Schedule of Accreditation

issued by

United Kingdom Accreditation Service

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

9865

Accredited to ISO 15189:2012

Manchester University NHS Foundation Trust

Issue No: 006 **Issue date:** 07 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
HUMAN BODY FLUIDS/TISSUE	Cytogenetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
Blood, amniotic fluid (AF), chorionic villus samples (CVS)	Chromosome analysis for: Prenatal diagnosis Reproductive medicine disorders Disorders of sexual development Developmental disorders	Metaphase preparation, microscopic examination and analysis of G-banded karyotypes using light microscopy or Metasystems image analysis system. Cell culture LP200 003, 011, LP110 015, 003, 012, LP120 012, 014, 015. G-banding analysis LP000 017, 008, 024, DOC2630
DNA from blood, mouthwashes, AF, CVS, foetal tissue	Chromosome profile analysis for: Prenatal diagnosis Developmental disorders (including 22q11, Williams syndrome, Angelman syndrome, Prader-Willi syndrome, etc.) Inheritance testing following proband microarray findings (targeted) Pregnancy loss	Microarray process using Beckman Coulter NX Span 8, Agilent microarray technology, OGT Constitutional V3 array, Agilent Microarray Scanner, Cytosure analysis software. DOC3097, 3236, LP130 011, LP430 004
DNA from blood, mouthwashes, AF, CVS, foetal tissue	Prenatal/Postnatal diagnosis of aneuploidy (chromosomes 13, 18, 21 or sex chromosomes) Maternal contamination testing	QF-PCR (Polymerase chain reaction) analysis using automated or manual DNA extraction (Instagene), in-house primer preparation, PCR (ABI9700), Genetic analyser (Applied Biosystems 3500XL), analysis (Genemapper software). LP500 001, 003, 005, 008, DOC868

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HUMAN BODY FLUIDS/TISSUE	Cytogenetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from blood	Identity testing	LP500 001, 003, 005, 008
DNA from blood	Graft v Host determination following bone marrow transplant	LP500 001, 003, 005, 008
Blood, AF, CVS, buccal swabs, tissue touch preparation slides	Macroscopic and microscopic detection and analysis of genetic rearrangements and/or genomic imbalance including confirmation and inheritance testing following proband microarray findings.	FISH using in-house procedures and commercial probes - including Kreatech (Leica), Abbott Molecular (Vysis), Cytocell (OGT), TCAG (Toronto, Canada), BlueGnome (Illumina), Pisces Scientific FISH probes (Empire Genomics). Thermobrite hybridisation station, fluorescence microscopes (Olympus), Metasystems Isis software. DOC3033, LP300 024
FFPE tissue slide	Detection of ALK and ROS1 gene rearrangements in non-small cell carcinoma of the lung. Detection of 1p/19q codeletions in Glioma.	DOC1411
Blood, AF, CVS, fresh tissue (including fibroblasts)	Cell culture for cryogenic storage, DNA/RNA extraction, export or immortalisation (lymphoblastoid cell lines)	DOC2285, LP160 015, 018, 022, 033, 041, 050, 043, 012, 015

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
Blood, mouthwashes, AF, CVS, solid tissues (products of conception, skin, placenta, umbilical cord, liver etc.), blood spots, FFPE tissue	DNA extraction and quality check for subsequent in house analysis (see below), referral to specialist centres and longterm storage	Automated JANUS Chemagic Prime system for DNA extraction (Perkin Elmer). DOC4355 Automated Chemagen DNA extraction (Chemagen 360). DOC2000 EZ1 DNA extraction kits. DOC905, 906, 907, 908, 913 COBAS FFPE extraction. DOC1300 iGENatal extraction. DOC2354 Cleaning DNA samples. DOC2047, DOC2347, LP400 002 Quality assessment. LP 000 239, Nanodrop LP 000 230, Qubit 2.0 DOC2356, DropSense DOC2828. DNA dilutions DOC752
Plasma	DNA extraction and quality check for subsequent in house analysis (see below), referral to specialist centres and longterm storage	Plasma preparation from blood using DOC2893, ctDNA and cfDNA extraction from plasma using DOC3183
PHA stimulated short term lymphocyte cultures; melanocyte cultures	RNA extraction and quality check for subsequent in house analysis (see below), referral to specialist centres and longterm storage	ENZ1 RNA extraction kit DOC992
DNA from primary samples	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs)	Next Generation Sequencing using: NextSeq 500 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines DOC2258, 2047, 2146, 3234, 1146, 2176, 2204, 2175, 2608, 3087, 4056, DOC1146
DNA from blood	Breast and Ovarian Cancer familial (BRCA1 and 2)	MiSeq, LP 000 284, DOC2816

