

SciLifeLab Bioinformatics Platform

National Bioinformatics Infrastructure Sweden (NBIS)

Björn Nystedt

RNAseq

Uppsala 06.11.2017

To be a national hub for **molecular life sciences**



SciLifeLab

National service

The Swiss army knife for Swedish
Life Science researchers

Local scientific
center



Director: Olli Kallioniemi
Co-director: Siv Andersson

Vision:

To be an internationally leading center that develops, uses and provides access to advanced technologies for molecular biosciences with focus on health and environment.

2010: Strategic research initiative
2013: National resource
2015: New management/chairman

SciLifeLab platforms

SciLifeLab national service

VR

SNIC

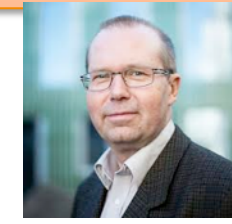
National Genomics Infrastructure

Diagnostics Development

Single-cell Biology

National Bioinformatics Infrastructure Sweden

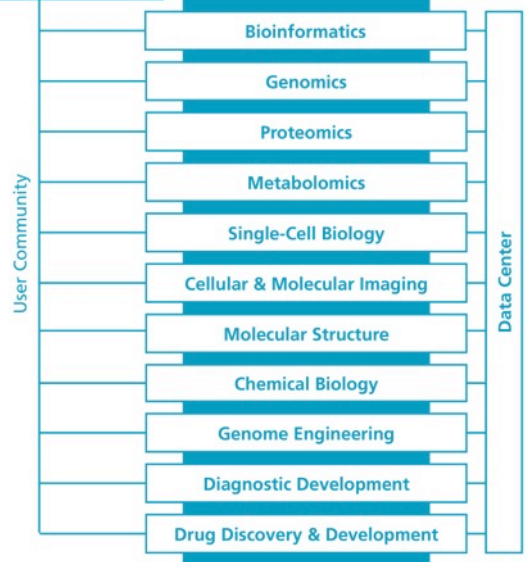
Bengt Persson



Computer resources free for Swedish researchers

Academia, Public Agencies, Healthcare & Industry

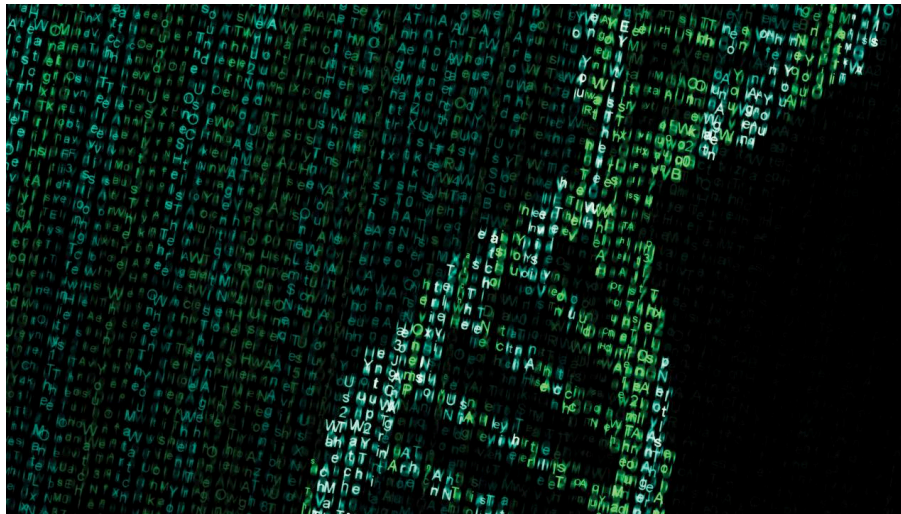
National Research Profiles



SciLifeLab Bioinformatics (NBIS) and Genomics (NGI) were both top-ranked in the recent VR evaluation (September 2017).

“NBIS is probably the largest genuinely national and fully established bioinformatics infrastructure in Europe.”

