Schedule of Accreditation

issued by

United Kingdom Accreditation Service

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

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Accredited to ISO 15189:2012

Oxford University Hospitals NHS Foundation Trust

Issue No: 011 **Issue date:** 30 October 2024

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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	Location code
Oxford Regional Genetics Laboratories Churchill Hospital Old Road Headington Oxford OX3 7LE	**Carolyn Campbell** (contact details above)	Molecular Genetics CytoGenetics	CH
Level 4 John Radcliffe University Hospital Headley Way Headington Oxford OX3 9DU	**Carolyn Campbell** (contact details above)	Molecular Haematology	JRH

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DETAIL OF ACCREDITATION

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY FLUIDS	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer DNA 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 and automated DNA extraction and quantification using:**	CH
		Automated or semi automated extraction:	**CH**
Blood		Tecan Evo-HSM Robot with ReliaPrep™ Large Volume HT gDNA Isolation kit DNA SOP 2011 62, 440, 2005-0159	
Blood Bone Marrow Fresh Tissue Cultured Cells Saliva/Buccal (Oragene collections) Urine		Promega Maxwell RSC platfrom with Maxwell RSC Blood DNA Kit DNA SOP 2011 62, 2005-0159, 607	
FFPE fixed tissue Blood Spots		Promega Maxwell RSC platfrom with Maxwell RSC FFPE DNA Kit DNA SOP 2005-0159, DNA SOP 2019 609, DNA SOP 2019 608	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY FLUIDS Bloods Pleural Effusions Urine	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer	Documented in house procedures incorporating manufacturer's instructions (where relevant) **Manual DNA extraction** InstaGENE Matrix CYTO SOP 376, DNA SOP 2011 62, 2005-0159	CH
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source		DNA Quantification for QC purposes: Qubit fluorometer, Nanodrop (ND-2000) and Glomax SOPs: CYTO SOP 456, DNA SOP 038, 040, 083.	CH
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Detection of specific SNVs and indels (including confirmatory testing) [Definitive list as per GEN DOC 926]	**Fragment Analysis** Non-fluorescent PCR amplification followed by restriction enzyme digest using: Thermal cycler and gel electrophoresis equipment SOPs DNA SOP 002, 049 090, 107	
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Presence/absence of DNA regions (including confirmatory testing) [Definitive list: GEN DOC 926]	Non-fluorescent PCR amplification followed by agarose gel electrophoresis using: Thermal cyclers and gel electrophoresis equipment SOPs DNA SOP 002. 049, 062, 090, 184	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY 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 or received as primary sample type from external source	Detection of specific SNVs and smallindels (including confirmatory testing) [Definitive list as per GEN DOC 926]	Fluorescent PCR amplification followed by Restriction enzyme digest using fluorescently tagged primers using: thermal cyclers ABI 3730 Analysis using Gene Mapper software SOPs DNA SOP 002, 049	CH
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Detection of expansions including triplet repeats, microsatellite marker analysis, detection of specific SNVs and indels [Definitive list :GEN DOC 926]	Fluorescent PCR amplification (including repeat primed PCR and, Devyser) and fragment analysis using commercially available kits or in-house designed assays using: thermal cyclers ABI 3730 Analysis using Gene Mapper software SOPs CYTO SOP 376, DNA SOP 079, 081, 090, 126, 184, 2006-0014, 2010 34, 049, 225	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY FLUIDS	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer	Documented in house procedures incorporating manufacturer's instructions (where relevant)	CH
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Quantification of mtDNA copy number [Definitive list as per GEN DOC 926]	Real-time PCR using commercially available kits or in-house designed assays Using: ABI 7500 Analysis using 7500 software SOPs DNA SOP 052, 013, 002, GEN SOP 014, 2008 4	CH
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection and quantification of specific SNVs and indels [Definitive list: GEN DOC 926]	<u>Pyrosequencing</u> using: Standard primer design methodology SOP: DNA SOP 731 in-house methods and Qiagen PyroMark ID System SOPs DNA SOP 002, 026, 101, 2010 34	
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Targeted detection and quantification of known germline and somatic variants; SNVs, CNVs [Definitive list: DOC 926]	**Digital Droplet PCR** Reaction setup using in-house or commercial primer/probe mixes using: Techne PCR Machines, Biorad AutoDG for droplet generation and Biorad QX200 Droplet Reader. Analysis using QX Manager Software. SOP 665, 666, 667, 941, 944	CH

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY FLUIDS Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer Detection of nucleic acid sequence variants - SNVs and small indels [Definitive list:GEN DOC 926]	Documented in house procedures incorporating manufacturer's instructions (where relevant) Sanger sequencing using: Standard Primer Design methodology: Robot: Biomek NX-MC(96) ABI 3730 with analysis using Mutation surveyor software and Alamut SOPs DNA SOP 023, 049, 140, 731, GEN SOP 156	CH

