Services offered at the Clinical Genomics facility, SciLifeLab, Uppsala

ID	Name	Description	Clinically validated
WP1_1	Tissue processing (solid tumors)	All tissue processing prior to DNA extraction, including sectioning, staining, slide scanning and determination of tumor cell content.	
WP1_2	Colon cancer panel	HaloPlex based resequencing of all coding exons of 13 genes + 15 markers associated with colorectal cancer.	Yes
WP1_3	Lung cancer panel	HaloPlex based resequencing of all coding exons of 18 lung cancer related genes.	Yes
WP1_4	Pan cancer panel	HaloPlex based resequencing of all coding exons of 32 genes associated with various solid tumor types, including lung, colon, gastric and melanoma.	Yes
WP1_5	GIST/melanoma panel	HaloPlex based resequencing of hotspot regions of 8 genes associated with various solid tumor types, including GIST, melanoma, lung and colon.	Yes
WP1_6	BRCA1/BRCA2 panel	Multiplex PCR (Accel-Amplicon) based resequencing of all coding exons of *BRCA1* and *BRCA2.*	Yes
WP1_7	cfDNA, EGFR pathway panel	Multiplex PCR (Accel-Amplicon) based resequencing of hotspot mutations in *EGFR* , *KRAS* , *NRAS* and *BRAF* , on cell free circulating DNA.	Yes
WP1_8	cfDNA, lung cancer panel	Multiplex PCR (Accel-Amplicon) based resequencing of hotspot mutations in 17 lung cancer related genes, on cell free circulating DNA.	Yes
WP1_9	NanoString lung cancer fusion gene panel	Detection of ALK, RET and ROS1 fusion genes + MET exon 14 skipping (RNA based).	Yes
WP1_10	NanoString predesigned panels	E.g. PanCancer immune profiling panel, PAM50, BC360 (RNA based).	
WP2_1	TruSight Myeloid panel	Illumina TruSight Myeloid panel (54 genes).	Yes
WP2_2	BCR-ABL1 PacBio	Detection of resistance mutations in BCR-ABL1 fusion gene using PacBio.	Yes
WP2_3	Archer Heme fusion gene panel	Archer FusionPlex based detection of fusions of 87 genes (RNA based).	Yes
WP2_4	KLL panel	In silico panel of TP53, SF3B1 and NOTCH1 extracted from TruSight Myeloid panel.	Yes
WP2_5	Archer Myeloid panel	Multiplex PCR (Archer VariantPlex) based resequencing of 75 genes associated with myeloid malignancies.	
WP2_6	Digital PCR	Sensitive detection of specific genetic variants using Bio-Rad droplet digital PCR.	

WP2_7	NanoString lymphoma classification	Classification of lymphoma (RNA based).	
WP3_6	Whole exome for inherited disorders	SureSelect Clinical Research Exome.	Yes
WP3_7	Custom constitutional panel (CCP17)	SureSelect resequencing panel of ca 5700 genes.	Yes
WP3_8	Craniosynostosis panel	In silico panel of 9 genes connected to craniosynostosis extracted from CCP17.	Yes
WP3_9	Rasopathies panel	In silico panel of 19 genes connected to Rasopathies extracted from CCP17.	Yes
WP3_10	Connective tissue disorder panel	In silico panel of 42 genes connected to connective tisue disorders extracted from CCP17.	Yes
WP3_11	Thrombocytopenia panel	In silico panel of 66 genes connected to thrombocytopenia extracted from CCP17.	Yes
WP3_12	Arrythmia panel	In silico panel of 39 genes connected to Arrhythmia extracted from CCP17.	Yes
WP3_13	Ichthyosis panel	In silico panel of 71 ichthyosis and palmoplantar keratosis genes extracted from Agilent Clinical Research Exome.	Yes
WP3_14	Inherited hematology panel	In silico panel of 112 genes connected to inherited hematological malignancies extracted from CCP17.	Yes
WP_U	Development projects	The facility can undertake clinical sequencing projects related to solid tumors, hematology and hereditary diseases.	
WP_K	NGS consulting	Consultation in matters regarding NGS (bioinformatics etc).	

We provide assistance with standard bioinformatic analyses. For clinically validated methods, we also provide clinical interpretation of variants.

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

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