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs)	Next Generation Sequencing using: NextSeq 500 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines. DOC2258, 2047, 2146, 3234, 1146, 2176, 2204, 2175, 2608, 3087, 4056, DOC1146
	Cardiac disorders: • Arrhythmia/ Cardiac Arrest • Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) • Brugada Syndrome • Cardiomyopathies • Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) • Familial Dilated Cardiomyopathy • Familial Hypertrophic Cardiomyopathy • Familial Thoracic Aortic Aneurysm • Long QT Syndrome • Molecular Autopsy	NextSeq, DOC2273
	Cystic fibrosis (CFTR; extended mutation testing)	MiSeq, DOC4032
	Familial Adenomatous Polyposis (APC) (FAP) and MUTYH-Associated Polyposis (MUTYH) (MAP)	MiSeq, LP 000 062
	Lynch Syndrome (HNPCC, familial colorectal cancer) (MLH1, MSH2, MSH6)	MiSeq, LP 000 050
	Fabry disease (GLA)	MiSeq, DOC761

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs)	Next Generation Sequencing using: NextSeq 500 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines. DOC2258, 2047, 2146, 3234, 1146, 2176, 2204, 2175, 2608, 3087, 4056, DOC1146
	Metabolic Disorders: • Amino acid metabolism and neurotransmission disorders (AA&NT • Carbohydrate metabolism disorders (CHO) • Disorders associated with hyperammonaemia, fatty acid oxidation defects and disorders of ketogeneis and ketolysis (AMN&FAOD&KET) • Lysosomal Storage Diseases and neuronal ceroid lipofuscinoses (LSD&NCL) • Niemann Pick C (NPC1, NPC2) • Organic acidaemias and vitamin cofactor disorders (OA&VIT) • Peroxisomal disorders (PER)	NextSeq, DOC2658

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs)	Next Generation Sequencing using: NextSeq 500 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines. DOC2258, 2047, 2146, 3234, 1146, 2176, 2204, 2175, 2608, 3087, 4056, DOC1146
	Inherited Cancer • Breast • Carney complex • Colorectal cancer and polyps • Cutaneous melanoma • CylindromatosisFanconi anemia • Gastric cancer • Hereditary diffuse gastric cancer • Hyperparathyroidism • Leukaemia • Melanoma • Neuroendocrine • Nevoid Basal Cell Carcinoma • Ovarian • Pancreatic • Paraganglioma / pheochromocytoma • Renal cell carcinoma • Schwannomatosis / meningioma	NextSeq, DOC4335

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs)	Next Generation Sequencing using: NextSeq 500 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines. DOC2258, 2047, 2146, 3234, 1146, 2176, 2204, 2175, 2608, 3087, 4056, DOC1146
	Ophthalmic disorders: • Anterior segment dysgenesis and glaucoma • Brittle cornea syndrome • Corneal abnormalities (including corneal dystrophy, BSC and other corneal genes) • Corneal dystrophy • Foveal hypoplasia & nystagmus • Lens abnormalities, including congenital cataracts • Lens abnormalities, non-cataract • Microphthalmia/ Coloboma • Ocular/ oculocutaneous albinism • Optic nerve disorders (including ocular/ oculocutaneous albinism, foveal hypoplasia & nystagmus, and optical atrophy) • Stickler syndrome	NextSeq, DOC3110
	Retinal Degeneration Conditions: Full gene panel Retinitis Pigmentosa, X-Linked (RPGR/ RP2) Macular Dystrophy (PRPH2/BEST1)	NextSeq, DOC1139
	Retinoblastoma (RB1)	MiSeq, LP 000 075
DNA from blood or fresh tissue	Overgrowth syndromes (PIK3CA/ PIK3R2/ AKT1/ AKT3 /mTOR/ CCND2/ PTEN)	MiSeq, DOC2671