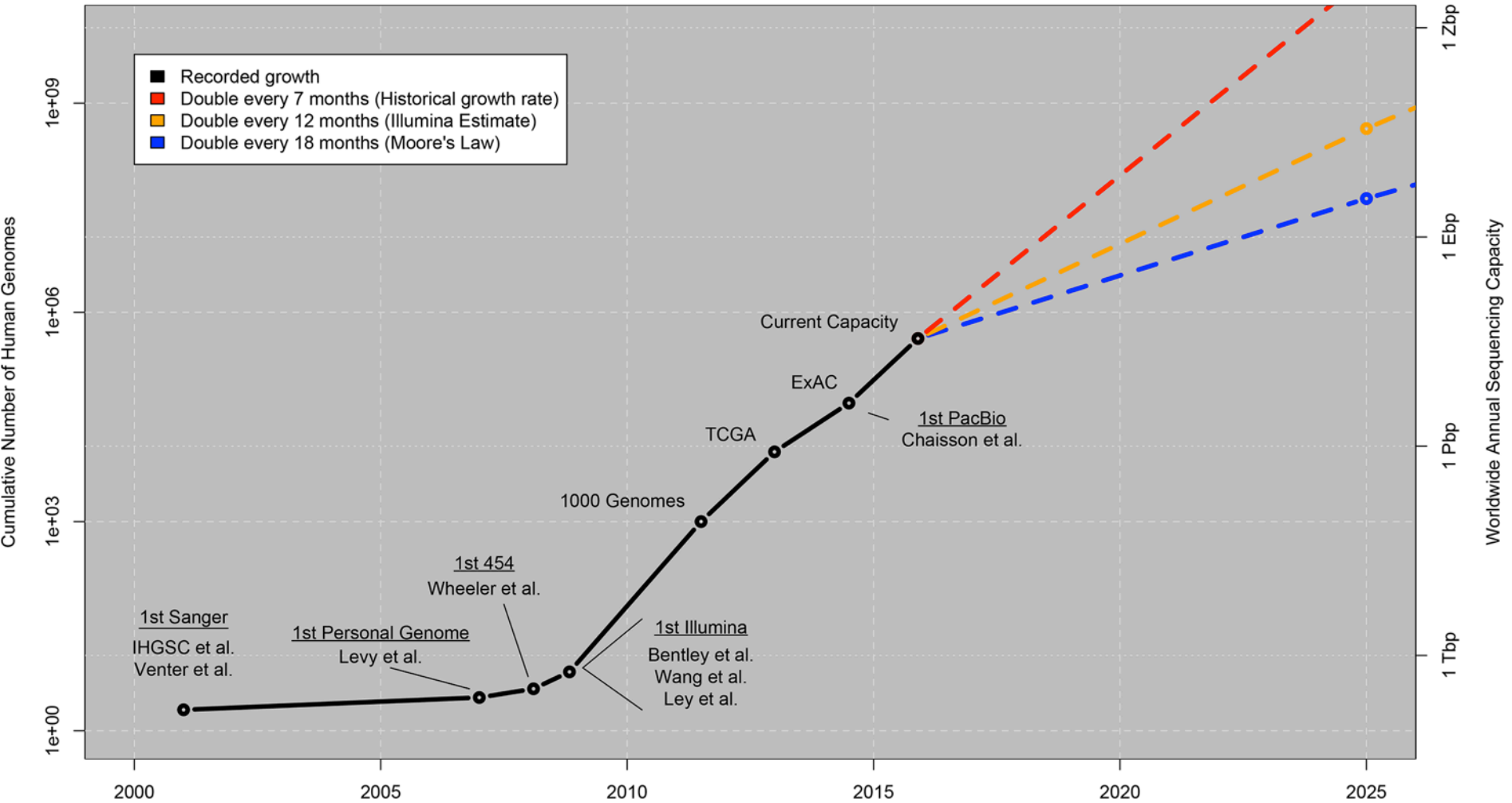
“[NBIS..] is crucial to the future competitiveness of Sweden in data-driven life sciences research, and is helping to keep Sweden in the European forefront in the area.”



