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.801+1delG

CA16613026

404140 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: d8470abc-cd84-4dd7-aa9d-def2c9c878aa
Approved on: 2017-10-18
Published on: 2018-12-10

HGVS expressions

NM_000314.6(PTEN):c.801+1delG
NM_000314.6:c.801+1delG
NC_000010.11:g.87958020del
CM000672.2:g.87958020del
NC_000010.10:g.89717777del
CM000672.1:g.89717777del
NC_000010.9:g.89707757del
NG_007466.2:g.99582del
NM_000314.5:c.801+1del
NM_000314.6:c.801+1del
NM_001304717.2:c.1320+1del
NM_001304718.1:c.210+1del
NM_000314.7:c.801+1del
NM_001304717.5:c.1320+1del
NM_001304718.2:c.210+1del
NM_000314.8:c.801+1del
ENST00000371953.7:c.801+1del
ENST00000472832.2:n.228+1del

Pathogenic

Met criteria codes 3

PVS1 PM2 PS4_Supporting

Not Met criteria codes 19

PM6 PM4 PM1 PM5 BA1 BS2 BS1 BS3 BS4 BP4 BP2 BP5 BP7 PS3 PS1 PS2 PP2 PP3 PP1

Evidence Links 1

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.801+1delG (IVS7+1delG) 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 PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (PMID 23335809)

Met criteria codes

PVS1	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4_Supporting	✔	Identified in patient #122, CC score = 34, 1 phenotype specificity point. Mucocutaneous lesions, facial papules, skin oral mucosal papillomatosis, acral keratoses, macrocephaly, GI polyps, lipomas/vascular lesions, GU lesions, benign thyroid disease, benign breast disease, malignant breast, melanoma. Personal communication from Dr. Longy: Cranial perimeter at 43 years measured 61 cm. Notion of glycogenic acanthosis of the esophagus, glandulocystic polyps of the stomach and hamartomatous polyps of the colon. PubMed:23335809

Not Met criteria codes

PM6	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4	✖	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
PM5	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS3		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4		✖	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
BP2		✖	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
PS3		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3		✖	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

Curation History

Showing 1 to 1 of 1 rows

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