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs)	Next Generation Sequencing using: NextSeq 500 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines. DOC2258, 2047, 2146, 3234, 1146, 2176, 2204, 2175, 2608, 3087, 4056, DOC1146
DNA/RNA from blood/melanocytes	Neurofibromatosis type 1 (NF1)	MiSeq, LP 000 250
DNA from FFPE tissue sections, slides or shavings	Breast and Ovarian Cancer treatment focussed (BRCA1 and 2) and germline mutations	DOC3340
DNA from FFPE tissue sections, slides or shavings	Cancer screening for somatic mutations (including malignant melanoma, non-small cell lung cancer, colorectal cancer and GIST)	MiSeq, DOC3338
	Glioma / CNS cancer panel screening for somatic mutations	MiSeq, DOC4253
	Glioma / CNS cancer panel screening for somatic mutations - Meningioma/schwannoma subpanel (NF2, SMARCB1, SMARCE1, SMARCA4, LZTR1)	MiSeq, DOC4763
	Colorectal cancer - tumour samples for somatic mutations (KRAS, NRAS, BRAF, PIK3CA)	MiSeq, DOC3338
	Colorectal cancer - tissue samples for somatic and germline mutations (APC, BMPR1A, CDH1, CTNNB1, MSH6, SMAD4, MLH1, MSH2, MUTYH, POLD1, POLE, PTEN and STK11)	MiSeq, DOC3339
RNA from FFPE samples	Lung cancer fusion gene panel (ALK, ROS1, NTRK3, RET)	QIAseq Targeted RNAScan Panel, MiSeq, DOC4764

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs)	Next Generation Sequencing using: NextSeq 500 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines. DOC2258, 2047, 2146, 3234, 1146, 2176, 2204, 2175, 2608, 3087, 4056, DOC1146
DNA from FFPE tissue sections, slides or shavings	Schwannomatosis in tumour samples (NF2, LZTR1, SMARCB1, SMARCE1, SMARCA4)	MiSeq LP 000 249, LP 000 069
DNA from blood, fresh tissue, AF, CVS	Schwannomatosis; Atypical Familial Rhabdoid Tumour (SMARCB1/ LZTR1)	MiSeq, LP 000 249
DNA from blood or FFPE tissue sections, slides, shavings, fresh tissue, AF, CVS	Neurofibromatosis type 2 (NF2)	MiSeq, LP 000 069
	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels	Next Generation Sequencing using:
DNA from blood	Exomes Panel (Clinical)	NextSeq, DOC3256
	Exomes Panel (Learning Difficulties)	NextSeq, DOC3196
DNA from blood or FFPE tissue sections, slides, shavings, fresh tissue, AF, CVS	Detection of clinical relevant nucleic acid sequence and copy number variants for confirmation of NGS results or family studies related to validated methods	Sanger sequencing using Thermo Thermal Cyclers and ABI3730XL sequencers, DOC1585, LP 000 236 Multiplex Ligation-dependant Probe Amplification (MLPA) or MS-MLPA using ABI3500XL analyser and DOC955, DOC1030, LP 000 162

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples	Mutation detection for the purpose of clinical diagnosis, carrier detection and predictive testing in the following:	PCR amplification and Sanger sequencing using Thermo Thermal Cyclers and ABI3730XL sequencers, DOC1585, LP 000 236
	Rare disease mutation confirmation on request	DOC1284
DNA from blood	Angelman Syndrome/ Prader-Willi Syndrome (UBE3A, SNRPN)	LP 000 054
	Doyne Honeycombe Retinal Dystrophy (DHRD; EFEMP1)	LP 000 246
	Late onset retinal dystrophy (LORD; C1QTNF5)	LP 000 247
	Legius Syndrome (SPRED1)	LP 000 251
	LCHAD common mutation (HADHA)	LP 000 264
	Macular Dystrophy (PROM1 c.1117C>T p.(Arg373Cys))	DOC1426
	Malignant melanoma tumour (KIT)	LP 000 232
	Schwannomatosis 1 (SMARCE1)	DOC1590
	Sorsby Fundus Dystrophy (SFD) (TIMP3)	LP 000 222
DNA from blood, AF, CVS	Congenital adrenal hyperplasia due to 21 hydroxylase deficiency(CYP21A2)	LP 000 082
	Retinitis Pigmentosa, X-Linked (RPGR, exon ORF15 only)	DOC2245
DNA from blood, AF, CVS, blood spots	MCADD common mutation (ACADM)	LP 000 268

