

Variant: NM_000314.7(PTEN):c.1A>G (p.Met1Val)

Version: 1.0

CA377781751 [↗](#)

484600 (ClinVar) [↗](#)

Gene: PTEN ([HGNC:5728](#))
Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))
Inheritance Mode: Autosomal dominant inheritance
UUID: b5f8e9e7-3f37-42d6-9844-c91a9757b411
Approved on: 2019/11/22
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HGVS expressions

NM_000314.7(PTEN):c.1A>G (p.Met1Val)
NM_000314.7:c.1A>G
NC_000010.11:g.87864470A>G
CM000672.2:g.87864470A>G
NC_000010.10:g.89624227A>G
CM000672.1:g.89624227A>G
NC_000010.9:g.89614207A>G
NG_007466.2:g.6032A>G
NG_033079.1:g.3968T>C
ENST00000700029.2:c.1A>G
ENST00000710265.1:c.1A>G
ENST00000472832.3:c.1A>G
ENST00000688922.2:c.1A>G
ENST00000700021.1:c.1A>G
ENST00000700022.1:c.1A>G
ENST00000706954.1:c.1A>G
ENST00000706955.1:c.1A>G
ENST00000686459.1:c.1A>G
ENST00000688158.1:c.1A>G
ENST00000688308.1:c.1A>G
ENST00000693560.1:c.520A>G
ENST00000371953.8:c.1A>G
ENST00000371953.7:c.1A>G
ENST00000462694.1:n.3A>G
ENST00000487939.1:n.22A>G
ENST00000610634.1:c.-102A>G
NM_000314.5:c.1A>G
NM_000314.6:c.1A>G
NM_001304717.2:c.520A>G
NM_001304718.1:c.-705A>G
NM_001304717.5:c.520A>G
NM_001304718.2:c.-705A>G
NM_000314.8:c.1A>G

Pathogenic

PS4_Moderate

PM6

PM2

PVS1

PTEN VCEP

Not Met criteria codes 22

PS1

PS2

PS3

BP5

BP7

BP4

BP3

BP1

BP2

PP1

PP2

PP3

PP4

BA1

PM1

PM3

PM5

PM4

BS1

BS4

BS3

BS2

Evidence Links 2

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel			
PTEN VCEP			
PTEN c.1A>G (p.M1?) 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). PVS1: Null variant predicted to result in nonsense-mediated decay or causing truncation/frameshift at or 5’ to c.1121 (NM_000314.4). PM2: Absent in large sequenced populations (PMID 27535533). PM6: Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. (PMID 29752200) PS4_M: Probands with phenotype specificity score of 2-3.5. (PMID 29752200, 28774669; internal laboratory contributor ClinVar Organization ID: 26957)			
Met criteria codes			
PS4_Moderate			3 phenotype specificity points (see PP4 section).
PM6			Identified in a M Japanese patient (patient 6) as de novo, HC 60 @ 5yrs (+6 SD), sever DD, autism, distinctive facial features. Parents were healthy, but father had Graves' disease. They did not mention how the alteration was confirmed as de novo. CC score of 5 (Kato 2018 PubMed: 29752200)
PubMed:29752200			
PM2			Not present in gnomAD, coverage >90X.
PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
Not Met criteria codes			
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			PubMed:29752200
PS3			

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

BP5			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
BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			Missense
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			Rolled into PS4. Pre-teen M with autism, macrocephaly +5SD (1 point). CC score of 5. (1 point) (GDx). PubMed:28774669 
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

