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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 TISSUE AND FLUIDS Whole Blood Amniotic Fluid CVS Foetal blood Products of conception Tissue biopsy	<u>Cytogenetics</u> Chromosome analysis for: Constitutional disorders Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders Chromosome Breakage Disorders	Documented in-house methods for Chromosome culture by in-house procedures CYT PND 002, 005, 006, 007, 008, 014, 015, 021, 023; CYT BLO 002, 003 CYT INST 001-006 Followed by: Preparation of material for chromosome analysis by manual harvest using procedures CYT PND 019, CYT BLO 004, CYT INST 001-006

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Cytogenetics (cont'd)	
Whole Blood Amniotic Fluid CVS Foetal blood Products of conception Tissue biopsy (cont'd)	Chromosome analysis for: Constitutional disorders Prenatal Diagnosis Reproductive Medicine Disorders Developmental Disorders Chromosome Breakage Disorders (cont'd)	Documented in-house methods for Chromosome culture by in-house procedures CYT PND 002, 005, 006, 007, 008, 021; CYT BLO 002 CYT INST 001-006 The above followed by any combination of the following and Applied Spectral Imaging Image Analysis System SOP : CYT ANAL 003, 004, 005 a) G Banding - Microscopic detection using procedures CYT GEN 013 b) Solid Staining and SCE staining with Microscopic detection using procedures CYT GEN 008 CYT INST007 c) Fluorescence In situ Hybridisation (FISH) Documented in house methods using commercial kits from Abott (Vysis), Cytocell, Kreatech, and TCAG and in-house manual processing - Microscopic detection CYT FISH 018
Whole Blood Culture fibroblasts Chorion Skin Muscle Foetal material dissected from POC (products of conception)	Constitutional disorders	Documented in-house methods for DNA extraction using in-house procedures MOL EXTR 009, 019
Amniotic Fluid CVS		iGENATAL DNA extraction SOP: MOL EXTR 021

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
	Molecular Pathology	
HUMAN TISSUE AND FLUIDS Solid Tumours Bone marrow Lymph node Whole blood Pleural Infusions Any malignant solid body tumour or fluid	Chromosome analysis for haemato-oncology and other oncology disorders	Chromosome culture by in-house procedures using commercial media (HAMS F10) CYT ACQ 002, 014, 015 Followed by: Preparation of material for chromosome analysis by manual harvest using in-house methods CYT ACQ 009 and one or both of the following: a) G-Banding - Microscopic detection CYT GEN 013b) Fluorescence In situ Hybridisation (FISH) by documented in house methods using Abott (Vysis), Cytocell and Kreatech kits and in-house manual processing – Microscopic detection CYT FISH 018
Paraffin embedded tissue sections		Treatment of FFPE sections by documented in house manual method CYT FISH 020, 021, MOL EXTR 017 Followed by: Fluorescence In situ Hybridisation (FISH) using documented in house methods and Abott (Vysis), Cytocell and Kreatech kits with in-house manual processing – Microscopic detection CYT FISH 018

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Pathology (cont'd)	
Bone marrow Lymph node Whole blood Pleural Infusions Any malignant solid body tumour or fluid	DNA and RNA-based molecular analysis for haemato-oncology disorders	Documented methods for DNA and RNA extraction using in-house procedures for manual or robotic extraction using QIASymphony and QIA EZ1 instruments, Biorobot and EZI XL and QIASymphony and QIA EZ1 kits Manual , CYT ACQM 003 Robotic MOL EXTR 009, 017, 019
Plasma	cfDNA	Roche Cobas cfDNA preparation kit For downstream EGFR testing on Cobas SOPs: MOL EXTR 015, 016
	Mutation detection in the diagnosis and management of: AML / MDS CML / MPN ALL	PCR and RT PCR amplification of DNA / RNA (cDNA) using in house techniques. CYT ACQM 012, 002, 016, 018 Followed by one of the following: a) Agarose gel electrophoresis MOL ELECT 001 b) Fragment analysis by capillary electrophoresis on ABI3730 MOL EQUIP 009 Followed by Data analysis using GeneMarker CYT ACQM 016, 023, 024, 025
Bone marrow Lymph node Whole blood Pleural Infusions Any malignant solid body tumour or fluid	CML / MPN ALL	(RT-q) PCR amplification of DNA / RNA (cDNA) using in house method and Qiagen kit with RotorGene 6000 CYT ACQM 006, 019
cDNA	MYD88 L265P Mutation	Realtime PCR and RotorgeneQ SOP:-CYT ACQM 020

