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_000314.6(PTEN):c.534_535delTAinsAT (p.Tyr178Ter)

CA000509

7824 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: ebef4811-dc36-4625-bd07-ea516d1cad7e
Approved on: 2018-04-06
Published on: 2018-12-10

HGVS expressions

NM_000314.6:c.534_535delTAinsAT
NM_000314.6(PTEN):c.534_535delTAinsAT (p.Tyr178Ter)
NC_000010.11:g.87952159_87952160delinsAT
CM000672.2:g.87952159_87952160delinsAT
NC_000010.10:g.89711916_89711917delinsAT
CM000672.1:g.89711916_89711917delinsAT
NC_000010.9:g.89701896_89701897delinsAT
NG_007466.2:g.93721_93722delinsAT
NM_000314.5:c.534_535delinsAT
NM_000314.6:c.534_535delinsAT
NM_001304717.2:c.1053_1054delinsAT
NM_001304718.1:c.-58_-57delinsAT
NM_000314.7:c.534_535delinsAT
NM_001304717.5:c.1053_1054delinsAT
NM_001304718.2:c.-58_-57delinsAT
ENST00000371953.7:c.534_535delinsAT

Pathogenic

Met criteria codes 3

PM2 PVS1 PS4_Supporting

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.534_535delTAinsAT (p.Y178X) 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). PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (PMID 9832032, PMID 23335809)

Met criteria codes

PM2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4_Supporting	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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