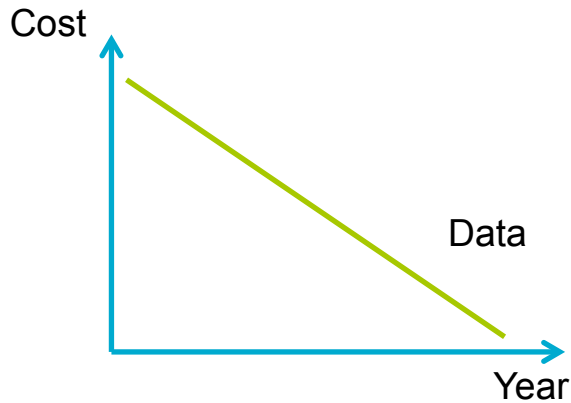
Scientific impact: 7/7 (“*Outstanding*”)
Overall score: 7/7 (“*Outstanding*”)

Data growth

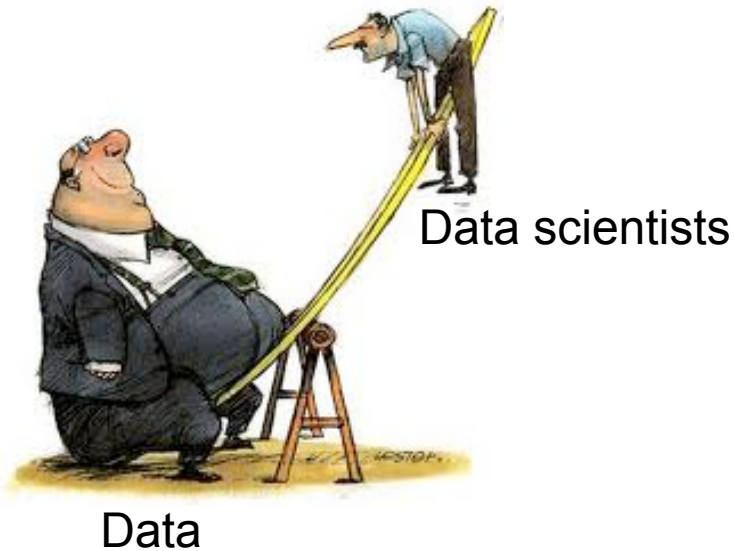
Growth of DNA Sequencing



Stephens ZD, Lee SY, Faghri F, Campbell RH, Zhai C, et al. (2015) Big Data: Astronomical or Genomical?. PLoS Biol 13(7): e1002195. doi:10.1371/journal.pbio.1002195
<http://127.0.0.1:8081/plosbiology/article?id=info:doi/10.1371/journal.pbio.1002195>

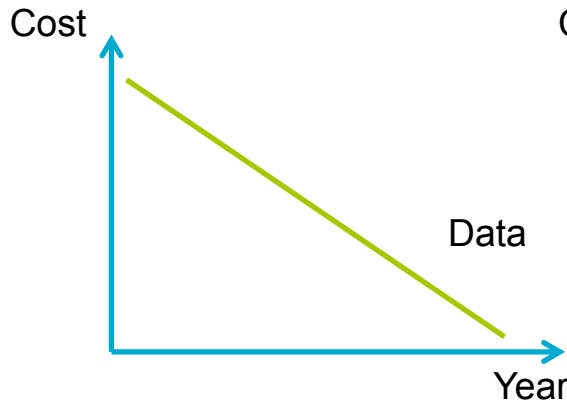


“Per base”

