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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 A. Blood B. Tissue (fresh, frozen, fixed and paraffin embedded) C. Saliva D. Amniotic fluid E. Choronic villus samples F. Cultured cells G. Urine H. Bone Marrow Aspirate I. Cerebrospinal fluid (CSF) J. Blood spots K. Plasma	<u>Molecular Genetics examination activities for the purpose of clinical diagnosis</u> Detection of clinically relevant nucleic acid sequence variants	Documented in- house methods Sequencing, genotyping or electrophoresis supported by extraction and amplification as listed below. 1. Polymerase Chain Reaction (PCR) using the G-Storm GS1 & GS2 thermocyclers for PCR set-up robotically. GRI Tetrad 2 and Dyad thermocyclers for PCR set up manually (MG/SOP/MON044) (MGSOP/MON045) (MGSOP/MON054) (MG/SOP/HAE019) 2. Multiplex Ligation-dependant Probe Amplification (MLPA) using the Applied Biosystems Veriti thermocycler (MG/SOP/MON037) 3. DNA sequencing using the G-Storm GS2 thermocycler and ABI 3130 and 3730 capillary electrophoresis automated DNA sequencers (MG/SOP/ABI012) (MG/SOP/HAE016) 4. Taqman genotyping using the Applied Biosystems 7900HT fast real-time PCR system (MG/SOP/HAE017) (MG/SOP/HAE022) (MG/SOP/MON060)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd) As listed on Page 1	<u>Molecular Genetics</u> (cont'd) Detection of clinically relevant nucleic acid sequence variants	Documented in- house methods using one or a combination of the techniques below 5. Chemagic & Qiacube nucleic acid extraction robots (MG/SOP/SPE034) (MG/SOP/SPE007) (MG/SOP/SPE037) 6. Analysis of fluorescently labelled PCR and MLPA products using the ABI 3130 and 3730 capillary electrophoresis systems (MG/SOP/ABI009) (MG/SOP/PHA007) (MG/SOP/MON028) (MG/SOP/MON043) (MG/SOP/MON055) (MG/SOP/HAE005) (MG/SOP/HAE008) 7. Beckman Coulter Biomek Span 8, Beckman Coulter Biomek NXp & Eppendorf ep5075 Liquid handling robots (MG/SOP/LAB008) (MGSOP/ABI012) (MG/SOP/MON045) (MG/SOP/HAE017) 8. Manual DNA & RNA Extraction (MG/SOP/SPE010) (MG/SOP/SPE007) 9. Agarose gel electrophoresis using ABgene and Anachem gel electrophoresis rigs (MG/SOP/MON027) (MG/SOP/HAE002) (MG/SOP/HAE003)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd) As listed on Page 1	<u>Molecular Genetics</u> (cont'd) Detection of clinically relevant nucleic acid sequence variants	Documented in- house methods using one or a combination of the techniques below 10. Targeted next generation sequencing using Agilent SureSelect system and Illumina HiSeq2500 and NextSeq500. (MG/SOP/NGS003, MG/SOP/NGS001, MG/SOP/NGS005, MG/SOP/NGS013, MG/SOP/NGS019, MG/SOP/NGS007, MG/SOP/NGS021, MG/SOP/NGS025) 11. Exome sequencing- whole exome using Agilent Sureselect v6 and rare disease exome using Agilent focused exome on Illumina HiSeq2500 and NextSeq500. (MG/SOP/NGS001, MG/SOP/NGS005, MG/SOP/NGS022, MG/SOP/NGS020, MG/SOP/NGS018, MG/SOP/NGS025) 12. Droplet Digital PCR using the BioRad QX200 for low level mutation detection and for copy number variation detection (MG/SOP/MON058, MG/SOP/MON067, MG/SOP/MON059)

