Schedule of Accreditation

issued by

United Kingdom Accreditation Service

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

9307

Accredited to ISO 15189:2012

North Bristol NHS Trust

Issue No: 004 **Issue date:** 16 November 2020

Bristol Genetics Laboratory
Pathology Sciences Building
Southmead Hospital
Westbury on Trym
BS10 5NB

Contact: Rachel Butler
Tel: +44 (0)117 414 6168
E-Mail: rachel.butler@nbt.nhs.uk
Website: www.nbt.nhs.uk/severn-pathology/pathology-services/bristol-genetics-laboratory-bgl

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	<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):
	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:**
Tissue, CVS, Saliva, Cultured cells Peripheral Blood, PET, Prenatal Peripheral Blood Peripheral Blood and Bone Marrow Peripheral Blood, Prenatal, Bone Marrow, FFPE and Blood Spot		**DNA Extraction :** Manual extraction processes 17.6.28, 21.26, 17.6.32, 17.6.34 Qiagen Qiacube 17.6.33, 17.6.24, 17.6.36 Promega Tecan Freedom Evo 17.6.35 QIAsymphony using QIAsymphony DSP DNA Mini and Midi kit 17.6.37 EZ1 Biorobot 17.6.27
		DNA Quantification for QC purposes: Nanodrop ND2000 and ND8000 17.6.8
FFPE Peripheral Blood		**RNA extraction:** Manual process and automated using Qiacube 17.6.34, Automated using Qiacube 20.12
		RNA Quantification for QC purposes : Nanodrop ND2000 and ND8000 17.6.8

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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):
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 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
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources (cont'd)	Detection of nucleic acid sequence variants, small indels and/or determination of methylation status [Definitive list in APP15/60]	**Pyrosequencing** Using: Qiagen Pyromark Q96 Pyrosequencer PCR amplification using kits electrophoresis, Beckman Coulter NXp/FxP robots and Thermocyclers. 17.23.16
Genomic DNA and RNA extracted in house from the sample types listed above and received as primary samples from external sources (cont'd)	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: 17.23.12, 17.43.3, 20.27

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HUMAN TISSUE AND FLUIDS	<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):
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 (cont'd)	For the qualitative detection of common fusion transcripts associated with malignancy [Definitive list in APP15/60]	**Qualitative Reverse Transcriptase PCR (RTPCR)** Using: G Storm Thermocycler and gel electrophoresis and Genesnap visualisation and: 20.13
Genomic DNA and RNA extracted in house from the sample types listed above and received as primary samples from external sources (cont'd)	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: 17.23.12 20.15: (RQ-PCR for BCR-ABL transcripts) MRD/S2/003, MRD/S4/003, MRD/S4/004, MRD/S5/001, MRD/S5/002
Genomic DNA and RNA extracted in house from the sample types listed and samples received as primary samples from external sources (cont'd)	Gene screening of large gene panels for genetic variants [Definitive list in APP15/60]	**Next Generation Sequencing:**
	SNVs	**Library Preparation methods:** Haloplex Agilent Sureselect Illumina Trusight (Cancer and TST15)
	CNVs	Haloplex Agilent Sureselect
	Indels	Agilent Sureselect Illumina Trusight (Cancer and TST15)

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):
Genomic DNA and RNA extracted in house from the sample types listed and samples received as primary samples from external sources (cont'd)	Gene screening of large gene panels for genetic variants [Definitive list in APP15/60]	**Next Generation Sequencing:** Using: Diagen Biorupter, Agilent 2200 Tapestation, Qubit 2.0 Fluorometer, thermal cycler and Illumina MiSeq and/or NextSeq 17.55.3, 17.55.4, 17.55.5, 17.55.6, 17.55.14, 17.55.17, 17.55.21, 17.55.22, Analysis using: Clinical Exome and targeted panel pipeline 25.2 Somatic pipeline 25.5 Somatic Variants BGL/SOP/Analysis/1
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: G Storm thermocyclers (GS0004M) and Coulter CEQ8000 / ABI 3730 and 3500 Genetic Analysers using GeneMarker and Genemapper analysis software 17.23.4, 17.23.14, ,17.23.2,20.17, BGL/TECH/FRAG/SOP/3 17.4.1, 17.4.6, 17.7.9, 17.46.1,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: G Storm thermocyclers, Beckman CEQ8000 and GeneMarker and Coffalyser data analysis software 17.23.18, 17.23.4, 12.55
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 XO Graph, and Chemiluminescence detection 17.8.1

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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):
Genomic DNA extracted in house from peripheral blood, bone marrow and FFPE tissues	Detection of single nucleotide variants and small indels [Definitive list in APP15/60]	**Droplet digital PCR** Using : BioRAD QX200 Droplet Reader BioRAD AutoDroplet Generator BioRAD PCR Plate Reader and:
Whole blood Amniotic fluid CVS Foetal Blood Products of conception Bone Marrow Whole Blood Tissues/Skin Biopsies Buccal cells	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)	**Culturing and processing of human tissue/cells to provide interphase cells:** Automated process using StemCell Robosep fully automated cell separator using: SOP 3.30 And 2.5, 2.7, 2.9 (Prenatal) 3.4 (Constitutional – blood) 3.7, 3.11 (Oncology) 3.12, 3.30, 4.0, 4.6, 4.20 (Solid tissue) Cell Harvesting: Automated process to provide material suitable for analysis using Freedom EVO 100 Tecan Liquid Handling Robot and: 3.15 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: 13.3, 10 and 10.2 7.1, 7.3, 7.5, 7.10, 3.22, 4.12, 4.13 Manual analysis using: 10, 10.2,7.4, 7.5, 7.7

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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):
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Cytogenetic examinations for diagnosing postnatal disorders prenatal diagnosis, neoplastic genetics including haemato-oncology and solid tumours loss of pregnancy by detection of sub microscopic chromosomal imbalance (gains and losses) expressed as changes to copy number	**Array Comparative Genomic Hybridisation (aCGH)** processing Competitive hybridisation of patient and control DNA using hybridisation oven, MiVac and Agilent array scanner and Scigene Little Dipper slide washer. Analysis and interpretation of genetic imbalances using Cytosure Interpret software 21.24 21.11
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 Fluorescent microscope and Carl Zeiss Metasystems Image analysis suite. and: SOP13.3
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