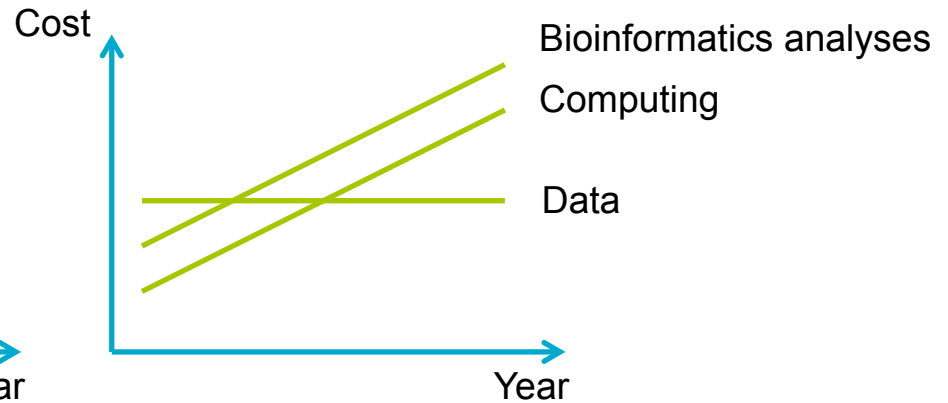


Our role

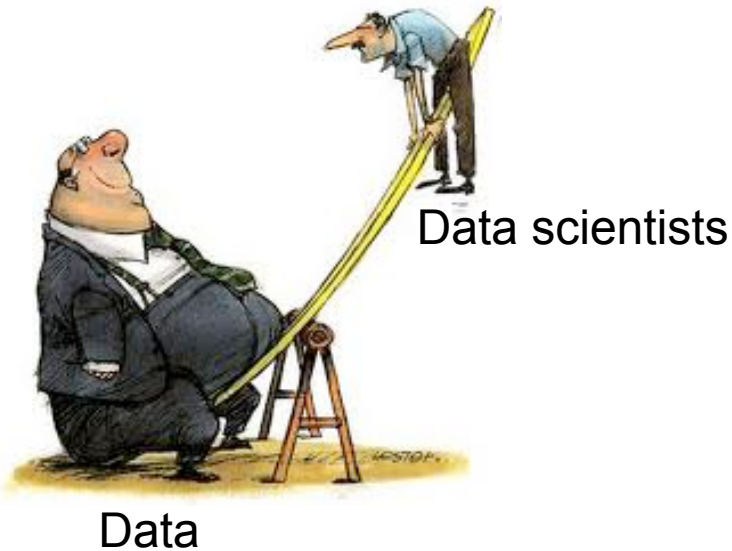
We want to help the Swedish Life Science community to **build knowledge** in large-scale data analysis, and to make bioinformatics **easily accessible** for all.



“Per base”



“Per project”



Our role

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NBIS activities

Support, tools and training



Support



Tools



Training



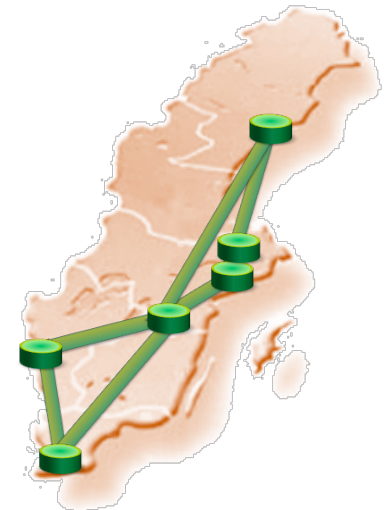


Custom-tailored support

www.nbis.se

www.scilifelab.se/platforms/bioinformatics/

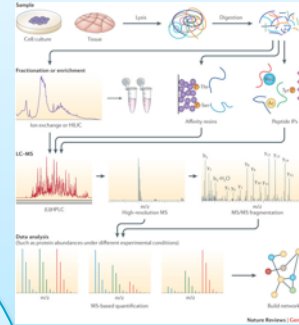
- Study design consultation (free)
www.nbis.se/support/supportform/index.php
+ drop-in sessions every week @ all 6 sites
- Support (User fee 800 kr/h)
www.nbis.se/support/supportform/index.php
- Long-term support and systems biology
(500h, free, scientific evaluation)
www.nbis.se/support/supportform/index.php?form=longterm



