Schedule of Accreditation

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Great Ormond Street Hospital for Children NHS Foundation Trust

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Rare & Inherited Disease Laboratory
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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 Rare & Inherited Disease Laboratory NHS North Thames Genomic Laboratory Hub Levels 4-6, Barclay House 37 Queen Square London WC1N 3BH	**Local contact** Deborah Morrogh	Rare and Inherited diseases Molecular Genetics Cytogenetics Acquired genomics as part of SIHMDS service	RGL

DETAIL OF ACCREDITATION

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
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
Whole Blood Bone marrow CSF Fresh tissue CSF Frozen tissue Body fluid FFPE sections Amniotic Fluid CVS Urine Blood spots Saliva Cultured cells from chorionic villus samples Cultured cells from amniotic fluid Cultured cells from skin biopsy	Extraction of DNA and RNA	RGS MOT9035 DNA Extraction from Blood/BM/Tissue using Maxwell RSC Automated system RGS MOT9038 SIHMDS-AG: Extraction of DNA from FFPE using Maxwell RGS Automated system RGS MOT9036 SIHMDS-AG: RNA Extraction using the Maxwell RSC Automated System RGS MOT0103 - DNA salting out method for CVS RGS MOT0112 - DNA Extraction from prenatal and tissue samples using the iGENatal Extraction Kit RGS MOT0110 DNA Extraction from Tissue/cells/urine using FujiQuickGene-Mini80 RGS MOT0138 DNA extraction using the Chemagic 360 RGS MOT0137 DNA – EXTRACTION FROM BLOOD SPOTS BY SEMI-AUTOMATION (EZ1 ADVANCED XL) RGS MOT1001 - RNA Extraction and cDNA Synthesis from Whole Blood RGS MOT0118 Quantification of DNA using Qubit fluorometer RGS MOT0140 DNA NORMALISATION USING BIOMEK I5 RGS MOT0139 DNA quantification using Lunatic RGS MOT0115 Measuring Optical Density of DNA Samples

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	
Whole Blood	Cell Free Fetal DNA (cffDNA) extraction from maternal plasma.	ffDNA Sample collection and separation of plasma DNA extraction based on Magnetic bead technology using the Qiagen QiaSymphony SP DNA extractor RGS MOT0201 ffDNA Sample Collection and Separation of Plasma RGS MOT0224 Using the QIAsymphony SP for cffDNA extraction RGS MOT0235 Using the QIAsymphony SP for cffDNA extraction (CircDNA2000)
DNA and RNA. Extracted from primary samples as detailed above	DNA and RNA profiling for detection of abnormal sequences associated with acquired cancer	Fragment analysis using ABI3730XLGenetic analyser RGS LAB0102 - ABI3730 XL General Use RGS LAB0713 Equipment - Thermal Cyclers and PCR systems RGS MOT 9045 SIHMDS-AG: Mutation detection by Fragment AnalysisRGS ANA9046 SIHMDS-AG: Use of GeneMapper software
	DNA and RNA profiling for detection of abnormal sequences for common and rare inherited/predisposing genetic disease condition	Fragment analysis using ABI3730XLGenetic analyser RGS LAB0102 - ABI3730 XL General Use RGS LAB0713 Equipment - Thermal Cyclers and PCR systems PCR using CFEU2 / CFEU4 kit RGS DGE0304 - Elucigene CF-EU2 Set Up and Analysis RGS DGE0305 - Elucigene CF-HT4 - set up and analysis

