UKGTN Testing Criteria

Test name: **Brain Channelopathy 11 Gene Panel**	
Approved name and symbol of disorder/condition(s): Brain Channelopathy See Appendix 1 **Approved name and symbol of gene(s):** See Appendix 1	**OMIM number(s):** See Appendix 1 **OMIM number(s):** See Appendix 1

Patient name:	**Date of birth:**
Patient postcode:	**NHS number:**
Name of referrer:	
Title/Position:	**Lab ID:**

Referrals will only be accepted from one of the following:	
Referrer	**Tick if this refers to you.**
Consultant Neurologist	
Consultant Paediatric Neurologist	
Consultant Clinical Geneticist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
A clear clinical history of an episodic ataxia **OR**	
Hemiplegic migraine with the following features: • Fulfils criteria for Migraine with Aura (MA) • Aura including some degree of hemiparesis and may be prolonged • At least one first-degree relative with identical attacks **OR**	
Paroxysmal dyskinesia	

Additional Information:
For panel tests: At risk family members where familial mutation is known do not require a full panel test but should be offered analysis of the known mutation

If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.

Testing Criteria Appendix 1

Conditions in panel test

OMIM standard name of condition and symbol	OMIM number	HGNC standard name and symbol of the gene	HGNC number	OMIM number	References or locus specific database links
MIGRAINE, FAMILIAL HEMIPLEGIC, 2; FHM2	602481	ATPase, Na+/K+ TRANSPORTING, ALPHA-2 POLYPEPTIDE; ATP1A2	**800**	182340	http://www.LOVD.nl/ATP1A2
ALTERNATING HEMIPLEGIA OF CHILDHOOD 1; AHC1	104290	ATPase, Na+/K+ TRANSPORTING, ALPHA-2 POLYPEPTIDE; ATP1A2	**800**	182340	http://www.LOVD.nl/ATP1A2
DYSTONIA 12; DYT12	128235	ATPase, Na+/K+ TRANSPORTING, ALPHA-3 POLYPEPTIDE; ATP1A3	**801**	182350	11 families with mutations in this gene have been described on OMIM (de Carvalho Aguiar et al 2004; Brashear et al 2007 and Blanco-Arias et al 2009).
ALTERNATING HEMIPLEGIA OF CHILDHOOD 2; AHC2	614820	ATPase, Na+/K+ TRANSPORTING, ALPHA-3 POLYPEPTIDE; ATP1A3	**801**	182350	Heinzen et al. (2012) Nature Genet. 44: 1030-1034 estimate that pathogenic mutations in this gene may account for up to 74% of patients with sporadic, typical AHC
EPISODIC ATAXIA, TYPE 2; EA2	108500	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT; CACNA1A	**1388**	601011	http://www.LOVD.nl/CACNA1A
MIGRAINE, FAMILIAL HEMIPLEGIC, 1; FHM1	141500	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT; CACNA1A	**1388**	601011	http://grenada.lumc.nl/LOVD2/FHM/home.php?select_db=CACNA1A
EPISODIC ATAXIA, TYPE 5; EA5	613855	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-4 SUBUNIT; CACNB4	**1404**	601949	1 family described on OMIM Escayg et al. 2000; 1 index case identified in house
EPILEPSY, IDIOPATHIC GENERALIZED, SUSCEPTIBILITY TO, 9; EIG9	607682	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-4 SUBUNIT; CACNB4	**1404**	601949	2 families described on OMIM Escayg et al. 2000
EPISODIC ATAXIA, TYPE 1; EA1	160120	POTASSIUM CHANNEL, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 1 (EPISODIC ATAXIA WITH MYOKYMIA); KCNA1	**6218**	176260	15 index cases with different pathogenic mutations are described on OMIM
MIGRAINE, WITH OR WITHOUT AURA, SUSCEPTIBILITY TO, 13; MGR13	613656	POTASSIUM CHANNEL, SUBFAMILY K, MEMBER 18;	**19439**	613655	1 family reported on OMIM Lafreniere et al 2010; 2 index cases identified in house

OMIM standard name of condition and symbol	OMIM number	HGNC standard name and symbol of the gene	HGNC number	OMIM number	References or locus specific database links
		KCNK18			
PAROXYSMAL NONKINESIGENIC DYSKINESIA 1; PNKD1	118800	PAROXYSMAL NONKINESIGENIC DYSKINESIA; PNKD	**9153**	609023	14 families with pathogenic mutations reported on OMIM
CONVULSIONS, FAMILIAL INFANTILE, WITH PAROXYSMAL CHOREOATHETOSIS; ICCA	602066	PROLINE-RICH TRANSMEMBRANE PROTEIN 2; PRRT2	**30500**	614386	>63 families described on OMIM
EPISODIC KINESIGENIC DYSKINESIA 1; EKD1	128200	PROLINE-RICH TRANSMEMBRANE PROTEIN 2; PRRT2	**30500**	614386	>47 families described on OMIM
SEIZURES, BENIGN FAMILIAL INFANTILE, 2; BFIS2	605751	PROLINE-RICH TRANSMEMBRANE PROTEIN 2; PRRT2	**30500**	614386	>57 families described on OMIM
DRAVET SYNDROME	607208	SODIUM CHANNEL, NEURONAL TYPE I, ALPHA SUBUNIT; SCN1A	**10585**	182389	http://www.molgen.ua.ac.be/SCN1AMutations/Home/Default.cfm
GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2; GEFSP2	604403	SODIUM CHANNEL, NEURONAL TYPE I, ALPHA SUBUNIT; SCN1A	**10585**	182389	http://www.molgen.ua.ac.be/SCN1AMutations/Home/Default.cfm
MIGRAINE, FAMILIAL HEMIPLEGIC, 3; FHM3	609634	SODIUM CHANNEL, NEURONAL TYPE I, ALPHA SUBUNIT; SCN1A	**10585**	182389	http://www.molgen.ua.ac.be/SCN1AMutations/Home/Default.cfm 5 families described on OMIM
EPISODIC ATAXIA, TYPE 6; EA6	612656	SOLUTE CARRIER FAMILY 1 (GLIAL HIGH AFFINITY GLUTAMATE TRANSPORTER), MEMBER 3; SLC1A3	**10941**	600111	2 families reported on OMIM Jen et al 2005; de Vries et al 2009
GLUT1 DEFICIENCY SYNDROME 2; GLUT1DS2	612126	SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1; SLC2A1	**11005**	138140	6 families described on OMIM Weber et al 2008; Suls et al 2008; Schneider et al 2010 6 index cases identified in house
EPILEPSY, IDIOPATHIC GENERALIZED, SUSCEPTIBILITY TO, 12; EIG12	614847	SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1; SLC2A1	**11005**	138140	2 families reported on OMIM Suls et al 2008; Striano et al 2012 1 index case identified in house
GLUT1 DEFICIENCY SYNDROME 1; GLUT1DS1	606777	SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1; SLC2A1	**11005**	138140	6 families described on OMIM Seidner et al 1998; Wang et al 2000; Klepper et al 2001; Brockmann et al 2001 3 index cases identified in house