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Viapath Analytics LLP

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Genetics Centre
5th Floor Tower Wing
Guy's Hospital
London
SE1 9RT

Contact: Jeremy Skinner
Tel: +44 (0)207 188 7373
E-Mail: jeremy.skinner@viapath.co.uk
Website: www.viapath.co.uk

Testing performed by the Organisation at the locations specified below

Locations covered by the organisation and their relevant activities

Laboratory locations:

Location details		Activity	Location code
Molecular Genetics 5th Floor Tower Wing & 7th Floor Borough Wing (analysis only) Guy's Hospital London SE1 9RT	Jeremy Skinner (contact details above)	Monogenics, Prenatal and Reproductive Genetics and Developmental Disorder testing for a range of genetic disorders.	GL
Pre-implantation Genetic Diagnosis (PGD) Laboratory 11th Floor Tower Wing Guy's Hospital London SE1 9RT	Jeremy Skinner (contact details above)	Molecular Genetics testing of pre-implantation embryos to identify familial mutations or abnormalities arising from parental chromosome rearrangements	PGD
Biochemical Genetics Laboratory 5th Floor Tower Wing Guy's Hospital London SE1 9RT	Jeremy Skinner (contact details above)	Enzymology, metabolite analysis and molecular genetic testing for a range of genetic biochemical disorders	BG
Molecular Oncology Unit 4th Floor Southwark Wing Guy's Hospital London SE1 9RT	Jeremy Skinner (contact details above)	Molecular oncology testing	MOU
Epidermolysis Bullosa Laboratory St. John's Institute of Dermatology Guy's Hospital Great Maze Pond London SE1 9RT	Jeremy Skinner (contact details above)	Molecular Pathology Frozen Sections Immunofluorescence	GEB

DETAIL OF ACCREDITATION

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS	Molecular Genetics		
	Detection of nucleic acid sequence variants and/or copy number changes for the purpose of clinical diagnosis	Documented in- house methods for sequencing, genotyping or electrophoresis supported by extraction and amplification as listed below.	
Blood Saliva Paraffin embedded tissue Buccal cells Dried bloodspots Muscle		DNA Extraction In-house procedures: LP-D-15 LP-D-24 LP-D-26 LP-D-29 LP-D-164 LP-D-165 LP-D-234 using commercial kits and Chemagen 360-D automated DNA extractor	GL
Hair roots Muscle		RNA Extraction In-house procedures: LP-D-165 using commercial kits and manual extraction	GL
Blood Bone marrow FFPE tissue Cell suspension (peripheral blood and bone marrow cells stored in RLT buffer)		Nucelic Acid Extraction In house procedures: LP-MO-63 Using commercial kits and QIAsymphony semi-automated extractor	GL
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source Dried bloodspots		Multiple Displacement Amplification Whole genome amplification of low copy number of cells using in-house procedures: LP-D-53 in conjunction with manufacturer's instructions using: MJ Research Dyad thermal cycler	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants and/or copy number changes as indicated for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: Alport syndrome (COL4A3, COL4A4, COL4A5, COL4A6, NPHS2) Alzheimer disease (PS1) Amyotrophic lateral sclerosis (SOD1, C9orf72, TARDBP, FUS) Ashkenazi screen (HEXA c.1274_1277dupTATC p.(Tyr427fs), c.805G>A p.(Gly269Ser), c.1421+1G>C; IKBKAP c.2087G>C p.(Arg696Pro), c.2204+6T>C; ASPA c.693C>A p.(Tyr231X), c.854A>C, p.(Glu285Ala) c.914C>A p.(Ala305Glu); FANCC c.456+4A>T; MCOLN1 c.1_788del, c.406-2A>G; SMPD1 c.911T>C p.(Leu304Pro), c.1493G>T p.(Arg498Leu), c.996delC p.(Phe333fs) ; G6PC c.247C>T p.(Arg83Cys); BLM c.2207delATCTGAinsTAGATTC; CFTR c.1521_1523delCTT	PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping Using In-house procedures: LP-D-2 LP-D-3 LP-D-21 LP-D-23 LP-D-25 LP-D-32 LP-D-51 LP-D-105 LP-D-162 LP-D-163 LP-D-191 LP-D-194 LP-D-258 LP-D-530 and Automated or manual methods in conjunction with manufacturer's instructions using: Gene Sequence Manager software NanoDrop® ND-1000 Spectrophotometer M2 Plate reader Hamilton STARlet robotBiorad Tetrad 2 MJ Research Dyad thermal cycler Nanodrop II nanodispenser Beckman Coulter Biomek NX ABI 3730 DNA Analyser	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants and/or copy number changes as indicated for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) Ashkenazi screen (cont'd) p.