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	
DNA and RNA. Extracted from primary samples as detailed above	Fragment analysis for repeat expansion disorders	Fragment analysis using ABI3730XLGenetic analyser RGS LAB0102 - ABI3730 XL General Use RGS LAB0713 Equipment - Thermal Cyclers and PCR systems PCR using CFEU2 / CFEU4 kit RGS DGE0304 - Elucigene CF-EU2 Set Up and Analysis RGS DGE0305 - Elucigene CF-HT4 - set up and analysis Direct PCR, Long PCR and Triplet Primed PCR Detection is using fragment analysis on a capillary sequencer or ABI3730XL as documented in RGS LAB0102 - ABI3730 XL General Use RGS DGE0501 Fragile X PCR RGS DGE0506 Fragile X - Asuragen Amplidex FMR1 PCR RGS DGE1100 Myotonic dystrophy – TPPCR testing RGS DGE1201 Denatorubropallidoluysian Atrophy (DRPLA) RGS DGE1202 Friedreich Ataxia (FRDA) Long PCR RGS DGE1203 Friedreich Ataxia (FRDA) TP-PCR RGS DGE1204 Huntington's Disease (HD) RGS DGE1205 Junctophilin3 (JPH3) RGS DGE1206 Androgen Receptor (AR) / X-Linked Bulbospinal Neuronopathy / RGS DGE1207 Spinocerebellar Tethering PCR (SCA 1 7) RGS DGE1208 Spinocerebellar Ataxia 12 and 17 (SCA 12 and 17) RGS DGE1210 C9orf72 Repeat-Primed and Sizing PCR PCRs for RFC1 pentanucleotide expansions RGS DGE1212 PCRs for NOP56 hexanucleotide expansions RGS DGE1213

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	
DNA and RNA. Extracted from primary samples as detailed above	DNA and RNA profiling for detection of repetitive sequences to exclude maternal cell contamination and identity testing	RGS DGE0912 PCR using Power Plex16 HS assay
	DNA profiling for detection of abnormal sequences by Multiplex ligation dependent Probe Analysis (MLPA).	Multiplex ligation dependent Probe Analysis (MLPA). Performed by manual in-house method or using the Biomek NK span-8 system. RGS MOT0401 Automated MLPA Set-up RGS MOT0406 Manual MLPA set up (molecular) RGS LAB0102 ABI3730 XL General Use RGS ANA0205 Analysis of MLPA results using Genemarker Software
DNA Extracted from primary samples as detailed above	DNA profiling for detection of abnormal sequences associated with acquired cancer by next-generation sequencing (Paediatric cancers including haematological disease and solid tumours)	Split pathway for DNA Next Generation Sequencing: RGS LAB9654 SIHMDS-AG Sending Material to External Laboratories Wet lab work performed by the Specialist Pathology Laboratory at The Royal Marsden NHS Foundation Trust. DNA Next generation sequencing analysis: Analysis SOP:RGS ANA9025 SIHMDS-AG: Sequence Analysis and Variant Classification
DNA and RNA. Extracted from primary samples as detailed above	DNA profiling for detection of abnormal sequences associated with acquired cancer	Sanger sequence analysis using RGS LAB0713 Equipment - Thermal Cyclers and PCR systemsExoProStar clean up of PCR products RGS MOT0305 Streamlined Robot PCR Set-Up Using Mutation Surveyor Software to analyse DNA variants Analysis SOP: RGS ANA9037 SIHMDS-AG: Using Mutation Surveyor Software to analyse DNA variants Sequence Analysis and Variant Classification Analysis SOP:RGS ANA9025 SIHMDS-AG: Sequence Analysis and Variant Classification

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	
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	Long range PCR Long range PCR assay for STRC RGS DDE0015 Polycystic kidney disease – Long range PCR for PKD1 pseudogene region RGS DRE0501 LONG-RANGE PCR FOR PMS2 PSEUDOGENE REGIONS RGS DCA0011 LONG-RANGE PCR FOR CHEK2 PSEUDOGENE REGIONS RGS DCA0010
DNA and RNA Extracted from primary samples as detailed above	DNA and RNA profiling for detection of abnormal sequences associated with acquired cancer	Methylation arrays using Bisulfite conversion and FFPE restore SOP: RGS MOT 9053 SIHMDS-AG: Bisulfite modification of DNA & FFPE Restore Methylation array analysis using bCHAPs pipeline, Analysis SOP:RGS ANA9055 SIHMDS-AG: Tumour Methylation Profiling Methylation Array Pipeline and the use of a downloaded controlled, local copy of the Heidelberg Classifiers ('brain_classifier_v12.8' and 'sarcoma_classifier_v12.2') managed and supported by GOSH bioinformatics team.
DNA Extracted from primary samples as detailed above.	DNA profiling for detection of abnormal sequences for common and rare inherited/predisposing genetic disease condition	Droplet digital PCR (ddPCR) RGS MOT1101 RGS MOT1103 – Mitochondrial depletion ddPCR RGS MOT1104 - Droplet digital PCR for TPSAB1 copy number

