# Schedule of Accreditation

**issued by**

# United Kingdom Accreditation Service

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

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| **8688**<br><br>**Accredited to ISO 15189:2012** | **Synnovis Analytics LLP**<br><br>**Issue No:** 027 **Issue date**: 23 December 2024 |
| **Genetics Centre**<br>**5th Floor Tower Wing**<br>**Guy's Hospital**<br>**London**<br>**SE1 9RT** | **Contact: Ashley Kilner**<br>**Tel: +44 (0)207 188 7373**<br>**Email: Ashley.kilner@nhs.net**<br>**Website: www.synnovis.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<br>5th Floor Tower Wing &<br>7th Floor Borough Wing (analysis only)<br>Guy's Hospital<br>London<br>SE1 9RT | Ashley Kilner (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<br>11th Floor Tower Wing<br>Guy's Hospital<br>London<br>SE1 9RT | Ashely Kilner (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<br>5th Floor Tower Wing<br>Guy's Hospital<br>London<br>SE1 9RT | Ashley Kilner (contact details above) | Enzymology, metabolite analysis and molecular genetic testing for a range of genetic biochemical disorders | BG |
| Molecular Oncology Unit<br>4th Floor Southwark Wing<br>Guy's Hospital<br>London<br>SE1 9RT | Ashley Kilner (contact details above) | Molecular oncology testing | MOU |
| Epidermolysis Bullosa Laboratory<br>St. John's Institute of Dermatology<br>Guy's Hospital<br>Great Maze Pond<br>London<br>SE1 9RT | Ashley Kilner (contact details above) | Frozen Sections<br>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<br>Saliva<br>Paraffin embedded tissue<br>Buccal cells<br>Dried bloodspots<br>Muscle | | DNA Extraction<br>In-house procedures:<br>LP-D-15<br>LP-D-24<br>LP-D-26<br>LP-D-29<br>LP-D-164<br>LP-D-165<br>LP-D-234<br>LP-D-505<br>using commercial kits and Chemagen 360-D automated DNA extractor | GL |
| Paraffin embedded tissue samples | | DNA Extraction<br>In house procedures:<br>LP-D-497<br>Using Qiagen EZ1 and EZ2 tissue DNA extraction kits and Qiagen EZ1 Advanced XL extraction platform | GL |
| Hair roots<br>Muscle | | RNA Extraction<br>In-house procedures:<br>LP-D-165 using commercial kits and manual extraction | GL |
| Blood<br>Bone marrow<br>FFPE tissue<br>Cell suspension (peripheral blood and bone marrow cells stored in RLT buffer) | | Nucelic Acid Extraction<br>In house procedures:<br>LP-MO-63<br>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<br>Dried bloodspots | | Multiple Displacement Amplification<br>Whole genome amplification of low copy number of cells using in-house procedures:<br>LP-D-53<br>in conjunction with manufacturer's instructions using:<br>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: | PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping<br>Using In-house procedures:<br>LP-D-2<br>LP-D-3<br>LP-D-21<br>LP-D-23<br>LP-D-25<br>LP-D-32<br>LP-D-51<br>LP-D-105<br>LP-D-163<br>LP-D-191<br>LP-D-194<br>LP-D-258<br>LP-D-530<br>and<br>Automated or manual methods in conjunction with manufacturer's instructions using:<br>Gene Sequence Manager software<br>NanoDrop® ND-1000 Spectrophotometer<br>M2 Plate reader<br>Hamilton STARlet robotBiorad Tetrad 2<br>MJ Research Dyad thermal cycler<br>Nanodrop II nanodispenser<br>Beckman Coulter Biomek NX<br>ABI 3730 DNA Analyser | GL |
