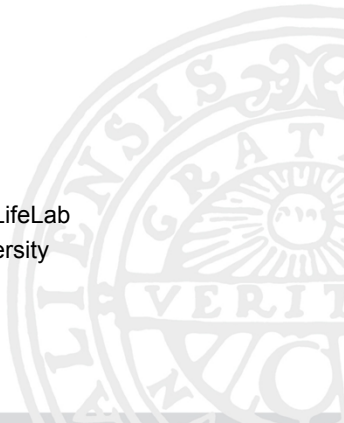



Status of the exome sequencing in Prediction ADR

Ulrika Liljedahl
 Head of Sequencing
 SNP&SEQ Technology Platform
 National Genomics Infrastructure hosted by SciLifeLab
 Department of Medical Sciences, Uppsala University

NGS instrumentation and applications

5 HiSeqX instruments

- Human whole genome sequencing

4 HiSeq2500 instruments

- All possible sequencing applications.

1 MiSeq instrument



- Amplicons, small genomes, target panels.

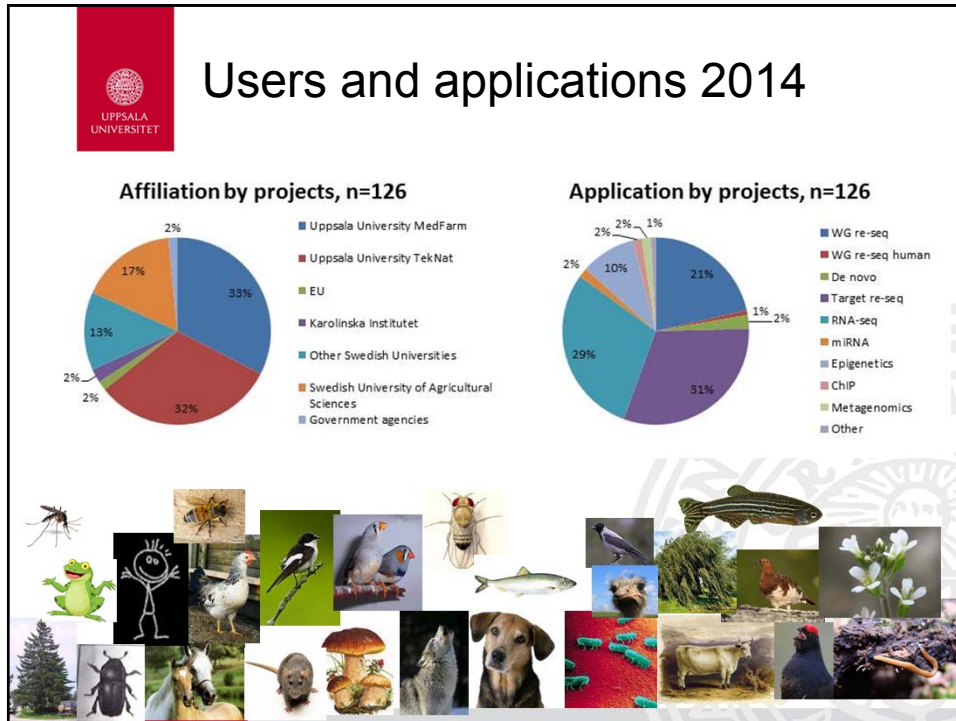
DNA

- Whole genome sequencing (WGS)
 - De novo
 - Re-seq
- Targeted Sequencing
 - Exome
 - Custom Panels
 - Amplicons
- Epigenetics
 - Whole genome bisulfite sequencing (WGBS)
 - Reduced representation bisulfite sequencing (RRBS)
 - Targeted Panels
 - Chromatin Immunoprecipitation sequencing (ChIP-seq)


RNA



















- Total RNA (Ribosomal RNA depletion)
- mRNA (poly-A selection)
- smallRNA (miRNA and other small regulatory RNAs)




Sequencing staff




					
Ulrika Liljedahl, PhD Head of sequencing, Project coordinator	Jessica Nordlund, PhD Head of R&D	Johanna Lagensjö Project coordinator	Ellenor Devine, PhD Project coordinator	Jon Ramsell, PhD Lab coordinator, sample manager	Project and lab management
					Library preparation and MiSeq runs.
Heidur Loftsdottir Research engineer	Helena Fällmar, PhD Research engineer	Maria Hägglund, PhD Research engineer	Olof Wadell Research engineer	Johanna Manninen Research engineer	
					HiSeq2500, HiSeqX and MiSeq runs.
Ingvar Örn Thorsteinsson Research engineer	Katarina Tegnér Research engineer	Sara Ekberg Research engineer	Marie Lindersson Research engineer	Elin Övernäs, PhD Research engineer	
					Library preparation, HiSeq2500, HiSeqX and MiSeq runs.
	Ann-Christin Wiman Research engineer	Karin Sollander Research engineer	Magnus Lindell, PhD Research engineer		

