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_000314.7(PTEN):c.610C>G (p.Pro204Ala)

Version: 2.0

CA000535

189415 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: b5627c7e-eb93-4a97-95aa-58fd645e077c
Approved on: 2023/10/11
Published on: 2023/10/18

HGVS expressions

NM_000314.7:c.610C>G
NM_000314.7(PTEN):c.610C>G (p.Pro204Ala)
NC_000010.11:g.87952235C>G
CM000672.2:g.87952235C>G
NC_000010.10:g.89711992C>G
CM000672.1:g.89711992C>G
NC_000010.9:g.89701972C>G
NG_007466.2:g.93797C>G
ENST00000700029.2:c.610C>G
ENST00000710265.1:c.610C>G
ENST00000472832.3:c.610C>G
ENST00000688158.2:n.1345C>G
ENST00000688922.2:c.*440C>G
ENST00000700021.1:c.565C>G
ENST00000700022.1:c.493-5618C>G
ENST00000700023.1:n.1768C>G
ENST00000700024.1:n.2002C>G
ENST00000700025.1:n.1379C>G
ENST00000700029.1:c.444C>G
ENST00000706954.1:c.610C>G
ENST00000706955.1:c.*645C>G
ENST00000686459.1:c.*196C>G
ENST00000688158.1:c.*721C>G
ENST00000688308.1:c.610C>G
ENST00000688922.1:c.531C>G
ENST00000693560.1:c.1129C>G
ENST00000371953.8:c.610C>G
ENST00000371953.7:c.610C>G
ENST00000472832.2:c.37C>G
NM_000314.5:c.610C>G
NM_000314.6:c.610C>G
NM_001304717.2:c.1129C>G
NM_001304718.1:c.19C>G
NM_001304717.5:c.1129C>G
NM_001304718.2:c.19C>G
NM_000314.8:c.610C>G

Pathogenic

Met criteria codes 6

PM6_Strong | PS3_Moderate | PP2 | PP3 | PM2_Supporting | PS4_Supporting

Not Met criteria codes 20

PVS1 | PM1 | PM3 | PM5 | PM4 | BA1 | BS2 | BS1 | BS4 | BS3 | BP4 | BP3 | BP1 | BP2 | BP5 | BP7 | PS1 | PS2 | PP1 | PP4

Evidence Links 2

Expert Panel

PTEN VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.0.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

PTEN VCEP

PTEN c.610C>G (p.Pro204Ala) 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 3.0.0). Please see a summary of the rules and criteria codes in the "PTEN ACMG Specifications Summary" document (assertion method column). PM6_S: One proband with presumed de novo occurrence (maternity/paternity not confirmed) for a patient with highly specific phenotype. (internal laboratory contributor SCV000222125.7) PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -1.675 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (internal laboratory contributor SCV000222125.7) PM2_P: Absent in large sequenced populations (PMID 27535533). 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 = 0.881)

Met criteria codes

PM6_Strong	✔	GDx #1 assumed de novo; patient with high phenotype specificity (meets PS4_P), data combined to upweight here.
PS3_Moderate	✔	Mighell score hypomorphic Protein abundance score 0.52, "possibly low" range. PubMed:29785012 Score -1.675 in hypomorphic range. PubMed:29706350
PP2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3	✔	Score = 0.881
PM2_Supporting	✔	Absent gnomAD
PS4_Supporting	✔	GDx #1: F child extreme macro and autism, peds score = 5, 1 phenotype point. GDx #2: M child macro NOS and DD; mother pos macro 59cm and thy nod CC score 10. Assumed de novo, PS4_P wrapped into PM6_S upweight. GDX #2:

male child with extreme macroceph (+5.5SD), delays. Inherited from HET mother with macrocephaly, thyroid nodules. Meets PS4_P, 1 meiosis towards PP1.

Not Met criteria codes

PVS1	✖	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
PM4	✖	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
BS4	✖	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
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
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
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
PP1		✖	1 meiosis from GDx #2, described in PS4_P
PP4		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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