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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	Genomic data analysis for the purpose of clinical diagnosis of rare neurological disease	
Receipt of Sanger sequencing data	Variant detection in genes and variant confirmation and carrier/ predictive testing for variants detected by NGS 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
	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 (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 nuclear genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype (cont'd)	
Receipt of MLPA data	MLPA analysis for the following neurological disorders: 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	Reporting of MLPA data as described in NGEN-LP-GeneMarker MLPA for MLPA analysis :

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 ofthe human nuclear genome (or part thereof) to detect pathogenic variation that results in a clinical phenotype (cont'd)	
Data extracted following Next Generation Sequencing Analysis	Variant interpretation	Bioinformatic analysis with reference to: Output from in house pipeline
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: 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
Receipt of Fragment Analysis data	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	Confirmation of pathogenic variation	Using Mutation Surveyor software as described in NGENS ANA0020 for Sanger sequencing

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