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd)	Molecular Genetics (cont'd)	Documented in- house methods using one or a combination of the techniques below
As listed on Page 1	Detection of clinically relevant nucleic acid sequence variants Name of disease with name of analysed gene(s) in brackets	See Pages 1 - 3
A, B, C, D, E, F	Aarskog-Scott syndrome (FGD1)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Acrodysostosis (PRKAR1A & PDE4D)	1, 3, 5, 7, 8, 10
A, H	Acute promyelocytic leukaemia (t(15;17))	1, 4, 5, 8
A, B, C, D, E, F	Alagille syndrome (JAG1, NOTCH2)	1, 2, 3, 5, 7, 8, 10
A, B, C, D, E, F	Alveolar Capillary with Misalignment of Pulmonary Veins (FOXF1)	1, 2, 3, 5, 7, 8, 10
A, B, C, D, E, F	Aortic Valve disease (NOTCH1)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Atypical Progeriod Syndrome (LMNA)	1, 3, 4, 5, 8, 10
A, B, H, I	B-cell clonality (IgH, IgK, IgL)	1, 5, 6, 8
A, B, H	BCR-ABL kinase domain sequencing (BCR-ABL)	1, 3, 5, 8
A, B, C, D, E, F	Bosley-Salih-Alorainy syndrome (HOXA1, HOXB1)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Brain Small-Vessel Disease with Haemorrhage (COL4A1 & COL4A2)	1, 3, 5, 7, 8, 10
A, B, C, F	CADASIL (NOTCH3)	1, 3, 5, 7, 8, 10
A, B, C, F	Carney Complex (PRKAR1A)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Charcot-Marie-Tooth hereditary neuropathy type 2B1 (LMNA)	1, 3, 5, 7, 8
A, B, C, D, E, F	Chondrodysplasia punctata (EBP, ARSE, PEX7, GNPAT, AGPS)	1, 3, 5, 7, 8, 10
A, H	Chronic Neutrophilic Leukaemia and Atypical CML (CSF3R)	1, 3, 5, 7, 8

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd)	Molecular Genetics (cont'd)	Documented in- house methods using one or a combination of the techniques below
As listed on Page 1	Detection of clinically relevant nucleic acid sequence variants Name of disease with name of analysed gene(s) in brackets (cont'd)	See Pages 1 - 3
A, B, H	Chronic lymphocytic leukaemia (TP53, RB1 / DLEU / MIRN15A-16 region, ATM, trisomy 12)	1, 2, 5, 7 ,6 ,8
A, B, C, F	Cole Disease (ENPP1)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Combined Pituitary Hormone deficiency (HESX1, POU1F1, PROP1, LHX3, LHX4)	1, 3, 5, 7, 8, 10
A, B, C, F	Cone Rod Dystrophy (GUCY2D)	1, 3, 5, 7, 8
A, B, C, D, E, F	Congenital abnormalities of the kidney and urinary tract and VACTERL (TRAP1)	1, 3, 5, 7, 8, 10
A, B, C, F	Congenital Fibrosis of the extraocular muscles (KIF21A, PHOX2A, TUBB3)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Congenital Generalised lipodystrophy type 1 (AGPAT2, BSCL2, CAV1, PPARG, PTRF)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Congenital hemidysplasia with ichthyosiform nevus and limb defects (CHILD) syndrome (NSDHL)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Congenital hypothyroidism (FOXE1, NKX2-1, PAX8, TSHR, TPO, TG, DUOX2, THRA)	1, 3, 5, 7, 8, 10
A, B, C, F	Cystic Fibrosis (CFTR) – screen for 38 mutations	1, 3, 5, 7, 8
A, B, C, F	Dilated Cardiomyopathy type 1A (LMNA)	1, 3, 5, 7, 8, 10
A, B, C, F	Duane radial ray syndrome (Okihiro syndrome) (SALL4)	1, 3, 5, 7, 8, 10
A, B, C, F	Duane Retraction Syndrome (CHN1)	1, 3, 5, 7, 8, 10

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd)	Molecular Genetics (cont'd)	Documented in- house methods using one or a combination of the techniques below
As listed on Page 1	Detection of clinically relevant nucleic acid sequence variants Name of disease with name of analysed gene(s) in brackets (cont'd)	See Pages 1 - 3
A, B, C, F	Emery Dreifuss Muscular Dystrophy (LMNA)	1, 3, 5, 7, 8, 10
A, B, C, D, E	Exome sequencing	1, 3, 7, 11, 12
A, B, C, F	Familial Hepatic Adenomas due to bi-allelic inactivation of HNF1A (HNF1A)	1, 2, 3, 5, 7, 8, 10
A, B, C, D, E, F	Familial Glucocorticoid deficiency (MC2R, MCM4, MRAP, NNT, STAR)	1, 3, 5, 7, 8, 10
A, B, C, F	Familial Hyperparathyroidism (MEN1, CASR, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, RET)	1, 2, 3, 5, 7, 8, 10
A, B, C, F	Familial hypocalciuric hypercalcaemia (AP2S1, GNA11, CASR)	1, 3, 5, 7, 8, 10
A, B, C, F	Familial Hypoparathyroidism (CASR, GCM2, GNA11, PTH)	1, 3, 5, 7, 8, 10
A, B, C, F	Familial Isolated Pituitary Adenoma (AIP)	1, 2, 3, 5, 7, 8, 10
A, B, C, D, E, F	Familial partial lipodystrophy (LMNA, PPARG, PLIN1)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Familial Porencephaly (COL4A1, COL4A2, JAM3)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Fanconi-Bickel syndrome (SLC2A2)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Feingold Syndrome (MYCN, MIR17HG)	1, 2, 3, 5, 7, 8, 10
A, B, H	Follicle centre cell lymphoma (t(14;18))	1, 3, 5, 6, 8, 9
A, B, C, D, E, F	Gastrointestinal atresia (CFAP53, CHD7, FANCB, FANCC, GLI3, MID1, MYCN, RFX6, SOX2, TTC7A, EFTUD2, FOXF1)	1, 2, 3, 5, 7, 8, 10