| | Prader-Willi Syndrome and Angelman Syndrome | MS-MLPA Laboratory Procedure LP-D-543 | 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)<br><br>Breast/ovarian cancer (R208)<br><br>Congenital muscular dystrophy (R79)) | 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)<br><br>DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source | Molecular Genetics (cont'd)<br><br>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)<br><br>Congenital myopathy (R81)(<br><br>Cystic fibrosis (CFTR common variants)<br><br>Direct Test (confirmation of Whole Exome research results e.g. DDD project – varied genes)<br><br>Duchenne/Becker muscular dystrophy (dystrophin) | PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping<br>(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)<br><br>DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source | Molecular Genetics (cont'd)<br><br>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)<br><br>Familial hypercholesterolaemia R135(<br><br>Fanconi anaemia R229 (<br><br>Fragile X syndrome (FMR1)<br><br>Huntington disease (HTT) | PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping<br>(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)<br><br>DNA/RNA extracted in-house from the sample types listed above or received as primary sample type from external source | Molecular Genetics (cont'd)<br><br>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)<br><br>Lynch syndrome (MSI analysis)<br><br>Melanoma (BRAF exon 15)<br><br>Nucleotide Excision Repair disease R227( | PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping<br>(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)<br><br>Skeletal dysplasia; Stickler, Achondrogenesis type II and hypochondrogenesis, SEDC R24(Spondyloepiphyseal dysplasia congenital) and Kniest dysplasial, Achondroplasia, Thantophoric dysplasia (FGFR3)<br><br>Spinal muscular atrophy (SMN1) | PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping<br>(cont'd) | GL |
| Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source | Rare diseases and oncology | Next Generation Sequencing:<br>DNA normalisation<br>Data analysis of samples<br>In-house procedures:<br>LP-D-650<br>LP-D-433<br>LP-D-558<br><br>In conjunction with manufacturer's instructions using:<br>Hamilton Athena liquid handling robot<br>Illumina NextSeq 550<br>Congenica DSS (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 (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)<br><br>X-inactivation | PCR amplification (inc. Quantitative fluorescent (QF)-PCR & Multiplex Ligation-Dependent Amplification (MLPA)) of DNA/RNA, Capillary electrophoresis, Sanger sequencing and genotyping<br>(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:<br><br>Combined panel for:<br><br>Familial hypercholesterolaemia (VCP1)<br><br>Breast/ovarian cancer (VCP2p1) | Next Generation Sequencing and data analysis<br>In-house procedures:<br>59<br>LP-D-567<br><br>In conjunction with manufacturer's instructions using:<br>Twist Human Core Exome EF Multiplex kitQubit fluorometer<br>TWIST Bravo automated preparation NGS TWIST robot Life Tech Veriti Model# 9902<br>Savant Speed Vac Centrifuge<br>Agilent TapeStation 2200<br>Illumina NextSeq 550<br>Congenica DSS (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<br><br>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><br><br>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)<br><br>CM/CMD, DMD and NERD (VCP3)<br><br>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)<br><br>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) | 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)<br><br>Nucleotide Excision Repair disease R227(MPLKIP, DDB1, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, GTF2H5, POLH, XPA, XPC, LIG4, XRCC4, NHEJ1, UVSSA, CDKN2A, PTCH1)<br><br>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<br>In-house procedures<br>LP-D-422<br>LP-D-423<br>In conjunction with manufacturer's instructions using:<br>Elucigene kit<br>ABI 3730 DNA analyser<br>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) | <u>Molecular Genetics (cont'd)</u> | | |
