Schedule of Accreditation

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NHS Tayside

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Testing performed by the Organisation at the locations specified below

Locations covered by the organisation and their relevant activities

Laboratory locations:

Location details		**Activity**
Address Level 6 Ninewells Hospital Dundee DD1 9SY	**Local contact** Dr David Baty 01382 496271	Molecular Genetic Cytogenetic Molecular Pathology Molecular Haematology testing

DETAIL OF ACCREDITATION

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS	Genomic analysis for the purposes 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:
		Manual extraction processes:
Peripheral Blood, Bone Marrow		Flexigene (MGM085)
Saliva		Oragene (MGM106)
FFPE LMD tissue		Qiagen Microkit (MGM264)
Amniotic Fluid		Igenatal (MGM287)
		Automated DNA Extraction Processes:
Peripheral Blood		Qiagen QiaSymphony (MGE053, MGM230)
Peripheral Blood FFPE Fresh Tissue		EZ1 & EZ1 Advanced XL (MGE023, MGE063, MGM084, MGM247 MGM097 MGM235)
Urine FFPE		Maxwell RSC (MGE126, MGM311, MGM316 & MGM320)
FFPE		(MGM316 & MGM320 FFPE) Biorobots

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd) Peripheral Blood Bone Marrow	<u>Genomic analysis for the purposes of clinical diagnosis of rare disease and cancer</u> (cont'd) DNA/RNA Extraction, quantification and quality check for subsequent in-house analysis (see below), referral to specialist centres and long term storage	Documented in house procedures incorporating manufacturer's instructions (where relevant) **Manual, semi-automated and automated DNA /RNA extraction and quantification using:** **DNA Quantification for QC purposes:** Nanodrop ND-2000 (MGE014) Qubit Fluorometer (MGM299) Automated RNA Extraction processes EZ1 and RNA Cell Mini Kit (MGM242) **RNA Quantification for QC purposes**: Nanodrop ND-2000 (MGE014)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)		Documented in house procedures incorporating manufacturer's instructions (where relevant)
Genomic DNA extracted in house from the sample type listed above and received as primary samples from external sources	Detection of nucleic acid sequence variant - SNVs and Indels [definitive list to be held by this laboratory]	**Sanger Sequencing Using:** PCR Amplification using: documented in-house procedures on Veriti thermocyclers. (MGM001, MGM144, MGM207, MGM229, MGM237, MGM243, MGE036, MGM313) Sequencing of products using: Standard primer design methodology (Primer Design (SOP GEN084), PCR amplification, gel electrophoresis, capillary electrophoresis, Beckman Biomeck robot, HamiltonStar Robot, Veriti Thermocyclers, Applied Biosystems ABI 3730 DNA analyser and examination and analysis of sequence data using Mutation Surveyor software package). (MGM313 Sequencing –PCR guide, MGE035, MGE067, MGM220, MGM238, MGM012, MGM103, MGE045, MGE128, MGM096, MGM277 MGM229, MGM319)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (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 and DNA from external sources	Detection of nucleic acid sequence variants andsmall indels [definitive list held by this laboratory].	**Pyrosequencing** Using: Qiagen PyroMarkQ24 platform using in-house procedures and Qiagen therascreen kits as per: SOPs MGM174, MGM175, MGM178, MGM228, MGM241, MGM257, MGM258, MGM270, MGM272, MGM286, GEN064.
Peripheral Blood	Quantitative analysis of known gene fusions events for the monitoring of malignancies [definitive list of fusions and transcripts to be held by this laboratory]	**Quantitative Real Time PCR** Using: Fully automated nucleic acid extraction, reverse transcription and QPCR by Cephid GeneXpert BCR-ABL Ultra System (MGM302)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS		Documented in house procedures incorporating manufacturer's instructions (where relevant) Next Generation Sequencing:
Genomic DNA extracted in house from the sample types listed and samples received as primary samples and DNAfrom external sources (cont'd)	Gene screening of large targeted gene panels for nucleic acid SNVs and small indels –[definitive list of panels and genetic variant types to be held by this laboratory] **SNV/indels**	For targeted panels: Library Preparation Methods: QIASeq Targeted DNA Panel (MGM315) Qubit 2.0 Fluorometer, thermal cycler and, Illumina MiSeq platform SOPs MGM299, MGE036, MGE130 Analysis using: Targeted panel pipeline Qiagen Biomedical Genomics Workbench and Qiagen Clinical Insight Interpret (QCI). SOPs MGM307, MGM317, MGM322.
Genomic DNA extracted in house from the sample types listed and samples received as primary samples from external sources.	Detection of fragment length size, deletions, known mutations, repeat expansions, short tandem repeats (definitive list to be held by this laboratory]	**Fragment Length Analysis** Using: CFTR Analysis by capillary electrophoresis (as detailed beloq) using Elucigene CFEU2v1 Amplification Refractory Mutation System PCR Kit. SOP MGM308

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (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.	Detection of fragment length size, deletions, known mutations, repeat expansions, short tandem repeats (definitive list to be held by this laboratory]	**Fragment Length Analysis** Using: BIOMED2 IdentiClone Assays using capillary electrophoresis (as detailed above). SOPs MGM244, MGM271. Amplification refractory mutation detection system (ARMS) using in-house agarose gel electrophoresis or capillary electrophoresis (as detailed above). SOPs MGM101, MGM252, MGM308. Manual resolution: Gel electrophoresis to separate DNA fragments using documented in-house procedures. SOPs MGE055, MGM314, MGM151 Automated resolution: Capillary electrophoresis for fragment separation using the ABI3730 genetic analyser. Analysis using GeneMarker software SOP GEN057. SOPs MGE031, MGE045, MGM025, MGM029, MGM042, MGM044, MGM054, MGM137, MGM257, MGM303.

