Services offered at the SciLifeLab unit Clinical Genomics Uppsala

Services within solid tumors

Category	Name	Description	Clinically validated
Large-scale NGS	Whole exome for tumor tissue	Twist exome on FFPE or fresh frozen tumor samples and paired normal samples.	No
	RNA-seq for tumor tissue	RNA sequencing on fresh frozen samples.	No
Solid tumor gene panels	BRCA1/BRCA2 panel	Multiplex PCR (xGen Amplicon) based resequencing of all coding exons of *BRCA1* and *BRCA2*.	Yes
	cfDNA EGFR pathway panel	Multiplex PCR (xGen Amplicon) based resequencing of hotspot mutations in *EGFR* , *KRAS* , *NRAS* and *BRAF* , on cell free circulating DNA.	Yes
	cfDNA Lung cancer panel	Multiplex PCR (xGen Amplicon) based resequencing of hotspot mutations in 17 lung cancer related genes, on cell free circulating DNA.	Yes
	Colon cancer panel	HaloPlex based resequencing of all coding exons of 13 genes + 15 markers associated with colorectal cancer.	Yes
	Lung cancer panel	HaloPlex based resequencing of all coding exons of 18 lung cancer related genes.	Yes
	Pan-cancer panel (GMS560)	Hybrid capture based comprehensive genomic profiling. DNA capture: 544 genes, RNA capture: 44 genes. FFPE and fresh frozen tissue. SNVs, indels, CNVs, fusion genes, MSI and TMB analysis.	Yes
	Tumor actionable mutations panel	HaloPlex based resequencing of hotspot regions of 8 genes associated with various solid tumor types, including GIST, melanoma and MTC.	Yes
NanoString expression panels	NanoString breast cancer Prosigna	Molecular subtyping of breast cancer and risk score for recurrence.	Yes
	NanoString lung cancer fusion gene panel	Detection of ALK, RET and ROS1 fusion genes + MET exon 14 skipping (RNA based).	Yes
	NanoString predesigned panels	E.g. PanCancer immune profiling panel, PAM50, BC360 (RNA based).	No
Sensitive detection	Digital PCR plasma	TERT promotor, EGFR, KRAS and customized probes on request.	No
Long-read sequencing	Whole genome long-read sequencing	Whole genome sequencing of fresh frozen samples on ProtmethION sequencer from Oxford nanopore technologies	No
Other molecular methods	Single cell analysis	Single-cell multi-omics (DNA, RNA and protein) using the Tapestri instrument from Mission Bio.	No
Other services	Tissue processing	All tissue processing prior to DNA extraction, including sectioning, staining, slide scanning and determination of tumor cell content. Only in connection to other services.	
	Consulting	Consultation in matters regarding study design and molecular methods.	
	Bioinformatics support	Assistance with standard and tailored bioinformatic analyses.	
	Clinical interpretation	Clinical interpretation of variants is available for clinically validated methods.	
	Development projects	Collaborative projects aimed at improved molecular diagnostics.	

