

List of products at Clinical Genomics facility, SciLifeLab, Uppsala

Product ID	Product name	Product description
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	Diagnostic sequencing, colon cancer gene panel	HaloPlex based targeted resequencing of all coding exons of 13 genes + 15 markers associated with colorectal cancer
WP1_3	Diagnostic sequencing, lung cancer gene panel	HaloPlex based targeted resequencing of all coding exons of 18 lung cancer related genes
WP1_4	Diagnostic sequencing, Pan cancer gene panel	HaloPlex based targeted resequencing of all coding exons of 32 genes associated with various solid tumor types, including lung, colon, gastric and melanoma
WP1_5	Diagnostic sequencing, GIST/melanoma	HaloPlex based targeted resequencing of hotspot regions of 8 genes associated with various solid tumor types, including GIST, melanoma, lung and colon
WP1_6	Diagnostic sequencing, BRCA1/BRCA2 panel	Multiplex PCR (Accel-Amplicon) based resequencing of all coding exons of <i>BRCA1</i> and <i>BRCA2</i>
WP1_7	Diagnostic sequencing of cfDNA, EGFR pathway panel	Multiplex PCR (Accel-Amplicon) based resequencing of hotspot mutations in <i>EGFR</i> , <i>KRAS</i> , <i>NRAS</i> and <i>BRAF</i> , on cell free circulating DNA
WP2_1	Diagnostic sequencing, TruSight Myeloid panel	Illumina TruSight Myeloid panel (54 genes). Sequencing and clinical interpretation of variants.
WP2_2	Diagnostic sequencing, BCR-ABL1 PacBio	BCR-ABL1 fusion gene detection using PacBio. Sequencing and clinical interpretation of variants.
WP2_3	Diagnostic sequencing, Archer Heme fusion gene detection	Archer™ FusionPlex™ Heme Panel. Sequencing and clinical interpretation of variants.
WP3_1	Diagnostic sequencing, Ichthyosis <i>in silico</i> panel Inherited disease	Gene panel of 38 ichthyosis genes extracted from Agilent Inherited disease. Sequencing and clinical interpretation of variants.
WP3_2	Inherited disease reduced exome	Sequencing and analysis of Inherited disease reduced exome (approximately 2700 genes) from Agilent.
WP3_3	Diagnostic sequencing, ARVC/LQT gene panel	Haloplex based gene panel of 10 genes connected to Arrhythmogenic right ventricular cardiomyopathy and Long QT. Sequencing and clinical interpretation of variants.
WP3_4	Diagnostic sequencing, Connective tissue gene panel	Haloplex based gene panel of 31 genes connected to connective tissue disorders. Sequencing and clinical interpretation of variants.
WP3_5	Diagnostic sequencing, Rasopathies gene panel	Haloplex based gene panel of 18 genes connected to Rasopathies.

		Sequencing and clinical interpretation of variants.
WP3_6	Diagnostic sequencing, Whole exome for inherited disorders	Agilent Clinical Research Exome. Sequencing and clinical interpretation of variants.
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).

Please contact us for further information about the products and how to set up a project.

<https://www.scilifelab.se/facilities/clinical-genomics-uppsala/>

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