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (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 and DNA from external sources	Determination of copy number changes and methylation status [definitive list of targets assessed to be held by this laboratory].	**Multiplex Ligation Probe Amplification (MLPA)** Using: MRC Holland kits, Veriti thermocyclers, ABI3730 and GeneMarker data analysis software to detect dosage abnormalities by capillary electrophoresis SOPS MGM112, GEN065, GEN070
Genomic DNA extracted in house from the sample types listed and samples received as primary samples and DNA from external sources	Rapid diagnosis of common trisomys, [definitive list of targets assessed to be held by this laboratory].	**QF-PCR** Using: QF-PCR using the Devyser Compact and XY Kits and Elucigene kits by capillary electrophoresis on ABI3730 (as detailed above). SOPS MGM282, MGM183
Genomic DNA extracted in house from the sample types listed and samples received as primary samples and DNA from external sources	Determination of copy number changes and identity matching [definitive list of targets assessed to be held by this laboratory].	Powerplex HS16 Kit for the detection of molar pregnancy by capillary electrophoresis on ABI3730 (as detailed above). SOP MGM280

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)		Documented in house procedures incorporating manufacturer's instructions (where relevant)
Genomic DNA extracted in house from peripheral blood	Confirmation of genomics gains and losses detected by array studies [definitive use to be held by this laboratory]	**Digital PCR** Using: Quantstudio digital PCR platform: Digital PCR Chip Loading: sample preparation: SOP MGE074 PCR using Proflex Digital PCR System: SOP MGE075 Chip scanning using Quantstudio 3D Digital PCR Imaging Instrument: SOP MGE076 Analysis using Quantstudio 3D software: SOP MGM305
Whole blood Amniotic fluid CVS Bone marrow Lymph node Fresh tissue samples	G-banding/Karyotyping: Detection of chromosomal rearrangements or aberrations arising from: *(e.g)* Prenatally detected Disorders Developmental disorders Reproductive medicine disorders Haematological/Oncology disorders [definitive list to be held by this laboratory] (preparative pre-examination steps listed first)	**Culturing and processing of human tissue/cells to provide interphase cells:** Cell culture Using in-house methods and commercial media:SOPs NGLSM009, NGLSM048 And NGLSM028, NGLSM029, NGLSM030, NGLSM031, (Prenatal) NGLSM013, NGLSM048 (oncology) NGLSM044 (solid tumour) **Cell Harvesting:**Manual harvesting using in-house methods: SOPs NGLSM011, NGLSM014, NGLSM033, NGLSM045, NGLSM050, NGLSM057.

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd) Cells harvested from: Whole blood Amniotic fluid CVS Bone marrow Lymph node Fresh tissue samples		Documented in house procedures incorporating manufacturer's instructions (where relevant) **Slide preparation and G Banding:** SOPs NGLSM024, NGLSM021 **Chromosome analysis** **Microscopic analysis of G banded chromosomes** Olympus light microscope and Cytovision Image analysis system and: Manual G banded bright field analysis analysis and karyotyping against considered normal pattern/profile using: SOP MGE078, GEN010, GEN013 Image analysis using: SOPs MGE081, NGLSM118
DNA	Clinical Exome TruSight Expaded One panel using Illumina DNA Prep with Enrichment (previously known as Nextera Flex for enrichment)	Using validated methods on Illumina NextSeq platform: VAL585, MGM323, MGM310 & MGE141
Blood (STRECK)	Non-invasive prenatal testing (NIPT) The VeriSeq NIPT Solution v2 – CE-IVD prenatal screening test used to detect fetal aneuploidies of chromosomes 21, 18 and 13 from maternal blood samples.	Using verified (VER065) CE-IVD method MGM325 on Hamilton Star robot MGE142 and Illumina NextSeq550DX platform MGE141
DNA	Targeted panel NGS using Illumina DNA Prep with Enrichment (previously known as Nextera Flex for Enrichment) using custom Twist panels	Using validated method on existing Illumina MiSeq platform: VAL615, MGM330 on existing MiSeq platform and MGM331.

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (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	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 [definitive list of referral reasons to be held by this laboratory]	**Array Comparative Genomic Hybridisation (aCGH)** processing Competitive hybridisation of patient and control DNA using Affymetrix platform: (Thermo Fisher Scientific) GeneChip Scanner 3000 7G: MGE122 hybridisation oven MGE124, Affymetrix CytoScan 750 - Processing: MGM289 – MGM296 - Data Processing, Analysis & Reporting: MGM297
Cultured & uncultured cells Paraffin embedded tissues (PETs) Tumour imprints Purified plasma cells	Detection of chromosomal aberrations in the diagnosis of haematological malignancy, bone marrow failure syndromes, non-haematological malignancies and constitutional disorders and solid tumours Break-apart probes Fusion products Deletion Insertion Copy Number / Amplification	**Fluorescence in situ hybridisation (FISH)** Using: Fluorescent in situ preparation and hybridisation (FISH) using using in-house methods and commercial probes (Vysis; Cytocell; Kreatech (now Leica Microsystems); (Illumina; Zytovision, CAMBIO; Cancer Genetics) Processing: SOPs NGLSM001, NGLSM002, NGLSM003, NGLSM123, MGE077, NGLSM074 Fluorescence microscopy and Cytovision Image Analysis Systen for signal detection and analysis SOPs GEN014, MGE078, MGE081
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