

## Variant: NM\_005249.5(FOXG1):c.430G>A (p.Glu144Lys)

Version: 1.0

CA206112 [↗](#)

211032 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UID:** 927bf4da-dc44-4398-9fdb-e3384aff5a5a

**Approved on:** 2025-08-27

**Published on:** 2025-10-01

### HGVS expressions

**NM\_005249.5:c.430G>A**

NM\_005249.5(FOXG1):c.430G>A (p.Glu144Lys)

NC\_000014.9:g.28767709G>A

CM000676.2:g.28767709G>A

NC\_000014.8:g.29236915G>A

CM000676.1:g.29236915G>A

NC\_000014.7:g.28306666G>A

NG\_009367.1:g.5629G>A

ENST00000706482.1:c.430G>A

ENST00000313071.7:c.430G>A

ENST00000313071.6:c.430G>A

NM\_005249.4:c.430G>A

Uncertain Significance

Met criteria codes **3**

PS4\_Supporting

BP4

PM2\_Supporting

Not Met criteria codes **4**

PVS1

PS1

PM1

PM5

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 5.0.0*

[↗](#) **Criteria Specification Approval History**







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

Evidence submitted by expert panel









### ***Rett and Angelman-like Disorders VCEP***

The p.Glu144Lys variant in FOYG1 is absent from gnomAD v4.1 (PM2\_Supporting). The p.Glu144Lys variant has been observed in at least 2 individuals with neurodevelopmental phenotypes (internal databases - GeneDx and University of Chicago) (PS4\_Supporting). The computational predictor REVEL gives a score of 0.112, which is below the threshold of 0.290, evidence that does not predict a damaging effect on FOYG1 function (BP4). In summary, the p.Glu144Lys variant in FOYG1 is classified as uncertain significance based on the ACMG/AMP criteria (PM2\_Supporting, PS4\_Supporting, BP4). (FOYG1 Specifications v.5.0.0; curation approved on 8/27/2025)

#### **Met criteria codes**

- |                       |   |   |  |
|-----------------------|---|---|--|
| <b>PS4_Supporting</b> |  |  | The p.Glu144Lys variant has been observed in at least 2 individuals with neurodevelopmental phenotypes (internal database - GeneDx and University of Chicago).             |
| <b>BP4</b>            |  |  | The computational predictor REVEL gives a score of 0.112, which is below the threshold of 0.290, evidence that does not predict a damaging effect on FOYG1 function (BP4). |
| <b>PM2_Supporting</b> |  |  | The p.Glu144Lys variant in FOYG1 is absent from gnomAD v4.1 (PM2_Supporting).  |

#### **Not Met criteria codes**

- |             |   |   |  |
|-------------|---|---|--|
| <b>PVS1</b> |    |    | No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline |
| <b>PS1</b>  |  |  | No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline |
| <b>PM1</b>  |  |  | No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline |
| <b>PM5</b>  |  |  | No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline |

#### **Curation History**

Showing 1 to 1 of 1 rows



The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

---

[ClinGen Terms of Use.](#)  
⌘ Powered by BCM's Genboree.