Additional staff


Bioinformatics and IT




Ann-Christine Syvänen
Director SNP&SEQ




Katarina Jonasson
Admin assistant



Kristina Larsson
Quality systems manager




Pontus Larsson, PhD
Head of bioinformatics




Lars Bäckström
Head of IT systems


R&D




Jessica Nordlund PhD
Head of R&D




Amanda Raine PhD,
Research engineer




Per Lundmark, PhD
Bioinformatician




Johan Dahlberg
Bioinformatician




Patrik Smeds
Bioinformatician




Matilda Åslin
Bioinformatician




Sofia Adolfsson
Engineer




Susanne Forsberg
Technician




Mathias Brännvall, PhD
EU project coordinator





Edvard Englund, PhD
Systems developer



Johan Hermansson
Systems developer

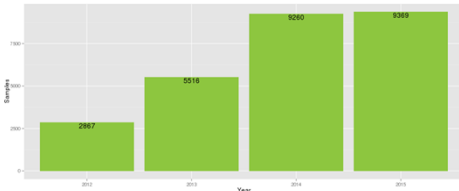


Steinar Sturlaugsson
Systems developer

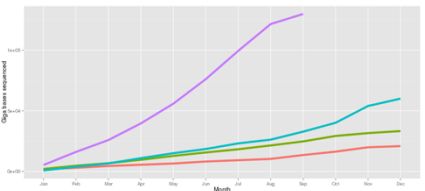



Data growth

Total number of samples per year



Cumulative GB per month




HiSeq2500 generates up to 1 Tb per instrument per week

- With 4 instruments we can produce up to 4 Tb per week

HiSeqX will produce 2 x 1.6 Tb per instrument and week

- With 5 instruments we will be producing 16 Tb per week!!

Year	Gbases
2011	10 077
2012	20 850
2013	32 425
2014	60 000
2015 jan-sept	>130 000




National Genomics Infrastructure


- one of the platforms at SciLifeLab

- To make a state-of-the-art infrastructure for massively parallel DNA sequencing and SNP genotyping available to researchers in Sweden
- To enable Swedish scientists to perform internationally competitive research in genomics
- To provide guidelines and support for sample collections, study design and protocol selection


Platform Directors



Joakim Lundeberg, KTH
Professor in Gene Technology
SciLifeLab Stockholm





Ulf Gyllenstein, UU
Professor in Medical Genetics
Uppsala Genome Center



Ann-Christine Syvänen, UU
Professor in Molecular Medicine
SNP&SEQ Technology Platform

Web Portal for submission of projects <https://portal.scilifelab.se/genomics>





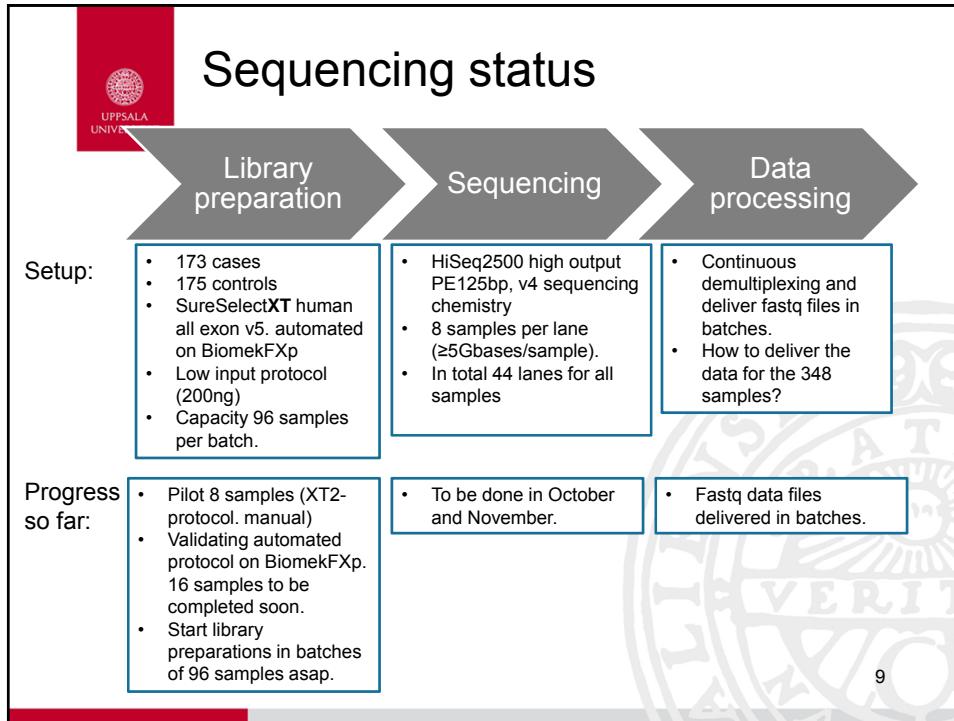
SciLifeLab
TECHNOLOGIES & SERVICES ▾
RESEARCH ▾
EDUCATION ▾
COLLABORATION ▾