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY FLUIDS	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer	Documented in house procedures incorporating manufacturer's instructions (where relevant)	CH
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Screening of large targeted single or multigene panels for nucleic acid sequence variants; SNVs, indels [Definitive list: GEN DOC 926]	Next generation sequencing with target enrichment using; Twist Biosciences generated probes: DNA SOP 0227, 603. Using: Veriti PCR Machines, GloMax®-Multi+' Fluorometer, Agilent 2200 Tape Station, Illumina NovoSeq (located within the West Midlands Regional Genetics Laboratory) or Illumina MiSeq. Analysis of DNA sequence data generated internally or externally using an in-house validated bioinformatics pipeline. Variant interpretation using Alamut SOPs: DNA SOP 2019 602, , 2019 606, 2020 621, 2020 622, 2020 628, 712	
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	SNVs, indels and CNVs [Definitive list:GEN DOC 926]	Illumina NexteraXT Using: Dyad PCR Machines Glomax Multi+ Fluorometer, Illumina MiSeq, Agilent 2200 Tape Station Analysis of DNA sequence data generated either internally or externally using an in-house validated bioinformatics pipeline DNA SOP 0227, 421, 2015 327, 2019 592 and 2019 593.	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY FLUIDS Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer SNVs, indels and CNVs [Definitive list:GEN DOC 926]	Documented in house procedures incorporating manufacturer's instructions (where relevant) SOPHiA Genetics custom panel kit DNA SOP 0460, 2015 316, 0227 Using: Veriti (Applied Biosystems) PCR Machines'GloMax®-Multi+' Fluorometer, Illumina MiSeq, Agilent 2200 Tape Station Analysis using the Sophia Genetics bioinformatics DDM pipeline Variant interpretation using Alamut DNA SOP 528, 2015 327.	CH
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from external source	Detection of whole or partial gene deletions, duplications probe specific SNVs and indels and methylation status	**Multiplex Ligation Probe Analysis (MLPA) and methylation specific MS-MLPA** Using: Multiplex Ligation-dependant Probe Amplification (MLPA) (using MRC Holland kit) or custom design MLPA (using a kit template supplied by MRC-Holland) Methylation specific MLPA kit (MS-MLPA) Using Thermal cyclers and ABI 3730 with analysis using Coffalyser software DNA SOP 2015 374, 050, 128, 204	CH

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY FLUIDS Products of conception Biopsies (includes skin) Tissues CSF Pleural Fluid, Ascites Whole Blood Bone Marrow	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer G-Banding/Karyotyping Detection of chromosomal rearrangements or aberrations arising from: *(e.g)* Developmental disorders Reproductive medicine disorders Haematological/Oncology disorders (Preparative Pre-examination steps listed first)	Documented in house procedures incorporating manufacturer's instructions (where relevant) <u>G-Banding/Karyotyping</u> Manual culturing using commercial media, processing (cell harvesting and slide preparation and banding) of human tissue to provide metaphase cells Using: Safety cabinets, incubators, centrifuges, hotplate, SOPs: CYTO SOP 67, 111, 185, 271, 414 Analysis: Karyotype analysis of metaphase chromosomes light microscopy using: Microscopes, Cytovision Image Capture System	CH
Products of conception Biopsies (includes skin) Tissues CSF Pleural Fluid, Ascites Whole Blood Bone Marrow Urine Buccal swabs	(Preparative Pre-examination steps for FISH)	Direct preparation (without culture) to provide fixed interphase cells and Processing of FFPE sections for material suitable for FISH Using: Hotplate, fume hood, water bath, and Hybrite/Thermobrite SOPs 67, 132, 271, 340, 352	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND BODY FLUIDS Fixed cultured cells as prepared as detailed in G-banding/Karyotyping section Fixed uncultured cells FFPE sections	Genomic analysis for the purpose of clinical diagnosis of rare disease and cancer Targeted analysis of the genome for copy number alterations, balanced chromosome rearrangements, mosaicism and formation of gene fusions. Using locus specific probes for: Translocations and/or inversion Deletions Fusions Copy Number [Definitive list:GEN DOC 926]	Documented in house procedures incorporating manufacturer's instructions (where relevant) Fluorescence in situ hybridisation (FISH) using: Hybrite/Thermobrite, UV light box, microfuge fume hood and waterbath. FISH analysis by use of fluorescently tagged probes and fluorescence microscopy to detect copy number and spatial location of specific regions of the genome using: SOPs CYTO SOP 67, 132, 271, 340, 352, 364, 412, 414 FISH analysis by use of fluorescently tagged probes and fluorescence microscopy to detect copy number and spatial location of specific regions of the genome using: Cytovision Image Capture System	CH