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
RNA from lymphocyte culture	Retinoblastoma (RB1)	LP 000 075
DNA from primary samples	Mutation detection for the purpose of clinical diagnosis, carrier detection and predictive testing in the following cont'd:	PCR amplification and Sanger sequencing using Thermo Thermal Cyclers and ABI3730XL sequencers, DOC1585, LP 000 236
DNA from FFPE tissue sections, slides or shavings	Non-Small Cell Lung Cancer (somatic mutations, - tumour samples) (EGFR)	LP 000 257
	Gastro intestinal Stromal tumours for familial GIST (KIT, PDGFRA)	LP 000 232
	Glioma- hTERT promoter	DOC4161
	Melanoma (KIT)	DOC3104 and LP 000 232
DNA from primary samples	Mutation detection for the purpose of clinical diagnosis, carrier detection and predictive testing in the following:	PCR using Thermo Thermal Cycler and Pyromark PQQ 96MA pyrosequencing, DOC887 and:
DNA from blood, AF, CVS	Congenital Adrenal Hyperplasia due to 21 hydroxylase deficiency (CYP21A2)	LP 000 082
DNA from FFPE tissue sections, slides or shavings	Colorectal cancer (MLH1 hypermethylation and BRAF codon 600)	DOC3389, DOC869, DOC3104
	Malignant melanoma (BRAF, NRAS)	DOC3104
	MGMT methylation analysis (Giloma)	DOC3349
	Glioma (MGMT hypermethylation and BRAF codon 600)	DOC3349, DOC869

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples	Detection of whole or partial gene deletions and duplications and probe specific mutations of the following genes/disease regions:	Multiplex Ligation-dependant Probe Amplification (MLPA) or MS-MLPA using ABI3500XL analyser and DOC955, DOC1030, LP 000 162 and:
DNA from blood	Angelman syndrome/ Prader-Willi Syndrome (UBE3A, SNRPN)	LP 000 054
	Breast and Ovarian cancer (BRCA1 and 2)	LP 000 284
DNA from primary samples	Familial Adenomatous Polyposis (APC) (FAP) and MUTYH-Associated Polyposis (MUTYH) (MAP)	LP 000 062
	Lynch Syndrome (HNPCC, familial colorectal cancer) (MLH1, MSH2, MSH6)	LP 000 050
	Niemann Pick C (NPC1, NPC2)	DOC4020
DNA from blood, AF, CVS	Cystic fibrosis (CFTR)	DOC4032
	Duchenne and Becker Muscular Dystrophy (DMD)	LP 000 063
DNA/RNA from blood, lymphocyte culture, melanocyte culture	Neurofibromatosis type 1 (NF1)	LP 000 250
DNA from whole blood, FFPE tissue sections, slides or shavings, fresh tissue, AF, CVS	Neurofibromatosis type 2 (NF2)	LP 000 069
	Retinoblastoma (RB1)	LP 000 075
DNA from whole blood, FFPE tissue sections, slides or shavings, fresh tissue	Schwannomatosis; Atypical Familial Rhabdoid Tumour (SMARCB1/ LZTR1)	LP 000 249
	Schwannomatosis 1 (SMARCE1)	DOC1590

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from FFPE tissue sections, slides or shavings	Quantitative detection of clinically relevant nucleic acid sequence variants for the following:	Real time PCR using Cobas Z 480* analyser DOC1185 or Digital Droplet PCR using Biorad QX200 Droplet Digital System^and:
	Colorectal cancer - tumour samples for sporadic mutations (BRAF, KRAS)	DOC1186*
	Malignant Melanoma - tumour samples (BRAF)	DOC1184*
DNA from: FFPE tissue sections, slides or shavings, and circulating tumour DNA (ctDNA)	Non-Small Cell Lung Cancer (somatic mutations, - tumour samples) (EGFR)	LP 000 257, DOC3065, DOC3312*^
RNA from FFPE samples	Glioma (KIAA1549: BRAF fusion, C11orf95: RELA fusion, EGFRvIII transcript)	DOC4206
DNA extracted from plasma	Non-Invasive Foetal Sex Determination (cffDNA)	Real time PCR using ABI Prism 7900 analyser and LP 000 253
DNA from primary samples	Mutation detection for the purpose of clinical diagnosis, carrier detection and predictive testing in the following:	PCR methods using Thermo Thermal Cycler and either: CE- Capillary electrophoresis using ABI3500XL genetic analyser, DOC955 or GE- Gel electrophoresis, LP 000 017 including:
Blood	Angelman/ Prader-Willi Syndrome (incl. UDP/parent of origin studies)	Methylation sensitive PCR, LP 000 054 using EZ DNA methylation kit, Zymo Research, LP 000 087 and GE Microsatellite analysis using CE, LP 000 054
	Breast and Ovarian cancer (BRCA1 and 2)	ARMS PCR and GE, LP 000 284 Microsatellite analysis using CE, LP 000 284
	Huntington Disease (HTT)	Fragment analysis using CE, LP 000 060