| Embryonic cells<br>Buccal cells<br>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:<br>QP-G-24<br>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<br><br>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<br>Buccal cells<br>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:<br>LP-D-109<br>LP-D-130<br>LP-D-131<br>LP-D-132<br>In conjunction with manufacturer's instructions using:<br>PCR6 hoods, PTC200 DNA Engine, MJ Dyad thermocycler | PGD |
| | | Polymerase chain reaction (PCR) using in-house procedures:<br>LP-D-112<br>LP-D-231<br>LP-D-241<br>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:<br>LP-D-32<br>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<br>Buccal cells<br>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:<br>LP-D-214<br>in conjunction with manufacturer's instructions using Biorad C1000 thermocyclers | PGD |

| 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 |
| | Immunodermatology | Examination using in-house procedures (listed) in conjunction with manufacturer's instructions for the following methods (where relevant) | GEB |
| Skin biopsies | | Manual processing<br>Cryostat sectioning using Leica CM1950 cryostat<br>Photography using Nikon Eclipse E600 microscope<br>and:<br>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 | <u>Mapping of the following antigens:</u> | Manual indirect immunofluorescence by microscopy using:<br>DME-SOP-42<br>and:<br>Nikon Eclipse E600 microscope<br>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)<br><br>1. Plasma<br>2. Chorionic villus<br>3. Skin fibroblasts<br>4. Amniotic cells<br>5. Dried bloodspots<br>6. Leucocytes<br>7. Tissues<br>8. Amniotic fluid<br>9. Serum<br>10. Liver<br>11. Urine<br>12. Lithium heparin blood | <u>Enzymology and metabolite analysis</u><br><br>Detection of, for the purpose of clinical diagnosis: | | |
| 11 | Oligosaccharides (Glycoprotein storage disorders) | Thin layer chromatography<br>In-house procedures:<br>LP-B-109 | BG |
| 11 | Glycosaminoglycans (GAGs) (diagnosis of mucopolysaccharidosis disorders) | Colorimetric enzyme assay<br>Quantitative measurement using in-house procedures:<br>LF-B-12<br>LP-B-44<br>in conjunction with manufacturer's instructions using:<br>Roche Cobas Bio | BG |
| 8, 11 | Glycosaminoglycans (GAGs) (mucopolysaccharidosis disorders) | 2D electrophoresis<br>In-house procedures:<br>LP-B-45 | BG |
| 2, 3, 4 | Free cholesterol accumulation (Niemann Pick C disease) | Filipin staining and fluorescence microscopy<br>In-house procedures:<br>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) | <u>Enzymology and metabolite analysis (cont'd)</u> | | |
| 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) | <u>Enzymology and metabolite analysis (cont'd)</u> | | |
| 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: BMG Lab Tech ClarioStar Fluorimeter | BG |
| 6, 9 | % Hexosaminidase A by heat inactivation for Tay-Sachs carrier testing | LP-B-50<br>LP-B-103 | |
| 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<br>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 | |
| 12 | Galactose-1-Phosphate Uridyl Transferase | Direct Spectrophotometry using Shimadzu UV-1900i Spectrophotometer LP-B-47 | 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) | Prenatal and reproductive genetics | | |
| Amniotic fluid<br>Chorionic villus<br>Blood<br>Fetal blood<br>Solid tissues / products of conception<br>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:<br>LP-G-2/ LP-G-PCR DNA extraction<br>LP-G-6/LP-G-aCGHQF-PCR tissue samples<br>LP-G-1/LP-G-PCR set up<br>LP-G-322/LP-G-PCR Run 3500<br>LP-G-8/LP-G-PCRAnalysisTrisomy<br>and<br>Chemagen 360-D automated DNA extractor Dyad & Eppendorf Thermocyclers<br>ABI 3500 Genetic analyser | GL |