(Phe508de)l, c.1624G>Tp.(Gly542X), c.3846G>A p.(Trp1282X),c.3909C>G p.(Asn1303Lys), c.3717+12192G>A , c.1585-1G>A.) Breast/ovarian cancer (BRCA1, BRCA2, TP53, STK11, CDH1, PTEN) Brown-Vialetto-Van Laere syndrome (SLC52A3, SLC52A2, SLC52A1) Congenital muscular dystrophy (COL6A1, COL6A2, COL6A3, COL12A1, FKRP, FKTN, LAMA2, LARGE, POMGNT1, POMGNT2, POMT1, POMT2, COL4A1, COL4A2, DAG1, DPM1, DPM2, DPM3, DOLK, ISPD, GMPPB, B3GALNT2, CHKB, PLEC1, SIL1, B3GNT1, POMK, ITGA7, ITGA9, TMEM5, MICU1)	PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping (cont'd)	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants and/or copy number changes as indicated for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) Congenital myopathy (ACTA1, BIN1, CFL2, DNM2, KBTBD13, MTM1, MYH2, MYH3, MYH7, MYH8, NEB, RYR1, SEPN1, TNNI2, TNNT1, TNNT3, TPM2, TPM3, ORAI1, STIM1, ECEL1, CCDC78, KLHL41, KLHL40, DNA2, SLC35A3, MYBPC1, PIEZO2, ZC4H2, VPS33B, LAMP2, VMA21, STAC3, LMOD3, MEGF10, EPG5) Cystic fibrosis (CFTR common variants) Direct Test (confirmation of Whole Exome research results e.g. DDD project – varied genes) Duchenne/Becker muscular dystrophy (dystrophin) Familial dysautonomia (IKBKAP)	PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping (cont'd)	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants and/or copy number changes as indicated for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) Familial hypercholesterolaemia (LDLR, APOB, LDLRAP1, PCSK9) Fanconi anaemia (FANC, FANCA common variants) Fragile X syndrome (FMR1) Glycogen storage disease (AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYG2, GYS1, GYS2, LAMP2, LDHA, LIPA, NHLRC1, PFKL, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG1, PHKG2, PPP1R2, PRKAG2, PYGL, PYGM, SLC37A4, SLC2A2) Hajdu-Cheney syndrome (NOTCH2) Hereditary spastic paraplegia (ATL, NIPA1, REEP1, SPAST) Huntington disease (HTT)	PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping (cont'd)	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants and/or copy number changes as indicated for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) Lynch syndrome (MSI analysis) Medium chain CoA dehydrogenase deficiency (ACADM) Melanoma (BRAF exon 15) Methylmalonic acidemia (ABCD4, ACSF3, AMN, CBS, CD320, CUBN, GIF, HCFC1, IVD, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, SLC46A1, SUCLA2, TCN2) Niemann-Pick disease (NPC1, NPC2) Nucleotide Excision Repair disease (MPLKIP, DDB1, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, GTF2H5, POLH, XPA, XPC, LIG4, XRCC4, NHEJ1, UVSSA)	PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping (cont'd)	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants and/or copy number changes as indicated for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) Ohdo syndrome, Genitopatellar syndrome (KAT6B) Primary pulmonary hypertension (BMPR2, ACVRL1, ENG) Skeletal dysplasia; Stickler, Achondrogenesis type II and hypochondrogenesis, SEDC (Spondyloepiphyseal dysplasia congenital) and Kniest dysplasial, Achondroplasia, Thantophoric dysplasia (COL2A1, FGFR3) Spinal muscular atrophy (SMN1) SMA with respiratory distress (IGHMBP2) Tay-Sachs disease (HEXA) Urea cycle defects (ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15) Vici syndrome (EPG5)	PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping (cont'd)	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants and/or copy number changes as indicated for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) X-inactivation X-linked Infantile Spasm syndrome (ARX) X-linked SMA (UBE1) X-Linked VACTERL with hydrocephalus syndrome (FANC B)	PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping (cont'd)	GL