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd)	Molecular Genetics (cont'd)	Documented in- house methods using one or a combination of the techniques below
As listed on Page 1	Detection of clinically relevant nucleic acid sequence variants Name of disease with name of analysed gene(s) in brackets (cont'd)	See Pages 1 - 3
A, B, C, D, E, F	Generalised Arterial Calcification of Infancy (ABCC6, ENPP1, NT5E)	1, 3, 5, 7, 8, 10
A, B, C, F	Glucose-Galactose Malabsorption (SLC5A1)	1, 3, 5, 7, 8, 10
A, C	Haemochromatosis (HFE)	4, 5, 7, 8
B	Hairy Cell Leukaemia (BRAF)	1, 3, 5, 7, 8
A, B, C, D, E, F	Hajdu-Cheney Syndrome (NOTCH2)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Hanac Syndrome (COL4A1)	1, 3, 5, 7, 8, 10
A, B, C, F	Hereditary Phaeochromocytoma / paraganglioma (RET, VHL, SDHB, SDHC, SDHD, SDHAF2, TMEM127, MAX, FH, SDHA)	1, 2, 3, 5, 7, 8, 10
A, B, C, D, E, F	Hirschsprung disease (RET)	1, 2, 3, 5, 7, 8, 10
A, C	HLA-B27	4, 5, 7, 8
A, B, C, D, E, F	Holoprosencephaly (GLI2, PTCH1, SHH, SIX3, TGIF1, ZIC2)	1, 2, 3, 5, 7, 8, 10
A, B, C, F	Horizontal gaze palsy with progressive scoliosis (ROBO3)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Hyperinsulinism (ABCC8, KCNJ11, HNF4A, GLUD1, GCK, HADH)	1, 2, 3, 5, 7, 8, 10
A, B, C, D, E, F	Hyperphoshatemic familial tumoral calcinosis (GALNT3, FGF23, KL, SAMD9)	1, 3, 5, 7, 8, 10
A, B, C, F	Hyperthyroidism & Congenital Hypothyroidism (TSHR)	1, 3, 5, 7, 8, 10
A, B, C, F	Hypophosphatemic rickets (PHEX, FGF23, DMP1, ENPP1)	1, 2, 3, 5, 7, 8, 10

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd)	Molecular Genetics (cont'd)	Documented in- house methods using one or a combination of the techniques below
As listed on Page 1	Detection of clinically relevant nucleic acid sequence variants Name of disease with name of analysed gene(s) in brackets (cont'd)	See Pages 1 - 3
A, B, C, F	Hypophosphatemic rickets with Hypercalciuria (SLC34A3)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	IPEX Syndrome (FOXP3)	1, 3, 5, 7, 8, 10
A, C	Inherited thrombophilia (F5, F2)	4, 5, 7, 8
A, B, C, D, E, F	Isolated Growth Hormone Deficiency (GH1, GHRHR)	1, 2, 3, 5, 7, 8, 10
A, B, C, D, E, F	Kallmann syndrome (KAL1, FGFR1, FGF8, PROKR2, PROK2)	1, 2, 3, 5, 7, 8, 10
A, B, C, D, E, F	Limb Girdle Muscular Dystrophy type 1b (LMNA)	1, 3, 5, 7, 8, 10
B	Malignant Melanoma (BRAF)	1, 3, 5, 7, 8
A, B, C, D, E, F	Mandibuloacral Dysplasia (ZMPSTE24, LMNA, POLD1)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Mandibulofacial dysostosis with microcephaly (EFTUD2, SF3B4)	1, 3, 5, 7, 8, 10
A, B, C	11p15 maternal loss of heterozygosity testing in pancreatic tissue	1, 5, 6, 8
A, B, C, D, E, F	Maternal Cell Contamination & Specimen Source Identification	1, 5, 6, 8
A, B, C, D, E, F	Mobius syndrome (PLXND1, REV3L)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Multiple Exostosis (EXT1, EXT2)	1, 3, 5, 7, 8, 10
A, B, H	Mantle cell lymphoma (t(11;14))	1, 3, 5, 6, 8, 9
A, H	Mantle cell lymphoma (Cyclin D1)	1, 4, 5, 8
A, B, C, D, E, F	Maturity-onset diabetes of the young (MODY) (GCK, HNF1A, HNF4A)	1, 2, 3, 5, 7, 8, 10

