Schedule of Accreditation

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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 TISSUE AND FLUIDS	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer	Documented in house procedures incorporating manufacturer's instructions (where relevant):
	DNA/RNA Extraction, quantification and quality check for subsequent in-house analysis (see below), referral to specialist centres and long-term storage	**Manual semi-automated and automated DNA /RNA extraction and quantification using:**
		DNA Extraction :
Tissue, CVS, Cultured cells		Manual extraction processes 17.6.28, 21.26
Peripheral Blood		Qiagen Qiacube 17.6.33
Peripheral Blood		Promega Tecan Freedom Evo 17.6.35
Peripheral Blood and Bone Marrow		QIAsymphony using QIAsymphony DSP DNA Mini and Midi kit 17.6.37
Peripheral Blood, Prenatal, Bone Marrow, FFPE, saliva, buccal brush and Blood Spot		EZ1 Biorobot 17.6.27
Solid tumour and tissue		Automated extraction processes QIAsymphony 17.6.41
FFPE		Promega Maxwell RSC 48 instrument and Promega MaxPrep, Maxwell RSC DNA/RNA FFPE Kit (DNA and RNA) BGL/TECH/EXTRACTION/SOP/4 17.6.42

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	<u>Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer</u> (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant):
	DNA/RNA Extraction, quantification and quality check for subsequent in-house analysis (see below), referral to specialist centres and long-term storage (cont'd)	Manual, semi-automated and automated DNA /RNA extraction and quantification using: **DNA Quantification for QC purposes** Nanodrop ND2000 and ND8000 17.6.8 Qubit 4.0 and Flex 17.55.5
Whole Blood, FFPE		Promega Maxwell RSC 48 instrument and Promega MaxPrep BGL/TECH/EXTRACTION/SOP/4 Maxwell RSC simplyRNA Blood Kit17.2.43) and Maxwell RSC DNA/RNA FFPE Kit17.6.42 **RNA Quantification for QC purposes**: Nanodrop ND2000 and ND8000, 17.6.8 Qubit 4.0 and Qubit Flex 17.55.5
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Preparation of DNA for referral to external provider labs	**Whole Genome Sequencing** Using: Beckman Coulter NXp/FxP Liquid Handlers and SWGLH/WGS/LABORATORY/SOP/1 BGL/EXPORT/WGS/SOP/1 SWGLH/WGS/LABORATORY/SOP/5 BGL/ADMIN/SOP/
Genomic DNA & RNA extracted in house from the sample types listed above and received as primary samples from external sources	Detection of nucleic acid sequence variant - SNVs and Indels [Definitive list in APP15/60]	**Sanger Sequencing** Using: Standard primer design methodology, PCR amplification, gel electrophoresis Beckman Coulter NXp/FxP robots and Eppendorf Nexus, Applied Biosciences MiniAmp Applied Biosciences Simply Amp Thermocyclers. Sanger Sequencing performed using Applied Biosystems ABI 3730 DNA analyser and Mutation Surveyor software 17.23.8, MRD/S3/003, 18.1, 17.23.5

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant):
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Detection of nucleic acid sequence variants, small indels and/or determination of methylation status [Definitive list in APP15/60]	**Pyrosequencing** Using: Qiagen Pyromark Q48Pyrosequencer PCR amplification using kits electrophoresis, Beckman Coulter NXp/FxP robots and Eppendorf Nexus, Applied Biosciences MiniAmp Applied Biosciences Simply Amp Thermocyclers BGL/TECH/PYRO/SOP/1
Genomic DNA and RNA extracted in house from the sample types listed above and received as primary samples from external sources	Qualitative Genotype analysis for SNVs, indels and fusion transcripts [Definitive list in APP15/60]	**Qualitative Real Time PCR** Using: Life Technologies 7500 and 7500 FAST real time analysers and Quantstudio 5 analysers and: 17.23.12, 20.27 BGL/TECH/REAL-TIME/SOP/1 BGL/TECH/REAL-TIME/SOP/10
Whole Blood	Genotyping [definitive list in APP15/60]	**Loop-mediated isothermal Amplification (LAMP) Melt Analysis** (LaCar) (Qualitative PCR) using Quantstudio 5 Manual or Automated Set Up using BioMek NxP liquid hander or FXP liquid handler BGL/TECH/REAL-TIME/SOP/2 Analysis using BGL/TECH/REALTIME/SOP/4
RNA extracted from peripheral blood and bone marrow (see above)	Generation of cDNA by reverse transcriptase for subsequent in-house analysis (see below)	**cDNA generation:** Manual method using Applied Biosystems cDNA Reverse Transcription Kit 17.6.38
RNA extracted in house from the sample types listed above and received as primary samples from external sources	For the qualitative detection of common fusion transcripts associated with malignancy [Definitive list in APP15/60]	**Qualitative Reverse Transcriptase PCR (RTPCR)** Using: Eppendorf Nexus, Applied Biosciences MiniAmp Applied Biosciences Simply Amp Thermocyclers and gel electrophoresis and Genesnap visualisation and: 20.13