Find more information and search for what you need on the page for Technologies & Services

What is the difference between national and regional facilities?

National facilities

<p style="font-weight: bold; margin: 0;">Affinity Proteomics</p> <p style="margin: 0;">Biobank Profiling Cell Profiling Fluorescence Tissue Profiling Mass Cytometry PLA Proteomics Protein and Peptide Arrays Tissue Profiling</p> <p style="font-weight: bold; margin: 0;">Bioimaging</p> <p style="margin: 0;">Advanced Light Microscopy Fluorescence Correlation Spectroscopy</p> <p style="font-weight: bold; margin: 0;">Bioinformatics</p> <p style="margin: 0;">Bioinformatics Compute and Storage (JPPNEX) Bioinformatics Long-term Support (WABI) Bioinformatics Short-term Support and Infrastructure (BLS)</p>	<p style="font-weight: bold; margin: 0;">Chemical Biology Consortium Sweden</p> <p style="margin: 0;">Laboratories for Chemical Biology Umeå (LCBU) The Laboratories for Chemical Biology at Karolinska Institutet (LCBKJ) Uppsala Drug Optimization and Pharmaceutical Profiling (UDOPP)</p> <p style="font-weight: bold; margin: 0;">Clinical Diagnostics</p> <p style="margin: 0;">Clinical Biomarkers Clinical Genomics Clinical Sequencing</p>	<p style="font-weight: bold; margin: 0;">Drug Discovery and Development</p> <p style="margin: 0;">ADME (Absorption Distribution, Metabolism Excretion) of Therapeutics (UDOPP) Biochemical and Cellular Screening Biophysical Screening and Characterization Human Antibody Therapeutics In Vitro and Systems Pharmacology Medicinal Chemistry – Hi2Lead Medicinal Chemistry – Lead Identification Protein Expression and Characterization</p> <p style="font-weight: bold; margin: 0;">Functional Genomics</p> <p style="margin: 0;">Eukaryotic Single Cell Genomics Karolinska High Throughput Center (KHTC) Microbial Single Cell Genomics Single Cell Proteomics</p>	<p style="font-weight: bold; margin: 0;">National Genomics Infrastructure</p> <p style="margin: 0;">NGI Stockholm (Genomics Applications) NGI Stockholm (Genomics Production) NGI Uppsala (SNP&SEQ Technology Platform) NGI Uppsala (Uppsala Genome Center)</p> <p style="font-weight: bold; margin: 0;">Structural Biology</p> <p style="margin: 0;">Protein Science Facility</p>
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Metrics from evaluation of automated library prep

Sample	% mapped	% at 20x coverage	% on target	% duplicates
CEPH10860 200ng	99.4	98.5	69.4	13.9
CEPH10860 3µg	99.5	97.3	72.7	12.4
CEPH11992 200ng	99.1	98.0	68.6	13.1
CEPH11992 3µg	99.0	97.7	69.2	16.5
CEPH11993 200ng	99.4	97.1	71.5	8.8
CEPH11993 3µg	99.5	98.2	72.8	12.9
Average from 8 pilot samples	95.3	87.1	59.5	5.4

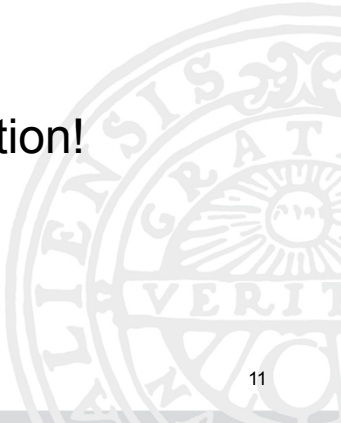
NB! More data generated per sample for CEPH samples compared to the 8 pilot samples.

10




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Thank you for your attention!



11



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