UniProtKB/Swiss-Prot P35498: Variant p.Tyr413Asn

Sodium channel protein type 1 subunit alpha
Gene: SCN1A
Chromosomal location: 2q24.3

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Variant information

Variant position: 413

Type of variant: Disease [Disclaimer]

Residue change: From Tyrosine (Y) to Asparagine (N) at position 413 (Y413N, p.Tyr413Asn).

Physico-chemical properties: Change from large size and aromatic (Y) to medium size and polar (N)

BLOSUM score: -2

Involvement in disease: Epileptic encephalopathy, early infantile, 6 (EIEE6) [MIM:607208]: A severe form of epileptic encephalopathy characterized by generalized tonic, clonic, and tonic-clonic seizures that are initially induced by fever and begin during the first year of life. Later, patients also manifest other seizure types, including absence, myoclonic, and simple and complex partial seizures. Psychomotor development delay is observed around the second year of life. Some patients manifest a borderline disease phenotype and do not necessarily fulfill all diagnostic criteria for core EIEE6. EIEE6 is considered to be the most severe phenotype within the spectrum of generalized epilepsies with febrile seizures-plus. {ECO:0000269|PubMed:11359211, ECO:0000269|PubMed:12083760, ECO:0000269|PubMed:12566275, ECO:0000269|PubMed:12754708, ECO:0000269|PubMed:12821740, ECO:0000269|PubMed:14504318, ECO:0000269|PubMed:14738421, ECO:0000269|PubMed:15087100, ECO:0000269|PubMed:15944908, ECO:0000269|PubMed:16122630, ECO:0000269|PubMed:16458823, ECO:0000269|PubMed:16713920, ECO:0000269|PubMed:17054684, ECO:0000269|PubMed:17054685, ECO:0000269|PubMed:17129991, ECO:0000269|PubMed:17347258, ECO:0000269|PubMed:17561957, ECO:0000269|PubMed:18413471, ECO:0000269|PubMed:18639757, ECO:0000269|PubMed:18930999, ECO:0000269|PubMed:19522081, ECO:0000269|PubMed:19563458, ECO:0000269|PubMed:19589774, ECO:0000269|PubMed:19783390, ECO:0000269|PubMed:20110217, ECO:0000269|PubMed:20431604, ECO:0000269|PubMed:20452746, ECO:0000269|PubMed:20522430, ECO:0000269|PubMed:20729507, ECO:0000269|PubMed:21248271, ECO:0000269|PubMed:21864321, ECO:0000269|PubMed:22092154, ECO:0000269|PubMed:22612257, ECO:0000269|PubMed:23195492}. Note=The disease is caused by mutations affecting the gene represented in this entry.

Variant description: In EIEE6.

Other resources:
- Variant rs121917967 [dbSNP | Ensembl]

Sequence information

Variant position: 413

Protein sequence length: 2009

Location on the sequence: RAAGKTYMIFFVLVIFLGSF Y LINLILAVVAMAYEEQNQAT

Residue conservation:

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Human                          RAAGKTYMIFFVLVIFLGSFYLINLILAVVAMAYEEQNQAT
Rat                            RAAGKTYMIFFVLVIFLGSFYLINLILAVVAMAYEEQNQAT
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Sequence annotation in neighborhood:

Type	Positions	Description
Chain	1 – 2009	Sodium channel protein type 1 subunit alpha
Transmembrane	400 – 425	Helical; Name=S6 of repeat I
Repeat	110 – 454	I

Literature citations

De-novo mutations of the sodium channel gene SCN1A in alleged vaccine encephalopathy: a retrospective study.
Berkovic S.F.; Harkin L.; McMahon J.M.; Pelekanos J.T.; Zuberi S.M.; Wirrell E.C.; Gill D.S.; Iona X.; Mulley J.C.; Scheffer I.E.;
Lancet Neurol. 5:488-492(2006)
Cited for: VARIANTS EIEE6 LEU-403; ASN-413; HIS-946; ASP-1238; GLY-1396 AND GLN-1645;

The spectrum of SCN1A-related infantile epileptic encephalopathies.
Harkin L.A.; McMahon J.M.; Iona X.; Dibbens L.; Pelekanos J.T.; Zuberi S.M.; Sadleir L.G.; Andermann E.; Gill D.; Farrell K.; Connolly M.; Stanley T.; Harbord M.; Andermann F.; Wang J.; Batish S.D.; Jones J.G.; Seltzer W.K.; Gardner A.; Sutherland G.; Berkovic S.F.; Mulley J.C.; Scheffer I.E.;
Brain 130:843-852(2007)
Cited for: VARIANTS CYS-393; PRO-395; GLU-422; GLY-626; VAL-1480; SER-1543; GLN-1636 AND HIS-1657; VARIANTS EIEE6 HIS-79; CYS-84; TRP-101; ARG-199; MET-226; THR-239; LEU-403; ASN-413; GLY-674; PRO-783; GLU-944; LEU-945; GLU-950; ASP-1238; MET-1390; GLY-1396; PRO-1441; VAL-1545; CYS-1596; GLN-1645; VAL-1707; ARG-1721 AND THR-1922; VARIANT GEFS+2 VAL-973;

De novo SCN1A mutations in Dravet syndrome and related epileptic encephalopathies are largely of paternal origin.
Heron S.E.; Scheffer I.E.; Iona X.; Zuberi S.M.; Birch R.; McMahon J.M.; Bruce C.M.; Berkovic S.F.; Mulley J.C.;
J. Med. Genet. 47:137-141(2010)
Cited for: VARIANTS EIEE6 CYS-84; GLN-101; LYS-171; THR-175; ASN-194; SER-227; PHE-406; ASN-413; PRO-783; GLU-944; LEU-945; HIS-946; GLU-950; GLY-1396; LYS-1450; VAL-1545; GLN-1645; ARG-1726 AND THR-1783; VARIANTS HIS-604; GLN-1636 AND HIS-1657;