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND FLUIDS	Molecular genetic analysis for genetic mutations and variants	Procedures documented in manufacturer's equipment manuals in conjunction with documented in- house procedures by the following methods:	JRH
Blood Bone Marrow CVS Amniotic Fluid FFPE tissue		Automated DNA extraction and quantification using the QIAGEN Symphony and in-house procedures: Haem SOP 940 HC 2531 HC 2271 HC 2315	JRH
Blood Bone Marrow		Semi-automated RNA extraction and cDNA preparation using the QIAGEN Qiacube and in house procedures HC 2302	
Blood Bone Marrow FFPE		Automated Paramegnetic particle nucleic acid extraction and purification using the Promega Maxwell system; Haem SOP 2599: DNA using AS1321 kit Haem SOP 2608: RNA using AS410 kit Haem SOP 2611: FFPE using RSC FFPE DNA plus kit	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND FLUIDS (cont'd)	Molecular genetic analysis for genetic mutations and variants (cont'd)	Procedures documented in manufacturer's equipment manuals in conjunction with documented in- house procedures by the following methods:	JRH
Blood DNA	Somatic myeloid disorder diagnostic gene panel. Mutations in:	NextSeq 550 Next Generation Sequencing and custom designed commercial panel (SeqCap EZ HyperCap (Roche)	
	ASXL1 (NM_015338.5), ATRX (NM_000489.3), CBL (NM_005188), CBLB (NM_170662.3), CBLC (NM_012116.3), CSF3R (NM_156039.3), DNMT3A (NM_022552), ETV6 (NM_001987.4), EZH2 (NM_004456.4), HRAS (NM_005343.2), IDH1 (NM_005896.2), IDH2 (NM_002168.2), FLT3 (NM_004119), JAK2 (NM_004972.3), KIT (NM_000222.2), KRAS (NM_033360.2), MPL (NM_005373), NPM1 (NM_002520.6), NRAS (NM_002524.4), PDGFRA (NM_006206.4), PHF6 (NM_032458.2), PTEN (NM_000314), RUNX1 (NM_001754.4), SETBP1 (NM_015338.5), SF3B1 (NM_012433.2), SRSF2 (NM_001195427.1), TET2 (NM_001127208.2), TP53 (NM_000546.5), U2AF1 (NM_001025203.1), WT1 (NM_024426.4) and ZRSR2 (NM_005089.3)	Haem SOP 2623 Haem SOP 2621 Haem SOP 2618 Haem SOP 2619	JRH

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND FLUIDS (cont'd)	Molecular genetic analysis for genetic mutations and variants (cont'd)	Procedures documented in manufacturer's equipment manuals in conjunction with documented in- house procedures by the following methods:	JRH
Next Generation Sequencing Data in the form of BAM files, variant lists, and gene- level coverage data received from Oxford Regional Genetics (UKAS 8694)	Rare anaemia diagnostic gene panel. Mutations in: ABCB7 (NM_004299.3), ALAS2 (NM_000032.4), ALDOA (NM_000034.3), ANK1 (NM_000037.3), C15orf41 (NM_001130010.1), CDAN1 (NM_138477.2), ENO1 (NM_001428.3), EPB41 (NM_004437.3), EPB42 (NM_000119.2), G6PD (NM_001042351.2), GATA1 (NM_002049.3), GATA2 (NM_032638.4), GCLC (NM_001498.3), GPI (NM_000175.3), GPX1 (NM_000581.2), GSR (NM_000637.3), GSS (NM_000178.2), HK1 (NM_000188.2), KIF23 (NM_138555.2), KLF1 (NM_006563.3), LPIN2 (NM_014646.2), NT5C3A (NM_016489.12), PFKM (NM_000289.5), PGK1 (NM_000291.3), PIEZO1 (NM_001142864.2), PKLR (NM_000298.5), RHAG (NM_000324.2), RPL11 (NM_000975.3), RPL26 (NM_000987), RPL27 (NM_000988.3), RPL35A (NM_000996.2), RPL5 (NM_000969.3), RPL9 (NM_000661.4), RPS10 (NM_001014.4), RPS17 (NM_001021.3)	Analysis and reporting of data only using Alamut Visual and IGV software HC 2154 HC 2152 Haem SOP 2457	JRH