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	Split pathway for RNA Next generation sequencing:
RNA. Extracted from primary samples as detailed above.	RNA profiling for detection of abnormal sequences associated with acquired cancer (Paediatric cancers including haematological disease and solid tumours)	RGS LAB9654 SIHMDS-AG Sending Material to External Laboratories Wet lab work is performed by the Specialist Pathology Laboratory at The Royal Marsden NHS Foundation Trust. RNA fusion (Next generation sequencing) analysis using the Illumina Trusight RNA Pan Cancer Panel: RGS ANA9603 SIHMDS-AG: RNA fusion analysis and interpretation Restriction enzyme digest performed manually according to in-house method described in RGS MOT0307 – Manual PCR using KCl, NH4 or Megamix buffers RGS MOT0310 – Agarose Gel preparation and Electrophoresis
Whole Blood Muscle Urine Brain CVS 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	RGS DGE1209 Mitochondrial DNA Rearrangement Long PCR

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	In-house documented methods and following Manufacturers instructions
Whole Blood Muscle Urine Brain CVS DNA. Extracted from primary samples as detailed above.	DNA dosage analysis of mtDNA	Quantitative fluorescent PCR and restriction digest followed by fragment analysis using ABI 3730XL Automated DNA Sequencer RGS MIT0004 Quantitation of 3 Common mtDNA Point Mutations
Whole Blood RNA. 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	RNA analysis of variants SOP RGS MOT1001 - RNA Extraction and cDNA Synthesis from Whole Blood SOP RGS ANA0801 – RNA Analysis of Variants cDNA synthesis Worksheet template - RGF MOT0901 RNA analysis manual conditions PCR worksheet - RGF MOT0902
DNA extracted from Whole Blood Amniotic Fluid CVS		Prenatal Exome sequencing by Next Generation Sequencing supported by PCR amplification or hybridisation. Sequencing carried out using the Illumina NextSeq sequencer. RGS MOT0118 Quantification of DNA using Qubit fluorometer RGS MOT0129 Quantity and Quality assessment using Agilent 2200 Tapestation RGS MOT0232 NextSeq for NGS Libraries RGS ANA0512 GOSHG2P Analysis RGS ANA0513 Genesis pipeline RGS ANA0213 – VariantSifter RGS ANA0214 – GeneticsStore RGP DEX002 Prenatal exomes RGS MOT0236 TWIST exomes manual library preparation RGF MOT0147 TWIST exomes manual library preparation worksheet - 2 captures RGF MOT0148 TWIST exomes manual library preparation worksheet – 1 capture

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	In-house documented methods and following Manufacturers instructions
DNA extracted from Whole Blood Amniotic Fluid CVS	DNA and RNA profiling for detection of abnormal sequences for common and rare inherited/predisposing genetic disease condition by TWIST exomes	Next Generation Sequencing supported by PCR amplification or hybridisation. Sequencing carried out using the Illumina NextSeq and NovaSeq sequencers. RGS MOT0118 Quantification of DNA using Qubit fluorometer RGS MOT0129 Quantity and Quality assessment using Agilent 2200 Tapestation RGS MOT0232 NextSeq for NGS Libraries RGS MOT0236 TWIST exomes manual library preparation RGF MOT0147 TWIST exomes manual library preparation worksheet - 2 captures RGF MOT0148 TWIST exomes manual library preparation worksheet – 1 capture NovaSeq Sequencing for NGS Libraries RGS MOT0239 RGS ANA0512 GOSHG2P Analysis RGS ANA0513 Genesis pipeline RGS ANA0213 – VariantSifter RGS ANA0214 – GeneticsStoreRGS MOT0238 Agilent 4200 TapeStation RGS MOT0237 NGS BRAVO TWIST AUTOMATED LIBRARY PREPARATION

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	In-house documented methods and following Manufacturers instructions
DNA, RNA, CVS, amniotic fluid, Tissue. 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	Rapid PCR-free whole genome sequencing (WGS) AUTOMATED ILLUMINA DNA PCR-FREE WHOLE GENOME SEQUENCING LIBRARY PREPARATION RGS MOT0240 KAPA LIBRARY QUANTIFICATION FOR ILLUMINA PLATFORMS RGS MOT0241

