		PHENOTYPE(S)	INHERITANCE	FUNCTIONAL EFFECT	CHANNEL	ACTIVATED BY	ION SELECTIVITY
	Neurological						
	Presenting with severe early-onset epilepsy						
	sodium channels						
(A)		Dravet syndrome	Dominant	LOF	α subunit of the type 1 neuronal voltage-gated sodium channel ($Na_v1.1$)	Voltage	Sodium
(B)		EOEE	Recessive	LOF	β1 auxiliary subunit for the type 1 neuronal voltage-gated sodium channel	Voltage	Sodium
(C)		Early infantile epileptic encephalopathy	Dominant	LOF	α subunit of the type 2 neuronal voltage-gated sodium channel ($Na_v1.2$)	Voltage	Sodium
		EIMFS	Dominant	LOF			
		Autism without other neurological features	Dominant	LOF			
(D)		EOEE	Dominant	GOF	α subunit of the type 6 neuronal voltage-gated sodium channel ($Na_v1.6$)	Voltage	Sodium
(E)		Epileptic encephalopathy with neuromuscular disease	Recessive	LOF	α subunit of the type 8 neuronal voltage-gated sodium channel ($Na_v1.8$)	Voltage	Sodium
	Potassium channels						
(F)		EOEE	Dominant	LOF and GOF	α2 subunit of Shaker family potassium channels ($K_v1.2$)	Voltage	Potassium
		EOEE	Dominant	LOF and GOF	Member 1 of the Shab family of potassium channels ($K_v2.1$)	Voltage	Potassium
		EOEE	Dominant	Unknown	Member 5 of the Ether-a-go-go family of potassium channels ($K_v10.2$)	Voltage	Potassium
(G)		EOEE	Dominant	LOF	Voltage-gated potassium channel, Q subfamily, member 2 ($K_v7.2$)	Voltage	Potassium
		Infantile spasms	Dominant	GOF			
(H)		EIMFS	Dominant	GOF	Calcium-activated potassium channel (subfamily T) member 1 ($K_{ca}4.1$)	Calcium	Potassium
	Calcium channels						
(I)		EOEE	Dominant	LOF	α1A subunit of the P/Q type voltage-gated calcium channel ($Ca_v2.1$)	Voltage	Calcium
		Epileptic encephalopathy	Recessive	LOF	α2δ2 auxiliary subunit of the P/Q voltage-gated calcium channel	Voltage	Calcium
	Glutamate receptors						
		Epileptic-dyskinetic encephalopathy	Recessive	LOF	Accessory protein to the neuronal AMPA receptor	Glutamate	Cations
		EOEE	Dominant	LOF	Subunit 1 of the neuronal NMDA receptor	Glutamate	Cations
		Focal epilepsy + speech difficulties	Dominant	LOF and GOF	Subunit 2A of the neuronal NMDA receptor	Glutamate	Cations
		Focal epilepsy + speech difficulties	Dominant	LOF and GOF	Subunit 2B of the neuronal NMDA receptor	Glutamate	Cations
		EOEE	Dominant	GOF	Subunit 2D of the neuronal NMDA receptor	Glutamate	Cations
	GABA Receptors						

(J)		EOEE	Dominant	LOF	α1 subunit of the neuronal GABA-A receptor	GABA	Chloride
		Epileptic encephalopathy	Dominant	LOF	β1 subunit of the neuronal GABA-A receptor	GABA	Chloride
		Early myoclonic encephalopathy	Dominant	LOF	B2 subunit of the neuronal GABA-A receptor	GABA	Chloride
(K)		EOEE	Dominant LOF	LOF	β3 subunit of the neuronal GABA-A receptor	GABA	Chloride
(L)		EOEE	Dominant	LOF	γ2 subunit of the neuronal GABA-A receptor	GABA	Chloride
	Other Channels						
(M)		EOEE	Dominant	LOF	α3 isoform of sodium/potassium ATPase	ATP	Sodium/Potassium
		EOEE	Dominant	LOF and GOF	Hyperpolarization-activated, cyclic nucleotide–gated (HCN) channel, type 1	Hyperpolarisation	Cations
		EIMFS	Recessive LOF	LOF	Potassium and chloride transporter (KCC2)	Potassium	Potassium and chloride
	Self-limiting familial epilepsies with onset in the neonatal or early infantile period						
(G)		Self-limiting familial neonatal seizures	Dominant	LOF	Voltage-gated potassium channel, Q subfamily, member 2 ($K_v7.2$)	Voltage	Potassium
(N)		Self-limiting familial neonatal seizures	Dominant	LOF and GOF	Voltage-gated potassium channel, Q subfamily, member 3 ($K_v7.3$)	Voltage	Potassium
