Schedule of Accreditation

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Belfast Health and Social Care Trust

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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 BODY FLUIDS / TISSUES	<u>Molecular Genetics</u>	
	Detection of nucleic acid sequence variants and/or copy number changes for the purpose of clinical diagnosis	Documented in-house procedures using techniques and kits in combination with manufacturers instructions
Whole blood Cultured cells CVS Cord blood Buccal cells Products of Conception Fresh / frozen tissue (including skin, muscle, spleen) Guthrie cards Saliva Urine Amniotic Fluid	DNA extraction, quantification and quality check for subsequent in-house analysis (see below), referral to specialist centres and longterm storage	Manual and Automated DNA Extraction and QuantificationManual Extraction Using: iGEN Biotech New iGENatal Kit/ Qiagen Gentra Puregene Blood Kit/ DNA Genotek prepIT.L2P Kit SOPs: LP 352 003 iGENatal LP 364 012 Puregene LP 364 014 prepIT.L2P Automated Extraction Using: Roche MagNA Pure Compact/ Perkin Elmer Chemagic MSM1 SOPs: LP 364 019 Compact LP 364 020 Chemagic DNA Quantification Using: IMPLEN Nanophotometer Pearl/ Invitrogen Qubit 4 Fluorometer SOPs: LI 366 020 Nanophotometer PD 305 307 Qubit

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES	Detection of nucleic acid sequence variants and/or copy number changes for the purpose of clinical diagnosis	Documented in-house procedures using techniques and kits in combination with manufacturers instructions
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	DNA amplification by PCR for fragment analysis or downstream use in sanger sequencing	DNA Amplification by PCR Using: In-house methods/ MegaMix Gold/ Eurogentec Primers/ Beckman Coulter Biomek NXP Automated Workstation/ Thermocyclers [Definitive list in Current Dept Equipment List]/ Syngene G:BOX F3 Gel Documentation System SOPs: LI 362 104 PCR Conditions LP 366 001 Gel Electrophoresis LP 366 013 PCR LP 366 016 G:BOX F3 LP 366 032 Biomek NXP

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Detection of nucleic acid sequence variants (SNVs and Indels) in genes specified in definitive list and those identified through research and/or family (trio) NGS sequencing work [Definitive list in RMDS Test Accreditation List held by this laboratory]	Sanger Sequencing Using: In-house methods/ Applied Biosystems BigDye Terminator Cycle Sequencing Kit/ Beckman Coulter Agencourt AMPure XP PCR Purification System/ Beckman Coulter Agencourt CleanSEQ Dye-Terminator Removal/ Thermocyclers [Definitive list in Current Dept Equipment List/ Beckman Coulter Biomek NXP Automated Workstation/ Applied Biosystems 3730 DNA Analyzer/ Applied Biosystems 3500xL Genetic Analyzer/ SoftGenetics Mutation Surveyor Software/ Documented in-house procedures using techniques and kits in combination with manufacturers instructions

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES (cont'd)	<u>Molecular Genetics (cont'd)</u>	Sanger Sequencing (cont)
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Detection of nucleic acid sequence variants (SNVs and Indels) in genes specified in definitive list and those identified through research and/or family (trio) NGS sequencing work [Definitive list in Test Accreditation List	Alamut Visual Software SOPs: LI 362 102 CleanSEQ LI 366 008 AMPure LP 366 008 Sanger Sequencing LP 366 028 Mutation Surveyor LP 366 032 Biomek NXP LP 366 037 ABI 3730 LP 366 038 ABI 3500xL LP 366 043 Variant Classification
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Determination of copy number changes and methylation specific (MS) dosage changes [Definitive list in RMDS Test Accreditation List held by this laboratory]	Multiplex Ligation Probe Amplification (MLPA) (to include MS-MLPA) Using: MRC Holland SALSA MLPA Reagents and Probemixes/ Thermocyclers [Definitive list in Current Dept Equipment List]/ Applied Biosystems 3500xL Genetic Analyzer/ SoftGenetics GeneMarker Software SOPs: LP 363 026 MS-MLPA LP 366 025 MLPA Set Up LP 366 026 MLPA Analysis LP 366 038 ABI 3500xL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES	<u>Molecular Genetics (cont'd)</u>	
	Detection of nucleic acid sequence variants and/or copy number changes for the purpose of clinical diagnosis	Documented in-house procedures using techniques and kits in combination with manufacturers instructions
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Detection of nucleic acid point variants, small insertions and deletions [Definitive list in RMDS Test Accreditation List held by this laboratory]	Amplification Refractory Mutation System (ARMS) Using: Elucigene CF4 & CF-EU2/ Thermocyclers [Definitive list in Current Dept Equipment List]/ Applied Biosystems 3730 DNA Analyzer/ Applied Biosystems 3500xL Genetic Analyzer/ Applied Biosystems GeneMapper Software SOPS: LI 362 081 CF Neonatal Reporting LP 362 083 CF Neonatal LP 363 032 CF Routine LP 366 034 GeneMapper LP 366 037 ABI 3730 LP 366 038 ABI 3500xL
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Detection of nucleic acid sequence repeat expansions [Definitive list in RMDS Test Accreditation List held by this laboratory]	Repeat Fragment Detection Using: In-house methods/ MegaMix Gold/ Eurogentec Primers / Asuragen AmplideX PCR/CE FMR1 Kit/ Thermocyclers [Definitive list in Current Dept Equipment List]/ Applied Biosystems 3730 DNA Analyzer/ Applied Biosystems 3500xL Genetic Analyzer/

