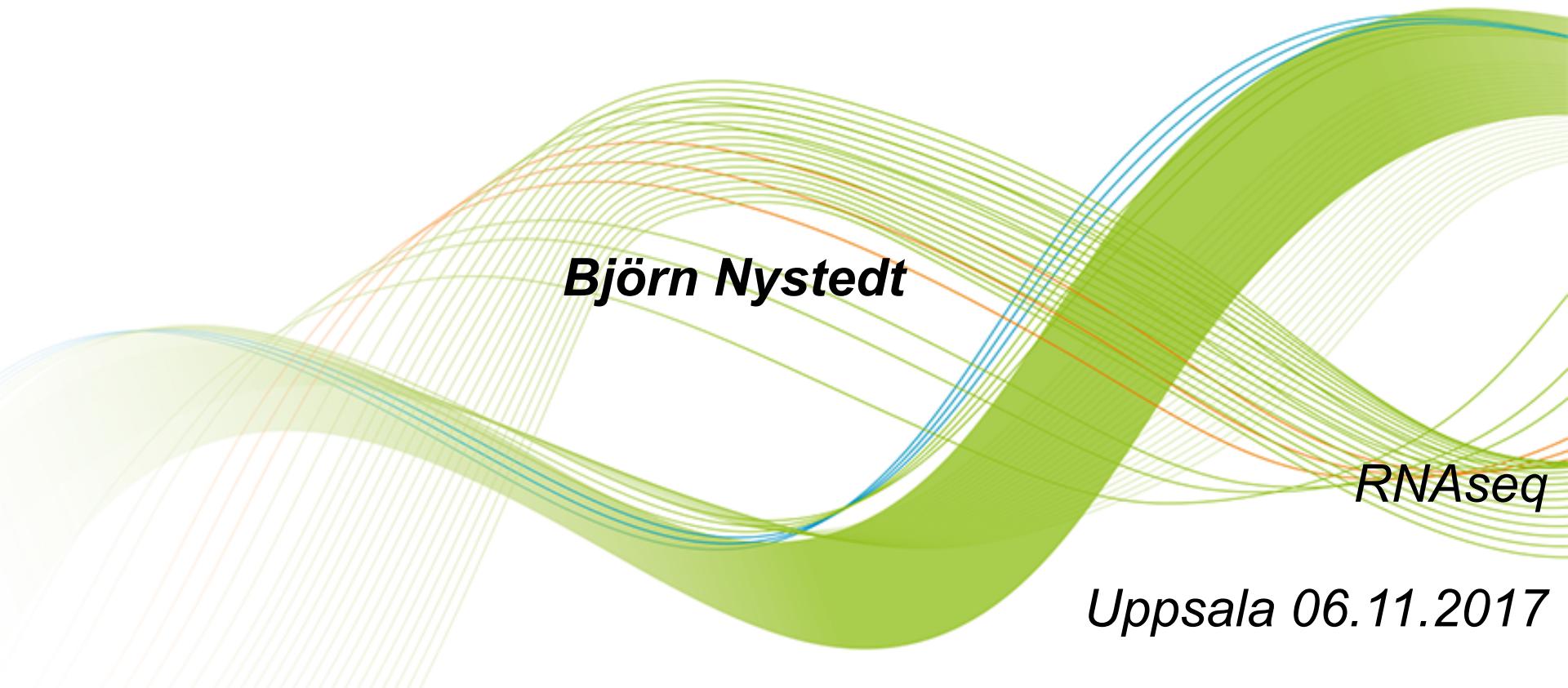


SciLifeLab Bioinformatics Platform

National Bioinformatics Infrastructure Sweden (NBIS)