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	<u>Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer</u> (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant):
Genomic DNA and RNA extracted in house from the sample types listed above and received as primary samples from external sources	Quantitative analysis of known gene fusions events for the monitoring of malignancies [Definitive list in APP15/60]	**Quantitative Real Time PCR** Using: Life Technologies 7500 and 7500 FAST real time analysers and Quantstudio 5 analysers and: 20.15: (RQ-PCR for BCR-ABL transcripts) MRD/S2/003, MRD/S4/003, MRD/S5/001 BGL/TECH/REAL-TIME/SOP/7
	Gene screening of large gene panels for genetic variants [Definitive list in APP15/60]	**Next Generation Sequencing:** Using: Agilent 4200 Tapestation, Qubit 4.0 and Qubit Flex Fluorometer, Eppendorf Nexus Applied Biosciences MiniAmp Applied Biosciences SimpliAmp thermal cycler, Myra liquid handling robots and Ilumina Novaseq X Plus sequencer 17.55.5, BGL/TECH/NGS/SOP/8, BGL/TECH/NGS/SOP/15
	SNVs, CNV and Indels	**Rare Disease Panel** Library preparation: Nextera Flex with Twist Biosciences BGL/TECH/NGS/SOP/1 Pipeline: DNAnexus Rare Disease Pipeline (SOP 25.14) Analysis using: BGL/ANALYSIS/SOP/3
	SNVs, Indels and Gene Amplifications	**Solid Tumour and Haemato-Oncology DNA panel** Library preparation: Roche DNA BGL/TECH/NGS/SOP/10 Pipeline: In-house DNAnexus somatic pipeline (SOP 25.10) Analysis using: BGL/ANALYSIS/SOP/1, BGL/ANALYSIS/SOP/5, BGL/SOLID TUMOUR/SOP/1 BGL/HAEMATO-ONCOLOGY/SOP/2

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant):
Genomic DNA and RNA extracted in house from the sample types listed above and received as primary samples from external sources (cont'd)	Gene Fusions	**Solid Tumour and Haemato-Oncology RNA panel** Library preparation: Roche RNA BGL/TECH/NGS/SOP/11 Pipeline: In-house DNAnexus somatic pipeline (SOP 25.10) Analysis using: BGL/ANALYSIS/SOP/1, BGL/SOLID TUMOUR/SOP/1, BGL/HAEMATO-ONCOLOGY/SOP/2
	Gene Screening Gene rearrangements at the immunoglobulin and T cell receptor loci for minimal residual disease (MRD) target identification and assessment of clonality	**Univ8 Euroclonality NGS DNA Capture (NDC[SG1.1]) panel** Library preparation: Roche BGL/TECH/NGS/SOP/10 Pipeline: In-house demultiplex process (SOP 25.13) followed by commercial Illumina EuroClonality-NDC App Analysis using: MRD/S3/004, BGL/ANALYSIS/SOP/10
	SNVs and MSI	**MSI-Plus (SBRI) panel** Using Ilumina MiSeq (SOP 17.55.6) Library preparation: PacBio/NGL Index Plate BGL/TECH/NGS/SOP/12 Pipeline: In-house MSI pipeline (SOP 25.12) Analysis using: BGL/SOLID TUMOUR/SOP/4
	Genomic screening for genetic variants causing deficiencies in the Homologous Recombination Repair pathway and for the detection of HRD from shallow WGS	**HRD Shallow Whole Genome Sequencing (sWGS)** Library preparation: Roche BGL/TECH/NGS/SOP/13 Pipeline: In-house demultiplex process (SOP 25.13) followed by commercial SOPHiA DDM "GIInger" pipeline Analysis using: BGL/SOLID TUMOUR/SOP/5
Whole genome sequencing data and variants identified by an external source	Analysis using whole genome sequencing data	**Whole Genome Sequencing** Using: SWGLH/WGS/LABORATORY/SOP/2 SWGLH/WGS/LABORATORY/SOP/3 SWGLH/WGS/LABORATORY/SOP/4

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant):
Genomic DNA extracted in house from the sample types listed and samples received as primary samples from external sources. Unless otherwise stated testing is undertaken on DNA.	Detection of fragment length size, deletions, known mutations, repeat expansions, linkage markers. [Definitive list in APP15/60]	**Fragment length analysis** Nucleic acid amplification Using: Eppendorf Nexus Applied Biosciences MiniAmp Applied Biosciences SimpliAmp thermal cycler[SG29.1] Beckman NXp liquid handling system ABI 3730 and 3500 Genetic Analysers using Amplidex analysis software by Assuragen 17.23.14, ,17.23.2, BGL/TECH/FRAG/SOP/3 BGL/RD3/SOP/4 BGL/TECH/FRAG/SOP/9 BGL/TECH/FRAG/SOP/1, BGL/TECH/FRAG/SOP/2, BGL/RD3/SOP/1
Genomic DNA extracted in house from the sample types listed and samples received as primary samples from external sources	Determination of copy number changes [Definitive list in APP15/60]	**Multiplex Ligation Probe Amplification (MLPA)** Using: Eppendorf Nexus Applied Biosciences MiniAmp Applied Biosciences SimpliAmp thermocyclers, ABI 3730, GeneMarker and Coffalyser data analysis software 17.23.18, 12.55

