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 TISSUES AND FLUIDS	Molecular genetic examinations for the purpose of clinical diagnosis	In-house documented methods incorporating manufacturer's instructions where relevant
Whole blood Bone marrow DNA RNA Tissue Mouthwash/saliva Buccal scrape Guthrie bloodspots	Molecular Genetics: for Detection of common and rare genetic conditions.	Sample preparation and DNA and RNA extraction Documented methods for DNA and RNA extraction using one or a combination of the techniques below by in-house procedures, using commercial kits and manual extraction. SOP 033057 (Chemagic 360D) SOP 061 (EZ1 BioRobot / EZ1 Advanced XL)) SOP 060 – Manual DNA extraction SOPS 031970, 0302088 - (QIAcube) - RNA extraction and processing SOP0384 Guthrie bloodspots (Qiacube) SOP 0018 (Nanodrop), 033501 (Qubit) DNA Measurement

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)	<u>Molecular genetic examinations for the purpose of clinical diagnosis</u> (cont'd)	In-house documented methods incorporating manufacturer's instructions where relevant
Genomic DNA extracted in-house from the sample types listed below or received as primary sample type from an external source Whole blood Wax-embedded tissue DNA Mouthwash/Saliva Bone marrow Tissue (fresh)	Molecular Genetics: for Detection of common and rare genetic conditions. For two distinct applications: (i) screening for unknown variants in multi-gene fixed and virtual panels [as per definitive list] . (1A)	(i) Next-generation sequencing with library preparation using a custom-designed commercial panel and analysed using Illumina MiSeq/MiniSeq technology. Screening for unknown variants in multi-gene panels (SOPs 032107 and 032100); followed by either Alissa Interpret (Agilent) NGS software (SOP 032492) or in-house excel macro (SOP 032107). analysis of NGS data using in-house validated bioinformatics pipeline (SOP 032576) followed by either Alissa Interpret (Agilent) NGS software (SOP 032492) or in-house excel macro (SOP032107). Variant classification is carried out using one or more of the ACMG, ACGS and relevant gene/disease-specific guidelines (SOP 032530).
	(ii) genotyping of specific mutations and genomic regions for both constitutional and acquired conditions[as per definitive list] (2A)	(ii)Genotyping of specific variants and genomic intervals by Next-generation sequencing (SOPs 032031 and 032101) using libraries prepared by Reverse Complement RPCR (RC-PCR) and analysed using in-house bioinformatics pipeline (SOP 032576)
Whole blood and Bone marrow DNA	Screening for unknown somatic variants in myeloid disorders in multi-gene fixed panel Illumina TruSight Myeloid Panel (1B)	Screening for unknown variants sequenced on an Illumina MiSeq (SOP 3309) using the Alissa Interpret (Agilent) NGS Software (SOP 033310)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd) RNA or genomic DNA extracted in-house from the sample types listed below or received as primary sample type from an external source Whole blood Amniotic Fluid CVS Wax-embedded tissue Bone marrow Tissue Mouthwash Buccal scrape	<u>Molecular genetic examinations for the purpose of clinical diagnosis</u> (cont'd) Molecular Genetics: Detection of abnormal sequences for common and rare genetic conditions For confirmation of variants identified as part of whole gene screens or identified in a non-accredited laboratory, screens for unknown varinats in part or all of a gene and family follow-up of specific varinats. [as per definitive list] (2B)	In-house documented methods incorporating manufacturer's instructions where relevant Sanger sequencing using ABI 3130XL/3500 sequencing reagents (SOPs 0012, 0344, 0456, 031652) analysed using ABI 3130XL/3500 (SOP 0481) and Mutation Surveyor software. Where necessary, variant classification is carried out using one or more of the ACMG, ACGS and relevant gene/disease-specific guidelines (SOP 032530).

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)	Molecular genetic examinations for the purpose of clinical diagnosis (cont'd)	In-house documented methods incorporating manufacturer's instructions where relevant
Genomic DNA extracted in-house from the sample types listed below or received as primary sample type from an external source Whole blood Amniotic Fluid CVS Wax-embedded tissue Bone marrow Tissue Mouthwash Buccal scrape	Molecular Genetics: Detection of constitutional and acquired repeat disorders [as per definitive list]	Fragment size analysis of fluorescent PCR using ABI 3130XL/3500 (SOP 032468 and 033540) and analysed using Gene Marker software (SOP 031683)
	Sizing analysis haemato-oncology disorders (bone marrow and whole blood only) [as per definitive list]	Non-fluorescent PCR products analysed by agarose gel electrophoresis (SOP 0169).
Genomic DNA extracted in-house from the sample types listed below or received as the primary sample type from an external source Whole blood Amniotic Fluid CVS DNA Tissue	Molecular genetics: For the determination of the X inactivation pattern in females to aid the interpretation of X-linked copy number variants or single nucleotide variants or where X-linked inheritance is suspected	Using enzyme digestion and fragment sizing on an ABI 3500 analyser (SOP 0267).
Genomic DNA extracted in-house from the sample types listed below or received as primary sample type from an external source Whole blood Amniotic Fluid CVS Tissue Mouthwash Buccal scrape	Molecular Genetics: Detection of whole exon deletions/duplications and specific microdeletion syndromes either as a stand-alone test or as part of a whole gene screen, and for index cases and family follow up [as per definitive list]	Multiplex Ligation Probe Analysis (MLPA) for the detection of whole exon deletions/duplications and specific microdeletion syndromes (SOP031686).