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd)	Molecular Genetics (cont'd)	Documented in- house methods using one or a combination of the techniques below
As listed on Page 1	Detection of clinically relevant nucleic acid sequence variants Name of disease with name of analysed gene(s) in brackets (cont'd)	See Pages 1 - 3
A, B, C, F, G	MELAS (mitochondrial m.3243A>G mutation)	4, 5, 7, 8
B	Metastatic colorectal cancer (KRAS, NRAS, BRAF)	1, 3, 5, 7, 8
A, B, C, F	Multiple Endocrine Neoplasia types 1 & 4 (MEN1, CDKN1B)	1, 2, 3, 5, 7, 8, 10
A, B, C, F	Multiple Endocrine Neoplasia type 2 (RET)	1, 3, 5, 7, 8, 10
A, B, C, F	Medullary Thyroid Carcinoma (RET, RAS)	1, 3, 5, 7, 8
A, C, F, H	Myeloproliferative disorders (JAK2, MPL, CALR)	1, 3, 4, 5, 7, 8, 12
A, B, C, D, E, F	Neonatal Diabetes (ABCC8, KCNJ11, INS)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Neonatal Diabetes with Pancreatic and cerebellar agenesis (PTF1A)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Neonatal Diabetes and Congenital Hypothyroidism (GLIS3)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Nestor-Guillermo Progeria Syndrome (BANF1)	1, 3, 5, 7, 8, 10
B	Non-Small cell lung cancer (EGFR)	1, 3, 4, 5, 6, 7, 8
A, B, C, D, E, F	Pontocerebellar hypoplasia (AMPD2, CASK, CHMP1A, CLP1, EXOSC3, PCLO, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VPS53, VRK1)	1, 3, 5, 7, 8, 10
A, B, C, F	Prediction of 5-fluorouracil toxicity (DPYD)	1, 3, 5, 7, 8
A, B, C, F	Prediction of Irinotecan toxicity (UGT1A1)	1, 3, 5, 7, 8

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd)	Molecular Genetics (cont'd)	Documented in- house methods using one or a combination of the techniques below
As listed on Page 1	Detection of clinically relevant nucleic acid sequence variants Name of disease with name of analysed gene(s) in brackets (cont'd)	See Pages 1 - 3
A, B, C, F	Primary pigmented nodular adrenocortical disease (PRKAR1A, PDE8B, PDE11A)	1, 3, 5, 7, 8, 10
A, B, C, F	Pseudoxanthoma elasticum (ABCC6 & ENPP1)	1, 3, 5, 7, 8, 10
A, B, C, F	Pseudohypoaldosteronism type 2 (WNK1, WNK4, CUL3, KLHL3)	1, 3, 5, 7, 8, 10
A, B, C, F	Pyridoxine dependent epilepsy (ALDH7A1)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Renal Cysts & Diabetes (RCAD) Syndrome (HNF1B)	1, 2, 3, 5, 7, 8, 10
A	Detection of Rhesus D fetal genotype using circulating cell free DNA	4, 5
A, B, C, D, E, F	Restrictive Dermopathy (ZMPSTE24, LMNA)	1, 3, 4, 5, 8, 10
A, B, C, D, E, F	SHORT syndrome (PIK3R1)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Spondylocostal Dysostosis (DLL3, MESP2, LFNG, HES7, TBX6, RIPPLY2)	1, 3, 5, 7, 8, 10
A, B, C, F, H	Systemic mastocytosis (KIT)	1, 3, 5, 7, 8, 12
A, B, H, I	T-cell clonality (TCRg, TCRb, TCRd)	1, 5, 6, 8
A, B, C, D, E, F	Thiamine Responsive Megaloblastic Anaemia (SLC19A2)	1, 3, 5, 7, 8
A, B, C, D, E, F	Thyroid hormone resistance (THRB)	1, 3, 5, 7, 8, 10
A, B, C, D, E, F	Visceral Heterotaxy (CFC1, ZIC3)	1, 3, 5, 7, 8, 10
A, B, C, F, H	Waldenstrom's Macroglobulinemia (MYD88)	1, 3, 5, 7, 8, 12

Materials/Products tested	Type of test/Properties measured/Range of measurement	Standard specifications/ Equipment/Techniques used
HUMAN BODY FLUIDS (cont'd) As listed on Page 1 A, B, C, D, E, F	<u>Molecular Genetics</u> (cont'd) Detection of clinically relevant nucleic acid sequence variants Name of disease with name of analysed gene(s) in brackets (cont'd) Werner Syndrome (WRN) X-linked Acrogigantism (GPR101)	Documented in- house methods using one or a combination of the techniques below See Pages 1 – 3 1, 3, 5, 7, 8, 10 1, 3, 5, 7, 8, 10, 12
END		