

Variant: NM_001330260.2(SCN8A):c.5795G>C
(p.Arg1932Pro)

Version: 1.0

CA318305 [↗](#)

207134 (ClinVar) [↗](#)

Gene: SCN8A ([HGNC:6334](#))

Condition: complex neurodevelopmental disorder ([MONDO:0100038](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 2a80ec23-1208-46a6-9e9c-740e0bd96689

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HGVS expressions

NM_001330260.2(SCN8A):c.5795G>C (p.Arg1932Pro)

NM_001330260.2:c.5795G>C

NC_000012.12:g.51807281G>C

CM000674.2:g.51807281G>C

NC_000012.11:g.52201065G>C

CM000674.1:g.52201065G>C

NC_000012.10:g.50487332G>C

NG_021180.2:g.221046G>C

NG_021180.3:g.222324G>C

ENST00000354534.11:c.5795G>C

ENST00000627620.5:c.5795G>C

ENST00000662684.1:c.5795G>C

ENST00000668547.1:c.5672G>C

ENST00000354534.10:c.5795G>C

ENST00000355133.7:c.5672G>C

ENST00000545061.5:c.5672G>C

ENST00000599343.5:c.5828G>C

ENST00000627620.2:c.5795G>C

NM_001177984.2:c.5672G>C

NM_014191.3:c.5795G>C

NM_001330260.1:c.5795G>C

NM_001369788.1:c.5672G>C

NM_014191.4:c.5795G>C

NM_001177984.3:c.5672G>C

Likely Benign

Met criteria codes **2**

BP4 BS1

Not Met criteria codes **4**

PS4 PP3 PM1 PM5

Evidence Links **0**

Expert Panel

Epilepsy Sodium Channel VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Epilepsy Sodium Channel
Expert Panel Specifications to the ACMG/AMP Variant
Interpretation Guidelines for SCN8A Version 2.0

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

Evidence submitted by expert panel

Epilepsy Sodium Channel VCEP

The c.5795G>C variant in SCN8A is a missense variant predicted to cause substitution of Arginine by Proline at amino acid 1932 (p.Arg1932Pro). The total allele frequency in GnomAD 4.1.1 is 0.006754% (109/1613768 alleles), which is higher than the ClinGen Sodium Channel VCEP threshold (0.0002%) for BS1, and therefore meets this criterion (BS1). The computational predictor REVEL gives a score of 0.248, which suggests that the variant does not impact SCN8A function (BP4). In summary, this variant meets the criteria to be classified as likely benign for autosomal dominant complex neurodevelopmental disorder based on the ACMG/AMP criteria applied, as specified by the ClinGen Epilepsy Sodium Channel VCEP: BS1, BP4 (VCEP SCN8A specifications v2.0.0; June 23, 2026).

Met criteria codes

BP4			Revel= 0.248
BS1			Total allele frequency in GnomAD 4.1.1 is 0.006754%, which is higher than the ClinGen Epilepsy Sodium Channel VCEP threshold (>0.0002%) for BS1, and therefore meets this criterion (BS1).

Not Met criteria codes

PS4			SCV002647299.2= Ambry- Likely Benign SCV006072489.1/SCV000633977.10= Labcorp- VUS SCV001247161.37= Tuebingen- VUS SCV000242925.11= GeneDX- Likely Benign
PP3			Revel= 0.248
PM1			Not in Pathogenic Enriched Region
PM5			Different missense variants at this codon in SCN8A or paralogous genes have been identified but do not meet the classification of pathogenic or likely pathogenic by the ClinGen Epilepsy Sodium Channel VCEP. SCN8A c.5795G>A (p.Arg1932Gln)= Likely benign (BS1+BP4) SCN2A c.5878C>T (p.Pro1960Ser)= VUS (PM2_sup) SCN3A c.5863C>T (p.Pro1955Ser)= VUS (PM2_sup)

Curation History [↗](#)

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