| Amniotic fluid<br>Chorionic villus<br>Blood<br>Products of conception<br>DNA extracted in-house from the sample types listed here or received as primary sample type from external source | Genome-wide copy number variation | Single Nucleotide Polymorphism SNP-array technology using in-house procedures:<br>LP-G-346 SNP Array Automated ProcessThermo Fisher CytoScan High-throughput cytogenomic array (HT-CMA) | GL |
| Blood<br>Fetal blood samples<br>Chorionic villus<br>Amniotic fluid | Karyotype | Culture and karyotype analysis using in-house procedures:<br>Prenatal:<br>LF-G-138/LP-G-PNHAR<br>LP-C-102/LP-C-AFSETUP<br>LP-G-194/LP-G-PNCVS Processing<br>Postnatal:<br>LP-C-70/LP-C-ProcBloodCult, LP-C-72/LPC-ProcHarvBlood, LP-C-125/LP-CBloodGBanding<br>and<br>Ikaros software on Metasystems | GL |
| Blood<br>Fetal blood samples<br>Chorionic villus<br>Amniotic fluid | Chromosome breakage studies | Culture and chromosome breakage screening using in-house procedures:<br>DOC3/LP-C-Atbldsu<br>DOC4/LP-C-Atbldharv<br>DOC9/LP-C-FABldDEB<br>DOC10/LP-C-FABldHarv<br>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<br>Fetal blood samples<br>Chorionic villus<br>Amniotic fluid<br>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:<br>LP-G-13/LP-G-PCR UPD<br>and<br>Dyad & Eppendorf Thermocyclers<br>ABI 3500 Genetic analyser | PGD |
| Blood<br>Fetal blood samples<br>Chorionic villus<br>Amniotic fluid<br>Solid tissues / products of conception<br>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:<br>LP-G-2001/LP-G-MCCtestingPCR set up<br>LP-G-13/LP-G-PCR UPD<br>and<br>Dyad & Eppendorf Thermocyclers<br>ABI 3500 Genetic analyser | GL<br>PGD |
| Blood<br>Fetal blood samples<br>Chorionic villus<br>Amniotic fluid<br>Solid tissues/ products of conception<br>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:<br>LP-G-1/LP-G-PCR set up<br>LP-G-200/LP-G-MCCtesting<br>and<br>Dyad & Eppendorf Thermocyclers<br>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<br>Saliva<br>Tissues | Genome-wide copy number variation | Single Nucleotide Polymorphism SNP-array technologyusing in-house procedures:<br>DOC188 / Array Wash and Scan<br>LP-G-356 SNP Array Analytics<br>And Thermo Fisher CytoScan High-throughput cytogenomic array (HT-CMA) | GL |
| | <u>Chromosome rearrangement PGD</u> | | |
| Trophectoderm biopsy samples from embryos | Unbalanced products from chromosome rearrangements | Single Nucleotide Polymorphism SNP-array technology using in-house procedures:<br>LP-G-356 SNP Array Analytics<br>LP-G-31 SNP Array analysis for PGT-SR samples<br>and<br>Thermo Fisher CytoScan High-throughput cytogenomic array (HT-CMA) | 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</u> | | |
| Blood<br>Bone marrow | Non-specific global chromosome analysis for disease types:<br><br>Acute myeloid leukaemia (AML)<br>Acute lymphoblastic leukaemia (or lymphoma) (ALL)<br>Chronic myeloid leukaemia (CML)<br>Myelodysplastic syndromes (MDS)<br>Myeloproliferative neoplasms (MPN)<br>Non-Hodgkin's lymphoma with bone marrow involvement | G-banded analysis using in-house procedures:<br>LP-G-191 CG Cyto - Analysis Guidelines | GL |
| Blood<br>Bone marrow | Analysis of Haematological malignancies caused by mutations within genes:<br><br>BCR/ABL<br>PDGFRβ<br>FGFR1<br>FIP1L1-PDGFRA<br>RUNX1/RUNX1T1<br>PML/RARα<br>RARα<br>CBFβ<br>KMT2A (MLL)<br>EGR1/D5S23, D5S721<br>D7S486/CEP7<br>D20S108<br>Chromosome 8 alpha satellite<br>ETV6<br>EVI<br>ETV6/RUNX1<br>KMT2A (MLL)<br>KMT2A/AFF1<br>TCF3(E2A) | Fluorescent in-situ hybridisation (FISH) analysis using in-house procedures:<br>LP-G-191 CG Cyto - Analysis Guidelines<br>and<br>Thermobrite hybridisation chamber<br>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> | | |
| Blood<br>Bone marrow | Analysis of Haematological malignancies caused by mutations within genes (cont'd):<br><br>Lymphoma probes:<br>IGH/MYC<br>IGH/CCND1<br>IGH/BCL2<br>IGH<br>ALK<br>BCL6<br>IGH/MALT1<br>BIRC3(API2)/MALT1<br>MYC | FISH analysis using in-house procedures:<br>LP-G-191 CG Cyto - Analysis Guidelines<br>and<br>Thermobrite hybridisation chamber<br>Fluorescence microscopy | GL |
