Schedule of Accreditation

issued by

United Kingdom Accreditation Service

2 Pine Trees, Chertsey Lane, Staines-upon-Thames, TW18 3HR, UK

8040

Accredited to ISO 15189:2012

University College London Hospitals NHS Foundation Trust

Issue No: 005 **Issue date:** -16 June 2022

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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
	Neurogenetics	
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	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>	
HUMAN BODY FLUIDS / TISSUES	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	Variant detection in genes and variant confirmation and carrier/ predictive testing for the following neurological disorders: Episodic ataxia: Type 1- KCNA1 Familial Amyloid polyneuropathy; FAP- TTR 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

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	<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 (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 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 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	
HUMAN BODY FLUIDS / TISSUES	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 (including MLPA)	DNA dosage analysis for:	Reporting of fragment analysis data (including MLPA) using Genemarker software as described in RGS ANA0205 and NGEN-LP-GeneMarkerMLPA
	Autosomal dominant Parkinson's disease with a-synuclein rearrangements (PARK1/4)- SNCA	
	Autosomal dominant Parkinson's disease-PARK8- LRRK2	

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	<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 (cont'd)	
Receipt of fragment analysis data (including MLPA)	Fragment analysis:	Fragment sizing using GeneMapper software as described in SOPs NGENS ANA0004
	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	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES	<u>Neurogenetics</u> Analysis of the human mitochondrial genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype (cont'd)	
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 NGENS DNG0018
Receipt of sequencing data	Detection of: 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	Reporting of outcomes of targeted variant screening using Mutation Surveyor as described in NGENS ANA0020 for Sanger sequencing and NGENS ANA0004 for GeneMapper

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
	Neurogenetics	
HUMAN BODY FLUIDS / TISSUES	Analysis of the human mitochondrial genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype (cont'd)	
Receipt of Sanger sequencing data and agarose gel images	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

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	<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 (cont'd)	
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: NGEN-LP-Alamut NGEN-LP-Custom NGS Panel Data Analysis for Pipeline NGEN-LP-NGS_Pipeline_Hands-on_Analysis
END		