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: Combined panel for: Alport syndrome (COL4A3, COL4A4, COL4A5, COL4A6, NPHS2) Cystinuria (SLC3A1, SLC7A9) Familial hypercholesterolaemia (LDLR, APOB, LDLRAP1, PCSK9) Breast/ovarian cancer (BRCA1, BRCA2, CHEK2 1100deC, PALB2, TP53)	Next Generation Sequencing and data analysis In-house procedures: 59 LP-D-351 LP-D-388 LP-D-389 DOC133 LP-D-273 In conjunction with manufacturer's instructions using: Twist Human Core Exome EF Multiplex kitQubit fluorometer Agilent NGS Workstation Life Tech Veriti Model# 9902 Savant Speed Vac Centrifuge Agilent TapeStation 2200 Illumina NextSeq 550 Qiagen Ingenuity (data analysis)	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	<u>Molecular Genetics (cont'd)</u> Detection of nucleic acid sequence variants for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) Congenital myopathy / Congenital muscular dystrophy (COL6A1, COL6A2, COL6A3, COL12A1, FKRP, FKTN, LAMA2, LARGE1, POMGNT1, POMGNT2 (GTDC2), POMT1, POMT2, COL4A1, COL4A2, DAG1, DPM1, DPM2, DPM3, DOLK, ISPD, GMPPB, B3GALNT2, CHKB, PLEC, SIL1, B4GAT1 (B3GNT1), POMK (SGK196), ITGA7, THEM5, MICU1, ACTA1, BIN1, CFL2, DNM2, KBTBD13, MTM1, MYH2, MYH3, MYH7, MYH8, NEB, RYR1, SEPN1, TNNI2, TNNT1, TNNT3, TPM2, TPM3, ORAI1, STIM1, ECEL1, CCDC78, KLHL41 (KBTBD10), KLHL40 (KBTBD5), MYBPC1, PIEZO2, ZC4H2, VPS33B, LAMP2, VMA21, STAC3, LMOD3, MEGF10, EPG5, TTN, ADAMTS15, CACNA1S, CNTN1, DOK7, GOLGA2, HACD1 (PTPLA), INPP5K, KLHL9, MSTO1, MTMR14, MYBPC3, MYL1, MYO18B, NUP88, SCN4A, SPEG, SRPK3, THEM38A, TRAPPC11)	<u>Next Generation Sequencing and data analysis (cont'd)</u>	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) Glycogen storage disease (AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYG2, GYS1, GYS2, LAMP2, LDHA, LIPA, NHLRC1, PFKL, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG1, PHKG2, PPP1R2, PRKAG2, PYGL, PYGM, SLC37A4, SLC2A2) Methylmalonic acidemia (ABCD4, ACSF3, AMN, CBS, CD320, CUBN, GIF, HCFC1, IVD, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, SLC46A1, SUCLA2, TCN2)	Next Generation Sequencing and data analysis (cont'd)	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of nucleic acid sequence variants for the purpose of clinical diagnosis. Name of disease with name of analysed gene(s) in brackets: (cont'd) Nucleotide Excision Repair disease (MPLKIP, DDB1, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, GTF2H5, POLH, XPA, XPC, LIG4, XRCC4, NHEJ1, UVSSA, CDKN2A, PTCH1) Urea cycle defects (ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15)	Next Generation Sequencing and data analysis (cont'd)	GL
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	CFTR	Fluorescent Amplification Refractory Mutation System (ARMS) multiplex PCR In-house procedures LP-D-422 LP-D-423 In conjunction with manufacturer's instructions using: Elucigene kit ABI 3730 DNA analyser GeneMarker analysis software	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of pathogenic gene variants	Flexible scope protocols: QP-G-24 in conjunction with documented in-house methods for Whole Exome Sequencing by Next Generation Sequencing using: Illumina NextSeq Confirmation by Sanger Sequencing using: ABI3730 genetic sequencer. Supported by extraction and amplification as listed above Flexible scope limited to the application of these methods for the detection of additional pathogenic gene variants listed in MF-D-13 New Test Log	GL
Embryonic cells Buccal cells DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Detection of pathogenic gene variants for inherited genetic disorders for the purpose of clinical diagnosis	Flexible scope protocols: QP-G-24 in conjunction with documented in-house methods for haplotype analysis using STR microsatellite markers or in combination with genotyping supported by in-house primer design, extraction and amplification methods as listed below Flexible scope limited to the application of these methods for the detection of additional pathogenic gene variants listed in LF-D-494 PGD Diseases	PGD

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
