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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 TISSUES & FLUIDS	<u>Genomics analysis for the purpose of clinical diagnosis of rare disease, inherited and acquired cancer</u> DNA extraction, quantification and quality check for subsequent in-house analysis, referral to other specialist centres and long term storage.	Documented in house procedures incorporating manufacturer's instructions (where relevant) Manual, semi automated and Automated DNA extraction and quantification using: DNA extraction: **Manual extraction processes:**
Amniotic Fluid.		Igenatal Genomic DNA extraction kit SOP GENE-WC87. **Semi-Automated and Automated extraction processes:**
Peripheral Blood, Bone Marrow, Mouth wash (Oragene Collection) and Buccal scrapes.		Chemagic 360 DNA extractor and Janus Robotic Workstation SOP GENE-WM321 and SOPGENE-WM43.
CVS, fresh tissue, frozen or fixed human tissue or cells, neonatal and Maternal peripheral bloods.		Qiagen EZ1 Advanced XL with DNA tissue kit and DNA blood kit SOP GENE-WC87. **DNA Quantification for QC purposes:**
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source.		Using: NanoDrop One and Promega Quantus Fluorometer SOP's GENE-WM363 and GENE-WM17.

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 or received as primary sample type from an external source. (cont'd)	Detection of genetic variants of SNVs and small indels. [definitive list to be held by this laboratory]	**Sanger Sequencing** Standard primer design methodology SOP's GENE-WM166 GENE-WM177 Semi- nested PCR design methodology SOP GENE-WM157 PCR amplification using in-house methodology using: For manual process PCR blocks, ABI3500xl Capillary electrophoresis instruments. For automated process Beckman Biomek NXp Liquid handler, PCR blocks. Sequencing of products by ABI3500xl Capillary electrophoresis instruments. Analysis using SoftGenetics Mutation Surveyor™ software and interpretation of variants by Alamut software. Procedures: SOP's GENE-WM42, GENE-WM136 GENE-WM16, GENE-WM28, GENE-WM19, GENE-WM12, GENE-WM8, GENE-WM57

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Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source. (cont'd)	Targeted screening for the detection of SNVs and small insertions/deletions using custom-designed gene panels. [definitive list to be held by this laboratory]	**Next Generation Sequencing** Library amplification and hybridisation performed using Illumina DNA prep kit. Enrichment with Twist Bioscience probes. Library amplification and hybridisation performed using Illumina DNA prep kit. Enrichment with Twist Bioscience or Illumina TruSight Capture probes (set up performed using the Hamilton Microlab STAR Liquid Handling System) or manually Paired-end next-generation sequencing performed on Illumnia MiSeq instruments. Using Applied Biosystems thermal cyclers, Promega Quantus fluorometer and Illumina MiSeq. Data analysis using SoftGenetics, NextGENe and Illumina Local Run Manager/ Variant Studio . SOP's GENE-WM342, GENE-WM167, GENE-WM2, GENE-WM158, GENE-WM57, GENE-WM16, GENE-WM383, GENE-WM162

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Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source. (cont'd)	Detection of fragment length size, deletions, known mutations, repeat expansions, short tandem repeats. [definitive list to be held by this laboratory] Determination of repeat size expansions. Detection of fragment length size, deletions, known mutations, repeat expansions, short tandem repeats. [definitive list to be held by this laboratory] Detection of Cystic fibrosis (CFTR) variants. Rapid detection of common trisomies.	**Fragment Length Analysis** Determination of repeat size using the Asuragen AmplideX kit with: Manual set up using Thermal cyclers, and ABI3500xl Capillary electrophoresis instrument. Analysis using SoftGenetics GeneMarker™ SOP's GENE-WM27, GENE-WM6, GENE-WM117,GENE-WM116. Detection of Cystic fibrosis (CFTR) variants using the YourGene Cystic Fibrosis Base kit Using thermal cycler and 3500XL Genetic Analysers. GENE-WM42 GENE-WM136 Analysis using SoftGenetics GeneMarker™ software. SOP's GENE-WM24, GENE-WM5 **Quantitative Fluorescence Polymerase Chain Reaction (QF-PCR)** PCR amplification using YourGene QST*R Base kit, YourGene QST*R plus XY kit, YourGene QST*R plus 13, 18, 21 kits, and YourGene Male Infertility Base kit and thermal cyclers with ABI 3500XL Genetic Analyser. Analysis and interpretation of aneuploidy results using GeneMarker software.

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Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source. (cont'd)		SOP's GENE-WC246, GENE-WC85, GENE-WC86, GENE-WC245.
	Determination of copy number changes (deletions and duplications).	**Multiplex Ligation-dependent Probe Amplification (MLPA)** Using
	Detection of fragment length size, deletions, known mutations, repeat expansions, short tandem repeats. [definitive list to be held by this laboratory]	In-house methods or MRC Holland kits, thermocyclers and ABI3500xl Genetic Analyser. Analysis using GeneMarker SOP's GENE-WM45, GENE-WM7.
	Determination of repeat size expansions.	**In-house designed** Flanking or repeat Primer PCR using in-house methods: Automated set up using Thermal cyclers, BiomekNXp liquid handling robot and ABI3500xl Capillary electrophoresis instrument. Analysis using SoftGenetics GeneMarker™ SOP's GENE-WM152, GENE-WM187, GENE-WM156, GENE-WM179, GENE-WM29, GENE-WM118, GENE-WM153, GENE-WM154, GENE-WM151, GENE-WM194, GENE-WM294. Standard in-house primer design methodology SOP GENE-WM204.