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
	Mutation detection for the purpose of clinical diagnosis, carrier detection and predictive testing in the following:	PCR methods using Thermo Thermal Cycler and either: CE- Capillary electrophoresis using ABI3500XL genetic analyser, DOC955 or GE- Gel electrophoresis, LP 000 017 including:
Blood, AF, CVS	Congenital Adrenal Hyperplasia due to 21 hydroxylase deficiency (CYP21A2)	Fragment analysis or Fluorescence PCR using CE, LP 000 082
Blood, AF, CVS	Fragile X Syndrome and associated disorders (FXTAS and FXPOI) (FMR1)	Fragment analysis (in-house or Asuragen Amplidex) using CE, LP 000 061, DOC1174
Blood, AF, CVS, blood spots	Cystic Fibrosis (CFTR)	ARMS PCR, DOC4032 or Fluorescence PCR, DOC850 using CE
FFPE tissue sections, slides or shavings	Microsatellite Instability (MSI) Testing	Fluorescence PCR using CE, DOC1332
	MLH1 promoter hypermethylation (MLH1)	Methylation using Qiagen Epitect Plus FFPE Bisulphite kit, DOC869 and pyrosequencing, DOC887
DNA from blood, FFPE tissue sections, slides or shavings, fresh tissue, AF, CVS	Neurofibromatosis type 2 (NF2; incl. LOH)	Microsatellite analysis for LOH or linkage using CE, LP 000 069 Mosaicism detection using ARMS PCR and GE, LP 000 069
	Retinoblastoma (RB1)	Microsatellite analysis for LOH or linkage using CE, LP 000 075
	Schwannomatosis (LZTR1, SMARCB1)	LP 000 249
DNA from blood and fresh tissue	Identity Testing for purposes of QC within the Genomics Medicine Workflow for Whole Genome Sequencing	Agena SNP panel test by PCR using Thermo Thermal Cyclers and Mass Spectrometry using Agena MassARRAY system Analysis using Typer 4.0.20 software SOP DOC4460

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular Haematology examinations for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from blood, AF, CVS	Detection of F8 intron 22 gene inversion mutation	Long PCR. DOC4392
	Detection of F8 intron 1 gene inversion mutation	PCR, DOC4393
	Detection of single nucleotide polymorphism of: Prothrombin G20210A FV Leiden HFE C282Y/H63D TPMT	PCR methods using Thermo thermal cyclers and mass spectrometry using Agena MassArrray DOC4434
	Detection of the following gene mutations:	PCR methods using Thermo thermal cyclers and Sanger sequencing using Applied Biosystems 3730XL sequencer and Alamut mutation surveyor software.
	F2	DOC4441
	F5	DOC4446
	F7	DOC4425
	F8	DOC4448
	F9	DOC4401
	F10	DOC4449
	F11	DOC4415
	F13A1, F13B	DOC4445
	GGCX, VKORC1	DOC4444
	TPMT	DOC4428
	Beta thalassemia (HBB and HBG2 promoter region)	DOC4409
	Alpha thalassemia (HBA1 and HBA2)	DOC4414
	Von Willebrand factor (VWF)	DOC4405
	Combined FVIII and FV deficiency analysis (LMAN1 and MCFD2)	DOC4450

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular Haematology examinations for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from blood	Detection of whole or partial gene deletions and duplications: F7 F8 F9 F10 F11 Alpha globin gene cluster deletions Beta globin gene cluster deletions	MLPA analysis using Applied Biosystems and Thermo thermal cyclers and Applied Biosystems AB 3500XL genetic analyser DOC4407
	Molecular Oncology examinations for the purposes of clinical diagnosis	
DNA or RNA from blood or bone marrow	Quantitative monitoring of gene expression for: BCR-ABL (t9;22) AML-MTG8 (t8;21) PML-RARA (t15;17) CBFB-MYH11 (Inv16 and t16;16) NPM1	Real time PCR using Applied Biosystems QuantStudio6 and DOC4453, DOC4454
DNA from blood or bone marrow	Detection of mutations in the following genes: CEBPA BCR-ABL TKD	PCR and Sanger sequencing using Thermo thermal cyclers and Applied Biosystems 3730XL genetic analysers DOC4439 DOC4443
DNA from blood or bone marrow	Detection of mutations in the following genes: CALR exon 9 FLT3-ITD JAK 2 (Exon 12) MPL (Exon 10) NPM1 (Exon 12) JAK2 V617F	PCR fragment analysis using Thermo thermal cyclers and Applied Biosystems AB 3500XL genetic analyser DOC4436 DOC4429 DOC4442 DOC4438 DOC4440 DOC4411