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND FLUIDS (cont'd)	Molecular genetic analysis for genetic mutations and variants (cont'd)	Procedures documented in manufacturer's equipment manuals in conjunction with documented in- house procedures by the following methods:	JRH
Next Generation Sequencing Data in the form of BAM files, variant lists, and gene- level coverage data received from Oxford Regional Genetics (UKAS 8694)	Rare anaemia diagnostic gene panel (cont). Mutations in: RPS19 (NM_001022.3), RPS24 (NM_033022.3), RPS26 (NM_001029.3), RPS27 (NM_001030.4), RPS29 (NM_001032.4), RPS7 (NM_001011.3), SBDS (NM_016038.2), SEC23B (NM_006363.4), SLC11A2 (NM_000617.2), SLC25A38 (NM_017875.2), SLC2A1(NM_006516.2), SLC4A1 (NM_000342.3), SPTA1 (NM_003126.2), SPTB (NM_000347.5), TMPRSS6(NM_153609.2), TPI1(NM_000365.5).	Analysis and reporting of data only using Alamut Visual and IGV software HC 2154 HC 2152 Haem SOP 2457	JRH
Blood Bone Marrow	BCR-ABL analysis (ALL, CML) Diagnosis and monitoring of MRD (p210, p190 only)	Published Multiplex RT-PCR method using Qiagen, Rotorgene, Commercial QPCR (Qiagen) kit for MRD, Pyrosequencing using QIAGEN Pyromark Q24 HC 2301 HC 2302 HC 2303 HC 2304 HC 2306	JRH

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND FLUIDS (cont'd)	Molecular genetic analysis for genetic mutations and variants (cont'd)	Procedures documented in manufacturer's equipment manuals in conjunction with documented in- house procedures by the following methods:	
	Acute Leukaemia Diagnosis:	RE-PCR using Biometra Thermocycler	
Blood Bone Marrow	FLT-3 D835	HC 2118	JRH
	NPM1	In house fragment analysis method using ABI instrumentation HC 2119	
Blood Bone Marrow	Acute Leukaemia MRD monitoring (PML-RARA, Inv16, AML-ETO)	Commercial QPCR (Qiagen) kit for MRD using QIAGEN Rotorgene, HC 2304	
Blood Bone Marrow Tissue	Lymphoma B Clonality (IgH rearrangements)	Commercial (Invivoscribe) fragment analysis using ABI instrumentation Haem SOP 1163	
	Lymphoma T Clonality (TCR re-arrangements)	HC 2268 HC 2267 Haem SOP 1163	
Blood Bone Marrow	Jak-2 mutation screen (V617F)	In house ARMS-PCR using Biometra thermocycler, Pyrosequencing using the QIAGEN Rotorgene HC 2261	JRH
Blood Bone Marrow	Chimerism STR pattern analysis (Chimerism) to detect mxed samples (e.g. Maternal contamination / Chimerism	Promega Powerplex Fusion commercial kit, and fragment analysis using ABI instrumentation HC 2381 HC 2141	
Blood Bone Marrow FFPE	Detecting L265P point mutation of the MYD88 gene	Allelic specific real-time PCR assay using hydrolysis probes ABI Real Time 7500 Haem SOP 2468.	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location
HUMAN TISSUES AND FLUIDS (cont'd)	Molecular genetic analysis for genetic mutations and variants (cont'd)	Procedures documented in manufacturer's equipment manuals in conjunction with documented in- house procedures by the following:	
FFPE DNA	EGFR common mutation screen	COBAS 4800 system, RT-PCR and commercial (COBAS) kits Haem SOP 578	JRH
FFPE DNA	BRAF mutation screen (V600E)	COBAS 4800 system, RT-PCR and commercial (COBAS) kits Haem SOP 579	
FFPE DNA	Somatic Cancer 50 gene diagnostic panel. Mutations in: TP53 NM_000546.5, PTEN NM_000314.4, PIK3CA NM_006218.2, PDGFRA NM_006206.4, KRAS NM_004985.3, NRAS NM_002524.4, KIT NM_000222.2, EGFR NM_005228.3, BRAF NM_004333.4. ERBB4, FGFR1, ERBB2, MET, FLT3, FGFR3, GNAS, SMARCB1, CTNNB1, CDKN2A, ABL1, NOTCH1, ATM, PTPN11, SMO, SMAD4, VHL, NPM1, MPL, CSF1R, HRAS, JAK3, AKT1, IDH1, CDH1, FGFR2, SRC, KDR, ALK, JAK2, RB1, MLH1, HNF1A, APC, RET, STK11, FBXW7, EZH2, GNA11, GNAQ and IDH2	Thermofisher Ampliqseq Next Generation Sequencing commercial panel and Ion Torrent next generation sequencing. Haem SOP 624 Haem SOP 491 Haem SOP 625 HC 2411 HC 2408	JRH
FFPE DNA	Acquired NGS panel using custom AmpliSeq primer pools and S5 Ion Torrent Sequencing DNA -AmpliSeq panel on S5 Ion Torrent	Custom designed Ampliseq Next Generation Sequencing Panels and Ion Torrent Next Generation Sequencing (ThermoFisher). Haem SOP 625 Haem SOP 2657 Haem SOP 491	
END			