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant):
Genomic DNA extracted in house from the sample types listed	Detection of large gene rearrangements [Definitive list in APP15/60]	**Southern Blotting** Using: Model 400 and Carbolite Hybridisation incubators, Stratolinker, and Azure 280 Chemiluminescence detection BGL/TECH/FRAG/SOP/8 Analysis using Azure Spot analysis software BGL/ANALYSIS/SOP/6
Genomic DNA extracted in house from peripheral blood, bone marrow and FFPE tissues	Detection of single nucleotide variants and small indels and gene amplification [Definitive list in APP15/60]	**Droplet digital PCR** Using: BioRAD QX200 Droplet Reader BioRAD AutoDroplet Generator BioRAD PCR Plate Reader Myra liquid handling system BGL/TECH/ddPCR/SOP/1 BGL/TECH/ddPCR/SOP/3
	Confirmation of CNV	Biorad EVAGreen chemistry: BGL/TECH/ddPCR/SOP/2

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd) Whole blood Amniotic fluid CVS Foetal Blood Products of conception Bone Marrow Whole Blood Tissues/Skin Biopsies Buccal cells	<u>Cytogenetics analysis for the purpose of clinical diagnosis of rare disease and cancer</u> G-banding/Karyotyping: Detection of chromosomal rearrangements or aberrations arising from: *(e.g.)* Prenatally detected Disorders Developmental disorders Reproductive medicine disorders Haematological/Oncology disorders (preparative pre-examination steps listed first)	Documented in house procedures incorporating manufacturer's instructions (where relevant): **Culturing and processing of human tissue/cells to provide interphase cells:** Automated process using StemCell Robosep fully automated cell separator using: SOP 3.30 APP07/06 And 2.5, 2.7, 2.8 (Prenatal) 3.4 (Constitutional – blood) 3.7, (Oncology) 3.30, 4.0, 4.6, 4.20 (Solid tissue) Cell Harvesting: Manual harvesting using 3.16, 3.28 Automated process using Genial Genetics Coverslip Harvester and: 2.12 **Chromosome analysis** **Microscopic analysis of G banded chromosomes** Carl Zeiss light microscope and Metasystems Image analysis suite and: BGL/TECH/CULTURE/SOP/210 and 10.2 7.1,7.5 11.14.13 Manual analysis using: 10, 10.2, 7.5

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	<u>Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer</u>	Documented in house procedures incorporating manufacturer's instructions (where relevant):
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Microarray examinations for the detection of sub-microscopic chromosomal imbalance (gains and losses) expressed as changes to copy number, the detection of uniparental disomy (UPD) and for confirming relatedness of individuals	**Illumina GSACyto SNP microarray** Using Qubit 4.0 and Qubit Flex fluorometer, hybridisation oven, MiVac, Infinium automated pipetting system, and Illumina iScan platform. BGL/TECH/ARRAY/INFINIUM/SOP/4 Analysis and interpretation for genetic imbalances using Bionano VIA software enterprise. BGL/ANALYSIS/SOP/2 BGL/ANALYSIS/SOP/8 BGL/ANALYSIS/SOP/9
Extracted DNA from FFPE and fresh tissue	Classification of brain tumours based on methylation status: CNS tumours	**EPIC Array** DNA extraction from FFPE, bisulphite modification methylation array set up using Inifinium methylation EPIC Bead chip using Qubit 4.0 and Qubit Flex Fluorometer, Illumina iScan array scanner and generation of methylation profile using GenomeStudio software and Heidelberg classifier BGL/TECH/ARRAY/INFINIUM/SOP/1 BGL/TECH/ARRAY/INFINIUM/SOP/2 BGL/TECH/ARRAY/INFINIUM/SOP/3 Heidelberg classifier software v12.8 accessed via https://app.epignostix.com/ BGL/SOLID TUMOUR/SOP/3

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HUMAN TISSUE AND FLUIDS (cont'd)	<u>Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer</u> (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant):
Formalin fixed paraffin embedded tissue (FFPE) Peripheral Blood Bone Marrow Fixed culture cells (more specific – cultured, uncultured PB, Marrow, AFs etc) Amniotic fluid CVS- Chorionic villus samples	Detection of chromosomal aberrations in the diagnosis of haematological malignancy, bone marrow failure syndromes, non-haematological malignancies and constitutional disorders, solid tumours and companion testing Break-apart probes Fusion products Deletion Insertion Copy Number / Amplification	**Fluorescence in situ hybridisation (FISH)** Fluorescent in situ hybridisation (FISH) using commercial and in house developed probes by Hybrite using: SOP0504, SOP0502, SOP0524 and analysis using Abbott VIP2000 automated processing unit Fluorescent microscope and Carl Zeiss Metasystems Image analysis suite. and: SOP13.3
END		