Embryonic cells Buccal cells DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Detection of pathogenic gene variants for inherited genetic disorders for the purpose of clinical diagnosis	Multiple Displacement Amplification whole genome amplification of low copy number of cells using in-house procedures: LP-D-109 LP-D-130 LP-D-131 LP-D-132 In conjunction with manufacturer's instructions using: PCR6 hoods, PTC200 DNA Engine, MJ Dyad thermocycler	PGD
		Polymerase chain reaction (PCR) using in-house procedures: LP-D-112 LP-D-231 LP-D-241 Manual or automated set up in conjunction with manufacturer's instructions using Cas1200 liquid handler followed by PCR using Biorad C1000 thermocyclers.	PGD
		Detection of fluorescently labelled PCR products using in-house procedures: LP-D-32 in conjunction with manufacturer's instructions using ABI 3730 DNA Analyser	PGD

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular Genetics (cont'd)		
Embryonic cells Buccal cells DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Sickle cell anaemia [HBB]	Multiple Displacement Amplification, PCR and Detection of fluorescently labelled PCR products (as above) plus Amplification Refactory Mutation System (ARMS) using in-house procedures: LP-D-214 in conjunction with manufacturer's instructions using Biorad C1000 thermocyclers	PGD
	Spinal muscular atrophy [SMN1]	Multiple Displacement Amplification, PCR and Detection of fluorescently labelled PCR products (as above) using in-house procedures: LP-D-198 in conjunction with manufacturer's instructions using Biorad C1000 thermocyclers	PGD
Embryonic cells DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Detection of inherited genetic disorders by SNP haplotype for the purpose of clinical diagnosis	Next Generation Sequencing for SNP haplotype analysis using in-house procedures: LP-D-PGD-OnePGT library prep LP-D-PGD-OnePGT data analysis In conjunction with manufacturer's instructions using: Agilent OnePGT Technology: Qubit fluorometer Sage PippinHT Agilent TapeStation 2200 Illumina NextSeq 550 Agilent Alissa (data analysis)	PGD, GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUES AND FLUIDS (cont'd)			
Whole blood Chorionic villi Amniotic fluid & cells Skin sections Saliva	<u>Molecular Pathology</u> Screening for the following genes/transcripts: KRT5 KRT14 TGM5 DST1e(BP230) PLEC1 EXPH5 CD151 COL17A1 ITGA6 ITGB4 ITGA3 ITGB1 LAMB3 LAMC2 LAMA3 COL7A1 DSC3 FERMT1 (KIND1) DSP1 JUP PKP1 DSG1 DSG2 DSG4 KRT1 KRT9 KRT10 ABCA12 TGM1 PORCN1 TP63 ECM1 SLC39A4 CX26 CX30 CX30.3 CX31 EDA EDAR EDARADD WNT10A	Examination using in-house procedures (listed) in conjunction with manufacturer's instructions for the following methods (where relevant) Manual DNA & RNA extraction using commercial kits and: DME-SOP-47 DME-SOP-8 DME-SOP-26 DME-SOP-53 Manual process for Prenatal diagnosis using: DME-SOP-45 Manual process for Maternal Cell Contamination using: DME-SOP-45 Polymerase Chain Reaction (PCR) using: ABI geneAmp 9700 thermal cycler and: DME-SOP-43 Automated capillary electrophoresis Sanger sequencing Using: ABI 3730 gene analyser* Chromas Pro software and: DME-SOP-23 DME-SOP-48 DME-SOP-56 DNA quantification using Qubit 4 and: DME-SOP-59	GEB

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUES AND FLUIDS (cont'd)		Examination using in-house procedures (listed) in conjunction with manufacturer's instructions for the following methods (where relevant)	GEB
Whole blood Chorionic villi Amniotic fluid & cells Skin sections Saliva	creening for the above genes/transcripts (cont'd)	Microsatellite analysis using: DME-SOP-50 Restriction Enzyme Digest PCR using: using: ABI geneAmp 9700 thermal cycler and: DME-SOP-43 DME-SOP-50 Restriction enzyme digest electrophoresis Sanger sequencing using: ABI 3730 gene analyser* Chromas Pro software and: DME-SOP-23 DME-SOP-48 DME-SOP-50 DME-SOP-56 Data analysis & reporting using: DME-SOP-56	
	Immunodermatology	Examination using in-house procedures (listed) in conjunction with manufacturer's instructions for the following methods (where relevant)	GEB
Skin biopsies		Manual processing Cryostat sectioning using Leica CM1950 cryostat Photography using Nikon Eclipse E600 microscope and: DME-SOP-42	GEB

