Variant: NM_005249.5(FOXG1):c.460GAG[1] (p.Glu155del)

Version: 1.0

CA613324873

547389 (ClinVar)

Gene: FOXG1
Condition: FOXG1 disorder (MONDO:0100040)
Inheritance Mode: Autosomal dominant inheritance
UUID: 8b7bfbae-4431-47f3-b6fd-5840cdd87f0c
Approved on: 2025-06-25
Published on: 2025-06-30

HGVS expressions

NM_005249.5:c.460GAG[1]
NM_005249.5(FOXG1):c.460GAG[1] (p.Glu155del)
NC_000014.9:g.28767742_28767744del
CM000676.2:g.28767742_28767744del
NC_000014.8:g.29236948_29236950del
CM000676.1:g.29236948_29236950del
NC_000014.7:g.28306699_28306701del
NG_009367.1:g.5662_5664del
ENST00000706482.1:c.463_465del
ENST00000313071.7:c.463_465del
ENST00000313071.6:c.463_465del
NM_005249.4:c.463_465del
NM_005249.5:c.463_465del

Benign

Met criteria codes 1

BA1

Not Met criteria codes 2

PM4 BP3

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 highest population minor allele frequency of the p.Glu155del variant in FOXG1 in gnomAD v4.1 is 0.0003552 in the African/African-American population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥0.0003) for BA1, and therefore meets this criterion (BA1). In summary, the p.Glu155del variant in FOXG1 is classified as Benign based on the ACMG/AMP criteria (BA1). (FOXG1 Specifications v.4.1; curation approved on [06/25/2025])

Met criteria codes

BA1	✔	The highest population minor allele frequency of the p.Glu155del variant in FOXG1 in gnomAD v4.1 is 0.0003552 in the African/African-American population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥0.0003) for BA1, and therefore meets this criterion (BA1).

Not Met criteria codes

PM4	✖	Did not assign since this variant occurs in a LCR.
BP3	✖	Located in LCR, but only 2 Glu repeats.

Curation History

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