(A)		GEFS+	Dominant	LOF and GOF	α subunit of the type 1 neuronal voltage-gated sodium channel ($Na_v1.1$)	Voltage	Sodium
(B)		Genetic epilepsy with febrile seizures GEFS+	Dominant	LOF	β1 auxiliary subunit for the type 1 neuronal voltage-gated sodium channel	Voltage	Sodium
(C)		Self-limiting familial neonatal-infantile seizures	Dominant	LOF	α subunit of the type 2 neuronal voltage-gated sodium channel ($Na_v1.2$)	Voltage	Sodium
(D)		Infantile convulsions choreoathetosis (ICCA) syndrome	Dominant	LOF	α subunit of the type 6 neuronal voltage-gated sodium channel ($Na_v1.6$)	Voltage	Sodium
(O)		GEFS+	Dominant	Unknown	α subunit of the type 7 neuronal voltage-gated sodium channel ($Na_v1.7$)	Voltage	Sodium
	Autosomal dominant nocturnal frontal lobe epilepsy						
		Nocturnal frontal lobe epilepsy	Dominant	GOF	α2 subunit of the nicotinic acetylocholine receptor	Acetylcholine	Cations
		Nocturnal frontal lobe epilepsy	Dominant	GOF	α4 subunit of the nicotinic acetylocholine receptor	Acetylcholine	Cations
		Nocturnal frontal lobe epilepsy	Dominant	GOF	α4 subunit of the nicotinic acetylocholine receptor	Acetylcholine	Cations
(H)		Nocturnal frontal lobe epilepsy	Dominant	GOF	Calcium-activated potassium channel (subfamily T) member 1 ($K_{ca}4.1$)	Calcium	Potassium
	Susceptibility to idiopathic generalised epilepsies						
(P)		Susceptibility to IGE	Dominant	GOF	α1H subunit of the T type calcium channel ($Ca_v3.2$)	Voltage	Calcium
(Q)		Susceptibility to JME	Dominant	Unknown	β_4 auxiliary subunit of the P/Q voltage-gated calcium channel	Voltage	Calcium
		Susceptibility to IGE	Dominant	LOF (microdeletions only)	α7 subunit of the nicotinic acetylocholine receptor	Acetylcholine	Cations

(R)		Susceptibility to IGE	Dominant	LOF	Type 2 voltage-gated chloride channel	Voltage	Chloride
(J)		Susceptibility to IGE	Dominant	LOF	α1 subunit of the neuronal GABA-A receptor	GABA	Chloride
(K)		Susceptibility to IGE	Dominant	LOF	β3 subunit of the neuronal GABA-A receptor	GABA	Chloride
		Susceptibility to IGE	Dominant	LOF	δ subunit of the neuronal GABA-A receptor	GABA	Chloride
(L)		Susceptibility to IGE	Dominant	LOF	γ2 subunit of the neuronal GABA-A receptor	GABA	Chloride
		Familial febrile seizures	Dominant	LOF			
	Other epilepsies						
		Variable epilepsy phenotype – ranging from epileptic encephalopathy to well-controlled seizures	X-linked recessive	LOF	Type 4 voltage-gated chloride channel	Voltage	Chloride
		Progressive myoclonic epilepsy	Dominant	LOF	Member 1 of the Shaw family of potassium channels ($K_v3.1$)	Voltage	Potassium
		Epilepsy and paroxysmal movement disorder	Dominant	GOF	α subunit of the large conductance calcium-sensitive potassium channel (BK)	Calcium	Potassium
(N)		Familial focal epilepsy	Dominant	LOF	Voltage-gated potassium channel, Q subfamily, member 3 ($K_v7.3$)	Voltage	Potassium
		Childhood-onset focal epilepsy	Dominant	LOF and GOF	α subunit of the type 3 neuronal voltage-gated sodium channel ($Na_v1.3$)	Voltage	Sodium
	Other neurological disorders						
	Movement disorders and related phenotypes						
		Familial hemiplegic migraine	Dominant	LOF	α2 isoform of sodium/potassium ATPase	ATP	Sodium/
		Familial basilar migraine	Dominant	LOF			Potassium
		Alternating hemiplegia of childhood	Dominant	LOF			
(M)		Alternating hemiplegia of childhood	Dominant	LOF	α3 isoform of sodium/potassium ATPase	ATP	Sodium/
		Rapid-onset Parkinsonism dystonia	Dominant	LOF			Potassium
		CAPOS syndrome	Dominant	GOF (unconfirmed)			
