Schedule of Accreditation

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Neurogenetics Unit
Rare & Inherited Disease Laboratory
NHS North Thames Genomic Laboratory Hub
Level 4-6 Barclay House
37 Queen Square
London
WC1N 3BH

Contact: Vaneesha Gibbons
Tel: +44 (0)207 829 8870
E-Mail: vaneeshagibbons@nhs.net
Website: www.uclh.nhs.uk/neurogeneticslab

Testing performed at the above address only

DETAIL OF ACCREDITATION

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
	<u>Neurogenetics</u>	
HUMAN BODY FLUIDS / TISSUES	Analysis of the human genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype	
Receipt of Sanger sequencing data	Variant detection in genes and variant confirmation and carrier/ predictive testing for the following neurological disorders: Andersen Tawil syndrome- KCNJ2 Familial British Dementia- ITM2B Amyloidosis, Finnish Type (FAF)- GSN Charcot-Marie-Tooth Neuropathy, Type 1B and CMT2- MPZ CMT X-Linked - CX32/ GJB1 Distal Hereditary Motor Neuropathy Type V; HMNV- BSCL2 Episodic ataxia: Type 1- KCNA1 Familial Amyloid polyneuropathy; FAP- TTR	Reporting of Sanger sequencing data using Mutation Surveyor software as described in NGENS ANA0020

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
	<u>Neurogenetics</u> (cont'd)	
HUMAN BODY FLUIDS / TISSUES (cont'd)	Analysis of the human genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype (cont'd)	
Receipt of Sanger sequencing data (cont'd)	Variant detection in genes and variant confirmation and carrier/ predictive testing for the following neurological disorders: Charcot-Marie-Tooth disease type 2K; CMT2K and Charcot-Marie-Tooth disease type 4A; CMT4A-GDAP1 Charcot-Marie-Tooth disease type 2F; CMT2F and distal hereditary motor neuropathy; HMN2B- HSPB1 Neuropathy, hereditary sensory and autonomic, type IA; HSAN1-SPTLC1 Paramyotonia Congenita- SCN4A Hypokalemic periodic paralysis-CACNA1S & SCN4A Hyperkalemic periodic paralysis-SCN4A Leber Optic Atrophy; m.3460G>A; m.11778G>A; m.14484T>C Mitochondrial mutations in: MT-TK including m.8344A>G and MT-ATP6 including, m.8993T>G/C	Reporting of Sanger sequencing data using Mutation Surveyor software as described in NGENS ANA0020

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
	Neurogenetics (cont'd)	
HUMAN BODY FLUIDS / TISSUES (cont'd)	Analysis of the human genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype (cont'd)	
Receipt of Sanger sequencing data (including MLPA)	Sanger Sequencing and MLPA analysis for the following neurological disorders:	Reporting of Sanger sequencing data and MLPA data as described in NGENS ANA0020 for Sanger sequencing and using Mutation surveyor software as described in RGS ANA0205 and NGEN-LP-GeneMarkerMLPA for MLPA analysis
	SCN1A related epilepsy- SCN1A	
	Charcot-Marie-Tooth Neuropathy, Type 2A2- MFN2	
	Dopa Responsive Dystonia; DRD-GCH1	
	Hereditary liability to pressure palsies- PMP22 del/PMP22 sequence mutations	
	Charcot-Marie-Tooth disease demyelinating type 1A; CMT1A-PMP22 Dup/PMP22 sequence mutations	
	Myotonia Congenita dominant and recessive forms- CLCN1	:
	Parkinson disease, juvenile, type 2 - PARK2	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
	Neurogenetics (cont'd)	
HUMAN BODY FLUIDS / TISSUES (cont'd)	Analysis of the human genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype (cont'd)	
Receipt of fragment analysis data	Fragment analysis: Autosomal Dominant Spino-cerebellar Ataxia :1,2,3,6,7, 12, 17- ATXN1; ATXN2; ATXN3; TBP, CACNA1A; ATXN7; PPP2R2B; Dentatorubropallidoluysian Atrophy; DRPLA- ATN1 Friedreich's Ataxia; FRDA- FXN Huntington Disease; HD- HTT Huntington Disease-Like 2; HDL2- JPH3 Primary Torsion Dystonia; DYT1- TOR1A X- linked Bulbospinal Neuropathy- AR	Fragment sizing using GeneMapper software as described in SOPs NGENS ANA0004
Receipt of fragment analysis data	Detection of Chromosome-9 linked ALS/FTD- C9orf72	Reporting of fragment analysis data using GeneMapper software as described in SOPs NGENS ANA0004
Receipt of sequencing data	Large scale rearrangements of the mitochondrial genome.	Reporting of fragment sizing data using long range PCR and mitochondrial WGS

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
	Neurogenetics (cont'd)	
HUMAN BODY FLUIDS / TISSUES (cont'd)	Analysis of the human mitochondrial genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype (cont'd)	
Receipt of sequencing data	Detection of:	Reporting of outcomes of targeted variant screening using Mutation Surveyor as described in NGENS ANA0020 for Sanger sequencing and NGENS ANA0004 for GeneMapper
	Leber Optic Atrophy: m.3460G>A; m.11778G>A; m.14484T>C	
	Mitochondrial common mutations: m.3243A>G, m.8344A>G, m.8993T>G/C	
Fragment Analysis	DNA dosage analysis of mtDNA	Reporting of Quantitative fluorescent PCR as described in NGENS DNG0019 and NGENS DNG0015
Receipt of NGS data	Mitochondrial whole genome sequencing	SOP: Whole mtDNA NGS data analysis as described in NGENS DNG0028 using in house pipeline
Receipt of Sanger data and MLPA data	Confirmation of pathogenic variation	Using Mutation Surveyor software as described in NGENS ANA0020 for Sanger sequencing and using Genemarker software as described in RGS ANA0205 and NGEN-LP-GeneMarkerMLPA for MLPA
Data extracted following Next Generation Sequencing Analysis	Variant interpretation	Bioinformatic analysis with reference to: Output from in house pipeline
END		