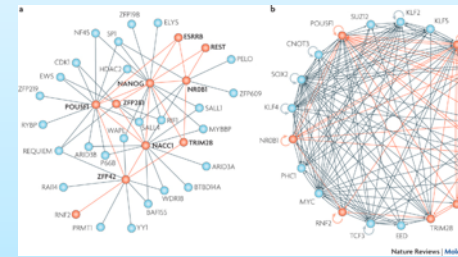
Bioinformatics support



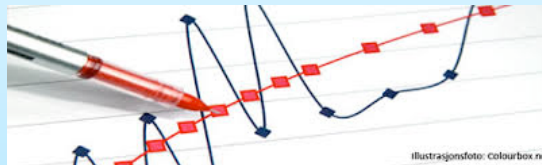
Genomics



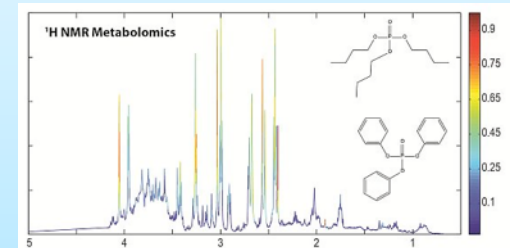
Proteomics



Systems biology



Biostatistics



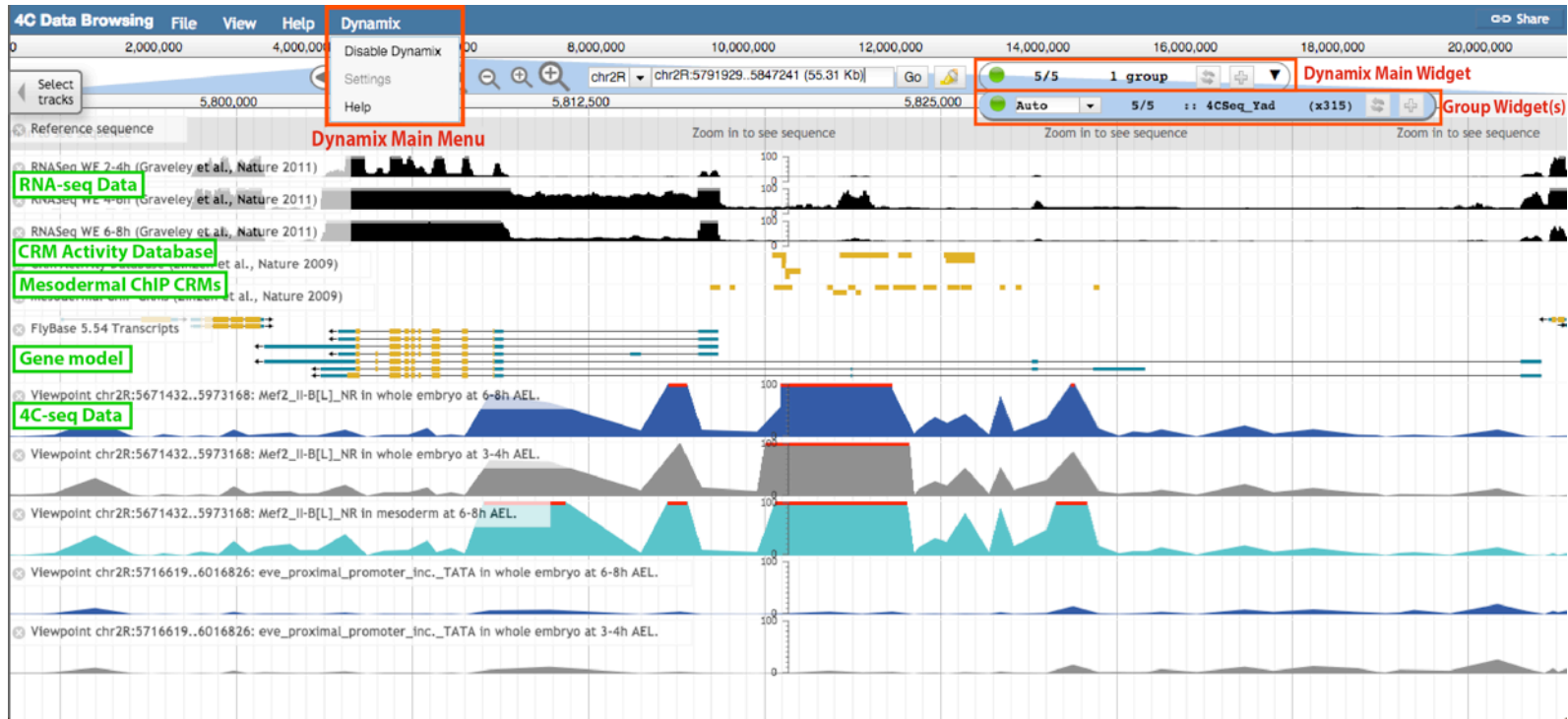
Metabolomics

2 tracks!

- Fee-for-service (800kr/h)
Rapid turnaround
- Scientific ranking (free)
“Long-term Support”
3 open calls/year

How to get support nbis.se

The screenshot shows a web browser window displaying the nbis.se website. The browser's address bar shows 'nbis.se'. The website's navigation menu includes 'Support', 'Infrastructure', 'Training', 'News', and 'About'. The 'Support' dropdown menu is open, listing the following options: 'General information', 'Consultation', 'Support', 'Long-term Support', 'Genome Assembly and Annotation', and 'Apply here!'. The 'Support' and 'Apply here!' options are circled in red. Below the navigation menu, the website features a large banner with the text 'NBIS NATIONAL BIOINFORMATICS INFRASTRUCTURE SWEDEN' and a sub-header: 'NBIS is a distributed national bioinformatics infrastructure, supporting life sciences in