(((SCN1A) OR (SCN2A) OR (SCN3A) OR (SCN4A) OR (SCN5A) OR (SCN8A) OR (SCN9A) OR (SCN10A) OR (SCN11A)) AND ((gain-of-function) OR (loss-of-function)) OR ((GOF) OR (LOF)) AND ((electrophysiology) OR (patch clamp) OR (voltage clamp))) AND (("2021/04/28"[Date - Publication] : "2021/07/13"[Date - Publication]))

Start date: 28 April 2021

54 results, acc. 13/JUL/2022 2.17pm

Incl. criteria: Missense variant in Nav with experimental evidence of functional effect and clear net LOF or GOF

15 studies

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Gene | Variant | Effect | Reference | Phenotype | Pred fuNCion | Pred SCION-MTL | Pred SCION-MTMKL | Winner |
| SCN2A | Q1531K | GOF | 35637276 | See Table | LOF 0.71 | GOF 0.81 | GOF 0.52 | SCION |
|  | L1563V | GOF |  |  | LOF 0.76 | GOF 0.76 | GOF 0.48 | SCION |
|  | E1321K | GOF |  |  | GOF 0.82 | GOF 0.59 | GOF 0.48 |  |
|  | V1325I | GOF |  |  | GOF 0.90 | GOF 0.63 | GOF 0.51 |  |
|  | D195G | LOF |  |  | LOF 0.73 | LOF 0.55 | LOF 0.73 |  |
|  | R853Q | LOF |  |  | LOF 0.84 | LOF 0.64 | LOF 0.85 |  |
| SCN5A | V281A | LOF | 34628415 | BrS | LOF 0.60 | LOF 0.85 | LOF 0.89 |  |
|  | L1582P | LOF |  |  | LOF 0.81 | LOF 0.69 | LOF 0.79 |  |
|  | F543L | LOF |  |  | GOF 0.52 | GOF 0.53 | LOF 0.73 | SCION |
|  | K1419E | LOF |  |  | LOF 0.60 | LOF 0.88 | LOF 0.87 |  |
| SCN5A | L1412F | LOF | 34649698 | ERS | LOF 0.66 | LOF 0.80 | LOF 0.66 |  |
|  | G452C | LOF |  | ERS | LOF 0.68 | LOF 0.75 | LOF 0.62 |  |
| SCN1A | A1783V | LOF | 34776868 | DS | GOF 0.69 | LOF 0.83 | LOF 0.88 | SCION |
| SCN5A | R893C | LOF | 35305865 | BrS | LOF 0.75 | LOF 0.81 | LOF 0.87 |  |
|  | L846R | LOF |  |  | GOF 0.51 | LOF 0.73 | LOF 0.78 | SCION |
|  | G897E | LOF |  |  | LOF 0.65 | LOF 0.90 | LOF 0.83 |  |
|  | R878H | LOF |  |  | LOF 0.78 | LOF 0.90 | LOF 0.86 |  |
|  | G1743E | LOF |  |  | LOF 0.79 | LOF 0.87 | LOF 0.81 |  |
| SCN5A | R893C | LOF | 35331424 | BrS | LOF 0.75 | LOF 0.81 | LOF 0.86 |  |
| SCN4A | R1463H | GOF | 33670307 | Non-dystrophic myotonia | GOF 0.73 | GOF 0.56 | GOF 0.53 |  |
| SCN5A | G1481V | GOF | 33778442 | LQT3 | GOF 0.79 | LOF 0.71 | GOF 0.80 |  |
|  | Q1491H | GOF |  |  | GOF 0.66 | LOF 0.70 | GOF 0.80 |  |
| SCN5A | V1667D | LOF | 33946750 | BrS | GOF 0.53 | LOF 0.55 | LOF 0.72 | SCION |
| SCN2A | V911A | GOF | 34093402 | ESES, DEE | LOF 0.60 | GOF 0.65 | GOF 0.55 | SCION |
|  | K1933M | GOF |  | ESES, DEE | LOF 0.60 | GOF 0.79 | GOF 0.52 | SCION |
|  | S863F | LOF |  | BFIS, ESES | LOF 0.59 | LOF 0.74 | LOF 0.84 |  |
| SCN5A | R104W | LOF | 34122134 | BrS | LOF 0.82 | LOF 0.83 | LOF 0.89 |  |
| SCN5A | P1310L | LOF | 34204499 | BrS | GOF 0.86 | LOF 0.83 | LOF 0.86 | SCION |
| SCN9A | E44Q | GOF | 34222704 | Paroxysmal pain | LOF 0.67 | GOF 0.78 | GOF 0.79 | SCION |
| SCN2A | R853Q | LOF | 34287911 | DEE | LOF 0.79 | LOF 0.61 | LOF 0.79 |  |
| SCN5A | G1712S | LOF | 34348284 | BrS | LOF 0.72 | LOF 0.88 | LOF 0.84 |  |