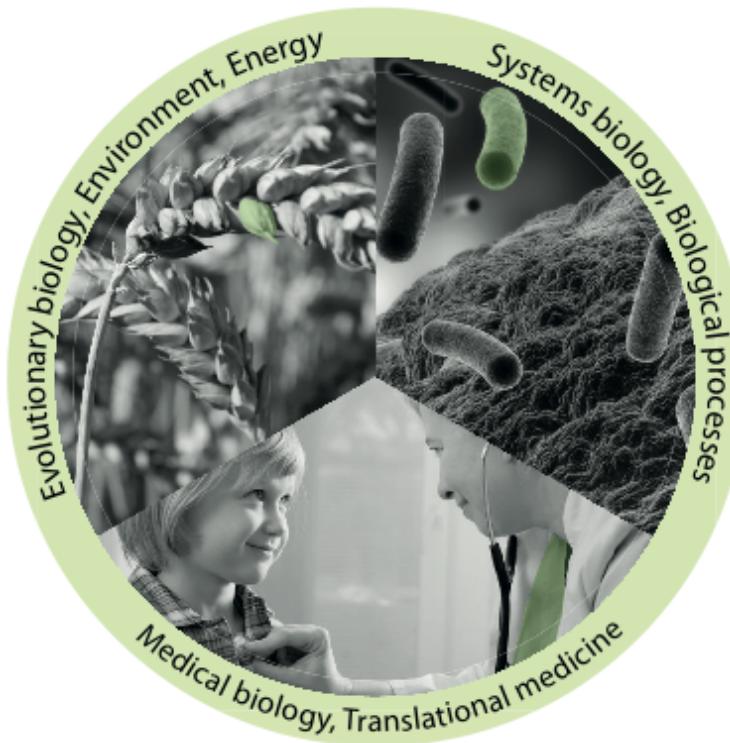


Björn Nystedt

RNAseq

Uppsala 06.11.2017

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



SciLifeLab

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

National
Genomics
Infrastructure

Diagnostics
Development

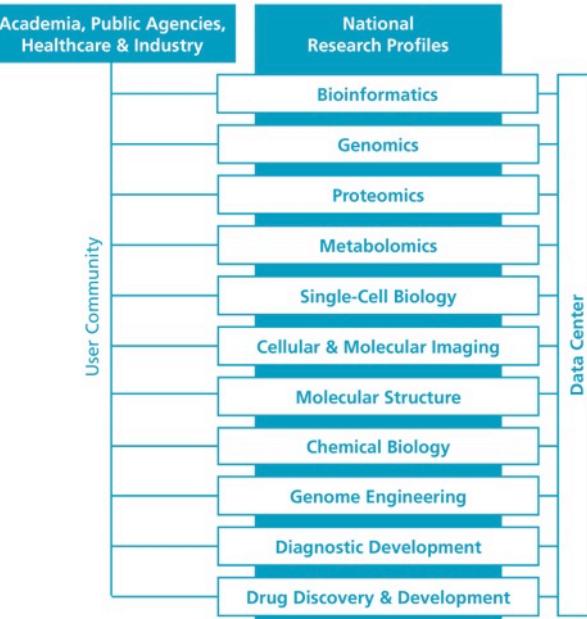
Single-cell
Biology

National
Bioinformatics
Infrastructure
Sweden

Bengt Persson



NBS
NATIONAL BIOINFORMATICS
INFRASTRUCTURE SWEDEN



VR

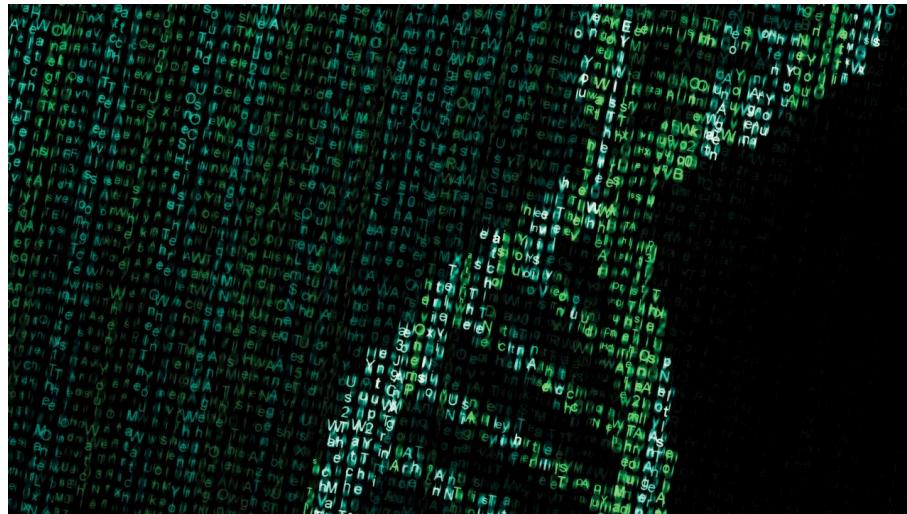
SNIC

Computer
resources
free for
Swedish
researchers

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.”