Sweden'. At the bottom of the page, there are three green buttons with icons representing people, a gear, and a graduation cap. The browser's address bar at the bottom left shows 'nbis.se/support'.



- 10 - 20 projects per year
- Highly specialized staff and robust pipelines
- Tight user interaction
- Numerous manual and semi-manual QC steps
- Supports ENA submission
- Editable user interface

Cost effective with high quality!

BigData/Integrative omics

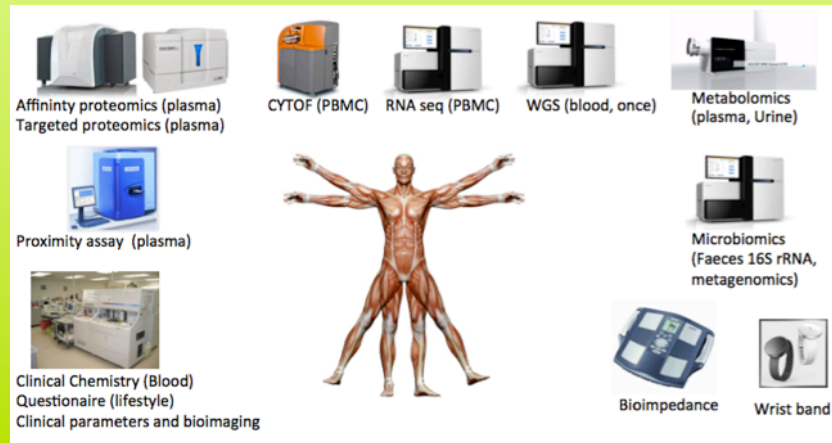
4 FTE, joint effort by Long-term Support and Systems Biology
Projects apply in the regular Long-term Support calls

Combine data from SciLifeLab platforms

- Building **tools** and **resources** for handling very large and/or complex biological data sets
- Typically performed in the context of longer support projects
- State-of-the-art analytical methods for integrating multi-modal biological data sets, eg
 - Machine learning/deep learning
 - Graph-based models
 - Genome-scale metabolic models

Support track for integrative projects

First call Feb 2016; First few projects initiated
Involves extensive integration of data