| Blood<br>Bone marrow | Analysis of CD138 cell separation:<br><br>CKS1B/CDKN2C<br>TP53/ATM<br>DLEU/13q34/CEP12<br>IGH<br>IGH/FGFR3<br>IGH/CCND1<br>IGH/MAF<br>IGH/MAFB<br>IGH/CCND3<br>D5S23,<br>D5S721/CEP9/CEP15 | FISH analysis using in-house procedures:<br>LP-G-191 CG Cyto - Analysis Guidelines<br>and<br>Thermobrite hybridisation chamber<br>Robosep<br>Fluorescence microscopy | GL |
| Blood<br>Bone marrow | Analysis of Chronic lymphocytic leukaemia genes:<br><br>TP53/ATM<br>DLEU/LAMP/CEP12 | FISH analysis using in-house procedures:<br>LP-G-191 CG Cyto - Analysis Guidelines<br>and<br>Thermobrite hybridisation chamber<br>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:<br><br>SS18 (SYT)<br>FUS<br>CHOP (DDIT3)<br>FOXO1A (FKHR)<br>EWSR1<br>ALK<br>ROS1<br>MET<br>MYB<br>MAML2<br>ETV6<br>HER2<br>Vysis Melanoma panel<br>Vysis UroVysion FISH panel | FISH analysis using in-house procedures:<br>LP-G-191 CG Cyto - Analysis Guidelines<br>and<br>Thermobrite hybridisation chamber<br>Fluorescence microscopy | GL |
| FFPE lymphoma samples | IGH/MYC<br>IGH/CCND1<br>IGH/BCL2<br>IGH<br>ALK<br>BCL6<br>IGH/MALT1<br>BIRC3(API2)/MALT1<br>MYC | FISH analysis using in-house procedures:<br>LP-G-191 CG Cyto - Analysis Guidelines<br>and<br>Thermobrite hybridisation chamber<br>Fluorescence microscopy | GL |
| FFPE tissues:<br>Lung<br>Colorectal | RET | FISH analysis using in-house procedures:<br>LP-G-191 CG Cyto - Analysis Guidelines<br>and<br>Thermobrite hybridisation chamber<br>Fluorescence microscopy | GL |
| Blood<br>Bone marrow | *JAK2* V617F quantification and determination of allelic burden | qPCR using in-house procedures:<br>LP-MO-61 JAK2 V617F quantification assay<br>And Qiagility<br>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 Qiagility 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 Qiagility Applied Biosystems QuantStudio 7 thermal cycler | MOU |
| Blood<br>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<br>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<br>Bone marrow | Detection of PMLRARA fusion transcripts | qPCR using in-house procedures: LP-MO-27 RQ PCR for the detection of PMLRARA And Qiagility, Applied Biosystems QuantStudio 7 thermal cycler | MOU |
| cDNA (from PB, BM, RLT, TRIZOL, cell suspension) | Core Binding Factor translocations QPCR MRD testing | qPCR using in house procedures: LP-MO-45 Core Binding Factor translocations by qPCR | 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) | <u>Molecular oncology (cont'd)</u> | | |
| Blood<br>Bone marrow | Nucleophohosmin Mutation detection | 1PCR using in-house procedures:<br>LP-MO-42 qNPM1 mutation detection And Qiagility,<br>Applied Biosystems QuantStudio 7 thermal cycler | MOU |
| Extracted DNA/RNA from FFPE | Sequencing of gene targets for the NHSE National Genomic Test Directory | Ion Torrent Genexus System Oncomine Precsision Assay (OPA)<br><br>LP-MO-79 Genexus™ FFPE DNA/RNA Oncomine™ Precision Assay GX<br>LP-MO-80 Genexus™ FFPE DNA/RNA Oncomine Precision Assay GX Reporting and Analysis | 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) | <u>Molecular oncology (cont'd)</u> | Next generation sequencing using TruSight™ Oncology 500 (TSO500) enrichment of DNA and sequencing on Illumina ® Novaseq<br>manual assay and high throughput assay using the Hamilton robot<br><br>Analysis and reporting of next generation sequencing data using integrated genomic viewer (IGV) | MOU |
| DNA | Detection of Single Nucleotide Variants (SNVs) and Indels (insertions/deletions) within genes covered by the pan cancer panel | LP-MO-63<br>LP-MO-171<br>LP-MO-74 | |
| END | | | |