

Variant: NM\_000314.7(PTEN):c.112C>T (p.Pro38Ser)

Version: 1.1

CA000297 [↗](#)

127688 (ClinVar) [↗](#)

**Gene:** PTEN ([HGNC:5728](#))

**Condition:** PTEN hamartoma tumor syndrome ([MONDO:0017623](#))

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** c18b6eab-2415-46f1-9018-781158bc9857

**Approved on:** 2024/2/9

**Published on:** 2024/3/1

### HGVS expressions

**NM\_000314.7(PTEN):c.112C>T (p.Pro38Ser)**

**NM\_000314.7:c.112C>T**

NC\_000010.11:g.87894057C>T

CM000672.2:g.87894057C>T

NC\_000010.10:g.89653814C>T

CM000672.1:g.89653814C>T

NC\_000010.9:g.89643794C>T

NG\_007466.2:g.35619C>T

ENST00000700029.2:c.112C>T

ENST00000710265.1:c.112C>T

ENST00000472832.3:c.112C>T

ENST00000688158.2:n.899+13619C>T

ENST00000688922.2:c.112C>T

ENST00000700021.1:c.112C>T

ENST00000700022.1:c.112C>T

ENST00000706954.1:c.112C>T

ENST00000706955.1:c.\*147C>T

ENST00000686459.1:c.112C>T

ENST00000688158.1:c.\*275+13619C>T

ENST00000688308.1:c.112C>T

ENST00000693560.1:c.631C>T

ENST00000371953.8:c.112C>T

ENST00000371953.7:c.112C>T

ENST00000462694.1:n.114C>T

ENST00000610634.1:c.10C>T

NM\_000314.5:c.112C>T

NM\_000314.6:c.112C>T

NM\_001304717.2:c.631C>T

NM\_001304718.1:c.-594C>T

NM\_001304717.5:c.631C>T

NM\_001304718.2:c.-594C>T

NM\_000314.8:c.112C>T



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

|     |                                                                                     |                                                                                     |                                                                                                                            |
|-----|-------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------|
| PS4 |    |    | GDx #1: adult F CC score = 14; GDx #2: adult F CC score = 1                                                                |
| 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 |
| PP4 |                                                                                     |  | No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline |
| PM6 |  |  | 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 |
| 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

|      |                                                                                   |                                                                                   |                                                                                                                            |
|------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------|
| 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 |
| BS2  |  |  | 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 |

Curation History 



Showing 1 to 2 of 2 rows

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