Schedule of Accreditation

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Great Ormond Street Hospital for Children NHS Foundation Trust

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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
Address Regional Genetics Laboratory Barclay House (Levels 5 & 6) 37 Queen Square London WC1N 3BH	**Local contact** Lucy Jenkins	Constitutional and rare diseases Molecular genetics Cytogenetics	RGL
Address SIHMDS – Acquired Genomics Level 2 Camelia Botnar Laboratories Great Ormond Street Hospital for Children NHS Foundation Trust London WC1N 3JH	**Local contact** Dariusz Ladon	Acquired genomics as part of SIHMDS service Molecular Genetics Cytogenetics	CBL

DETAIL OF ACCREDITATION

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS	Molecular Genetics examination procedures for the purposes of clinical diagnosis	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	
Blood / bone marrow / CSF/ fresh tissue / CSF / frozen tissue / body fluid	Extraction	Using Maxwell 16 and Nanodrop one (spectrophotometer) And in house SOP: HSOP 782 And RGS MOT9035	CBL
FFPE section rolls	Extraction	Using Maxwell 16 Nanodrop one (spectrophotometer) And in house SOP: HSOP 782 And RGS MOT9038	CBL
Blood / bone marrow / fresh tissue / frozen tissue	Extraction	Using Maxwell 16 and Nanodrop one (spectrophotometer) And in house SOP: HSOP 782 And RGS MOT9036	CBL
FFPE section rolls	Extraction	Using Maxwell 16 and Nanodrop one (spectrophotometer) And in house SOP: HSOP 782 And RGS MOT9039	CBL
	DNA and RNA profiling for detection of abnormal sequences associated with acquired cancer	In-house documented methods and following maufacturers' instructions	
DNA and RNA. Extracted from primary samples as detailed above		Allele specific Polymerase chain reaction (AS-PCR) and reverse transcription using 7500 real-time PCR system Process SOP: RGS MOT9048 Analysis SOP: RGS ANA9049	CBL
DNA. Extracted from primary samples as detailed above		Fragment analysis using 3500 Genetic analyser Tetrad thermal cycler Process SOP FSOP 045 (RGS MOT 9045) Anaysis SOP: FSOP 046 (RGS ANA9046)	CBL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	<u>Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)</u> DNA and RNA profiling for detection of abnormal sequences associated with acquired cancer (cont'd)	In-house documented methods and following maufacturers' instructions	
DNA. Extracted from primary samples as detailed above		High resolution Melt curve analysis using 7500 real-time PCR system (ARMs based) Process SOP: FSOP 032 (RGS MOT9032) Analysis SOP: RGS ANA9034	CBL
PCR product		Sanger sequence analysis using Tetrad thermal cycler Exasapit clean up of PCR products SOP: RGSMOT9009 Cycle sequencing SOP: RGS MOT9005 Clean up of big dye products SOP: RGS MOT9040 Using Mutation Surveyor Software to analyse DNA variants Analysis SOP: RGS ANA9037 Sequence Analysis and Mutation Classification Analysis SOP: FSOP 025 (RGS ANA9025)	CBL
RNA		Reverse transcription-PCR using Thermal cycler Reverse transcription SOP: RGS MOT9013 Reporting Agarose Gel Electrophoresis Analysis SOP: RGS ANA9060 Electrophoresis gel tank and power pack Agarose Gel Analysis of DNA and RNA SOP: HSOP 646 (RGS MOT9646)	CBL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	<u>Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)</u> DNA and RNA profiling for detection of abnormal sequences associated with acquired cancer (cont'd)	In-house documented methods and following maufacturers' instructions	
DNA		Methylation arrays using Bisulfite conversion and FFPE restore SOP: RGS MOT 9053 Methylation array analysis using bCHAPs pipeline Analysis SOP: RGS ANA9055	CBL
DNA		Multiplex Ligation Probe Amplification (MLPA) using Tetrad thermal cycler Multiplex Ligation Probe Amplification (MLPA) Process SOP: RGS MOT9015 MLPA Analysis using GeneMarker Software Analysis SOP: RGS ANA9027	CBL
DNA		Next Generation Sequencing NGS panel and Sequencing analysis using the Trusight Myeloid Panel SOP: FSOP 659 (RGS MOT9659) NGS analysis of Myeloid panel using Variant studio Trusight myeloid NGS panel Panel of 54 genes / hotspots	CBL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	Documented methods for DNA and RNA extraction using commercial kits and extraction	