Services within hematology/hematopathology

Category	Name	Description	Clinically validated
Large-scale NGS	Whole genome sequencing	PCR free whole genome sequencing for cancer (both T-only and T/N paired samples)	No
	Whole exome for tumor tissue	Twist exome on FFPE or fresh frozen tumor samples and paired normal samples.	No
	RNA-seq for tumor tissue	RNA sequencing on fresh frozen samples and bone marrow.	No
Hematology gene panels	Archer Heme fusion gene panel	Archer FusionPlex based detection of fusions of 87 genes (RNA based).	Yes
	RUNX1 panel	Twist based sequencing panel for comprehensive characterization of the *RUNX1* gene (SNV, indels, larger deletions and insertions).	No
	Swift myeloid panel	Swift Biosciences Myeloid panel (30 genes).	No
	Twist myeloid panel	Twist based myeloid panel (195 genes).	Yes
Sensitive detection	Digital PCR for gene variants	Sensitive detection of specific genetic variants using Bio-Rad droplet digital PCR. Over 400 validated assays including cancer hotspots available. Customized probes on request.	Yes
	Digital PCR for gene fusions	Sensitive detection of specific fusion transcripts using Bio-Rad droplet digital PCR. Over 20 validated assays including fusion genes available. Customized probes on request.	Yes
	Lymphotrack	Detection of clonal rearrangements in leukemias.	Yes
	SuperRCA	Sensitive detection of specific genetic variants using super rolling circle amplification. Validated assays for cancer hotspots. Customized probes on request.	No
Long-read sequencing	Whole genome long-read sequencing	Whole genome sequencing on ProtmethION sequencer from Oxford nanopore technologies	No
Other molecular methods	BCR-ABL1 kinase domain mutation analysis	Detection of resistance mutations in *BCR*::*ABL1* fusion gene using Nextera library prep.	Yes
	Devyser Chimerism analysis	NGS based method for post-transplant monitoring	Yes
	Single cell analysis	Single-cell multi-omics (DNA, RNA and protein) using the Tapestri instrument from Mission Bio.	No
Other services	Cell separation	Antibody-based cell separation. Only in connection with other services.	
	Consulting	Consultation in matters regarding study design and molecular methods.	
	Bioinformatics support	Assistance with standard and tailored bioinformatic analyses.	
	Clinical interpretation	Clinical interpretation of variants is available for clinically validated methods.	
	Development projects	Collaborative projects aimed at improved molecular diagnostics.	

Services within inherited diseases

Category	Name	Description	Clinically validated
Large-scale NGS	Whole genome sequencing	PCR free whole genome sequencing for inherited disorders.	No
	Whole exome sequencing	Twist Comprehensive Exome panel.	Yes
	RNA-seq for blood	RNA sequencing on blood samples.	No
Inherited disorder gene panels	Alport syndrome panel	In silico panel of 6 genes connected to Alport syndrome extracted from Twist exome.	Yes
	Amyolidosis, Transthyetin	In silico panel of *TTR* from Twist exome.	Yes
	Arrythmia and cardiomyopathy panel	In silico panel of 100 genes connected to arrhythmia and cardiomyopathy extracted from Twist exome.	Yes
	Bartter and Gitelman syndrome	In silico panel of 13 genes connected to Gitelman, Bartter and Liddle syndrome from Twist exome.	Yes