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)	<u>Molecular genetic examinations for the purpose of clinical diagnosis</u> (cont'd)	In-house documented methods incorporating manufacturer's instructions where relevant
Whole blood Amniotic Fluid CVS Guthrie bloodspots	Molecular Genetics: Detection of 50 specific variants in the *CFTR* gene for the diagnosis and carrier testing of cystic fibrosis and the screening of four common mutations from neonatal bloodspots	Fluorescent ARMS (Amplification Refractory Mutation System) allele-specific amplification technology on an ABI 3500 (SOP 033540) using the Elucigene CF-50 (CF-EU2v1) kit (SOP0007) and SOP 0384 for screening from neonatal bloodspots
Genomic DNA extracted in-house from the sample types listed below or received as primary sample type from an external source Whole blood Amniotic Fluid CVS Tissue Mouthwash/saliva Buccal scrape	Molecular Genetics: For detection an analysis of imprinting disorders [as per definitive list]	MS-MLA using ABI 3500 (SOP 031495) and analysed using Coffalyser (SOP 033513) or Gene Marker software (SOP 033325). .
RNA extracted in-house from the sample types listed below or Genomic RNA or cDNA received as primary sample type from an external source Whole blood RNA cDNA	Molecular Genetics: Assessment of the impact of previously - reported sequence variants on splicing	RNA analysis using reverse transcription for detection of splicing abnormalities for common and rare genetic conditions. RNA analysis using cDNA (SOP 032088) and specific oligonucleotide primers (SOP 031721). Analysis using Sanger sequencing and/or gel electrophoresis sizing.
Whole blood and Bone marrow DNA	Qualitative detection of the KIT D816V mutation	ddPCR using the BioRad QX200 Droplet Digital PCR System (SOP 033359)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)	<u>Cytogenetics examinations for the purpose of clinical diagnosis</u>	In-house documented methods incorporating manufacturer's instructions where relevant
Whole blood Bone marrow Solid tumours External cell suspensions	Detection of chromosome rearrangements in comparison with considered normal pattern (ISCN). Detection of chromosome abnormalities associated with: Reproductive disorders Sex chromosome disorders Haemato-oncology disorders Confirmation of genomic rearrangements detected using alternative technologies [as per definitive list]	Conventional Karyotyping Examination of G-banded metaphase chromosomes. Cell culture by in-house procedures using commercial media and reagents. Standard cell harvesting and slide making procedure; Giemsa/Wright's chromosome staining Setting up samples- SOPs: oncology 0363, blood 0167. Cell harvesting by in-house methods -SOPs manual harvesting: oncology 0369, blood 0064 Chromosome preparation SOPs:oncology 0370, bloods, 0114, FISH 0023. Chromosome banding SOPs: Bloods and perinatal 0038, oncology 0371 Automated cell scan, capture and analysis - Cytovision GLS 120 automated karyotyper SOP 018, Cytovision 061, oncology 0372, analysis guidelines 032102.

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)	<u>Cytogenetics examinations for the purpose of clinical diagnosis</u> (cont'd)	In-house documented methods incorporating manufacturer's instructions where relevant
Whole Blood Bone marrow Solid tumours Buccal smears External cell suspensions	Detection and analysis of genomic rearrangements and imbalances. Confirmation of genomic rearrangements detected using alternative technologies. Family follow-up studies	Fluorescence in-situ hybridisation (FISH) by microscopy. FISH - commercial probe kits for constitutional and acquired abnormalities using Thermobrite Statspin hybridisation station.
	Detection of acquired chromosome rearrangements from patients with haemato-oncology disorders[as per definitive list]	SOPs: probe prep 0206 constitutional FISH protocol 0023 & 031960, oncology FISH protocol 0373. Automated cell scan/ capture / analysis Cytovision GLS 120 SOP 032123, scanner(SOP 018), cytovision manual FISH capture(SOP 032123)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)	Cytogenetics examinations for the purpose of clinical diagnosis (cont'd)	In-house documented methods incorporating manufacturer's instructions where relevant
Blood Products of conception Solid tissues	Clinical Cytogenetics: Detection of aneuploidy, maternal cell contamination and confirmation of genetic sex.	QF-PCR. Amplification of microsatellite markers using quantitative fluorescent PCR (Thermal cycler) and fragment analysis using ABI 3500 SOPs: DNA extraction and QF-PCR set up/ABI set up 031677/89 Setting up QSTAR kit 031678, QF-PCR analysis 031682

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND FLUIDS (cont'd)	Cytogenetics examinations for the purpose of clinical diagnosis (cont'd)	In-house documented methods incorporating manufacturer's instructions where relevant
Bone marrow Leukaemic blood pellets RNA cDNA	Detection of cryptic chromosome rearrangements in acute leukaemia	Molecular leukaemic analysis of fusions transcripts. RNA extraction from bone marrow/blood pellets - SOP032088. Generation of cDNA by RT-PCR followed by multiplex nested PCR amplification with specific primers for 28 common fusion transcripts in acute leukaemia and analysis of various breakpoints resulting from BCR-ABL1 fusions (Hemavision kits; SOP 0053). PCR products analysed by agarose gel electrophoresis (SOP 0169).
Bone marrow	CD138 cell separation for Multiple Myeloma	Magnetic separation using the EasySep TM Human CD 138 Positive Selection Kit 11 and EasySep TM Magnet (SOP 031731 Oncology-Culture Hub-Myeloma Plasma Cell Purification)
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