

Variant: *NM_005249.5(FOXG1):c.706A>G (p.Asn236Asp)*

Version: 1.0

CA389475686 [↗](#)

1878806 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: cce556e1-f5a0-4274-9858-77bc2aae1c45

Approved on: 2025/2/28

Published on: 2025/3/26

HGVS expressions

NM_005249.5:c.706A>G

NM_005249.5(FOXG1):c.706A>G (p.Asn236Asp)

NC_000014.9:g.28767985A>G

CM000676.2:g.28767985A>G

NC_000014.8:g.29237191A>G

CM000676.1:g.29237191A>G

NC_000014.7:g.28306942A>G

NG_009367.1:g.5905A>G

ENST00000706482.1:c.706A>G

ENST00000313071.7:c.706A>G

ENST00000313071.6:c.706A>G

NM_005249.4:c.706A>G

Likely Pathogenic

Met criteria codes **4**

PM2_Supporting

PS2

PP3

PM1

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 3.0.0*

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

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

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The c.706A>G (p.Asn236Asp) variant in FOXG1 has been identified as a de novo occurrence with confirmed parental relationships in 1 individual with FOXG1 disorder (GeneDx: internal data) (PS2). The p.Asn236Asp variant occurs in the well-characterized forkhead functional domain (aa 181-275) of FOXG1 (PM1). The computational predictor REVEL gives a score of 0.955, which is above the threshold of 0.75, evidence that correlates with impact to FOXG1 function (PP3). The p.Asn236Asp variant in FOXG1 is absent from gnomAD v4.1 (PM2_Supporting). In summary, the p.Asn236Asp variant in FOXG1 is classified as Likely Pathogenic for FOXG1 syndrome based on the ACMG/AMP criteria (PS2, PM1, PP3, PM2_supporting) (FOXG1 specifications version 3.0; curation approved on 2/28/2025).

Met criteria codes			
PM2_Supporting			The p.Asn236Asp variant in FOXG1 is absent from gnomAD v4.1 (PM2_Supporting).
PS2			This variant has been identified as a de novo occurrence with confirmed parental relationships in 1 individual with FOXG1 disorder (GeneDx: internal data) (PS2).
PP3			The computational predictor REVEL gives a score of 0.955, which is above the threshold of 0.75, evidence that correlates with impact to FOXG1 function (PP3).
PM1			The p.Asn236Asp variant occurs in the well-characterized forkhead functional domain (aa 181-275) of FOXG1 (PM1).

Curation History 	
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Showing 1 to 1 of 1 rows	
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