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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.

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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 genetic variants of SNVs and small indels managed under flexible scope to add and remove Sanger Sequencing Targets as defined in GENE-W86 Flexible Scope policy and on Genetics Flexible Scope list GENE-W82	**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

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. (cont'd)	Targeted screening for the detection of SNVs and small insertions/deletions using custom-designed gene panels under flexible scope to add and remove from existing Twist Bioscience panels, and extend the list to include new custom Twist Bioscience panels as defined in GENE-W86 Flexible Scope policy and on Genetics Flexible Scope list GENE-W82	**Next Generation Sequencing** Library amplification and hybridisation performed using Illumina DNA prep kit. Enrichment with Twist Bioscience 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

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)	Detection of repeat expansions in target regions of the FMR1 and C9ORF72 genes	**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 repeat expansions in target regions of AR, ATN1, ATXN1, ATXN2, ATXN3, ATXN7, CACNA1A, DMPK, FXN, HTT, JPH3, PRNP and TBP genes	Flanking and/or Repeat primed PCR using in-house methods using Thermal cyclers and ABI3500xl Capillary electrophoresis instrument, set up manually or automated with a BiomekNXp liquid handling robot. Analysis using SoftGenetics GeneMarker™ SOP's GENE-WM29, GENE-WM118, GENE-WM151, GENE-WM152, GENE-WM153, GENE-WM154, GENE-WM156, GENE-WM179, GENE-WM187, GENE-WM194, GENE-WM294, GENE-WM386, GENE-WM387, GENE-WM436. 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)	<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. (cont'd)	Detection of Cystic fibrosis (CFTR) variants.	**Detection of Cystic fibrosis (CFTR) variants** 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
Genomic DNA extracted in-house from the sample types listed above or received as primary sample type from an external source. (cont'd)	Rapid detection of common trisomies.	**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 SOP's GENE-WC246, GENE-WC85, GENE-WC86, GENE-WC245.

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)	Determination of copy number changes (deletions and duplications) using In-house methods or under flexible scope using MRC Holland kits as defined in GENE-W86 Flexible Scope policy and on GeneticsFlexible Scope list GENE-W82	**Multiplex Ligation-dependent Probe Amplification (MLPA)** Using In-house methods or MRC Holland kits, thermocyclers and ABI3500xl Genetic Analyser. Analysis using GeneMarker SOP's GENE-WM45, GENE-WM7.
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 F8 intron 22 inversions by inverse PCR. Detection of F8 intron 1 inversions by long range PCR.	**Detection of F8 gene inversions** In-house designed primers, thermocyclers and ABI3500xl Genetic Analyser. Analysis using GeneMarker GENE- WM377. GENE-WM378, In-house designed primers, thermocyclers and Gel electrophoresis using E-Gel Power Snap electrophoresis device. GENE- WM377, GENE-WM379

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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. (cont'd)	Haplotyping using multiplex short tandem repeat (STR) loci for allele size determination under flexible scope to add and remove patient tailored haplotyping and direct test protocols for pre existing and new disorders as defined in GENE-W86 Flexible Scope policy and on Flexible Scope list GENE-W82	**Preimplantation Genetic Testing for Monogenic disorders (PGT-M)** Standard in-house primer design methodology SOP's GENE-WM204, GENE-W72

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DNA extracted from Blastocyst or trophectoderm cells.	Pre case workups are designed for specific disorders. Multiplex PCR of microsatellite repeat regions specific to each test. Whole genome amplification (WGA) via multiple displacement amplification (MDA) followed by multiplex PCR with pre designed assay [see above].	Manual PCR using Qiagen multiplex buffer GENE-WM114, EXT-588. Using thermal cycler and ABI 3500xl Genetic Analyser GENE-WM42, GENE-WM136 Haplotype analysis using Genemarker GENE-WM70 Qiagen REPLI-g single cell kit [EXT-1210, GENE-WM351]. Manual PCR using Qiagen multiplex buffer GENE-WM114, EXT-588. Using thermal cycler and ABI 3500xl Genetic Analyser GENE-WM42, GENE-WM136 Haplotype analysis using Genemarker GENE-WM70 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 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. (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.	**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. Post PCR purification performed manually or automated using either the Hamilton Microlab STARlet Liquid Handling system or the Biomek NXp robot. GENE-WC75 Array-Manual PCR product purification GENE-WC83 Array- Automated PCR product Purification (Biomek) GENE-WC319 Array- Automated purification of PCR products using the Hamilton MicroLAB STARlet GENE-WC317 Hamilton Starlet maintenance GENE-WC138 Hamilton Starlet maintenance log

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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)
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 Under flexible scope to add and remove commercial probes as defined in GENE-W86 Flexible Scope policy and on Flexible Scope list GENE-W82.	**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		