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUES AND FLUIDS (cont'd)		Examination using in-house procedures (listed) in conjunction with manufacturer's instructions for the following methods (where relevant)	GEB
Processed, frozen & sectioned skin biopsies	Mapping of the following antigens:	Manual indirect immunofluorescence by microscopy using: DME-SOP-42 and: Nikon Eclipse E600 microscope and the following antibodies/clones:	GEB
	Laminin 5 (Laminin-332)	GB3	
	Type VII collagen	LH7.2	
	Type IV collagen	COL-94	
	Keratin 14	LL002	
	Plectin C terminal	pC185	
	Plectin N terminal	pN645	
	Type XVII collagen	NC16A3	
	Beta4 integrin (CD104)	450-9D	
	Alpha6 integrin	GOH3	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUES AND FLUIDS (cont'd)	Immunodermatology	Examination using in-house procedures (listed) in conjunction with manufacturer's instructions for the following methods (where relevant)	GEB
Processed, frozen & sectioned skin biopsies	Mapping of the following antigens:	Manual indirect immunofluorescence by microscopy (cont'd) using the following antibodies/clones:	GEB
	Keratin 5	XM26	
	Keratin 10	DE-K10	
	Keratin 16	LL0025	
	Desmoplakin 1	DP2.17	
	Plakophilin-1	PP1-5C2 (15F11)	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd) 1. Plasma 2. Chorionic villus 3. Skin fibroblasts 4. Amniotic cells 5. Dried bloodspots 6. Leucocytes 7. Tissues 8. Amniotic fluid 9. Serum 10. Liver 11. Urine 12. Lithium heparin blood	<u>Enzymology and metabolite analysis</u> Detection of, for the purpose of clinical diagnosis:		
11	Oligosaccharides (Glycoprotein storage disorders)	Thin layer chromatography In-house procedures: LP-B-109	BG
11	Glycosaminoglycans (GAGs) (diagnosis of mucopolysaccharidosis disorders)	Colorimetric enzyme assay Quantitative measurement using in-house procedures: LF-B-12 LP-B-44 in conjunction with manufacturer's instructions using: Roche Cobas Bio	BG
8, 11	Glycosaminoglycans (GAGs) (mucopolysaccharidosis disorders)	2D electrophoresis In-house procedures: LP-B-45	BG
2, 3, 4	Free cholesterol accumulation (Niemann Pick C disease)	Filipin staining and fluorescence microscopy In-house procedures: LP-B-93	BG

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Enzymology and metabolite analysis (cont'd)		
Sample types 1 – 12 as listed above	Detection of, for the purpose of clinical diagnosis: (cont'd)	Fluorimetric enzyme assay using in-house procedures below in conjunction with manufacturer's instructions using: Perkin Elmer LS55 Fluorimeter	BG
2, 3, 4, 6	N-acetylgalactosamine -6-sulphatase (MPS IVA – Morquio A)	LP-B-77	
2, 3, 4	N-acetylglucosamine-6-sulphatase (MPS IIID – Sanfilippo D)	LP-B- 145	
1, 2, 3, 4, 6	α-N-acetylgalactosaminidase (Schindler's disease)	LP-B-57	
1, 2, 3, 4	α-N-acetylglucosaminidase (MPS IIIB – Sanfilippo B)	LP-B- 116	
2, 3, 4	N-acetyl-neuraminidase (Sialidosis)	LP-B-71	
2, 3, 4, 6	Arylsulphatase B (MPS VI – Maroteaux Lamy)	LP-B-99	
1, 2, 3, 4, 6	Aspartylglucosaminidase (Aspartylglucosaminuria)	LP-B-78	
1, 2, 3, 4, 6	α-fucosidase (Fucosidosis)	LP-B-65, LP-B-66	
1, 9	Chitotriosidase (marker of macrophage activity: grossly elevated in Gaucher disease/ can be raised in other disorders)	LP-B-62	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Enzymology and metabolite analysis (cont'd)		
Sample types 1 – 12 as listed above	Detection of, for the purpose of clinical diagnosis: (cont'd)	Fluorimetric enzyme assay using in-house procedures below in conjunction with manufacturer's instructions using: Perkin Elmer LS55 Fluorimeter	BG
12	Galactose 1-phosphate uridyl transferase	LP-B-47	
2, 3, 4, 6	Glucosamine N-acetyl transferase (MPS IIIC – Sanfilippo C)	LP-B-76	
2, 3, 4, 6, 7	α-glucosidase (GSD II, Pompe disease)	LP-B-88, LP-B-113	
2, 3, 4, 6	β-glucosidase (Gaucher disease)	LP-B-61	
1, 2, 3, 4, 6, 9	β-glucuronidase (MPS VII- Sly)	LP-B-55	
2, 3, 4, 6	Heparan sulphamidase (MPS IIIA – Sanfilippo A disease)	LP-B-74 (119)	
1, 2, 3, 4, 6, 9	Hexosaminidase A (MUGS) (GM2 gangliosidosis -Tay Sachs disease)	LP-B-52	
1, 2, 3, 4, 6, 8, 9	Hexosaminidase A & B Total (GM2 gangliosidosis- Sandhoff disease)	LP-B-84	