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Pathology (cont'd)	
DNA	FLT3-TKD mutations	Fragment length polymorphism-mediated PCR assay (PCR followed by enzyme EcoRVI digest and resolution by fragment analysis ABI3730) CYT ACQM 016, MOL EQUIP 009
	Clonality assessment of suspected lymphoproliferative disorders	Commercial kit (InVivoScribe) Fragment analysis on ABI3730 Data analysis using GeneMarker CYT ACQM 005
Paraffin embedded tissue sections	Mutation detection and screening: Lung cancer Colorectal cancer Melanoma Ovarian cancer Breast cancer	PCR amplification of DNA/RNA (cDNA) using in house methods and commercial kits MOL PCR 001 MOL PATH 003 Using a combination of techniques below: a) Fragment length analysis (FLA) using ABI 3730 capillary electrophoresis MOL EQUIP 009 MOL PATH 002 b) Sequence analysis on ABI3730 platform. Data analysis Mutation Surveyor MOL SEQ 001 MOL EQUIP 009 MOL PATH 002
DNA from FFPE	Micro satellite instability (MSI)	In house methodology for evaluation of colo-rectal tumour samples only SOP: MOL PATH 001, 004

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Pathology (cont'd)	
Paraffin embedded tissue sections		Mutation detection on the Cobas platform using kits as below Roche Cobas EGFR Roche Cobas KRAS Roche Cobas BRAF V600 MOL PATH 001, 003, 006
cfDNA		Roche Cobas EGFR SOPs: MOL PATH, 001, 003 , 006
	Molecular Genetics	
Whole Blood Foetal Blood Mouth washes Amniotic Fluid CVS Products of Conception Fresh tissue (tumour, muscle, liver, lymph nodes etc) Bone Marrow Wax embedded tissue Slide sections DNA	DNA and RNA (cDNA)-based detection of abnormal sequences for common and rare genetic disease conditions	Documented in-house methods for DNA and RNA extraction using in-house procedures using commercial kits and manual or robotic extraction using QIASymphony and QIA EZ1 instruments and kits Manual MOL EXTR 005, 006 Robotic MOL EXTR 009, 017, 019 PCR amplification of DNA/RNA (cDNA) using in house methods and commercial kits MOL PCR 001 MOL QFPCR 015, MOL EXTR 018 Followed by (as appropriate): Capillary electrophoresis by in house methods using ABI 3730 genetic analyser MOL SEQ 001 MOL EQUIP 009

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd) Whole Blood, Foetal Blood Mouth washes, Amniotic Fluid CVS, Products of Conception Fresh tissue (tumour, muscle, liver, lymph nodes etc), Bone Marrow Wax embedded tissue, Slide sections, DNA	<u>Molecular Genetics</u> (cont'd) DNA and RNA (cDNA)-based detection of abnormal sequences for common and rare genetic disease conditions (cont'd) • Rapid Prenatal diagnosis of Trisomies	Using Elucigene kits as below QST*R plus QST*R 13 QST*R 18 QST*R 21 MOL QFPCR 015
	DNA profiling • Detection of maternal cell contamination (MCC)	Promega Powerplex using MOL QFPCR 015
	Deletion and duplication detection • Breast/ovarian cancer • HMSN/HLPP • Hyperlipidaemia • Li-Fraumeni • PTEN Hamartoma Tumour syndrome (PHTS)	Multiplex Ligation Probe Amplification (MLPA) to detect characteristic mutations and larger intragenic deletions and duplications using MRC-Holland kit MOL MLPA 001, 002
DNA extracted from whole blood (EDTA preserved), amniotic fluid, chorionic villus sampling, saliva.	Detection of specific wild type and mutant alleles in the CFTR gene	Fragment length analysis on ABI3730 platform with data analysis on GeneMarker MOL CF 004
Whole Blood, Foetal Blood Mouth washes, Amniotic Fluid CVS, Products of Conception Fresh tissue (tumour, muscle, liver, lymph nodes etc), Bone Marrow Wax embedded tissue Slide sections, DNA	Unstable repeat measurement and Specific mutation detection: • Myotonic Dystrophy types 1 and 2, • Fragile X • Torsion dystonia • GRA	Fragment length analysis (FLA) ABI 3730 capillary electrophoresis MOL EQUIP 008, 009 MOL DM 003, MOL DM 004 MOL FRAX 001 MOL DYT 001 MOL GRA 001