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES (cont'd)	Molecular Genetics (cont'd)	Documented in-house procedures using techniques and kits in combination with manufacturers instructions
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Detection of nucleic acid sequence repeat expansions [Definitive list in RMDS Test Accreditation List held by this laboratory]	Repeat Fragment Detection (cont) Applied Biosystems GeneMapper Software/ SOPs: LP 363 020 Friedreich Ataxia LP 363 021 Huntington Disease LP 363 022 Kennedy Syndrome LP 363 023 Fragile X Syndrome LP 363 025 Myotonic Dystrophy LP 363 039 Dominant Ataxias LP 366 034 GeneMapper LP 366 037 ABI 3730 LP 366 038 ABI 3500xL
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Detection of nucleic acid short tandem repeats	Short Tandem Repeat Detection Using: Promega PowerPlex 16 System/ Thermocyclers [Definitive list in Current Dept Equipment List]/ Applied Biosystems 3730 DNA Analyzer/ Applied Biosystems 3500xL Genetic Analyzer/ Applied Biosystems GeneMapper Software SOPs: LP 363 036 Promega PowerPlex 16 LP 366 034 GeneMapper LP 366 037 ABI 3730 LP 366 038 ABI 3500xL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES (cont'd)	Molecular Genetics (cont'd)	Documented in-house procedures using techniques and kits in combination with manufacturers instructions
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Gene screening of large gene panels for genetic variants: SNVs CNVs Indels [Definitive list in RMDS Test Accreditation List held by this laboratory]	Next Generation Sequencing (NGS) Library Preparation Using: Roche KAPA HyperPlus Kit/ Hamilton Microlab STAR Liquid Handling System/ Invitrogen Qubit 4 Fluorometer/ Agilent 4200 Tapestation System/ Roche LightCycler480 Instrument II/ Thermocyclers [Definitive list in Current Dept Equipment List] SOPs: LF 362 109 Library Quantification LF 366 047 Library Preparation LI 362 109 Library Quantification LP 366 047 Library Preparation

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
		Next Generation Sequencing Set Up Using: Illumina MiSeq Reagent Kit/ Illumina MiSeqDx System SOPs: LF 366 047 Library Preparation PD 305 299 MiSeqDx PD 305 300 MiSeq Libraries Guide Next Generation Sequencing Analysis Using: Germline Variant Calling and Annotation Pipeline (benchmarked against NHSE Pipeline Tool using Coriell Reference DNA)/ Congenica (for tertiary analysis)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES (cont'd)	Molecular Genetics (cont'd)	Documented in-house procedures using techniques and kits in combination with manufacturers instructions Next Generation Sequencing (NGS) (cont) SOPs: LP 306 010 Targeted Cancer Panel LP 307 004 GitHub LP 307 005 Microsoft Azure LP 307 007 Germline Pipeline LP 307 008 Congenica Analysis LP 307 009 Genome Builds

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Diagnosis of aneuploidy in products of conception, intrauterine deaths, neonatal deaths, stillbirths and by rapid prenatal diagnosis	Quantitative Fluorescence-Polymerase Chain Reaction (QF-PCR) Products of Conception, Intrauterine Deaths, Neonatal Deaths and Stillbirths Using: Elucigene QST*R-PL Kit/ Thermocyclers [Definitive list in Current Dept Equipment List]/ Applied Biosystems 3500xL Genetic Analyzer/ SoftGenetics GeneMarker Software SOPs: LP 352 001 QF-PCR Tissue Set Up LP 352 002 QF-PCR Tissue Analysis LP 366 038 ABI 3500xL Rapid Prenatal Diagnosis Using: Elucigene QST*R Plus Kit/ Elucigene QST*R-13 Kit/ Elucigene QST*R-18 Kit/ Elucigene QST*R-21 Euplex Kit/ Elucigene QST*R-XY Kit/ Applied Biosystems 3500xL Genetic Analyzer/ SoftGenetics GeneMarker Software/