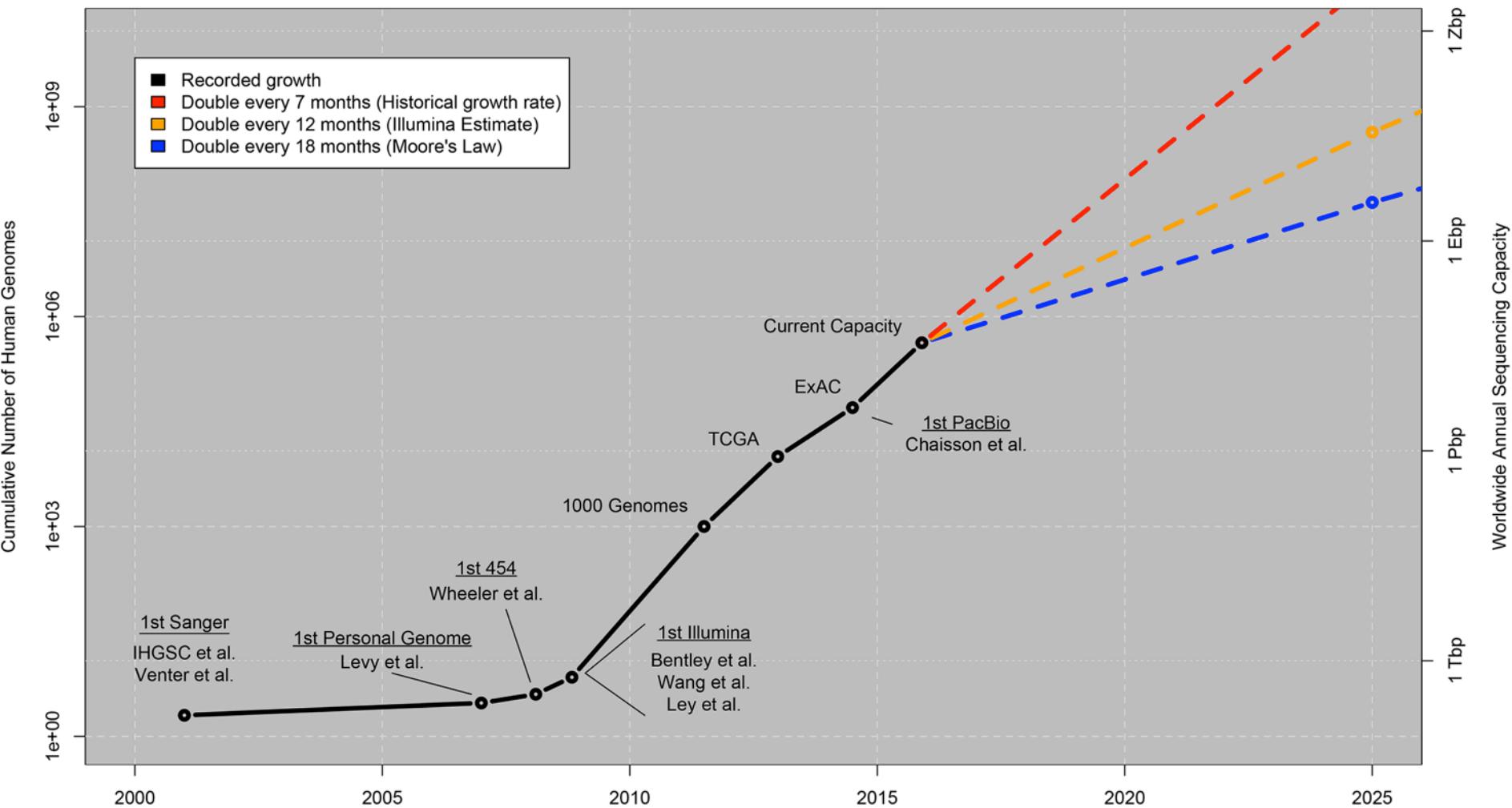


Scientific impact: 7/7 (“Outstanding”)

Overall score: 7/7 (“Outstanding”)