1, 2, 3, 4, 6, 9	Iduronate sulphatase (MPS II – Hunter disease)	LP-B-73	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Enzymology and metabolite analysis (cont'd)		
Sample types 1 – 12 as listed above	Detection of, for the purpose of clinical diagnosis: (cont'd)	Fluorimetric enzyme assay (cont'd)	BG
2, 3, 4, 6	α-iduronidase (MPS I – Hurler /Scheie disease)	LP-B-72	
1, 2, 3, 4, 6, 9	α-mannosidase (α-mannosidosis)	LP-B-67, LP-B-68	
1, 2, 3, 4, 6, 9	β-mannosidase (β-mannosidosis)	LP-B-56	
2, 3, 4, 6	Palmitoyl protein thioesterase (Infantile neuronal ceroid lipofuscinosis – NCL1)	LP-B-79	
2, 3, 4, 6	Tripeptidyl peptidase I (Classic late infantile neuronal ceroid lipofuscinosis – NCL2)	LP-B-80	
Sample types 1 – 12 as listed above	Detection of:	Fluorimetric enzyme assay using in-house procedures below in conjunction with manufacturer's instructions using: Perkin Elmer LS55 Fluorimeter and Fluostar Optima platereader	BG
1, 2, 3, 4, 5, 6	Acid esterase / lysosomal acid lipase. (Wolman/ cholesterol ester storage disease: LIPA deficiency)	LP-B-94	
1, 2, 3, 4, 5, 6, 9	α-galactosidase A (Fabry disease)	LP-B-69, LP-B-121, LP-B-138	
2, 3, 4, 5, 6	β-galactosidase (GM1 gangliosidosis & Morquio B disease)	LP-B-138, LP-B-60	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Enzymology and metabolite analysis (cont'd)		
Sample types 1 – 12 as listed above	Detection of, for the purpose of clinical diagnosis: (cont'd)	Fluorimetric enzyme assay using in-house procedures below in conjunction with manufacturer's instructions using: Roche Cobas Bio	BG
6, 9	% Hexosaminidase A by heat inactivation for Tay-Sachs carrier testing	LP-B-50	
Sample types 1 – 12 as listed above	Detection of:	Colorimetric enzyme assay using manufacturer's instructions for: Fluostar Optima platereader and in-house procedures:	BG
2, 3, 4, 6, 7, 10, 12	Protein Determination	LP-B-3	
2, 3, 4, 6	Arylsulphatase A (Metachromatic Leucodystrophy)	LP-B-58	
Sample types 1 – 12 as listed above	Detection of:	Colorimetric enzyme assay using manufacturer's instructions for: Roche Cobas Bio and in-house procedures:	BG
11	Creatinine	LP-B-4	
10	Carbamyl phosphate synthetase (CPS - Carbamyl phosphate synthetase deficiency)	LP-B-96	
1, 8, 9	Arylsulphatase A: screen for I-cell (mucolipidosis II/ III)	Qualitative assay LP-B-81 (115)	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Enzymology and metabolite analysis (cont'd)		
Sample types 1 – 12 as listed above	Detection of, for the purpose of clinical diagnosis: (cont'd)	Colorimetric enzyme assay (cont'd)	BG
10	Ornithine transcarbamylase (OTC - Ornithine transcarbamylase deficiency)	LP-B-96	
Sample types 1 – 12 as listed above	Detection of:	Radiolabelled enzyme assay using manufacturer's instructions for Scintillation counter Hidex 300SL and in-house procedures:	BG
2, 3, 4	14C-citrulline incorporation (citrullinaemia and argininosuccinic aciduria)	LP-B-94	
2, 3, 4	Branched chain ketoacid decarboxylase (MSUD Maple syrup urine disease)	LP-B-95	
2, 3, 4, 6	Dihydroxyacetone phosphate acyl transferase (Zellweger syndrome /other generalised peroxisomal disorders)	LP-B-89	
2, 3, 4, 6	Galactocerebrosidase (Krabbe leucodystrophy)	LP-B-59	
12	Galactokinase (Galactokinase deficiency)	LP-B-49	
2, 3, 4	3-methylcrotonyl-CoA carboxylase (3-methylcrotonyl-CoA carboxylase/ multiple carboxylase deficiency)	LP-B-97	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Enzymology and metabolite analysis (cont'd)		
Sample types 1 – 12 as listed above	Detection of, for the purpose of clinical diagnosis: (cont'd)	Radiolabelled enzyme assay (cont'd)	BG
2, 3, 4	Propionyl-CoA carboxylase (Propionic acidaemia)	LP-B-97	
2, 3, 4	Pyruvate carboxylase (Pyruvate carboxylase deficiency)	LP-B-97	
2, 3, 4, 6	Sphingomyelinase (Niemann Pick A/B disease)	LP-B-63	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Prenatal and reproductive genetics		
Amniotic fluid Chorionic villus Blood Fetal blood Solid tissues / products of conception DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Aneuploidy detection	QF-PCR using in-house procedures: LP-G-2/ LP-G-PCR DNA extraction LP-G-6/LP-G-aCGHQF-PCR tissue samples LP-G-1/LP-G-PCR set up LP-G-322/LP-G-PCR Run 3500 LP-G-8/LP-G-PCRAnalysisTrisomy and Chemagen 360-D automated DNA extractor Dyad & Eppendorf Thermocyclers ABI 3500 Genetic analyser	GL