Whole Blood Amniotic Fluid CVS Tissue Samples	Extraction of DNA	RGS MOT0101 – DNA extraction from cultured cells RGS MOT0102 – DNA salting out method for cell pellets RGS MOT0103 - DNA salting out method for CVS RGS MOT0104 - DNA salting out method for tissue RGS MOT0106 – Extraction of DNA from cells/blood spots by EZ1 RGS MOT0110 – DNA extraction from tissue using FUJIQuickGene-Mini80 RGS MOT0111 – DNA extraction from blood or saliva using FUJIQuickGene-610L RGS MOT0112 - DNA extraction using the iGENatal Extraction kit RGS MOT0128 – DNA extraction using the Chemagic Star	RGL
DNA. Extracted from primary samples as detailed above.	DNA and RNA profiling for detection of abnormal sequences for common and rare inherited/predisposing genetic disease condition	Sanger Sequencing techniques and targeted testing using ABI 3730XL, ABI 3130XL, Beckman NX Robotic Workstations, Tetrad Thermal Cyclers and Shimadzu MultiNA in accordance with SOPs: - RGS MOT0114 – Excel Instructions for streamlined robot PCR & Sequencing RGS MOT0301 – Robot PrePCR Setup RGS MOT0302 – Robot pre-PCR (96) –cleanse/AMPure setup RGS MOT0303 – PCR purification, sequencing & manual purification RGS MOT0304 – Streamlined sequencing system RGS MOT0305 – Streamlined Robot PCR setup RGS MOT0308 – Use of MicroChip Electrophoresis System (MCE) MultiNa	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
		RGS LAB0107 – DYAD/TETRAD PCR Machines	
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	In-house documented methods and following maufacturers' instructions	
DNA. Extracted from primary samples as detailed above.	DNA and RNA profiling for detection of abnormal sequences for common and rare inherited/predisposing genetic disease conditions (cont'd)	OLA PCR using GeneAmp. PCR products analysed using ABI 3130 and results analysis using GeneMapper according to SOP(s): RGS DGE0301 – Cystic Fibrosis – OLA v3 PCR Summary RGS LAB0106 – GeneAmp 9700PCR Machine SOP RGS LAB0101 - ABI 3130xl General Use SOP	RGL
DNA extracted from primary samples as detailed above.		Direct PCR and Amplidex FMR1 PCR Kits (P/N 76008 VH Bio) to amplify across the *FMR1* gene triplet repeat by Triplet Primed PCR. Detection is using fragment analysis on a capillary sequencer (ABI 3130XL) as documented in SOP(s): RGS DGE0501 Fragile X PCR RGS DGE0506 Fragile X - Asuragen Amplidex FMR1 PCR	RGL
DNA. Extracted from primary samples as detailed above.		Restriction enzyme digest performed manually according to in-house method described in SOP(s): RGS MOT0307 – Manual PCR using KCl, NH4 or Megamix buffers RGS MOT0308 - RGS MOT0308 – Use of MicroChip Electrophoresis System (MCE) MultiNa RGS MOT0310 – Agarose Gel preparation and Electrophoresis	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	<u>Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)</u>	In-house documented methods and following maufacturers' instructions	
DNA extracted from primary samples as detailed above.	DNA and RNA profiling for detection of abnormal sequences for common and rare inherited/predisposing genetic disease condition (cont'd)	Multi-ligation Probe Analysis (MLPA). Performed by manual in-house method or using the Biomek NK span-8 system. SOP(s): RGS MOT0401 Setting up MLPA using the Automated system RGS MOT0406 Manual MLPA set up (molecular) RGS ANA0201 Capillary Electrophoresis of MLPA and QF-PCR products using the ABI 3730 Genetic Analyser RGS ANA0205 Analysis of MLPA using GeneMarker Software (Molecular)	RGL
DNA extracted from primary samples as detailed above		Next Generation Sequencing supported by PCR amplification or hybridisation. Sequencing is carried out using the Illumina MiSeq sequencer. SOPs: RGS MOT0216 MiSeq Operation RGS MOT0214 Library Preparation for Illumina Custom Amplicon Panels RGS MOT0226 SureSelect Library Preparation RGS MOT0228 SureSelect Library Prep Using the NGS Express RGS ANA0502 NGS Bioinformatics pipeline: Variant calling RGS ANA0503 NGS bioinformatics pipeline: Coverage Checker RGS ANA0504 NGS analysis: Excel instructions to generate results	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	Non-Invasive Prenatal examination procedures for the purposes of clinical diagnosis and antenatal screening		
Maternal Blood plasma	Cell Free Fetal DNA (cffDNA) extraction from maternal plasma.	RGS MOT0201 – ffDNA – Sample collection and separation of plasma DNA extraction based on Magnetic bead technology using the Qiagen Symphony SP DNA extractor RGS MOT0224 for cffDNA extraction	RGL
DNA extracted from Maternal Plasma	Non-invasive prenatal diagnosis (NIPD)	Real Time PCR analysis of cffDNA to determine fetal gender performed on the ABI7300 as documented in SOP(s): RGS MOT0201 ffDNA Sample Collection and Separation of Plasma; RGS DGE 0908 - ffDNA RQ-PCR Sexing	RGL