	CADASIL panel	In silico panel of *NOTCH3* extracted from Twist exome.	Yes
	Cerebral microangiopathy	In silico panel of 36 genes connected cerebral microangiopathy from Twist exome.	Yes
	Connective tissue disorder panel	In silico panel of 44 genes connected to connective tisue disorders extracted from Twist exome.	Yes
	Craniosynostosis panel	In silico panel of 65 genes connected to craniosynostosis extracted from Twist exome.	Yes
	Cystisk Fibrosis	In silico panel of *CFTR* extracted from Twist exome.	Yes
	Dystonia	In silico panel of 67 genes connected to dystonia extracted from Twist exome.	Yes
	Epidermolysis bullosa panel	In silico panel of 21 genes connected to epidermolysis bullosa extracted from Twist exome.	Yes
	Epilepsy panel	In silico panel of 371 genes connected to epilepsy extracted from Twist exome.	Yes
	Familial Hypocalciuric Hypercalcemia (FHH)	In silico panel of 3 genes connected to FHH extracted from Twist exome.	Yes
	Hereditary hemorrhagic telangiectasia panel	In silico panel of 6 genes connected to hereditary hemorrhagic telangiectasia extracted from Twist exome.	Yes
	Hereditary spastic paraplegia	In silico panel of 87 genes connected to spastic paraplegia extracted from Twist exome.	Yes
	Hyperparathyroidism	In silico panel of 7 genes connected to hyperparathyroidism extracted from Twist exome.	Yes
	Hypoparathyroidism	In silico panel of 9 genes connected to hypoparathyroidism extracted from Twist exome.	Yes
	Ichthyosis panel	In silico panel of 71 ichthyosis and palmoplantar keratosis genes extracted from Twist exome.	Yes
	Inherited cancer	Twist Bioscience custom panel (28 genes).	Yes
	Inherited hematology panel	In silico panel of 215 genes connected to inherited hematological malignancies extracted from Twist exome.	Yes
	Macrocephaly and overgrowth syndrome	In silico panel of 50 genes connected to macrocephaly and overgrowth extracted from Twist exome.	Yes
	Multipel endocrine neoplasia type 1 (MEN1)	In silico panel of *MEN1* extracted from Twist exome.	Yes
	Multipel endocrine neoplasia type 2A (MEN2A)	In silico panel of *RET* extracted from Twist exome.	Yes
	Myotonia and Paramyotonia Congenita panel	In silico panel of *CLCN1* and *SCN4A* extracted from Twist exome.	Yes
	Nephronophthisis	In silico panel of 27 genes connected to nephronophthisis extracted from Twist exome.	Yes
	Nephrotic syndrome	In silico panel of 54 genes connected to nephrotic syndrome extracted from Twist exome.	Yes
	Neurofibromatosis type 1	In silico panel of *NF1* and *SPRED1* extracted from Twist exome.	Yes
	Neuromuscular diseases panel	In silico panel of 129 genes connected to neuromuscular diseases extracted from Twist exome.	Yes
	Neuropathy panel	In silico panel of 98 genes connected to neuropathy extracted from Twist exome.	Yes
	Obesity	In silico panel of 52 genes connected to obesity extracted from Twist exome.	Yes
	Osteogenesis imperfecta	In silico panel of 67 genes connected to osteogenesis imperfecta and fragile bones extracted from Twist exome.	Yes
	Paraganglioma and pheochromocytoma	In silico panel of 10 genes connected to paraganglioma and pheochromocytoma extracted from Twist exome.	Yes
	Periodic fever panel	In silico panel of 14 genes connected to periodic fever extracted from Twist exome.	Yes
	Periodic paralysis panel	In silico panel of 4 genes connected to periodic paralysis extracted from Twist exome.	Yes
	POLG-related disease	In silico panel of *POLG* extracted from Twist exome.	Yes
	Polycystic kidney disease	In silico panel of 13 genes connected to polycystic kidney extracted from Twist exome.	Yes
	Primary ciliary dyskinesia (PCD)	In silico panel of 42 genes connected to PCD extracted from Twist exome.	Yes
	Rasopathies panel	In silico panel of 19 genes connected to rasopathies extracted from Twist exome.	Yes
	Sotos panel	In silico panel of NSD1 and NFIX extracted from Twist exome.	Yes
	Thrombocytopenia panel	In silico panel of 127 genes connected to thrombocytopenia extracted from Twist exome.	Yes
	Thyroid hormone resistance panel	In silico panel of 21 genes connected to thyroid hormone resistance extracted from Twist exome.	Yes
	Tuberous sclerosis panel	In silico panel of *TSC1* and *TSC2* extracted from Twist exome.	Yes
	Von Hippel-Lindau (VHL) disease	In silico panel of *VHL* extracted from Twist exome.	Yes
Long-read sequencing	Whole genome long-read sequencing	Whole genome sequencing on ProtmethION sequencer from Oxford nanopore technologies	No
Other molecular methods	Non Invasive Prenatal Test (NIPT)	Illumina NIPT test of plasma samples	Yes
Other services	Consulting	Consultation in matters regarding study design and molecular methods.	
	Bioinformatics support	Assistance with standard and tailored bioinformatic analyses.	
	Clinical interpretation	Clinical interpretation of variants is available for clinically validated methods.	
	Development projects	Collaborative projects aimed at improved molecular diagnostics.	

Services within microbiology

Category	**Name**	**Description**	**Clinically validated**
Microbiology	Amplicon Microorganisms	all kind of amplicons as for instance 16S, 18S, ITS, virulence, resistance	No
	Bacterial WGS (long read)	Whole genome long read Sequencing of bacteria, optional assembly	No
	Metagenomics Bacteria (long read)	Shotgun metagenomics, optional bioinformatics	No
	SARS-CoV-2	Whole genome sequencing of SARS-CoV-2 inkl. assembly	Yes
Other services	Consulting	Consultation in matters regarding study design and molecular methods.	
	Bioinformatics support	Assistance with standard and tailored bioinformatic analyses.	
	Clinical interpretation	Clinical interpretation of variants is available for clinically validated methods.	
	Development projects	Collaborative projects aimed at improved molecular diagnostics.	

Please contact us or visit our website for further information about our services and how to set up a project.
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