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Molecular Oncology examinations for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
DNA from blood or bone marrow	Targeted detection of specific variants associated with acute myeloid leukaemia	Agena SNP panel PCR using Thermo thermal cyclers, iPLEX single base extension and Agena MassArray system DOC4447
Blood and bone marrow	DNA extraction	Maxwell DNA extraction system DOC4410
	RNA extraction	Qiagen Qiacube RNA extraction system and cDNA synthesis using Superscript DOC4422, DOC4413

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Biochemical genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
	Detection of plasma and white cell lysosomal storage enzymes:	Enzyme assay using Perkin Elmer fluorescent spectrophotometer LS55 and LS30, DOC 578 and:
Leucocytes, CVS, cultured cells	Acid esterase (liposomal acid lipase) Wolman Disease/ CESD/ LAL-D	DOC630
	a-fucosidase (Fucosidosis)	DOC559
	b-galactosidase (GM 1 gangliosidosis/ MPS IV B/ Morquio B/ Galactosialidosis	DOC570
	b-glucosidase (Gaucher disease)	DOC591, DOC571, DOC602
	b-glucuronidase (MPS VII, Sly disease)	DOC572
	a-iduronidase (Hurler disease / MPS I/ Scheie disease)	DOC554
Lymphocytes, CVS, cultured cells	a-glucosidase (Pompe) confirmatory	DOC560, DOC563
Leucocytes, cultured cells	Galactocerebrosidase (screen) Krabbe leukodystrophy	DOC579
Leucocytes	N-acetyl-a-galactosaminidase (Schindler disease)	DOC566
	NCL screen (neuronal ceroid lipofuscinosis/ Batten's)	DOC2688, DOC2864
Plasma, leucocytes, CVS, cultured cells	b-hexosaminidase A (Tay-Sachs disease)	DOC573
	b-hexosaminidase A & B (Sandhoff's disease)	DOC574
	a-mannosidase (alpha Mannosidosis)	DOC565

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Biochemical genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
Plasma, leucocytes, CVS, cultured cells	Detection of plasma and white cell lysosomal storage enzymes cont'd:	Enzyme assay using Perkin Elmer fluorescent spectrophotometer LS55 and LS30, DOC 578 and:
	b-mannosidase (beta Mannosidosis)	DCO575, 603
	a-N-acetylglucosaminidase (MPS III B/ Sanfilippo III B disease)	DOC633, DOC588
Plasma, leucocytes	a-galactosidase (Fabry disease) confirmatory	DOC562
Plasma, cultured cells	Multiple hydrolases (I-cell disease/ mucolipidosis II/III) screen. [battery composition depends on sample type]	DOC574
Plasma, leucocytes	Aspartylglucosaminidase (Aspartylglucosaminuria)	DOC634
Plasma	Chitotriosidase	DOC576
Leucocytes, cultured cells	Arylsulphatase A (Metachromatic leukodystrophy)	Enzyme assay with colorimetric detection using CECIL CE1011 spectrophotometer and DOC567
Leucocytes, CVS, cultured cells	Arylsulphatase B (MPS VI, Maroteaux-Lamy)	Enzyme assay with fluorescence detection using Perkin Elmer LS 30. DOC948, DOC600
	Detection of the following:	Enzyme assay with fluorescent detection using Biotech Synergy Microplate reader and:
Leucocytes, CVS, cultured cells	Galactose-6-sulphatase (MPS IV A, Morquio A)	DOC2760
Leucocytes, CVS, cultured cells	Heparan sulphamidase (MPS III A, Sanfilippo A)	DOC577