Compute and storage of sensitive data

- Local EGA
- ePouta integration pilot
- microMosler
- Pouta Blueprints
- web-servers with EGI cloud vo.NBIS.se

WGS tools and resources

- SweGen 1000 genomes
- WGS somatic variant calling WF
- WGS structural variation WF

Software maintenance

- MrBayes
- Structure prediction web services

Assembly and annotation

- Falcon on Milou
- ENA submission help

Other tools and resources

- Human Metabolic Atlas (HMA)
- Haloplex variant calling pipeline
- WhatsHap: Genomic phasing
- IgDiscover: Immunorepertoire

NBIS Support ▾ Infrastructure ▾ Training ▾ News Events About ▾ SciLifeLab

Tools

The current tools under development and maintenance by NBIS staff are listed below. For more detailed information on the tools, follow the links. See also the [SciLifeLab Open Source website](#) for tools developed at other SciLifeLab platforms.

CAW URL: http://opensource.scilifelab.se/projects/caw/ CAW (Cancer Analysis Workflow) is a complete open source pipeline to detect somatic variants from WGS data.	Software type: Command-line tool, Workflow Platform: Linux Development status: Active Released: Yes Contact: maxime.garcia@scilifelab.se Affiliation: scilifelab.se
ChIP-seq QC pipeline URL: https://github.com/ewels/clusterflow Pipeline for quality control of ChIP-seq data, implemented as a plug-in for the workflow manager ClusterFlow. See pipelines/chipseq_qc.config in the ClusterFlow repository on GitHub.	Software type: Workflow Platform: Linux Development status: Maintained Released: Yes Contact: jakub.westholm@scilifelab.se Affiliation: nbis.se
EMBLmyGFF3	Software type: Command-line tool Platform: Linux, Mac

<http://nbis.se/infrastructure/tools/>

SweGen Variant Frequency Database

- 950 twin registry + 50 Northern Sweden
- Deep coverage WGS (30X)
- ExAC browser interface
- Data Beacon
- Full SNP frequency table download



<https://swefreq.nbis.se/#/>
1st release October 2016

Variant: 22:46615880 T / C

Filter Status: PASS
dbSNP: rs1800234
Allele Frequency: 0.0035
Allele Count: 7 / 2000
UCSC: 22-46615880-T-C
ClinVar: [Click to search for variant in ClinVar](#)

Genotype Quality Metrics
Site Quality Metrics

Annotations

This variant falls on 7 transcripts in 1 genes:

missense
• PPARA - Transcripts

intron
• PPARA - ENST00000434345

non coding transcript exon
• PPARA - ENST00000493286

Note: This list may not include additional transcripts in the same gene that the variant does not overlap.

Population Frequencies

Population	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
SweGen	7	2000	0	0.0035
Total	7	2000	0	0.0035

Read Data

This interactive IGV.js visualization shows reads that went into calling this variant.

Note: Read data is not available for this variant.

chr22:46,615,730-46,616,030 301 bp

Funding: SciLifeLab
Sequencing: NGI
Variant calling: NGI
QC: NBIS
Data access interface: NBIS



SciLifeLab course curriculum 2018-2020

Course	Location
Advanced Molecular Technology and Instrumentation for Proteome Analyses	Uppsala
Biophysical methods in drug discovery	Uppsala / Sthlm
Cellular profiling within the Human Protein Atlas/Spatial Proteomics	Stockholm
Chemical Proteomics	Stockholm
Cryo-EM sample preparation and data collection	Umeå
Cryo Electron Tomography and image processing	Stockholm
Intermediate level R for Bioinformatics. Summer school.	Gotland
Introduction to bioinformatics using NGS data (4 x / year)	UU, LiU, LU, UmU, GU
Opportunities for Affinity Proteomics	Stockholm
Single cell genomics - a practical and theoretical workshop	Uppsala
Single Particle Cryo-EM image processing	Stockholm

Date	Training
Application Nov/ Dec	The Swedish Bioinformatics Advisory Program (1-2 years)

