The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000314.8(PTEN):c.801G>T (p.Lys267Asn)

Version: 1.0

CA377485115

536549 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 5c1f44d0-ab30-4090-8d98-dfaf2d2fd444
Approved on: 2025/12/5
Published on: 2025/12/17

HGVS expressions

NM_000314.8:c.801G>T
NM_000314.8(PTEN):c.801G>T (p.Lys267Asn)
NC_000010.11:g.87958019G>T
CM000672.2:g.87958019G>T
NC_000010.10:g.89717776G>T
CM000672.1:g.89717776G>T
NC_000010.9:g.89707756G>T
NG_007466.2:g.99581G>T
ENST00000700029.2:c.801G>T
ENST00000710265.1:c.801G>T
ENST00000472832.3:c.801G>T
ENST00000688158.2:n.1536G>T
ENST00000688922.2:c.*631G>T
ENST00000700021.1:c.756G>T
ENST00000700022.1:c.*140G>T
ENST00000700023.1:n.1959G>T
ENST00000700024.1:n.2193G>T
ENST00000700025.1:n.1570G>T
ENST00000700026.1:n.438G>T
ENST00000700029.1:c.635G>T
ENST00000706954.1:c.801G>T
ENST00000706955.1:c.*836G>T
ENST00000686459.1:c.*387G>T
ENST00000688158.1:c.*912G>T
ENST00000688308.1:c.801G>T
ENST00000688922.1:c.722G>T
ENST00000693560.1:c.1320G>T
ENST00000371953.8:c.801G>T
ENST00000371953.7:c.801G>T
ENST00000472832.2:c.228G>T
NM_000314.5:c.801G>T
NM_000314.6:c.801G>T
NM_001304717.2:c.1320G>T
NM_001304718.1:c.210G>T
NM_000314.7:c.801G>T
NM_001304717.5:c.1320G>T
NM_001304718.2:c.210G>T

Pathogenic

Met criteria codes 3

PM2 PS3 PM6_Strong

Not Met criteria codes 2

PP3 PS4

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.801G>T (p.Lys267Asn) 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). PS3: RNA, mini-gene, or other assay shows impact on splicing. (internal laboratory contributor(s)) PM6_S: One proband with presumed de novo occurrence (maternity/paternity not confirmed) for a patient with highly specific phenotype. (PMID: 30528446) PM2_Supporting: Absent in large sequenced populations (PMID 27535533).

Met criteria codes

PM2	✔	Variant absent from gnomADv4
PS3	✔	RNA data shared by Ambry Genetics shows exon 7 skipping, r.635_801del167 (p.N212RFS*30) Allele specific expression is consistent with complete splice impact
PM6_Strong	✔	6mo female with HC +2.9SD, with facial dysmorphism, DD/ID, vascular malformations and abnormal findings on MRI including enlarged perivascular spaces and short and thin corpus callosum.

Not Met criteria codes

PP3	✖	REVEL score 0.62 – PP3 not met
PS4	✖	2 patients shared by Gene DX do not quite meet weight

Curation History

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