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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
, Amniotic fluid (AF), chorionic villus samples (CVS)	Chromosome analysis for: Prenatal diagnosis	Metaphase preparation, microscopic examination and analysis of G-banded karyotypes using light microscopy or Metasystems image analysis system. Cell culture, LP110 015, , 012, LP120, 014,DOC4855. G-banding analysis LP000 017, 008, 024, DOC2630
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
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, fresh tissue (including fibroblasts)	Cell culture for cryogenic storage, DNA/RNA extraction, export or immortalisation (lymphoblastoid cell lines)	DOC2285, LP160 012, 015, 018, 022, 033, 034, 035, 041, 050, 043,

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	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, bone marrow	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. DOC, 906, 907, 908, 913 COBAS FFPE extraction. DOC1300 QiaAMP FFPE extraction. DOC2637 iGENatal extraction. DOC2354 Cleaning DNA samples. DOC2047, , 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, COBAS 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	EZ1 RNA extraction kit DOC992
FFPE	RNA extraction and quality check for subsequent in house analysis (see below), referral to specialist centres and longterm storage	RecoverAll Total Nucleic Acid Isolation kit DOC4754

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	Molecular genetics examination for the purposes of clinical diagnosis (cont'd)	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 550 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines, Congenica rare disease software. DOC 4932, 4943, 2258, 2047, , 3234, , 4325, 2176, 2204, 2175, 2608, 3087, , 3210
	Cardiac disorders gene panels:	NextSeq, DOC 4945
	Metabolic disorders gene panels:	NextSeq, DOC4950
	Ophthalmic disorders gene panels:	NextSeq, DOC 4946
	Haematology disorders gene panels	NextSeq, DOC4947
	Immunology disorders gene panels	NextSeq, DOC4949
	Neurology disorders gene panels	NextSeq, DOC4951
	Hearing disorders gene panels	NextSeq, DOC4948
	Retinoblastoma (RB1)	MiSeq, LP 000 075
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 BRCA2) and germline mutations	DOC3340 DOC5719
	Cancer screening for somatic mutations (including malignant melanoma, non-small cell lung cancer, colorectal cancer and GIST)	MiSeq, NextSeq DOC3338
	Glioma / CNS cancer panel screening for somatic mutations	MiSeq, NextSeq DOC4253

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	Molecular genetics examination for the purposes of clinical diagnosis (cont'd)	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples (cont'd)	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs) (cont'd)	Next Generation Sequencing using: NextSeq 550 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines or Congenica rare disease software. DOC4932, 4943, 2258, 2047, 3234, 4325, , 2176, 2204, 2175, 2608, 3087, 3210,
DNA from FFPE tissue sections, slides or shavings (cont'd)	Glioma / CNS cancer panel screening for somatic mutations - Meningioma/schwannoma subpanel (NF2, SMARCB1, SMARCE1, SMARCA4, LZTR1)	MiSeq, NextSeq DOC4763
	Colorectal cancer - tumour samples for somatic mutations (KRAS, NRAS, BRAF, PIK3CA)	MiSeq, NextSeq DOC5682
	Colorectal cancer - tissue samples for somatic and germline mutations (APC, BMPR1A, CDH1, CTNNB1, MSH6, SMAD4, MLH1, MSH2, MUTYH, POLD1, POLE, PTEN and STK11)	MiSeq, NextSeq DOC3339
DNA from FFPE tissue sections, slides or shavings	Homologous recombination deficiency (HRD) / Mismatch repair (MMR) panel for various cancer types	Next Generation Sequencing using: NextSeq or MiSeq and QIAseq gene panel. DNA analysis using BWA bioinformatics pipelines. DOC3339, DOC3340, DOC4847, DOC4888
RNA from FFPE samples	Lung cancer fusion gene panel (ALK, ROS1, NTRK3, RET) Sarcoma and paediatric fusion panel	QIAseq Targeted RNAScan Panel, NextSeq 550, MiSeq, DOC5672 DOC4764. Analysis using CLC Genomics Workbench, DOC4775

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	Molecular genetics examination for the purposes of clinical diagnosis (cont'd)	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples (cont'd)	Mutation detection of sequence variants causing genetic diseases and disorder: Single Nucleotide Variants (SNVs) Indels Copy Number Variations (CNVs) (cont'd)	Next Generation Sequencing using: NextSeq 550 or MiSeq and SureSelect XT, Nextera XT or GeneRead/ TruSeq gene panels. Data Analysis using: BWA bioinformatics pipelines or Congenica rare disease software.. DOC4932, 4943 2258, 2047, , 3234, 4325, , 2176, 2204, 2175, 2608, 3087, , 3210
DNA from blood or FFPE tissue sections, slides, shavings, fresh tissue, AF, CVS	Neurofibromatosis type 2 (NF2)	MiSeq, LP 000 069
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 DOC4983, DOC4901,, DOC1585, LP 000 236 Multiplex Ligation-dependant Probe Amplification (MLPA) or MS-MLPA using ABI3500XL analyser and DOC4983, DOC4901, DOC4472, DOC1030, LP 000 162 DOC4616 Digital droplet PCR (ddPCR) using BioRad Automated Droplet Generator, Droplet Reader and DOC4304