NBIS Courses

- Python Programming (2x / year)
- R programming (2x / year)
- RNA-seq (2x /year)
- Single cell RNA-seq analysis
- CHIP-seq data analysis
- Genome Annotation
- *De novo* Genome Assembly
- Metagenomics

<https://www.scilifelab.se/education/courses/>

The Swedish Bioinformatics Advisory Program

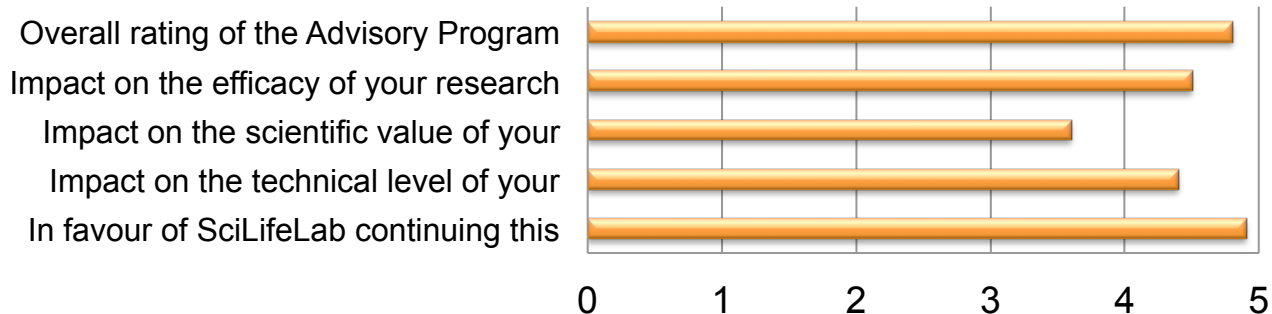
PhD students get a senior bioinformatician as a personal advisor during 2 years of their PhD. Monthly project meetings + two grand meetings per year to aid networking and knowledge transfer.

www.scilifelab.se/education/mentorship/the-swedish-bioinformatics-advisory-program/

Last call (2017/2018): 111 applicants for 15 places
Next call opens Nov 2017!

The Swedish Bioinformatics Advisory Program

Student evaluation, June 2015



- Mini-symposia
- Roadshows, Open House
- Scientific workshops and conferences
 - Swedish Bioinformatics Workshop, plasma profiling, microscopy, epigenetics and chromatin etc
 - <https://www.scilifelab.se/events/>

Attendance sheet

- Please sign morning and afternoon

Fika and lunches together every day

- 10, 12:00, 15
- Fika outside the lecture hall

Course dinner

- Tue 7/11, at 18:00, Meza Grill & Bar
- Sign up latest today before lunch
- Included: pre-ordered menu, 1 drink

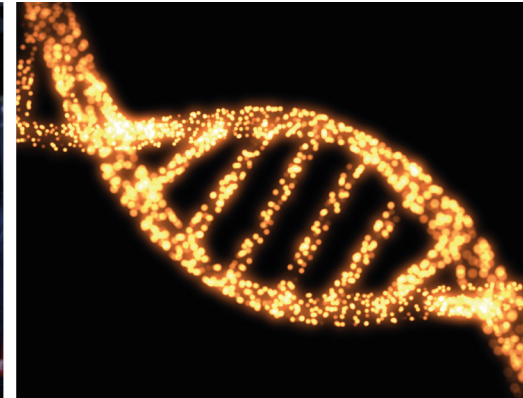
Science & SciLifeLab Prize



Join the Symposium, Dec 11, 2017!

<https://scienceprize.lpages.co/scientific-symposium/>

- A grand prize winner receives a prize of US \$30,000; and each of the three category winners will receive US \$10,000
- The grand prize winning essay will be published in *Science* and essays from the three category winners will be published online



“to incent our best and brightest to continue in their chosen fields of research”

We're here for you!
www.nbis.se

SciLifeLab



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NATIONAL BIOINFORMATICS
INFRASTRUCTURE SWEDEN