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES (cont'd)	Molecular Genetics (cont'd)	Documented in-house procedures using techniques and kits in combination with manufacturers instructions Rapid Prenatal Diagnosis (cont) Applied Biosystems GeneMapper Software SOPs: LP 352 003 QF-PCR AF Set Up LP 352 004 QF-PCR AF Analysis LP 366 038 ABI 3500xL

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES (cont'd)	<u>Germline and Somatic</u>	
	Chromosome analysis for the purpose of clinical diagnosis	Documented in-house procedures using techniques and kits in combination with manufacturers instructions:
Whole blood Amniotic fluid CVS CSF Cord blood Bone marrow Tissues	Detection of chromosomal rearrangements or aberrations arising from: Prenatally detected disorders Developmental disorders Reproductive medicine disorders Haematological/Oncology disorders	Karyotyping and G-Banding Cell Culturing and Harvesting Using: Manual methods SOPs: Prenatal LP 345 001 AF Culture LP 345 002 AF/Tissue/CVS Harvest LP 345 003 Solid Tissue Culture LP 345 031 CVS Culture Postnatal LP 345 016 Blood Culture LP 345 034 Blood Harvest Haematological/Oncology LP 345 029 BM & Blood Culture LP 345 030 BM & Blood Harvest

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
		Chromosome Preparation and G-Banding Using: ADSBiotec Hanabi-PVI Metaphase Spreader/ Thermo Scientific ClearVue Coverslipper SOPs: LP 345 033 Slide Making QA LP 346 011 G-Banding LP 351 005 Hanabi Microscopic Analysis of G-Banded Chromsomes Using: Leica Microsystems DM5500B Microscope/

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES (cont'd)	<u>Germline and Somatic</u> Chromosome analysis for the purpose of clinical diagnosis	Documented in-house procedures using techniques and kits in combination with manufacturers instructions: Microscopic Analysis of G-Banded Chromsomes (cont) Leica Biosystems GSL-120 Slide Loader/ Leica Biosystems CytoVision Image Analysis Capture System SOPs: LI 348 003 GSL120 Cancer Analysis LI 348 004 GSL120 Constitutional Analysis LP 347 033 DM5500B Use LP 348 005 GSL120 Set Up LP 348 008 Cancer Chromosome Analysis LP 348 011 Cytovision Scanning Classifiers LP 348 034 Constitutional Chromosome Analysis

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
Whole blood Amniotic fluid CVS CSF Cord blood Bone marrow Tissues Buccal cells Products of conception	Detection of chromosomal aberrations in the diagnosis of constitutional disorders and haematological malignancy using a range of probe types including: Break-apart probes Fusion probes Deletion probes Insertion probes Copy Number Amplification probes [Definitive list in RMDS FISH Probe List held by this laboratory]	Fluorescence In Situ Hybridisation (FISH) Using: A range of probe suppliers including: Abbott Vysis/ Cytocell/ Empire Genomics/ [Definitive list in FISH Probe List] Stemcell EasySep Human CD138 Positive Selection Kit/ Stemcell EasySep Human CD19 Positive Selection Kit/ Abbott StatSpin ThermoBrite S500/ Leica Microsystems DM5500B Microscope/

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS / TISSUES (cont'd)	<u>Germline and Somatic</u> Chromosome analysis for the purpose of clinical diagnosis	Documented in-house procedures using techniques and kits in combination with manufacturers instructions: Fluorescence In Situ Hybridisation (FISH) (cont) Leica Biosystems GSL-120 Slide Loader/ Leica Biosystems CytoVision Image Analysis Capture System SOPS: LI 347 007 ThermoBrite LI 347 034 Cytovision Spot Counting Analysis LP 345 037 CD138+ Selection LP 345 038 CD19+ Selection LP 347 001 Buccal Swabs Interphase FISH LP 347 002 FISH Set Up LP 347 003 Blood/BM FISH LP 347 005 Post Hybridisation FISH Washes LP 347 033 DM5500B Use LP 348 005 GSL120 Set Up LP 348 011 Cytovision Scanning Classifiers LP 348 038 FISH Analysis

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
Genomic DNA extracted in house from the sample types listed above and received as primary samples from external sources	Genomic analysis for detection of copy number changes or loss of heterozygosity arising from: Prenatally detected disorders Developmental disorders Reproductive medicine disorders	Single Nucleotide Polymorphism (SNP) Microarray Using: Illumina Infinium CytoSNP-850K BeadChip Kit/ Illumina NextSeq 550 Instrument/ VWR Hybridisation Oven/ Tecan Te-Flow Unit/ Thermocyclers [Definitive list in Current Dept Equipment List]/ Illumina BlueFuse Multi Software SOPs: LP 350 006 Microarray Set Up LP 350 012 Microarray Analysis PD 305 318 Recommendations for Microarray in Pregnancy
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