Variant: NM_005249.5(FOXG1):c.1273T>C (p.Ser425Pro)

Version: 1.0

CA314592

205476 (ClinVar)

Gene: FOXG1 (HGNC:2290)
Condition: FOXG1 disorder (MONDO:0100040)
Inheritance Mode: Autosomal dominant inheritance
UUID: 046eaace-29f2-4aab-81be-a16e8ecd7e7b
Approved on: 2024-04-18
Published on: 2024-07-01

HGVS expressions

NM_005249.5:c.1273T>C
NM_005249.5(FOXG1):c.1273T>C (p.Ser425Pro)
NC_000014.9:g.28768552T>C
CM000676.2:g.28768552T>C
NC_000014.8:g.29237758T>C
CM000676.1:g.29237758T>C
NC_000014.7:g.28307509T>C
NG_009367.1:g.6472T>C
ENST00000706482.1:c.1273T>C
ENST00000313071.7:c.1273T>C
ENST00000313071.6:c.1273T>C
NM_005249.4:c.1273T>C

Benign

Met criteria codes 2

BP5_Strong **BS2**

Not Met criteria codes 1

BS1

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 p.Ser425Pro variant in FOXG1 is present in one XX and one XY individual in gnomAD v2.1.1 (0.00176%) (not sufficient to meet BS1 criteria). The p.Ser425Pro variant is observed in at least 2 unaffected individuals (GeneDx internal database) (BS2). The p.Ser425Pro variant is found in at least 3 patients with an alternate molecular basis of disease (GeneDx internal database) (BP5_Strong). In summary, the p.Ser425Pro variant in FOXG1 is classified as benign based on the ACMG/AMP criteria (BS2, BP5_strong).

Met criteria codes

BP5_Strong	ℹ	✓	The p.Ser425Pro variant is found in at least 3 patients with an alternate molecular basis of disease (GeneDx internal database) (BP5_Strong).
BS2	ℹ	✓	The p.Ser425Pro variant is observed in at least 2 unaffected individuals (GeneDx internal database) (BS2).

Not Met criteria codes

BS1	ℹ	✗	The p.Ser425Pro variant in FOXG1 is present in one XX and one XY individual in gnomAD v2.1.1 (0.00176%) (not sufficient to meet BS1 criteria).

Curation History

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