(I)		Familial hemiplegic migraine	Dominant	LOF	α1A subunit of the P/Q type voltage-gated calcium channel ($Ca_v2.1$)	Voltage	Calcium
		Episodic ataxia type 2	Dominant	LOF			
		Progressive spinocerebellar ataxia (SCA6) – triplet repeat	Dominant	LOF			
		Hyperkalaemic periodic paralysis	Dominant	LOF	α1S subunit of the L type voltage-gated calcium channel ($Ca_v1.1$)	Voltage	Calcium
		Malignant hyperthermia	Dominant	LOF			
(Q)		Episodic ataxia type 5	Dominant	Unknown	β_4 auxiliary subunit of the P/Q voltage-gated calcium channel	Voltage	Calcium
(R)		Leukoencephalopathy with ataxia	Recessive	LOF	Type 2 voltage-gated chloride channel	Voltage	Chloride
		Hyperekplexia	Dominant or recessive	GOF	α1 subunit of the spinal glycine receptor	Glycine	Chloride

		Hyperekplexia	Dominant or recessive	GOF	β1 subunit of the spinal glycine receptor	Glycine	Chloride
		Episodic ataxia type 1	Dominant	LOF	α1 subunit of the Shaker family potassium channels (K_v1.1)	Voltage	Potassium
(F)		Hereditary spastic paraplegia and ataxia	Dominant	LOF	α2 subunit of the Shaker family potassium channels (K_v1.2)	Voltage	Potassium
		Spinocerebellar ataxia	Dominant	LOF	Member 3 of the Shaw family of potassium channels (K_v3.3)	Voltage	Potassium
(S)		Early-onset cerebellar ataxia, intellectual disability, oral apraxia, and epilepsy	Dominant LOF	LOF	Member 3 of the Shal family of potassium channels (K_v4.3)	Voltage	Potassium
		Spinocerebellar ataxia	Dominant LOF	LOF			
(A)		Familial hemiplegic migraine	Dominant GOF	GOF	α subunit of the type 1 neuronal voltage-gated sodium channel (Na_v1.1)	Voltage	Sodium
	<u>Neuromuscular disorders</u>						
		Slow channel congenital myasthenic syndrome	Dominant	GOF	α1 subunit of the nicotinic acetylcholine receptor	Acetylcholine	Cations
		Fast channel congenital myasthenic syndrome	Recessive	LOF			
		Multiple pterygium syndrome	Recessive	LOF (truncation)			
		Slow channel congenital myasthenic syndrome	Dominant	GOF	β1 subunit of the nicotinic acetylcholine receptor	Acetylcholine	Cations
		Congenital myasthenic syndrome associated with Ach receptor deficiency	Recessive	LOF (reduced expression)			
		Slow channel congenital myaesthenic syndrome	Dominant	GOF	δ polypeptide subunit of the nicotinic acetylcholine receptor	Acetylcholine	Cations
		Fast channel congenital myasthenic syndrome	Recessive	LOF			
		Congenital myasthenic syndrome associated with Ach receptor deficiency	Recessive	LOF (reduced expression)			
		Multiple pterygium syndrome	Recessive	LOF (truncation)			
		Slow channel congenital myasthenic syndrome	Dominant	GOF	ε polypeptide subunit of the nicotinic acetylcholine receptor	Acetylcholine	Cations
		Fast channel congenital myasthenic syndrome	Recessive	LOF			
		Congenital myasthenic syndrome associated with Ach receptor deficiency	Recessive	LOF (reduced expression)			
		Multiple pterygium syndrome (lethal and Escobar variants)	Recessive	LOF	γ subunit of the nicotinic acetylcholine receptor	Acetylcholine	Cations
		Dominant myotonia congenita	Dominant	LOF	Skeletal muscle voltage-gated chloride channel	Voltage	Chloride
		Recessive myotonia congenita	Recessive	LOF			

		Congenital contractures of the limbs and face, hypotonia, and developmental delay	Dominant Recessive	LOF	Non-selective non-inactivating cation channel	None	Cations
		Dominant central core disease Recessive central core disease Malignant hyperthermia Minicore myopathy with external ophthalmoplegia	Dominant Recessive Dominant Recessive	GOF LOF GOF LOF	Skeletal muscle Ryanodine receptor 1	Calcium	Calcium
