Schedule of Accreditation

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United Kingdom Accreditation Service

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

8840

Accredited to ISO 15189:2022

Cambridge University Hospitals NHS Foundation Trust

Issue No: 013 **Issue date:** 07 August 2026

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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 BODY FLUIDS	Genomic analysis for the purpose of clinical diagnosis of rare and inherited disease and cancer Sample preparation, 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) **Sample preparation:**
Whole Blood Bone Marrow		Red Cell lysis and storage in Qiagen Nucleon B, RLT or Tri-Reagent CU-EX-SOP-34, CU-EX-SOP-23, CU-EX-SOP-5 and CU-EX-SOP-8
Whole Blood		Separation of mononuclear cells from peripheral blood or bone marrow using Lymphoprep CU-EX-SOP-31 **Manual and automated DNA extraction and quantification using:** **Automated Extraction:**
Whole Blood,Buffy Coat CVS/Amniocentesis Tissues Guthrie Spots		Automated DNA extraction using Qiagen EZ1 CU-EX-SOP-9

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare and inherited disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Whole Blood Bone Marrow Body Fluid Tissue Lineage specific fractions		Automated DNA extraction using QIACube CU-EX-SOP-3 CU-EX-SOP-5
Whole Blood Bone Marrow Body fluid Human tissue (fresh, frozen or fixed) Formalin fixed paraffin embedded tissue		Automated DNA extraction using Promega Maxwell CSC48 CU-EX-SOP-43 CU-EX-SOP-44 CU-EX-SOP-46 CU-EX-SOP-47
		Manual extraction:
Body fluid and human tissue (fresh, frozen or fixed) Formalin fixed parrafin embedded tissue Bone Marrow		Manual DNA extraction usingQiagen DNEASY kit CU-EX-SOP-15 CU-EX-SOP-16 CU-EX-SOP-23
Bone Marrow		Extraction of DNA from DNA lysis buffer (Q-AL or nucleon B) CU-EX-SOP-22
Saliva and Buccal swabs		Manual DNA extraction using-Oragene Purifyer CU-EX-SOP-11
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: Nanodrop ND-1000 spectrophotometer and Qubit fluorometer CU-EQ-SOP-9 CU-EX-SOP-21
Genomic DNA extracted in-house from human body fluids and tissues or received as a primary sample type from an external source	DNA Quantification for QC purposes	DNA Quantification using the Hamilton STAR Liquid Handler and CLARIOstar Plate Reader CU-EX-SOP-28

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare and inherited disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
DNA Clean up		Qiagen QiaAmp DNA micro kit CU-EX-SOP12
Whole Blood, bone marrow, body fluid and human tissue (fresh, frozen or fixed)		**Automated Extraction:** Automated RNA extraction and conversion to cDNA using QIACube, NJ Research (Biorad) thermal cycler and Syngene Genius Bioimaging System CU-EX-SOP-3 CU-EX-SOP-19 CU-EX-SOP-32
	RNA extraction and preparation of cDNA, quantification and quality check for subsequent in-house analysis (see below), referral to specialist centres and long term storage	**Manual and automated RNA extraction, conversion to cDNA and quantification using:**
Human Tissues and body fluids, Whole Blood; Bone Marrow and human tissue (fresh, frozen or fixed) Formalin fixed paraffin embedded tissue		Automated RNA extraction using Promega Maxwell CSC48 CU-EX-SOP-43 CU-EX-SOP-44 CU-EX-SOP-46 CU-EX-SOP-47 **Manual Extraction**
cDNA prepared in-house from the sample types listed above or received as primary sample type from an external source	Quantification of RNA transcripts	Quantitative Real Time PCR Real time PCR using Rotogene 6000 series – QIAgen CU-UK-DTA-8

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare and inherited disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Genomic DNA and cDNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Detection of nucleic acid sequence variants - SNVs and small indels-	**Sanger Sequencing** Sanger Sequencing Analysis using Mutation Surveyor by SoftGenetics CU-TS-SOP-29
Genomic DNA and cDNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Detection of nucleic acid sequence variants - SNVs and small indels- See CU-UK-DTA-3	**Sanger Sequencing** Using: Standard Primer Design methodology: CU-BI-SOP-2 PCR/ Thermal cyclers, ABI 3730, Biomek robotics CU-CM-SOP-5 CU-CM-SOP-13 CU-TS-SOP-28 CU-CM-SOP-31 CU-TS-SOP-45 CU-TS-SOP-65 CU-TS-SOP-58, CU-TS-SOP-62
Genomic DNA and cDNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Next Generation Sequencing for Small Variants	TWIST WES Comprehensive exome panel CU-NG-SOP-8 CU-BI-MAN-2 CU-BI-MAN-1 CU-BI-MAN-6 CU-NG-SOP-14 CU-NG-SOP-10 CU-RP-SOP-9
Genomic DNA and cDNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Next Generation Sequencing	TWIST Myeloid Panel CU-NG-SOP-7 CU-RP-SOP-5 CU-BI-MAN-1

