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_005249.5(FOXG1):c.95A>G (p.Asn32Ser)

Version: 1.0

CA389474374

1144732 (ClinVar)

Gene: FOXG1 (HGNC:2290)
Condition: FOXG1 disorder (MONDO:0100040)
Inheritance Mode: Autosomal dominant inheritance
UUID: 342de681-0348-4ea5-9a5f-661c03750bd2
Approved on: 2025-06-25
Published on: 2025-06-30

HGVS expressions

NM_005249.5:c.95A>G
NM_005249.5(FOXG1):c.95A>G (p.Asn32Ser)
NC_000014.9:g.28767374A>G
CM000676.2:g.28767374A>G
NC_000014.8:g.29236580A>G
CM000676.1:g.29236580A>G
NC_000014.7:g.28306331A>G
NG_009367.1:g.5294A>G
ENST00000706482.1:c.95A>G
ENST00000313071.7:c.95A>G
ENST00000313071.6:c.95A>G
NM_005249.4:c.95A>G

Uncertain Significance

Met criteria codes 4

BS2_Supporting | BP5 | BP4 | PM2_Supporting

Evidence Links 0

Expert Panel

Rett and Angelman-like Disorders VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for FOXG1 Version 4.1.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Asn32Ser variant in FOXG1 is absent from gnomAD v4.1 (PM2_Supporting). The p.Asn32Ser variant is observed in at least 1 unaffected individual (internal database - Labcorp Genetics) (BS2_Supporting). The p.Asn32Ser variant is found in a patient with an alternate molecular basis of disease (internal database - Labcorp Genetics) (BP5). The computational predictor REVEL gives a score of 0.083, which is below the threshold of 0.290, evidence that does not predict a damaging effect on FOXG1 function (BP4). In summary, the p.Asn32Ser variant in FOXG1 is classified as a variant of unknown significance based on the ACMG/AMP criteria (PM2_Supporting, BS2_Supporting, BP5, BP4). (FOXG1 Specifications v.4.1; curation approved on [06/25/2025])

Met criteria codes

BS2_Supporting	✔	The p.Asn32Ser variant is observed in at least 1 unaffected individual (internal database - Labcorp Genetics) (BS2_Supporting).
BP5	✔	The p.Asn32Ser variant is found in a patient with an alternate molecular basis of disease (internal database - Labcorp Genetics) (BP5).
BP4	✔	The computational predictor REVEL gives a score of 0.083, which is below the threshold of 0.290, evidence that does not predict a damaging effect on FOXG1 function (BP4).
PM2_Supporting	✔	The p.Asn32Ser variant in FOXG1 is absent from gnomAD v4.1 (PM2_Supporting).

Curation History

Showing 1 to 1 of 1 rows