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Biochemical genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
	Detection of the following:	Enzyme assay with fluorescent detection using Biotech Synergy Microplate reader and:
Leucocytes	NCL screen (neuronal ceroid lipofuscinosis/ Batten's)	DOC2688, DOC2864
Plasma, leucocytes, CVS, cultured cells	Iduronate sulphatase (Hunter disease/ MPS II)	DOC564
Dry blood spot	a-galactosidase (Fabry disease) screen	DOC594
	a-glucosidase (Pompe) screen	DOC595
	Detection of the following:	Scintillation counting using Canberra Packard scintillation counter and
Leucocytes, CVS, cultured cells	Galactocerebrosidase (confirmatory)	DOC631
	Sphingomyelinase (Niemann Pick type A/B, acid sphingomyelinase deficiency, ASMD)	DOC596
Plasma, leucocytes, cultured cells	Multiple sulphatases (multiple sulphatase deficiency) [composition can comprise any of several sulphatases]	Enzyme assay using Perkin Elmer fluorescent spectrophotometer LS55 (or LS30), Biotech Synergy Microplate reader, CECIL CE1011 spectrophotometer and/or Lambda 25 and DOC564, 567, 580, 948, 2760
Cultured cells	Detection of stored cholesterol(Niemann-Pick Type C)	Fluorescent Filipin staining and microscopy using DOC673
Lymphocytes, cultured cells	a-glucosidase CRIM (Pompe)	Western blot using manual method and DOC2686

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Biochemical genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
Urine, amniotic fluid	Mucopolysaccharide urine screen	A manual method of different analyses (DOC643) including isolation (DOC555), colorimetric quantitation (DOC553), 2D electrophoresis (DOC552) and thin layer chromatography of oligosaccharides and sialic acid (DOC558).
Urine	Oligosaccharide/ sialic acid screen	Manual method of thin layer chromatography using DOC558
	Detection of other Metabolic disorders:	
Urine	Amino acid screen	Derivatisation, Waters Acuity Ultra performance liquid chromatography (UPLC) and DOC696
Blood spot	Amino acid quantitation	DOC4348
Plasma, urine, CSF	Amino acid quantitation	Derivitisation, ion exchange chromatography using Biochrom 30 and DOC1195
Blood, bile, plasma, serum, urine	Acyl carnitines and free carnitine	Derivitisation, MS/MS using Waters Quattro LC TMS and DOC666
	Detection of the following:	Liquid Chromatography-Mass Spectrometry LC-MS/MS using Waters Quattro LC and
Plasma, serum	Biotinidase	DOC2365
Plasma, serum, urine	Methylmalonic acid	DOC698
Urine	Orotic acid	DOC705
Urine	Quantitative detection of Creatinine	Isotope dilution TMS using Waters TQD and Waters Quattro-LC and DOC4132
Plasma	Oxysterol	LC-MS/MS using Waters TQD and DOC2378

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Biochemical genetics examination for the purposes of clinical diagnosis	In house documented methods incorporating manufacturers' instructions where relevant
	Detection of the following:	Gas chromatography-Mass Spectrometry, GC-MS using Agilent 6890N and:
Plasma	Cholestanol	DOC647
Plasma, serum, liver, spleen, amniotic fluid, CVS	7-dehydrocholesterol	DOC647
Plasma, serum	Phytanic acid and Pristanic acid	DOC644
	Very long chain fatty acids	DOC646
Erythrocytes	Plasmalogens	DOC645
	Detection of the following:	GC-MS using Shimadzu QP2010 SE and:
Urine	Organic acids	DOC688.
Urine	Succinylacetone (to investigate tyrosinameia type 1)	DOC688
Plasma	Total homocysteine	HPLC using JASCO LC2000 and DOC652
Urine	Sugar chromatography	Manual method using thin layer chromatography and DOC4368
Urine	Sulphite	Colorimetric assay using Mquant sulphite stick and DOC2277
Blood	Beutler screening test for classical galactosaemia (galactose-1-phosphate uridyl transferase)	Enzyme assay with fluorescence detection using CAMAG spectrophotometer and DOC653
Blood spot	PKU monitoring	TMS using Waters TQD or Quattro Micro and DOC1123

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE	Biochemical genetics examination for the purposes of clinical diagnosis Newborn Screening:	In house documented methods incorporating manufacturers' instructions where relevant
Blood spot	Of IMD disorders: PKU, MCADD, MSUD, IVA, GA1, HCU	TMS using Waters TQD or Quattro Micro and DOC1410
END		