Title: *SCN8A*-Related Epilepsy with Encephalopathy Table 3
Authors: Hammer MF, Wagnon JL, Mefford HC, Meisler MH
Initial posting: August 2016
Note: The following information is provided by the authors and has not been reviewed by *GeneReviews* staff.

**Table 3. Epilepsy-Associated Pathogenic Variants in *SCN8A* (published as of 12/1/15)**

| cDNA Position [1] | Protein Change | Inheritance | First Reported |
|---|---|---|---|
| c.629T>C | p.Phe210Leu | *de novo* | PMID 25818041 |
| c.641G>A | p.Gly214Asp | *de novo* | PMID 23934111 |
| c.643A>G | p.Asn215Arg | *de novo* | PMID 25533962 |
| c.643A>G | p.Asn215Arg | *de novo* | PMID 25568300 |
| c.647T>A | p.Val216Asp | *de novo* | PMID 24888894 |
| c.667A>G | p.Arg223Gly | *de novo* | PMID 25239001 |
| c.779T>C | p.Phe260Ser | *de novo* | PMID 26297079 |
| c.779T>C | p.Phe260Ser | *de novo* | PMID 25568300 |
| c.1221G>C | p.Leu407Phe | *de novo* | PMID 26235739 |
| c.1228G>C | p.Val410Leu | *de novo* | PMID 25568300 |
| c.1588C>T | p.Arg530Trp | inherited - mosaic father | PMID 25914188 |
| c.2003C>T | p.Thr767Ile | *de novo* | PMID 24874546 |
| c.2537T>C | p.Phe846Ser | *de novo* | PMID 24888894 |
| c.2549G>A | p.Arg850Gln | *de novo* | PMID 26235739 |
| c.2624T>A | p.Leu875Gln | *de novo* | PMID 23934111 |
| c.2668G>A | p.Ala890Thr | *de novo* | PMID 26235739 |
| c.2668G>A | p.Ala890Thr | *de novo* | PMID 25568300 |
| c.2879T>A | p.Val960Asp | *de novo* | PMID 25568300 |
| c.2952C>G | p.Asn984Lys | *de novo* | PMID 25725044 |
| c.3868C>G | p.Leu1331Val | inherited - mosaic father | PMID 23708187 |
| c.3979A>G | p.Ile1327Val | *de novo* | PMID 25799905 |
| c.3979A>G | p.Ile1327Val | *de novo* | PMID 24352161 |
| | p.Pro1428_Lys1473del | *de novo* | PMID 25568300 |
| c.4351G>A | p.Gly1451Ser | *de novo* | PMID 25725044 |
| c.4397A>C | p.Asn1466Thr | *de novo* | PMID 24888894 |
| c.4398C>A | p.Asn1466Lys | *de novo* | PMID 24888894 |
| c.4435A>G | p.Ile1479Val | *de novo* | PMID 25568300 |
| c.4774G>C | p.Val1592Leu | *de novo* | PMID 25568300 |

| cDNA Position [1] | Protein Change | Inheritance | First Reported |
|---|---|---|---|
| c.4787C>G | p.Ser1596Cys | *de novo* | PMID 26297079 |
| c.4787C>G | p.Ser1596Cys | *de novo* | PMID 26235739 |
| c.4813A>G | p.Ile1605Arg | *de novo* | PMID 25568300 |
| c.4850G>A | p.Arg1617Gln | *de novo* | PMID 25046240 |
| c.4850G>A | p.Arg1617Gln | *de novo* | PMID 26235739 |
| c.4850G>A | p.Arg1617Gln | *de novo* | PMID 25568300 |
| c.4850G>A | p.Arg1617Gln | *de novo* | PMID 24888894 |
| c.4850G>A | p.Arg1617Gln | *de novo* | PMID 23020937 |
| c.4862T>G | p.Leu1621Trp | *de novo* | PMID 26235738 |
| c.4873G>A | p.Gly1625Arg | *de novo* | PMID 25533962 |
| c.4948G>A | p.Ala1650Thr | *de novo* | PMID 25568300 |
| c.4948G>A | p.Ala1650Thr | *de novo* | PMID 24888894 |
| c.5302A>G | p.Asn1768Asp | *de novo* | PMID 22365152 |
| c.5401C>G | p.Gln1801Glu | *de novo* | PMID 25568300 |
| c.5610A>T | p.Glu1870Asp | *de novo* | PMID 26297079 |
| c.5614C>T | p.Arg1872Trp | *de novo* | PMID 25568300 |
| c.5614C>T | p.Arg1872Trp | *de novo* | PMID 25568300 |
| c.5614C>T | p.Arg1872Trp | *de novo* | PMID 24888894 |
| c.5614C>T | p.Arg1872Trp | *de novo* | PMID 25951352 |
| c.5615G>A | p.Arg1872Gln | *de novo* | PMID 25568300 |
| c.5615G>A | p.Arg1872Gln | *de novo* | PMID 25568300 |

1. All cDNA positions are based on transcript NM_014191.3.