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics examination procedures for the purposes of clinical diagnosis (cont'd)	In-house documented methods and following Manufacturers' instructions
Whole Blood Amniotic Fluid CVS DNA. Extracted from primary samples as detailed above.	SNP genotyping	RGS DGE0913 SNP genotyping for sample identity tracking RGS ANA0301 Fluidigm Genotyping analysis RGS LAB0132 Use and maintenance of the Fluidigm Juno system RGS LAB0135 Use and maintenance of the Fluidigm EP1 system
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)	Next Generation Sequencing supported by PCR amplification or hybridisation. Sequencing is carried out using the Illumina MiSeq and NextSeq sequencers. RGS MOT0118 Quantification of DNA using Qubit fluorometer Quantity and Quality assessment using Agilent 2200 Tapestation RGS MOT0129 RGS MOT0216 MiSeq operation and loading RGS MOT0232 NextSeq for NGS Libraries RGS ANA0512 GOSHG2P Analysis RGS ANA0513 Genesis pipeline RGS ANA0213 – VariantSifter RGS ANA0214 – GeneticsStore RGS MIT0001 Mitochondrial NGS Library Prep Using Illumina DNA Prep Kit
Whole Blood Muscle Urine Brain CVS DNA. Extracted from primary samples as detailed above.	Mitochondrial whole genome sequencing	

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd) DNA, RNA, Tissue Extracted from primary samples as detailed above	<u>Molecular Genetics examination procedures for the purposes of clinical diagnosis</u> (cont'd) DNA and RNA profiling for detection of abnormal sequences for common and rare inherited/predisposing genetic disease condition	In-house documented methods and following Manufacturers' instructions Analysis pipeline specifically developed to detect low-level variants RGS ANA0809 MosaicMiner Analysis Pipeline Whole Genome Sequencing Data received from an external source following Whole Genome Sequencing Analysis of nuclear-encoded and mitochondrially-encoded genes RARE DISEASE WHOLE GENOME SEQUENCING (WGS) WORKFLOW RGS WGS0001 WHOLE GENOME SEQUENCING (WGS) - GEL1001 RGS WGS0003 Rare disease whole genome sequencing (WGS) Duty Scientist RGS WGS0004 NGIS ADMIN SOP RGS WGS0006 WGS analysis and reporting RGS WGS0007

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Non-Invasive Prenatal examination procedures for the purposes of clinical diagnosis and antenatal screening	
DNA extracted from Maternal Plasma	Non-invasive prenatal diagnosis (NIPD)	Real Time PCR analysis of cffDNA to determine fetal gender performed on the Applied Biosystems StepOne Plus Real-Time PCR Machine as documented in RGS DGE 0908 - ffDNA RQ-PCR Sexing RGS LAB0133 Applied Biosystems StepOne Plus Real-Time PCR Machine
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 RGS MOT0215 - Using the QIAcube for MinElute® PCR Purification RGS MOT0216 – MiSeq operation and loading RGS DGE0911 Non-invasive prenatal diagnosis for de novo and paternal mutations (MiSeq) RGS ANA0519 - NIPD and low level mosaicism analysis

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
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 and Cell free foetal DNA (cffDNA)	Relative Haplotype Dosage Analysis (RHDO)	Relative Haplotype Dosage Analysis (RHDO) using the Roche HyperCap target enrichment kit in combination with the KAPA library preparation kits for targeted enrichment of multiple panels of SNPs to sequence on the Illumina MiSeq and NextSeq for NIPD of single gene disorders Cell free foetal DNA (cffDNA) extracted from maternal blood samples for library preparation and Non-Invasive Prenatal Diagnosis (NIPD) using RGS MOT0118 Quantification of DNA using Qubit fluorometer RGS MOT0232 NextSeq for NGS Libraries RGS MOT0216 – MiSeq operation and loading RGS MOT0129 Quantity and Quality assessment using Agilent 2200 Tapestation RGS ANA0514 Relative Haplotype Dosage Analysis RGS NIP0002 Relative Haplotype Dosage Analysis (RHDO) using Roche KAPA HyperCap custom probes RGS ANA0520 RHDO Analysis Pipeline RGF MOT0154 KAPA HyperCap RHDO worksheet for 1 family RGF MOT0155 KAPA HyperCap RHDO worksheet for 2 family