		Hyperkalaemic periodic paralysis Hypokalaemic periodic paralysis Paramyotonia congenita Congenital myaesthenic syndrome	Dominant Dominant Dominant Recessive	LOF LOF LOF LOF	α subunit of the type 4 neuronal voltage-gated sodium channel ($Na_v1.4$)	Voltage	Sodium
(X)		Carbamazepine-responsive cramp-fasciculation syndrome	Dominant	LOF	Transient receptor potential cation channel, family A, member 1	Various ligands, activated by noxious stimuli	Cations
	Peripheral nerve disorders						
(R)		Peripheral nerve hyperexcitability	Dominant	LOF	Voltage-gated potassium channel, Q subfamily, member 2 ($K_v7.2$)	Voltage	Potassium
(O)		Erytheromelalgia and related neuropathic pain syndromes Insensitivity to pain	Dominant Recessive	GOF LOF	α subunit of the type 7 neuronal voltage-gated sodium channel ($Na_v1.7$)	Voltage	Sodium
(E)		Episodic pain syndrome	Dominant	GOF	α subunit of the type 8 neuronal voltage-gated sodium channel ($Na_v1.8$)	Voltage	Sodium
		Familial episodic pain syndrome Hereditary sensory and autonomic neuropathy	Dominant GOF Dominant GOF	GOF GOF	α subunit of the type 9 neuronal voltage-gated sodium channel ($Na_v1.9$)	Voltage	Sodium
(X)		Familial episodic pain syndrome	Dominant	GOF	Transient receptor potential cation channel, family A, member 1	Various ligands, activated by noxious stimuli	Cations
	Deafness						
		Hearing loss	Dominant	LOF	Voltage-gated potassium channel, Q subfamily, member 4 ($K_v7.4$)	Voltage	Potassium
	Miscellaneous neurological						
		Intellectual disability, ataxia, hypotonia, microcephaly, and seizures	Dominant Recessive	GOF LOF	Ionotropic glutamate receptor 6	Glutamate	Cations
	Cardiac disease						
	Sodium channels						

(B)		Familial atrial fibrillation	Dominant	LOF	β auxiliary subunit for the type 1 neuronal voltage-gated sodium channel	Voltage	Sodium
		Brugada syndrome	Dominant	LOF			
		Nonspecific cardiac conduction defect	Dominant	LOF			
		Familial atrial fibrillation	Dominant	LOF	β auxiliary subunit for the type 2 neuronal voltage-gated sodium channel	Voltage	Sodium
		Familial atrial fibrillation	Dominant	LOF	β auxiliary subunit for the type 3 neuronal voltage-gated sodium channel	Voltage	Sodium
		Brugada syndrome	Dominant	LOF			
		Familial atrial fibrillation	Dominant	LOF	β auxiliary subunit for the type 4 neuronal voltage-gated sodium channel	Voltage	Sodium
		Long QT syndrome	Dominant	LOF			
		Familial atrial fibrillation	Dominant	LOF and GOF	α subunit of the type 5 neuronal voltage-gated sodium channel ($Na_v1.5$)	Voltage	Sodium
		Brugada syndrome	Dominant	LOF			
		Dilated cardiomyopathy	Dominant	LOF			
		Heart block	Dominant	LOF			
		Long QT syndrome	Dominant	LOF			
		Polymorphic ventricular tachycardia	Dominant	GOF			
		Sick sinus syndrome	Recessive	LOF			
		Brugada syndrome	Dominant	LOF	α subunit of the type 8 neuronal voltage-gated sodium channel ($Na_v1.8$)	Voltage	Sodium
		Prolonged QT syndrome	Dominant	LOF			
	Potassium channels						
(T)		Ventricular tachycardia and dilated cardiomyopathy	Dominant	LOF	ATP-sensitive potassium channel (K_{ATP})	ATP	Potassium
		Familial atrial fibrillation	Dominant	LOF	α5 subunit of the Shaker family potassium channels ($K_v1.5$)	Voltage	Potassium
(S)		Brugada syndrome	Dominant	GOF	Member 3 of the Shal family of potassium channels ($K_v4.3$)	Voltage	Potassium
		Atrial fibrillation	Dominant	LOF			
		Long QT syndrome	Dominant	LOF	Member 2 of the Ether-a-go-go (EAG) type potassium channels ($K_v10.2$)	Voltage	Potassium
