Schedule of Accreditation

issued by

United Kingdom Accreditation Service

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

8652

Accredited to ISO 15189:2012

Sheffield Children's Hospital NHS Foundation Trust

Issue No: 005 **Issue date:** 28 July 2020

Sheffield Genetics Service
Sheffield Children's Hospital
Western Bank
Sheffield
S10 2TH
United Kingdom

Contact: Karen Bennett
Tel: +44 (0)1142717240
Fax: +44 (0)1142706121
E-Mail : karen.bennett15@nhs.net
Website: www.sheffieldchildrens.nhs.uk/our-services/laboratory-medicine/

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 TISSUE AND FLUIDS	Cytogenetics examination activities for the purpose of clinical diagnosis	In-house documented procedures based on equipment manuals as relevant
Blood Chorionic villi sample Amniotic fluid Fetal blood Products of Conception Tissues/Skin Biopsy Bone Marrow Tumour	Chromosome analysis for:- Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders Haemato/Oncology Disorders Chromosome Breakage Disorders	Preparation of material for chromosome analysis by in-house methods SOPs 501.003, 501.004, 501.005, 501.006, 501.007, 326.4.428 , 501.009, 406.097 Analysis using Leica GSL120 image capture system, Leica cytovision analysis software. SOPs 501.010, 501.025, 501.031, 501.032, 501.044, 501.049, 501.050 501.028 501.051 Reporting 602.210 and 602.011

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Cytogenetics examination activities for the purpose of clinical diagnosis (cont'd)	In-house documented procedures based on equipment manuals as relevant
Fixed cell slides from tissue and fluids listed above and Paraffin embedded sections	Detection of chromosomal aberrations in the diagnosis of haematological malignancy, bone marrow failure syndromes, non-haematological malignancies and constitutional disorders, solid tumours and companion testing Amplification probes Break apart probe Break apart rearrangement probe Centromeric hetro chromatin Deletion/ Duplication probe Dual fusion rearrangement probe Duplication/ Deletion/Break Apart Probe Hetrochromatin Probe Paint Sub Telomere	Fluorescent in situ Hybridisation (FISH) using commercial and in house developed probes (and including probe validation/verification) using PTC 200 hybridisation station ; FISH microscopes FISH Technical SOPs 501.008506.004, 506.007 Constitutional FISH Analysis SOPs 401.075, 501.031, 501.032, 501.033, 501.034, 501.035, 501.049, 501.051, 501.051b, 501.052, 501.050, 501.044 Oncology FISH analysis SOPs 501.010, 501.028a-b, 501.031, 501.032, 501.033, 501.044, 501.049, 501.050, 501.051, 501.029, 501.052, 506.008 – Diagnostic Probes Reporting 602.210 and 602.011

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMANS TISSUES AND FLUIDS (cont'd)	Genetics examination activities for the purpose of clinical diagnosis	Documented in-house methods incorporating manufacturers' instructions where relevant:
Whole Blood Bone Marrow Amniocytes CVS Guthrie spots Paraffin embedded tissue Slide sections Sperm Saliva Buccal swabs Foetal blood Products of Conception Tissue Fibroblasts Lyophilised cells Leucocyte cones DNA external (checked and processed)		Pre-examination step for molecular methods. Extraction, quantification, cell separation and synthesis of DNA, RNA, cDNA and CD3 & CD138 using a combination of nanodrop, Chemagen 360 and EZ1 biorobot, Qubit and manual methods. SOPs 401.083, 401.253, 401.254, 401.263, 401.274, 401.276, 601.002, 601.003, 601.006, 601.007, 601.009, 601.173, 601.175, 601.178, 601.229, 601.230, 601.248, 601.268, 601.303, 601.304, 601.376, 601.380, 601.315, 606.179
Extracted DNA	Detection of aneuploidy of chromosomes 13, 18, 21 or sex chromosomes	QF-PCR using ABI 3730xl Thermal cycler, analysis by genemapper Software SOPs 401.006, 606.035, 401.026
Extracted DNA	Genome wide screen for copy number gain or loss of DNA by 8x60k	Microarray profiling using OGT/Agilent technology, Thermal Cycler, Agilent Scanner, Hybridisation Oven, DNA Concentrator SOPs 401.063, 401.073, 406.094 Analysed using Cytosure Interpret software SOPs 401.243, 401.076, 401.074 Reporting 602.210 and 602.011