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES & FLUIDS (cont'd)	Genomics analysis for the purpose of clinical diagnosis of rare disease, inherited and acquired cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source. (cont'd)	Manual multiplex short tandem repeat (STR) PCR.	Manual multiplex short tandem repeat (STR) PCR. Using thermal cyclers and ABI3500XL genetic analyser. Analysis of marker multiplexes and haplotyping by SoftGenetics GeneMarker™. SOP's GENE-WM114 and GENE-WM70
	Detection of F8 intron 22 inversions by inverse PCR.	In-house designed primers, thermocyclers and ABI3500xl Genetic Analyser. Analysis using GeneMarker GENE- WM377. GENE-WM378,
	Detection of F8 intron 1inversions by long range PCR.	In-house designed primers, thermocyclers and Gel electrophoresis using E-Gel Power Snap electrophoresis device. GENE- WM377, GENE-WM379

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES & FLUIDS (cont'd)	Genomics analysis for the purpose of clinical diagnosis of rare disease, inherited and acquired cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
DNA extracted from Blastocyst or trophectoderm cells.	Haplotyping using multiplex short tandem repeat (STR) loci for allele size determination. [definitive list to be held by this laboratory]	**Preimplantation Genetic Testing** Standard in-house primer design methodology SOP's GENE-WM204. Whole genome amplification (WGA) of DNA from biospies using Qiagen REPLI-g kit and thermal cycler. Manual multiplex short tandem repeat (STR) PCR using PCR using using thermal cyclers and ABI3500XL genetic analyser. Analysis of loci and haplotyping by SoftGenetics GeneMarker™. SOP's GENE-WM72, GENE-WM351, GENE-WM114, GENE-WM70.

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
Whole blood Amniotic fluid CVS Bone marrow Lymph node Fresh tissue samples	G-banding/Karyotyping: Detection of chromosomal rearrangements or aberrations arising from: (e.g) Prenatally detected disorders Developmental disorders Reproductive medicine disorders Haematological/Oncology disorders. [definitive list to be held by this laboratory] (Preparative Pre-examination steps listed first)	**Manual culturing and processing of human tissue to provide metaphase cells:** using commercial media and: GENE-WC63: GENE-WC56: GENE-WC62: GENE-WC49: SOPs. Preparation of Myeloma cells using CD138+ve selection GENE-WC132: Cell Culture protcols GENE-WC1, GENE-WC95, GENE-WC96, GENE-WC57, GENE-WC58, GENE-WC64 Cell Harvest Manual in-house Protocols GENE-WC2, GENE-WC99, GENE-WC66 Slide Preparation: Slide preparation and banding techniques using Thermotron Humidity Chamber and Gemini Automated Stainer SOP's GENE-WC108, GENE-WC37 and GENE-WC38 Analysis: G:banding analysis using the Bioview imaging system SOP's GENE-WC23, GENE-WC105, GENE-WC311, GENE-WC312 GENE-WC315, GENE-WC313

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES & FLUIDS (cont'd)	<u>Genomics analysis for the purpose of clinical diagnosis of rare disease, inherited and acquired cancer</u> (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source.	Cytogenetic examinations for diagnosing postnatal disorders, prenatal diagnosis, neoplastic genetics including haemato-oncology and solid tumours, and loss of pregnancy. By detection of microscopic chromosomal imbalance (gains and losses) expressed as changes to copy number. [definitive list to be held by this laboratory]	**Microarray** Processing using the Affymetrix Cytoscan 750K and HD array chips on a GeneChips Fluidics workstation. Analysis of array data using ChAS software. Processing using Cytoscan 750K and HD array chips on a GeneChips Fluidics workstation SOP's GENE- WC70 to WC104. Analysis of array data using ChAS software for postnatal, prenatal and oncology samples. GENE-WC107, GENE-WC123, GENE-WC243.

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN TISSUES & FLUIDS (cont'd)	Genomics analysis for the purpose of clinical diagnosis of rare disease, inherited and acquired cancer (cont'd)	Documented in house procedures incorporating manufacturer's instructions (where relevant)
Cultured & uncultured cells Paraffin embedded tissues (PETs) Tumour imprints Purified plasma cells	Detection of chromosomal aberrations in the diagnosis of haematological malignancy, bone marrow failure syndromes, non-haematological malignancies and constitutional disorders and solid tumours. Break-apart probes Fusion products Deletion Insertion Copy Number / Amplification [definitive list to be held by this laboratory]	**Fluorescent in-situ hybridisation (FISH)** Preparation of PETS slides by Fluorescent in situ hybridisation for: (FISH) using VIP2000 processor GENE-WC15: Documented in house methods using commercial probes. FISH protocols for processing slides SOP's GENE-WC6, GENE-WC13, GENE-WC14, GENE-WC130
END		