Open Peer Review on Qeios

SCN1A wt Allele

National Cancer Institute

Source

National Cancer Institute. SCN1A wt Allele. NCI Thesaurus. Code C102448.

Human SCN1A wild-type allele is located in the vicinity of 2q24.3 and is approximately 84 kb in length. This allele, which encodes sodium channel protein type 1 subunit alpha, plays a role in voltage-gated sodium transport. Mutation of the gene is associated with generalized epilepsy with febrile seizures plus type 2, Dravet syndrome, intractable childhood epilepsy with generalized tonic-clonic seizures, familial hemiplegic migraine type 3 and familial febrile convulsions type 3A.