Amniotic fluid Chorionic villus Blood Products of conception DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Genome-wide copy number variation	Array CGH (aCGH) using in-house procedures: LP-G-199/LP-G-Prenatal aCGH analysis DOC190/Puregene DNA Extraction and Agilent microarray scanner Agilent Genomic Workbench	GL
Blood Fetal blood samples Chorionic villus Amniotic fluid	Karyotype	Culture and karyotype analysis using in-house procedures: Prenatal: LF-G-138/LP-G-PNHAR LP-C-102/LP-C-AFSETUP LP-G-194/LP-G-PNCVS Processing Postnatal: LP-C-70/LP-C-ProcBloodCult, LP-C-72/LPC-ProcHarvBlood, LP-C-125/LP-CBloodGBanding and Ikaros software on Metasystems	GL
Blood Fetal blood samples Chorionic villus Amniotic fluid	Chromosome breakage studies	Culture and chromosome breakage screening using in-house procedures: DOC3/LP-C-Atbldsu DOC4/LP-C-Atbldharv DOC9/LP-C-FABldDEB DOC10/LP-C-FABldHarv DOC11/LP-C-FABldSU	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	<u>Prenatal and reproductive genetics</u>		
Blood DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Y chromosome microdeletion	PCR analysis using in-house procedures: LP-G-1/LP-G-PCR set up LP-G-14/LP-G-Ydel and Dyad and Eppendorf Thermocyclers ABI 3500 Genetic analyser	GL PGD
Blood Fetal blood samples Chorionic villus Amniotic fluid DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Uniparental disomy of chromosomes 7, 14, 15	PCR analysis using in-house procedures: LP-G-13/LP-G-PCR UPD and Dyad & Eppendorf Thermocyclers ABI 3500 Genetic analyser	PGD
Blood Fetal blood samples Chorionic villus Amniotic fluid Solid tissues / products of conception DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Uniparental disomy of chromosomes 7, 14, 15	PCR analysis using in-house procedures: LP-G-2001/LP-G-MCCtestingPCR set up LP-G-13/LP-G-PCR UPD and Dyad & Eppendorf Thermocyclers ABI 3500 Genetic analyser	GL PGD
Blood Fetal blood samples Chorionic villus Amniotic fluid Solid tissues/ products of conception DNA extracted in-house from the sample types listed here or received as primary sample type from external source	Maternal cell contamination	PCR analysis using in-house procedures: LP-G-1/LP-G-PCR set up LP-G-200/LP-G-MCCtesting and Dyad & Eppendorf Thermocyclers ABI 3500 Genetic analyser	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	<u>Developmental disorder genetics</u>		
Blood Saliva Buccal swab Tissues	Genome-wide copy number variation	aCGH using in-house procedures: LP-142 / LP-C-LabellingWithBravo LP-149 / Array Clean-up and Hyb DOC188 / Array Wash and Scan LP-154 / Array Analytics DOC50 / Array Analysis LP-157 / Checking Array Results and Agilent microarray scanner Agilent Genomic Workbench	GL
	<u>Chromosome rearrangement PGD</u>		
Trophectoderm biopsy samples from embryos	Unbalanced products from chromosome rearrangements	aCGH using in-house procedures: LP-142 / LP-C-LabellingWithBravo LP-149 / Array Clean-up and Hyb DOC188 / Array Wash and Scan LP-154 / Array Analytics LP-188 PGD Array and Agilent microarray scanner Agilent Genomic Workbench	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Cancer genetics		
Blood Bone marrow	Non-specific global chromosome analysis for disease types: Acute myeloid leukaemia (AML) Acute lymphoblastic leukaemia (or lymphoma) (ALL) Chronic myeloid leukaemia (CML) Myelodysplastic syndromes (MDS) Myeloproliferative neoplasms (MPN) Non-Hodgkin's lymphoma with bone marrow involvement	G-banded analysis using in-house procedures: LP-G-191 CG Cyto - Analysis Guidelines	GL
Blood Bone marrow	Analysis of Haematological malignancies caused by mutations within genes: BCR/ABL PDGFRβ FGFR1 FIP1L1-PDGFRA RUNX1/RUNX1T1 PML/RARα RARα CBFβ KMT2A (MLL) EGR1/D5S23, D5S721 D7S486/CEP7 D20S108 Chromosome 8 alpha satellite ETV6 EVI ETV6/RUNX1 KMT2A (MLL) KMT2A/AFF1 TCF3(E2A)	Fluorescent in-situ hybridisation (FISH) analysis using in-house procedures: LP-G-191 CG Cyto - Analysis Guidelines and Thermobrite hybridisation chamber Fluorescence microscopy	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Cancer genetics (cont'd)		