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	<u>Cytogenetics examination procedures for the purposes of clinical diagnosis</u>	
DNA extracted from: Blood, Saliva, Chorionic villus tissue, Uncultured amniocytes, Cultured cells from chorionic villus samples, Cultured cells from amniotic fluid, Saliva, Thymus Pregnancy loss tissue, Bone Marrow, Tissue, FFPE sections	Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders Acquired cytogenomic analysis	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 using the Illumina Beadchip CytoSNP- 850k array Platform RGS MOT0118 Quantification of DNA using Qubit fluorometer RGS MOT0607 Automated Processing of Illumina Beadchip CytoSNP-850kRGS MOT0609 Manual Processing of Illumina Beadchip CytoSNP-850K RGS ANA0408 Microarray Analysis and Checking using InfoQuant Fusion and cnTrack Equipment includes: High Speed Microplate shaker, Hybridisation Oven, Vortex, pulse centrifuge, heat block, vacuum desiccator, TECAN Evo. RGS ANA0410 Production and processing of Beadchip Data files using NextSeq550 and Beeline software Equipment includes: NextSeq 550, Beeline software SNP array for acquired genomic imbalance RGS ANA9601 SIHMDS-AG: Analysis of SNP arrays for acquired genomic imbalance RGP ANA9608 SNP array policy for acquired genomic imbalance

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
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)	Cytogenetics examination procedures for the purposes of clinical diagnosis (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

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
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 CUT0113 Harvesting blood cultures using the Hanabi-PI metaphase harvester RGS CUT0114 Slide making using the hanabi-p5 metaphase auto spreader mini RGS CUT0201 Preparation of amniotic fluid samples for microarray, culture, QF-PCR and other tests RGS CUT0202 Preparation of chorionic villus samples for microarray, culture, QF-PCR and other tests

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
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,tissue and SIHMDS cultures RGS CUT0211 Prenatal and Tissue Tracking 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

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
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 Bone Marrow CSF Body Fluid Tissue FFPE sections	Chromosome analysis for; - Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders Chromosome analysis for acquired cancers: - Haematological malignancies and solid tumours	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 FISH 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 RGS LAB9601 SIHMDS-AG: Setting up samples for cytogenetic analysis RGS LAB9604 SIHMDS-AG: Harvesting of cultured fluid specimens RGS LAB9605 SIHMDS-AG: Slidemaking from Fixed cell suspensions Following in house documented procedures: RGS LAB9630 SIHMDS-AG: Fluorescence in-situ hybridisation (FISH) using directly-labelled probes and whole chromosome paints Analysis is performed using fluorescence microscopes. SOP(s): RGS ANA0407 FISH analysis

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Cytogenetics examination procedures for the purposes of clinical diagnosis (cont'd)	Karyotyping analysis is performed manually using microscopes or using the Cytovision system.
Whole Blood Amniotic Fluid CVS Tissue Samples	G-band analysis using Leica Cytovision Karyotyping station and in house procedures	RGS CUT0213 AUTOMATED SCANNING OF SLIDES INTO CYTOVISION RGS ANA0402 Chromosome Analysis - General Procedures RGS ANA9638 SIHMDS-AG: Use of the Cytovision for Chromosome Analysis System RGS LAB0501 Storage and disposal of fixed cell suspensions, slides and DNA – Cytogenetics RGS CUT0107 Manual slidemaking from blood culture cell suspensions
Bone Marrow, Peripheral Blood, culktured/fixed cell suspensions	Karyotyping (G-band) Chromosome analysis for acquired cancers: - Haematological malignancies	G banding and G-band analysis using Leica Cytovision Karyotyping station and in house procedures: RGS LAB9601 SIHMDS-AG: Setting up samples for cytogenetic analysis RGS LAB9604 SIHMDS-AG: Harvesting of cultured fluid specimens RGS LAB9605 SIHMDS-AG: Slidemaking from Fixed cell suspensions RGS ANA9638 SIHMDS-AG: Use of the Cytovision for Chromosome Analysis System
END		