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMANS TISSUES AND FLUIDS (cont'd)	<u>Genetics examination activities for the purpose of clinical diagnosis</u> (cont'd)	Documented in-house methods incorporating manufacturers' instructions where relevant:
Extracted DNA	Detection of characteristic pathogenic variants and larger intragenic deletions and duplications: Hereditary Cancers Connective Tissue Disorders Inborn errors of metabolism Neurogenetic disorders Haemostatic disorders Polycystic Diseases	MLPA using MRC Holland Kit Capillary Electrophoresis Instrument, Thermal cycler SOPs 401.062, 606.035, 401.026, 401.237, 601.372, 401.027 Assay List – 407.103 MLPA Analysis by MRC Holland Coffalyser Software SOP 401.071 MLPA Analysis Reporting 602.210 and 602.011

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMANS TISSUES AND FLUIDS (cont'd)	Genetics examinations for the purpose of clinical diagnosis (cont'd)	Documented in-house methods incorporating manufacturers' instructions where relevant:
Extracted DNA/RNA/cDNA	DNA and RNA profiling for detection of pathogenic variants for common and rare genetic disease conditions Inborn errors of metabolism Connective Tissue disorders Hereditary cancers Oncology Neurogenetic disorders Haemostatic disorders Developmental delay	PCR amplification and sanger sequencing including primer design, SOPs 601.181, 401.007, 401.059, 401.060, 401.082, 601.200, 601.278, 601.279 Assay List - 401.336 Gel based assay, PCR ampilification followed by acrylaminde or agarose gel. SOPs 601.192, 601.196, 606.043 Assay list - 407.101 Quantitative PCR (Real time) using the Taqman system and the ABI 7500 and Lightcycler LC480. SOPs 406.089, 401.094, 606.048 and 601.037. ddPCR using Droplet Generator SOPs 401.285; 401.286; 406.136 Assay List - 407.102 Fragment sizing analysis using Capillary Electrophoresis via ABI 3730. SOPs 401.026, 606.035, 601.192, 401.093, 601.196, 601.197, 601.184 Assay List - 407.100 Analysis using mutation surveyor software and genemapper software and Alamut interpretation software SOPs 401.057, 401.062, 606.035, 401.026, 401.237, 601.235, 601.314, 402.006, 601.038 Reporting 602.210 and 602.011

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMANS TISSUES AND FLUIDS (cont'd)	Genetics examinations for the purpose of clinical diagnosis (cont'd)	Documented in-house methods incorporating manufacturers' instructions where relevant:
Extracted DNA/RNA/cDNA	Detection of DNA sequence variants for common and rare genetic disease conditions: Inborn Errors of Metabolism Connective tissue disorders Hereditary Cancers Polycystic diseases Neurogenetic Disorders Oncology Haematology Disorders	Next Generation Sequencing based on the Illumina HiSeq 2500 or Thermo Ion Torrent S5 Prime platforms. Sample preparation for the HiSeq is performed using the Agilent SureSelect Target Enrichment System and for the S5 using Thermo reagent kits. Additional equipment includes MiSeq, Covaris E220 shearing device, robotics - Biomek FX and NX, TapeStation and Qubit.SOPs 401.241; 406.091; 401.042; 401.043; 406.311; 406.071; 406.030, 401.058, 401.188, 401.189,401.191, 401.193, 401.194 Analysis SOP 401.047 Assay List – 413.004 Reporting 602.210 and 602.011
Extracted DNA referred to Newcastle.	Processing, analysis and reporting of SNP array data	SNP Array analysis and reporting using BlueFuse Multi software SOP 401.340
END		