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare and inherited disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Genomic DNA and cDNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Next Generation Sequencing	**TWIST Core, Endocrine, Neurology (CEN)** CU-NG-SOP-14 CU-BI-MAN-1 CU-NG-SOP-8 CU-BI-MAN-2 CU-RP-SOP-9
Genomic DNA and cDNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Detection of SNVs and small indels See CU-UK-DTA-8	**Real Time PCR** **Using Applied biosystems Realtime 7500 analyser** CU-CM-SOP-21 Using Rotogene 6000 series – QIAgen CU-CM-SOP-16 CU-CM-SOP-26 CU-CM-SOP-22
cDNA prepared in-house from the sample types listed above or received as primary sample type from an external source	Detection of fusion genes CU-UK-DTA-8	**Quantitative Real Time PCR** Real time PCR using Rotogene 6000 series – QIAgen CU-CM-SOP-51
cDNA prepared in-house from the sample types listed above or received as primary sample type from an external source	Detection of gene fusions CU-UK-DTA-8	Quantification Real Time PCR using Applied Biosystems Realtime 7500 analyser CU-CM-SOP-29 CU-CM-SOP-9
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source	Detection of SNVs and small indels CU-UK-DTA-9	High Resolution Melt Analysis using the Rotorgene 6000 series – QIAgen. CU-CM-SOP-1 CU-COM-SOP-38 CU-TS-SOP-74

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare and inherited disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Formalin fixed paraffin embedded (FFPE) tissue	Sample preparation, DNA and RNA extraction, quantification and quality check for subsequent in-house analysis, referral to specialist centres and long term storage.	**Manual or Automated Extraction of nucleic acids from formalin fixed paraffin embedded (FFPE) tissue using MagMax** CU-EX-SOP-33
Genomic DNA extracted in-house or received as primary sample type from an external source	Determination of methylation status	**Pyrosequencing** Pyrosequencing of Bisuplite modified DNA using Pyromark Q24 Pyrosequencer and Pyromark PCR kit. MGMT methylation status CU-CM-SOP-17 CU-CM-SOP-19

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd) Genomic DNA extracted in-house or received as primary sample type from an external source	<u>Genomic analysis for the purpose of clinical diagnosis of rare and inherited disease and cancer</u> (cont'd) Determination of fragment length size and detection of deletions, fusion deletions, known SNVs and indels, microsatellites, repeat expansion, linkage markers for the purpose of genotyping at specific known gene loci See CU-UK-DTA-8	Documented in house procedures incorporating manufacturer's instructions (where relevant) **Fluorescent Based Fragment Analysis** **Using:** Fluorescent Fragment Analysis using ABI 3730 and ABI 3500 xl genetic analysers and GeneMarker software CU-TS-SOP-34 Commercial Methods: Fluorescent Fragment Assays: Fluorescent ARMS using the Elucigene kits CU-TS-SOP-17 and CU-TS-SOP-9 QF-PCR kit (QF-PCR) - Devyser Kits CU-TS-SOP-44 CU-TS-SOP-43 And: In house Fluorescent Fragment Assays using PCR and allele specific PCR and PCR of linked markers CU-CM-SOP-14 CU-TS-SOP-21 CU-TS-SOP-23 CU-TS-SOP-30 CU-TS-SOP-36 CU-TS-SOP-41 Fluorescent Fragment Analysis using and 3730, 3500 genetic analysers and GeneMarker or Genemapper software CU-TS-SOP-34 and CU-RP-SOP-29

