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Variant: NM_005249.5(FOXG1):c.565C>G (p.Leu189Val)

Version: 1.0

CA389475372

547390 (ClinVar)

Gene: FOXG1 (HGNC:2290)
Condition: FOXG1 disorder (MONDO:0100040)
Inheritance Mode: Autosomal dominant inheritance
UUID: e62185d2-30c7-40d4-9a85-7f2c329113b1
Approved on: 2023/4/14
Published on: 2023/6/16

HGVS expressions

NM_005249.5(FOXG1):c.565C>G (p.Leu189Val)
NM_005249.5:c.565C>G
NC_000014.9:g.28767844C>G
CM000676.2:g.28767844C>G
NC_000014.8:g.29237050C>G
CM000676.1:g.29237050C>G
NC_000014.7:g.28306801C>G
NG_009367.1:g.5764C>G
ENST00000706482.1:c.565C>G
ENST00000313071.7:c.565C>G
ENST00000313071.6:c.565C>G
NM_005249.4:c.565C>G

Likely Pathogenic

Met criteria codes 4

PP3 PM1 PM5 PM2_Supporting

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 p.Leu189Val variant occurs in the well-characterized Forkhead functional domain of the FOXG1 gene (PM1). The p.Leu189Val variant in FOXG1 is absent from gnomAD (PM2_supporting). Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3). A pathogenic missense variant (p.Leu189Phe) has been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID 28661489, GeneDx internal database)(PM5). In summary, the p.Leu189Val variant in FOXG1 is classified as likely pathogenic for a FOXG1-related disorder based on the ACMG/AMP criteria (PM1, PM2_supporting, PP3, PM5).

Met criteria codes

PP3		✔	Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).
PM1		✔	The p.Leu189Val variant occurs in the well-characterized Forkhead functional domain of the FOXG1 gene (PM1).
PM5		✔	A pathogenic missense variant (p.Leu189Phe) has been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID 28661489, GeneDx internal database)
PM2_Supporting		✔	The p.Leu189Val variant in FOXG1 is absent from gnomAD (PM2_supporting).

Curation History

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