

Variant: *NM_005249.5(FOXG1):c.201G>T (p.Pro67=)*

Version: 1.0

CA290947 [↗](#)

137388 (ClinVar) [↗](#)

Gene: FOXG1 ([HGNC:2290](#))

Condition: FOXG1 disorder ([MONDO:0100040](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 87862518-0606-414b-8d0c-81b9cd260088

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HGVS expressions

NM_005249.5(FOXG1):c.201G>T (p.Pro67=)

NM_005249.5:c.201G>T

NC_000014.9:g.28767480G>T

CM000676.2:g.28767480G>T

NC_000014.8:g.29236686G>T

CM000676.1:g.29236686G>T

NC_000014.7:g.28306437G>T

NG_009367.1:g.5400G>T

ENST00000706482.1:c.201G>T

ENST00000313071.7:c.201G>T

ENST00000313071.6:c.201G>T

NM_005249.4:c.201G>T

Benign

Met criteria codes **2**

BP7 **BA1**

Evidence Links **0**

Expert Panel

[Rett and Angelman-like Disorders VCEP](#) [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The allele frequency of the p.Pro67= variant in FOXG1 is 0.055% in European (Non-Finnish) sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BA1). The silent p.Pro67= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Pro67= variant in FOXG1 is classified as benign based on the ACMG/AMP criteria (BA1, BP7).

Met criteria codes

BP7



The silent p.Pro67= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide

BA1



The allele frequency of the p.Pro67= variant in FOXG1 is 0.055% in European (Non-Finnish) sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions.

Curation History [↗](#)

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Showing 1 to 1 of 1 rows

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