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd)	Genomic analysis for the purpose of clinical diagnosis of rare and inherited disease and cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Genomic DNA extracted in-house or received as primary sample type from an external source	Determination of fragment size length, Detection of deletions, fusion deletions, known SNVs, repeat expansions and inversions for the purpose of genotyping at specific known gene loci - See CU-UK-DTA-4	**Gel electrophoresis based Fragment Analysis** In house methods for: Long range PCR, MS-PCR, , multiplex PCR followed by gel electophoresis. CU-CM-SOP-44 CU-TS-SOP-23
Genomic DNA extracted in-house from human body fluids and tissues or received as primary sample type from an external source	Determination of fragment length size and detection of deletions, fusion deletions, known SNVs and indels, microsatellites, repeat expansion, linkage markers for the purpose of genotyping at specific known gene loci in gene C9ORF72.	Fluorescent fragment analysis using ABI 3500 and ABI 3730 genetic instruments: Frontotemporal Dementia (FTD) and/or Amyotrophic Lateral Sclerosis (ALS) (C9ORF72 gene) CU-TS-SOP-34 CU-RP-SOP-33

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd)	Cytogenetic examinations , neoplastic genetics including haemato-oncology, - detection of sub microscopic chromosomal imbalance (gains and losses) expressed as changes to copy number and copy neutral loss/absence of heterozygosity	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Sequencing data received from internal and external sources	Detection of genomic variation with potential clinical utility, including small variants (SNV & indel) and structural variants (CNV, insertion, inversions and translocations resulting in fusion, ITD and PTD)	Detection of variants for clinical reporting of patients with rare disease and cancer: alignment and variant calling. GH-BI-POL-04
Whole blood Amniotic fluid Chorionic villus samples Bone Marrow Biopsy Material Body Fluids	G-Banding/Karyotyping Detection of chromosomal rearrangements or aberrations arising from: Prenatally detected Disorders Developmental disorders Reproductive medicine disorders Haematological/Oncology disorders (Preparative Pre-examination steps listed first)	**Manual culturing and processing of human tissue to provide metaphase cells:** using commercial media and: CU-CT-SOP-2. CU-CT-SOP-22, CU-CT-SOP-8,CU-CT-SOP-5 CU-CT-SOP-17, CU-CT-SOP-14, CU-CT-SOP-20 Manual harvesting using in house methods : CU-CT-SOP-3 (whole blood and prenatal and tissue cultures CU-CT-SOP-7). CU-CT-SOP-13 Slide Preparation, coverslipping and storage : CU-CT-SOP-4 and CU-CT-SOP-19 **Microscopic Analysis and reporting of G banded chromosomes:** Using: Brightfield microscopes and GenASIs image capture and anlaysis software CU-CT-SOP-25, CU-CT-SOP-26, CU-CT-SOP-27, CU-RP-SOP-38, CU-CT-SOP-15 and CU-RP-SOP-21, CU-RP-SOP-27

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES AND BODY FLUIDS (cont'd)	Cytogenetic examinations (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Genomic DNA extracted in house and received as primary samples from external sources	Detection of pathogenic and likely pathogenic DNA copy number variants and copy number neutral loss of heterozygosity (in diagnostic samples)	**SNP Array** Using Infinium Global Screening Array (GSA) v 3 kit with Illumina Infinium HTS Assay and Tecan Robot Analysis using Illumina iScanner with Illumina Genome Studio software and NxClinical software CU-MA-SOP-1 CU-RP-SOP-4
Genomic DNA extracted in house and received as primary samples from external sources	Detection of whole exon deletions/duplications and specific methylation abnormalities See CU-UK-DTA-5	**Multiplex Ligation Probe Analysis (MLPA) and methylation specific MS-MLPA** using: in house or commercial MRC Holland kits Thermal cyclers and 3500 / 3730 Genetic Analysers Analysis using GeneMarker CU-TS-SOP-37 CU-TS-SOP-16 CU-TS-SOP-42
Fixed cell slides from human tissues and body fluids: Amnoitic fluid CVS Stemcells Wholeblood Bone Marrow CSF Biopsy Material Body fluids FFPE Tissue	Detection and analysis of genomic rearrangements and imbalances. Confirmation of genomic rearrangements - using locus specific probes: Deletion Copy Number/Amplification See CU-UK-DTA-2	**Fluorescent in-situ hybridisation (FISH)** Using: Cytocell and Vysis commercial kits, Vysis HYBrite hotplate and applied imaging (Leica) Analysis using: Cytovision analysis software and and fluorescent microscope system CU-FI-SOP-6, CU-FI-SOP-5, CU-FI-SOP-12, CU-FI-SOP-11, CU-FI-SOP-4, and CU-FI-SOP-8
END		