DNA extracted from Maternal Plasma	Non-invasive prenatal diagnosis (NIPD)	Next Generation Sequencing performed on the MiSeq according to the in house method(s) to carry out non-invasive prenatal diagnosis for single gene disorders where the mutation is paternally derived, or *de novo* as documented in SOP(s): RGS MOT0201 ffDNA Sample Collection and Separation of Plasma; RGS MOT0215 - Using the QIAcube for MinElute® PCR Purification RGS MOT0216 – MiSeq operation RGS DGE0911 Non-invasive prenatal diagnosis for de novo and paternal mutations (MiSeq)	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	Non-Invasive Prenatal examination procedures for the purposes of clinical diagnosis and antenatal screening (cont'd)		
DNA extracted from Maternal Plasma	Non-invasive Prenatal Testing for Aneuploidy (NIPT) – Detection of trisomies 21, 13 & 18 and monosomy X	Analysis of cell free fetal DNA in the maternal blood by Next Generation Shotgun Sequencing to detect variation from normal in the copy number for these chromosomes that can be attributed to fetal aneuploidy. Documented in SOPs: RGS MOT0222 HiSeq Rapid Run Mode RGS MOT0223 HiSeq Rapid Run – Library Prep Dilutions RGS MOT0221 Non Invasive prenatal testing for aneuploidy - Agilent XT2 Bravo Prep RGF LAB0227 - HiSeq run mode switch over (Rapid>High Output>Rapid) RGS ANA0507 - RAPID-R analysis RGS ANA0506 - HiSeq data transfer and backup RGF LAB0226 HiSeq Maintenance Log RGF MOT0113 HiSeq rapid sequencing lab tracking form	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd) DNA extracted from: Blood, Saliva, Chorionic villus tissue, Uncultured amniocytes, Cultured cells from chorionic villus samples, Cultured cells from amniotic fluid, Saliva, Thymus	<u>Cytogenetics examination procedures for the purposes of clinical diagnosis</u> Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders	Documented methods for DNA extraction, quantification, labelling, hybridisation, identification, data analysis and reporting using one or a combination of the techniques below by in-house procedures and/or commercially verified procedures: - Microarray analysis by Affymetrix 750K SNP Array supported by: Extraction of DNA as documented in SOPs: RGS MOT0101 DNA extraction from cultured cells RGS MOT0111 DNA extraction from blood/saliva (FujiFilm QuickGene-610L) RGS MOT0128 DNA extraction using the Chemagic Star RGS MOT0115 Measuring Optical Density of DNA Samples RGP SAB0001 Sample acceptance guidelines (Cytogenetics) RGS MOTO606 Processing of Affymetrix Cytochip Microarrays RGS MOTO122 Feature extraction for Affymetrix Microarrays using ChAS RGS MOTO124 Upload of Microarray data files into InfoQuant cnTrack RGS ANA0408 Microarray analysis and checking using InfoQuant Fusion and cnTrack RGS ANA0404 Microarray analysis and checking using InfoQuant CGHFusion RGS REP0101 Microarray Reporting including the preparation of supplementary report	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
		documentation	
HUMAN TISSUE AND FLUIDS (cont'd) DNA extracted from: Blood, Saliva, Chorionic villus tissue, Uncultured amniocytes, Cultured cells from chorionic villus samples, Cultured cells from amniotic fluid, Saliva, Thymus (Cont'd)	<u>Cytogenetics examination procedures for the purposes of clinical diagnosis</u> (cont'd)	Quantative PCR (qPCR) is performed using the StepOnePlus Real Time PCR system to confirm copy number variation detected by microarray and NGS. SOP(s): RGS MOT0701 Setting up a qPCR using StepOnePlus Real-Time PCR System Analysis by ABI StepOne software. Interpretation by manual in-house method documented in SOP(s): RGS MOT0702 Primer Design for qPCR RGS ANA0204 Real Time qPCR Data Processing, Analysis and Checking using StepOne Software	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	Cytogenetics examination procedures for the purposes of clinical diagnosis (cont'd)		
Whole Blood Amniotic Fluid CVS Tissue Samples	Chromosome analysis for Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders	Chromosome analysis - samples are processed either by the Hanabi automated system or by manual in-house method documented in SOP(s): RGS CUT0101 Preparation of blood culture medium and tubes RGS CUT0102 Setting up blood cultures RGS CUT0104 Blocking, releasing, and adding Colcemid to blood cultures RGS CUT0106 Harvesting Blood Cultures Manually RGS CUT0107 Manual Slidemaking from blood culture cell suspensions RGS CUT0108 G-banding slides from blood cultures RGS CUT0110 Harvesting blood cultures using the Hanabi-PIII metaphase harvester RGS CUT0111 Slide Making using the Hanabi-PIV Metaphase Auto Spreader RGS CUT0112 Preparation of blood samples for rapid FISH aneuploidy screening and sex determination RGS CUT0201 Preparation of amniotic fluid samples for culture, QF-PCR and other tests RGS CUT0202 Preparation of chorionic villus samples for culture, QF-PCR and other tests	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	Cytogenetics examination procedures for the purposes of clinical diagnosis (cont'd)		