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	Molecular genetics examination for the purposes of clinical diagnosis (cont'd)	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples (cont'd)	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, DOC4983, DOC4901, DOC1585, LP 000 236
DNA from blood	Angelman Syndrome/ Prader-Willi Syndrome (UBE3A, SNRPN)	LP 000 054
DNA from blood, AF, CVS	Possible X-linked Retinitis Pigmentosa R33 (RPGR, exon ORF15 only)	DOC2245
DNA from blood, AF, CVS, blood spots	MCADD common mutation (ACADM)	LP 000 268
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:	PCR using Thermo Thermal Cycler and Pyromark PQQ 96MA pyrosequencing, DOC887 and:
DNA from FFPE tissue sections, slides or shavings	Colorectal cancer (MLH1 hypermethylation and BRAF codon 600)	DOC3389, DOC869,
	Malignant melanoma (BRAF, NRAS)	DOC6047
	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 (cont'd)	Molecular genetics examination for the purposes of clinical diagnosis (cont'd)	In house documented methods incorporating manufacturers' instructions where relevant
DNA from primary samples (cont'd)	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 DOC4472 , DOC1030, LP 000 162 and:
DNA from blood	Angelman syndrome/ Prader-Willi Syndrome (UBE3A, SNRPN)	LP 000 054
	Breast and Ovarian cancer (BRCA1 and BRCA2)	DOC4335
DNA from primary samples	Familial Adenomatous Polyposis (APC) (FAP) and MUTYH-Associated Polyposis (MUTYH) (MAP)	DOC4335
	Lynch Syndrome (HNPCC, familial colorectal cancer) (MLH1, MSH2, MSH6)	DOC4335
	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
	Meningiomas (SMARCE1)	DOC1590

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	Molecular genetics examination for the purposes of clinical diagnosis (cont'd)	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)	DOC1185 *
	Malignant Melanoma - tumour samples (BRAF)	DOC6047, 1185
DNA from: FFPE tissue sections, slides or shavings, and circulating tumour DNA (ctDNA)	Non-Small Cell Lung Cancer (somatic mutations, - tumour samples) (EGFR)	DOC4881, DOC1185*, DOC4344^, DOC4206^
RNA from FFPE samples	Glioma (KIAA1549: BRAF fusion, C11orf95: RELA fusion, EGFRvIII transcript)	DOC4253, DOC4344^, 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

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	Molecular genetics examination for the purposes of clinical diagnosis (cont'd)	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: (cont'd)	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	Fragile X Syndrome and associated disorders (FXTAS and FXPOI) (FMR1)	Fragment analysis (in-house or Asuragen Amplidex) using CE, LP 000 061, DOC1174
blood spots	Cystic Fibrosis (CFTR)	ARMS PCR, LP 000 238 DOC4873 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

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	<u>Molecular Haematology examinations for the purposes of clinical diagnosis</u>	In house documented methods incorporating manufacturers' instructions where relevant
	Detection of F8 intron 1 gene inversion mutation	PCR, DOC4910 DOC4947
	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.
	F5	DOC4947, DOC1585 DOC4432, DOC1585
	Beta thalassemia (HBB and HBG2 promoter region) Alpha thalassemia (HBA1 and HBA2) Von Willebrand factor (VWF)	DOC4432, DOC1585 DOC4947, DOC1585
DNA from blood	Detection of whole or partial gene deletions and duplications: 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 LP 000 162, analysed using Coffalyser Net software, DOC4616 DOC4432

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	<u>Molecular Oncology examinations for the purposes of clinical diagnosis</u>	In house documented methods incorporating manufacturers' instructions where relevant
DNA or RNA from blood or bone marrow	Quantitative monitoring of gene expression for: RUNX1/RUNX1T1 PML::-RARA (t15;17) CBFB::-MYH11 nv(16) and t16;16) NPM1	Real time PCR using Applied Biosystems QuantStudio6 and DOC4458, 5974
DNA from blood or bone marrow	Detection of mutations in the following genes: NPM1 (Exon 12) B and T cell Clonality (Ig, TCR) FLT3 ITD/TKD	PCR fragment analysis using Thermo thermal cyclers and Applied Biosystems AB 3500XL genetic analyser DOC4440 DOC4866 DOC4877

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	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 (cont'd)	Biochemical genetics examination for the purposes of clinical diagnosis (cont'd)	In house documented methods incorporating manufacturers' instructions where relevant
	Detection of plasma and white cell lysosomal storage enzymes (cont'd):	Enzyme assay using Perkin Elmer fluorescent spectrophotometer LS55 and LS30, DOC 578 and:
Plasma, leucocytes, CVS, cultured cells	b-mannosidase (beta Mannosidosis)	DOC 575, 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 Biotek Synergy HTX 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 (cont'd)	Biochemical genetics examination for the purposes of clinical diagnosis (cont'd)	In house documented methods incorporating manufacturers' instructions where relevant
	Detection of the following:	Enzyme assay with fluorescent detection using Biotek Synergy HTX 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), Biotek 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 (cont'd)	Biochemical genetics examination for the purposes of clinical diagnosis (cont'd)	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 TQD and
Plasma, serum	Biotinidase	DOC2365
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 (cont'd)	Biochemical genetics examination for the purposes of clinical diagnosis (cont'd)	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 the Chromsystems reagent kit, 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 TQS Micro Tandem MS and DOC1123

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS/TISSUE (cont'd)	Biochemical genetics examination for the purposes of clinical diagnosis (cont'd) 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 TQS Micro Tandem MS and DOC1410
END		