		Short QT syndrome	Dominant	GOF			
		Jervell and Lange-Nielsen syndrome	Recessive	LOF	Member 1 of the MinK subfamily of potassium channels	Voltage	Potassium
		Long QT syndrome	Dominant	GOF			
		Familial atrial fibrillation	Dominant	GOF	Member 1 of the MinK related peptide subfamily of potassium channels (MiRP1) (membrane subunit which assembles with $K_v10.2$)	Voltage	Potassium
		Long QT syndrome	Dominant	LOF			
		Brugada syndrome	Dominant	LOF	Member 2 of the MinK related peptide subfamily of potassium channels (MiRP1) (membrane subunit which assembles with $K_v10.2$)	Voltage	Potassium
		Familial atrial fibrillation	Dominant	GOF	Voltage-gated potassium channel, Q subfamily, member 1 ($K_v7.1$)	Voltage	Potassium
		Jervell and Lange-Neilsen syndrome	Recessive	LOF			
		Long QT syndrome	Dominant	LOF			
		Short QT syndrome	Dominant	GOF			

(U)		Andersen-Tawil syndrome	Dominant	LOF	Member 2 of the J family of inwardly rectifying voltage-gated potassium channels ($K_{ir}2.1$)	PIP_2	Potassium
		Familial atrial fibrillation	Dominant	GOF			
		Short QT syndrome	Dominant	GOF			
(V)		Long QT syndrome	Dominant	LOF	G-protein-activated inwardly rectifying voltage-gated potassium channel ($K_{ir}3.4$)	PIP_2	Potassium
	Calcium channels						
		Brugada syndrome	Dominant	LOF	α1C subunit of the L type voltage-gated calcium channel ($Ca_v1.2$)	Voltage	Calcium
		Timothy syndrome	Dominant	LOF			
		Brugada syndrome	Dominant	LOF	β2 subunit of the L type voltage-gated calcium channel	Voltage	Calcium
		Brugada syndrome	Dominant	LOF	α1δ subunit of the L type voltage-gated calcium channel	Voltage	Calcium
		Arrhythmogenic right ventricular dysplasia	Dominant	LOF	Cardiac ryanodine receptor 2	Calcium	Calcium
		Catecholaminegic polymorphic ventricular tachycardia	Dominant	LOF			
	Cation channels						
		Brugada syndrome	Dominant	LOF	Hyperpolarization-activated, cyclic nucleotide–gated (HCN) channel, type 4	Hyperpolarisation	Cations
		Sick sinus syndrome	Dominant	LOF			
	Renal Disease						
		Barrter's syndrome with sensorineural deafness	Recessive	LOF	β subunit of the renal chloride channel	N/A	Chloride
		Dent disease	X-linked recessive	LOF	Type 5 voltage-gated chloride channel	Voltage	Chloride
		Hypophosphataemic Rickets					
		Nephrolithiasis					
		Hypocalciuric nephrocalcinosis					
		Barrter's syndrome with sensorineural deafness	Digenic recessive	LOF	Renal chloride channel	N/A	Chloride
		Barrter's syndrome	Recessive	LOF	Renal outer-medullar potassium channel ($K_{ir}1.1$)	Voltage	Potassium
(W)		Liddle syndrome	Dominant	GOF	β subunit of the epithelial sodium channel	Voltage	Sodium
		Psedohyperaldosteronism	Recessive	LOF			
		Barrter's syndrome	Recessive	LOF	Type 1 renal sodium/potassium/chloride transporter (NKCC2)	N/A	Sodium, potassium and chloride
		Gitelman syndrome	Recessive	LOF	Type 3 renal sodium/potassium/chloride transporter (NKCC2)	N/A	Sodium, potassium and chloride
	Endocrine and Bone Disease						
		Permanent neonatal diabetes mellitus +/- neurologic features	Dominant	GOF	ATP-binding cassette of the sulphonylurea receptor	Sulphonylurea and ATP	Potassium

		Transient neonatal diabetes mellitus Persistent hyperinsulinaemic hypoglycaemia of infancy	Dominant Dominant and recessive	GOF LOF			
(T)		Cantú syndrome (hypertrichotic osteochondrodysplasia)	Dominant	GOF	ATP-sensitive potassium channel (K_{ATP})	ATP	Potassium
(P)		Familial hyperaldosteronism	Dominant	GOF	α1H subunit of the T type calcium channel ($Ca_v3.2$)	Voltage	Calcium
