The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_005249.5(FOXG1):c.701C>T (p.Ser234Phe)

Version: 1.0

CA314620

205491 (ClinVar)

Gene: FOXG1 (HGNC:2290)
Condition: FOXG1 disorder (MONDO:0100040)
Inheritance Mode: Autosomal dominant inheritance
UUID: c78214c2-6594-4a67-a350-7d60cc42c93c
Approved on: 2024/6/25
Published on: 2024/8/23

HGVS expressions

NM_005249.5:c.701C>T
NM_005249.5(FOXG1):c.701C>T (p.Ser234Phe)
NC_000014.9:g.28767980C>T
CM000676.2:g.28767980C>T
NC_000014.8:g.29237186C>T
CM000676.1:g.29237186C>T
NC_000014.7:g.28306937C>T
NG_009367.1:g.5900C>T
ENST00000706482.1:c.701C>T
ENST00000313071.7:c.701C>T
ENST00000313071.6:c.701C>T
NM_005249.4:c.701C>T

Pathogenic

Met criteria codes 4

PP3 | PS2_Very Strong | PM2_Supporting | PS4_Moderate

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.Ser234Phe variant in FOXG1 has been reported as a de novo occurrence (biological parentage confirmed) in at least 3 individuals with FOXG1-disorder (Internal database – Ambry, PMID 37308910) (PS2_Very Strong). The p.Ser234Phe variant has been observed in at least 3 individuals with FOXG1-disorder (Internal database – Invitae, PMID 26633542, 38539105) (PS4_Moderate). The p.Ser234Phe variant in FOXG1 is absent from gnomAD v4.1 (PM2_Supporting). Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3). In summary, the p.Ser234Phe variant in FOXG1 is classified as Pathogenic for FOXG1-disorder based on the ACMG/AMP criteria (PS2_Very Strong, PS4_Moderate, PM2_Supporting, PP3).

Met criteria codes

PP3	✔	Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).
PS2_Very Strong	✔	The p.Ser234Phe variant in FOXG1 has been reported as a de novo occurrence (biological parentage confirmed) in at least 3 individuals with FOXG1-related disorder (Internal database - Ambry, PMID 37308910).
PM2_Supporting	✔	The p.Ser234Phe variant in FOXG1 is absent from gnomAD v2.1 (PM2_Supporting).
PS4_Moderate	✔	The p.Ser234Phe variant has been observed in at least 3 individuals with FOXG1-disorder (Internal database - Invitae, PMID 26633542, 38539105).

Curation History

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