Whole Blood Amniotic Fluid CVS Tissue Samples	Chromosome analysis for Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders (cont'd)	RGS CUT0203 Processing amniocytes, chorionic villus and tissue culture cells in culture for DNA extraction or interphase FISH RGS CUT0204 Assessing amniotic fluid, chorionic villus and tissue cultures RGS CUT0205 Medium-changing, topping up and re-seeding amniotic fluid, chorionic villus and tissue cultures RGS CUT0206 Subculture and redistribution of amniotic fluid, chorionic villus and tissue cultures RGS CUT0207 Blocking amniotic fluid, chorionic villus and tissue cultures RGS CUT0208 Harvesting amniotic fluid, chorionic villus and tissue cultures RGS CUT0209 Slide-making from amniotic fluid, chorionic villus and tissue culture cell suspensions RGS CUT0210 G-banding slides from amniotic fluid, chorionic villus and tissue cultures RGS CUT0211 Completing culture sheets RGS CUT0212 Culture Media in Current Use and the Preparation of Complete Culture Media RGS CUT0301 Preparation of tissue samples for interphase FISH, touch preparations and setting up in culture	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	<u>Cytogenetics examination procedures for the purposes of clinical diagnosis</u> (cont'd)		
Whole Blood Amniotic Fluid CVS Tissue Samples	Chromosome analysis for; - Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders (cont'd)	Karyotyping analysis is performed manually using microscopes. SOP(s): RGS ANA0402 Chromosome Analysis - General Procedures	RGL
		Fluorescence in-situ Hybridisation (FISH) analysis. Probes hybridised manually and used to specific guidelines as documented in SOP(s): RGS MOT0801 Slide-making for FISH and preparation of the FISH process sheet RGS MOT0802 FISH setting up and hybridisation RGS MOT0803 Post hybridisation wash procedures, detection of indirectly labelled probes and counterstaining RGS MOT0804 FISH paperwork preparation and data recording RGS MOT0806 Molecular cytogenetic reagents and solutions - procedures for preparation	RGL
		Analysis is performed using fluorescence microscopes. SOP(s): RGS ANA0407 FISH analysis	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd)	Cytogenetics examination procedures for the purposes of clinical diagnosis (cont'd)		
Tissue Samples	Chromosome analysis for; - Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders (cont'd)	Multi-ligation Probe Analysis (MLPA). Performed by manual in-house method or using the Biomek NK span-8 system. SOP(s): RGS MOT0401 Setting up MLPA using the Automated system RGS MOT0402 Manually Setting Up an MLPA reaction using MRC Holland SALSA kits (cytogenetics) RGS ANA0201 Capillary Electrophoresis of MLPA and QF-PCR products using the ABI 3730 Genetic Analyser RGS ANA0208 MLPA data processing and analysis using GeneMarker Analysis Software (Cytogenetics)	RGL
Tissue Samples		Quantitative Fluorescence Polymerase Chain Reaction (QF-PCR). Analysis performed by manual in-house method documented in SOP(s): RGS MOT0404 Manually Setting Up a QF-PCR Reaction RGS ANA0201 Capillary Electrophoresis of MLPA and QF-PCR products using the ABI 3730 Genetic Analyser RGS ANA0203 QF-PCR data processing and analysis using Genemarker	RGL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used	Location Code
HUMAN TISSUE AND FLUIDS (cont'd) Bone Marrow, Peripheral Blood, CSF, Tissue sections	<u>Cytogenetics examination procedures for the purposes of clinical diagnosis</u> (cont'd) Chromosome analysis for: - Haematological malignancies including ALL, AML, CML, MPN, MDS, JMML, Lymphoma, solid tumours	G banding and G-band analysis using Leica Cytovision Karyotyping station and in house procedures: HSOP 632 RGS LAB9609 HSOP 614 RGS LAB9604 RGS LAB9601 HSOP 605 FISH analysis using Digital Scientific FISH analysis using system ISIS (Metasystem) Following in house documented procedures: RGS LAB9630 HSOP 632 HSOP 614 RGS LAB9604 RGS LAB9601	CBL
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