		Dominant osteopetrosis	Dominant	LOF	Type 7 chloride channel	N/A	Chloride
		Recessive osteopetrosis	Recessive	LOF			
(V)		Familial hyperaldosteronism	Dominant	LOF	G-protein-activated inwardly rectifying voltage-gated potassium channel ($K_{ir}3.4$)	PIP_2 /G-protein activation	Potassium
		Permanent neonatal diabetes, with neurological features (Delay, Epilepsy, Neonatal Diabetes, DEND syndrome) Persistent hyoerinsulinaemic hypoglycaemia of infancy Transient neonatal diabetes Permanent neonatal diabetes Maturity-onset diabetes if the young (MODY)	Dominant	GOF	Member 11 of the inward rectifier type of potassium channels ($K_{ir}6.2$)	ATP/PIP_2	Potassium
(O)		Osteogenesis imperfecta	Recessive	Unknown	α subunit of the type 7 neuronal voltage-gated sodium channel ($Na_v1.7$)	Voltage	Sodium
	Miscellaneous/multisystem Diseases						
		Primary aldosteronism, seizures, and neurologic abnormalities Sinoatrial node dysfunction and deafness	Dominant Recessive	GOF LOF	α1D subunit of the L type calcium channel ($Ca_v1.3$)	Voltage	Calcium
		Cystic fibrosis	Recessive	LOF	Cystic fibrosis transmembrane conductance regulator	ATP	Chloride
		Temple-Baraitser syndrome Zimmermann-Laband syndrome	Dominant Dominant	GOF GOF	Member 1 of the Ether-a-go-go (EAG) type potassium channels ($K_v10.1$)	Voltage	Potassium
(U)		Andersen-Tawil syndrome	Dominant	LOF	Member 2 of the J family of inwardly rectifying voltage-gated potassium channels ($K_{ir}2.1$)	PIP_2	Potassium
(V)		Andersen-Tawil syndrome (without dysmorphism)	Dominant	LOF	G-protein-activated inwardly rectifying voltage-gated potassium channel ($K_{ir}3.4$)	PIP_2 /G-protein activation	Potassium
		SESAME (**se**izures, **s**ensorineural deafness, **a**taxia, intellectual disability) or EAST (**e**pilepsy, **a**taxia, **s**ensorineural deafness and **t**ubulopathy) syndrome	Recessive	LOF	Member 10 of the inward rectifier type of potassium channels ($K_{ir}4.1$)	PIP_2	Potassium

		Familial pulmonary arterial hypertension	Dominant	LOF	Member 1 of the Task family of two pore potassium channels ($K_{2P}3.1$)	pH, oxygen tension, G-protein activation	Potassium
(W)		Bronchiectasis	Dominant	LOF	β subunit of the epithelial sodium channel	Voltage	Sodium
		Nonepidermolytic focal palmoplantar ketatoderma (Olmsted syndrome)	Dominant	GOF	Transient receptor potential cation channel, subfamily V, member 3	Temperature	Cations
		Brachyolmia	Dominant	GOF	Transient receptor potential cation channel, subfamily V, member 4, found in ciliated epithelial cells	Various physical, chemical, and hormonal stimuli	Calcium
		Familial digital arthropathy-brachydactyly	Dominant	LOF			
		Hereditary sensorimotor neuropathy	Dominant	LOF			
		Metatropic dysplasia	Dominant	GOF			
		Parastremmatic dwarfism	Dominant	Unknown			
		Scapuloperitoneal muscular atrophy	Dominant	LOF			
		Spondyloepiphyseal dysplasia	Dominant	GOF			
		Distal spinal muscular atrophy	Dominant	LOF			

Supplementary data – the known human channelopathies. Abbreviations used: EOEE – early-onset developmental and epileptic encephalopathy; EIMFS – epilepsy of infancy with migrating focal seizures; IGE: Idiopathic generalised epilepsy; JME – juvenile myoclonic epilepsy; CAPOS syndrome - **C**erebellar ataxia, **A**reflexia, **P**es cavus, **O**ptic atrophy, and **S**ensorineural deafness; GEFS+ - genetic epilepsy with febrile seizures plus; GOF – gain-of-function; LOF – loss-of-function; PIP_2 – phosphatidylinositol 4,5-bisphosphate

Genes in red text are auxiliary subunits to ion channels.