</p>			
Met criteria codes			
PS3_Supporting			Abnormal in vitro cellular assay or transgenic model with phenotype different from wild type that does not meet PS3. (PMID 21828076)
PS4_Moderate			Case 1 (PMID: 19265751): Two year old female who presented with developmental delay, oral motor and speech delay, and macrocephaly (OFC of 54.5 cm (+5.0 S.D.)). History of bilateral hernia repair at one month of age, mild GE reflux, and benign macrocrania diagnosed at age 2 after having a normal CT scan. Large café au lait spot on her left lower chest and upper abdomen. +5 Pediatric phenotype score, +1 proband point Case 2 (PMID: 24375884): 10 m old M, macrocephaly (>> 2SD), mild motor delay, Physical exam: mild frontal bossing, MRI: Posterior periventricular multifocal white matter abnormalities, Enlarged perivascular spaces with CSF isointense signal. +6 pediatric phenotype points, +1 proband point. Case 3 (PMID: 28526761): Pediatric female patient presented with macrocephaly (+7SD), ASD, developmental delay, cafe au lait spot, and MRI finding of heterotopia. +7 pediatric phenptype points, +1 proband point.
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 > 0.7 (score of this variant =0.96)
PM2_Supporting			Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort. (PMID 27535533).

PM6			One proband with presumed de novo occurrence (maternity/paternity not confirmed) for a patient with highly specific phenotype. (internal laboratory contributor(s))
PS2			At least two proven OR one proven plus two assumed de novo observations in a patient with the disease and no family history. (PMID 28475857)

Curation History 		
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