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics (cont'd)	
Whole Blood, Foetal Blood Mouth washes, Amniotic Fluid CVS, Products of Conception Fresh tissue (tumour, muscle, liver, lymph nodes etc), Bone Marrow Wax embedded tissue Slide sections, DNA (cont'd)	Mutation detection by DNA sequencing:	Sequence analysis on ABI3730 platform. Data analysis Mutation Surveyor MOL EQUIP 009 MOL SEQ 008 With specific reference to the procedures below as appropriate
	• Breast/ovarian cancer • Cardiac disorders	MOL IN CAN 001 MOL CARD 001 MOL PTEN 003
	• PTEN Hamartoma Tumour syndrome (PHTS)	Sequence analysis on ABI3730 platform. Data analysis Mutation Surveyor MOL EQUIP 009 MOL SEQ 008 With specific reference to the procedures below as appropriate
	• Haemochromatosis • HMSN (all types) • Hyperlipidaemia • Li Fraumeni • RR-MADD • Sickle cell mutation EB7 • Thrombophilia	MOL HFE 001 MOL HMSN 002 MOL FH 006 MOL APOE 001 MOL TRIG 001 MOL SICK 001 MOL THROM 001
	Rare disorders: • Non-syndromic X-linked mental retardation • Trimethylaminuria • Familial Mediterranean fever	MOL RARE 001 MOL TMA 001 MOL FMF 001

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics (cont'd)	
Whole Blood, Foetal Blood Mouth washes, Amniotic Fluid CVS, Products of Conception Fresh tissue (tumour, muscle, liver, lymph nodes etc), Bone Marrow Wax embedded tissue Slide sections, DNA (cont'd)	Mutation detection by DNA sequencing: (cont'd)	Sequence analysis on ABI3730 platform. Data analysis Mutation Surveyor MOL EQUIP 009 MOL SEQ 008 With specific reference to the procedures below as appropriate
DNA	Cardiac Arythmias Sub panels • Arrythmogenic Right Ventricular Cardiomyopathy • Atrial Fibrillation • Brugada Syndrome • Catecholaminergic polymorphic ventricular tachycardia (CPVT • Dilated Cardiomyopathy • Heart Block • Long QT • Long QT – Andersen syndrome • Sudden Cardiac Death	Custom Cardio Solution NGS panel for Cardiac Arrhythmias, sequencing by MiSeq and analysis of sub-panels through commercial Sophia DDM pipeline. MOL NGS 006, 016, 022, MOL EQUIP 015 SOP: MOL CARD 001
DNA	Hereditary Cancer Solution for breast and ovarian cancer,	Breast and ovarian cancer, sequencing by MiSeq and analysis through commercial Sophia DDM pipeline (Germline). SOP: MOL IN CAN 001 MOL NGS 006, 016, 022, MOL EQUIP 015
DNA	SPIDeR SEQ panel	(Scottish Primary Immune Deficiency and Rheumatology NGS SEQ panel) - custom design kit based on Twist Custom Design library preparation, sequencing by MiSeq and analysis by commercial Sophia DDM pipeline (CVID). SOP: MOL NGS 031 ,MOL NGS 022, MOL MIR 001

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics (cont'd)	
Whole Blood, Foetal Blood Mouth washes, Amniotic Fluid CVS, Products of Conception Fresh tissue (tumour, muscle, liver, lymph nodes etc), Bone Marrow Wax embedded tissue Slide sections, DNA (cont'd)	Mutation detection by DNA sequencing: (cont'd)	Sequence analysis on ABI3730 platform. Data analysis Mutation Surveyor MOL EQUIP 009 MOL SEQ 008 With specific reference to the procedures below as appropriate
DNA	Familial Hypercholesterolaemia	FH NGS panel sequencing by MiSeq and analysis through commercial Sophia DDM pipeline with reference to MOL FH 006, MOL NGS 022, 023

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUE AND FLUIDS (cont'd)	Molecular Genetics (cont'd)	
DNA	Myeloid Panel Detection of mutations in MDS and AML	Thermo Fisher Myeloid Oncomine NGS DNA panel Set up on Ion Chef) and sequencing by Ion Torrent S5. Analysis through commercial Ion Reporter Analysis Pipeline SOP: MOL NGS 003, 013, 015, 018
RNA /cDNA	Myeloid Panel Detection of fusion gene / translocation detection in haematological malignancies	Thermo Fisher Oncomine Myeloid RNA Assay Set up on Ion Chef and sequencing by Ion Torrent S5. Analysis through commercial Ion Reporter Analysis Pipeline. SOP: MOL NGS 003, 013, 015, 018
DNA	Fragment size data	Agilent Tapestation 4200 for QC and DNA NGS Libraries SOP: MOL NGS 017
END		