Blood Bone marrow	Analysis of Haematological malignancies caused by mutations within genes (cont'd): Lymphoma probes: IGH/MYC IGH/CCND1 IGH/BCL2 IGH ALK BCL6 IGH/MALT1 BIRC3(API2)/MALT1 MYC	FISH analysis using in-house procedures: LP-G-191 CG Cyto - Analysis Guidelines and Thermobrite hybridisation chamber Fluorescence microscopy	GL
Blood Bone marrow	Analysis of CD138 genes: CKS1B/CDKN2C TP53/ATM DLEU/13q34/CEP12 IGH IGH/FGFR3 IGH/CCND1 IGH/MAF IGH/MAFB IGH/CCND3 D5S23, D5S721/CEP9/CEP15	FISH analysis using in-house procedures: LP-G-191 CG Cyto - Analysis Guidelines and Thermobrite hybridisation chamber Robosep Fluorescence microscopy	GL
Blood Bone marrow	Analysis of Chronic lymphocytic leukaemia genes: TP53/ATM DLEU/LAMP/CEP12	FISH analysis using in-house procedures: LP-G-191 CG Cyto - Analysis Guidelines and Thermobrite hybridisation chamber Fluorescence microscopy	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	<u>Cancer genetics (cont'd)</u>		
FFPE solid tumour samples	Analysis of the following genes: SS18 (SYT) FUS CHOP (DDIT3) FOXO1A (FKHR) EWSR1 ALK ROS1 MET MYB MAML2 ETV6 HER2 Vysis Melanoma panel Vysis UroVysion FISH panel	FISH analysis using in-house procedures: LP-G-191 CG Cyto - Analysis Guidelines and Thermobrite hybridisation chamber Fluorescence microscopy	GL
FFPE lymphoma samples	IGH/MYC IGH/CCND1 IGH/BCL2 IGH ALK BCL6 IGH/MALT1 BIRC3(API2)/MALT1 MYC	FISH analysis using in-house procedures: LP-G-191 CG Cyto - Analysis Guidelines and Thermobrite hybridisation chamber Fluorescence microscopy	GL
FFPE tissues: Lung Colorectal	RET	FISH analysis using in-house procedures: LP-G-191 CG Cyto - Analysis Guidelines and Thermobrite hybridisation chamber Fluorescence microscopy	GL
Blood Bone marrow	*JAK2* V617F quantification and determination of allelic burden	qPCR using in-house procedures: LP-MO-61 JAK2 V617F quantification assay and Applied Biosystems QuantStudio 7 thermal cycler	GL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular oncology		
Blood	BCRABL quantitation (for CML p210 only)	qPCR using in-house procedures: LP-MO-2 BCRABL Quantitation qPCR and Applied Biosystems QuantStudio 7 thermal cycler	MOU
Blood	*JAK2* V617F Mutational status	qPCR using in-house procedures: LP-MO-3 JAK2 V617F Mutational Status by qPCR and Applied Biosystems QuantStudio 7 thermal cycler	MOU
Blood Bone marrow	Detection of CALR mutation	Fragment analysis using in house procedures: LP-MO-32 CALR Mutation and Applied Biosystems Veriti 96 thermal cycler ABI 3730 Sequencer	MOU
Blood Bone marrow	MPL exon 10 and/or *JAK2* exon 12 mutational status	HRM using in-house procedures: LP-MO-23 HRM amplification for MPL exon10 and/or Jak2 exon12 Mutational Status and Lightcycler 480	MOU
Blood Bone marrow	Detection of PMLRARA fusion transcripts	qPCR using in-house procedures: LP-MO-27 RQ PCR for the detection of PMLRARA and Applied Biosystems QuantStudio 7 thermal cycler	MOU

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISUES AND FLUIDS (cont'd)	Molecular oncology (cont'd)		
Blood Bone marrow	Nucleophohosmin Mutation detection	1PCR using in-house procedures: LP-MO-42 qNPM1 mutation detection and Applied Biosystems QuantStudio 7 thermal cycler	MOU
FFPE tissues: Lung Colorectal	Mutations within genes: EGFR (exons 18-21) KRAS (codons 12, 13, 59, 61, 117, 146) NRAS (codons 12, 13, 59, 61, 146) BRAF (codon 600)	Swift testing using in-house procedures: LP-MO-46 Cancer Genetics Swift EGFR Pathway panel analysis and reporting protocol LP-MO-58 Swift Accel-Amplicon NGS Panel and	MOU
FFPE solid tumour samples	Detection of Single Nucleotide Variants (SNVs) and Indels (insertions/deletions) within genes covered by the Accel-Amplicon 57G Pan cancer Profiling Panel	Manual DNA extraction using: QIAamp kit DNA measurement using: Promega Quantus fluorometer qPCR using: Applied Biosystems QuantStudio 7 thermal cycler PCR using Applied Biosystems Veriti 96 thermal cycler Next Generation Sequencing using: Illumina MiSeq	GL
END			