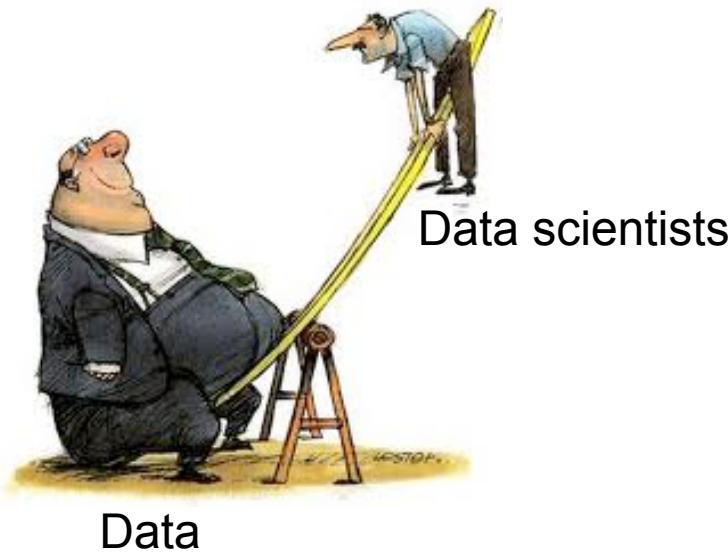
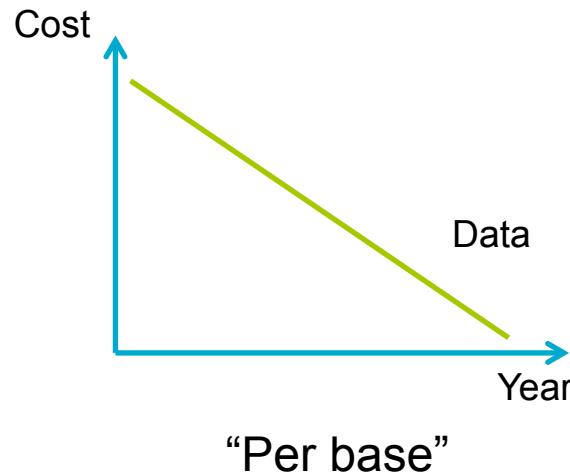
Data growth

Growth of DNA Sequencing



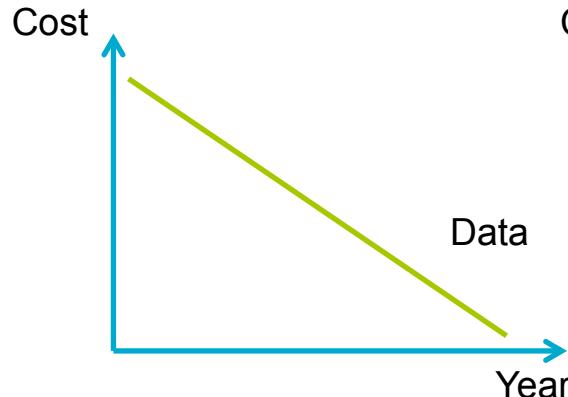
Stephens ZD, Lee SY, Faghri F, Campbell RH, Zhai C, et al. (2015) Big Data: Astronomical or Genomical?. PLoS Biol 13(7): e1002195. doi:10.1371/journal.pbio.1002195

<http://127.0.0.1:8081/plosbiology/article?id=info:doi/10.1371/journal.pbio.1002195>

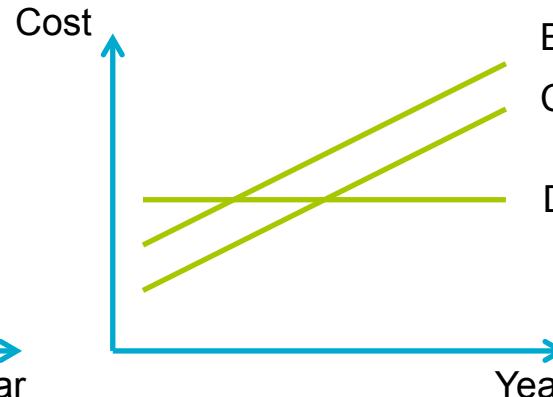


Our role

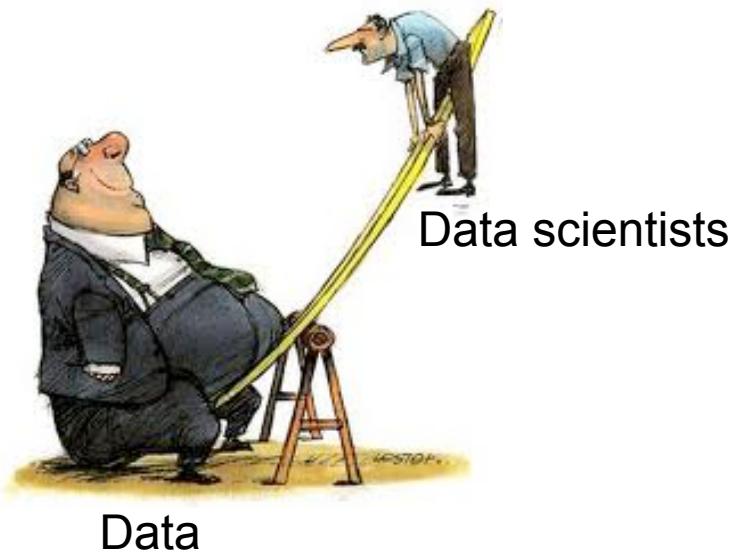
We want to help the Swedish Life Science community to **build knowledge** in large-scale data analysis, and to make bioinformatics **easily accessible** for all.



“Per base”



“Per project”

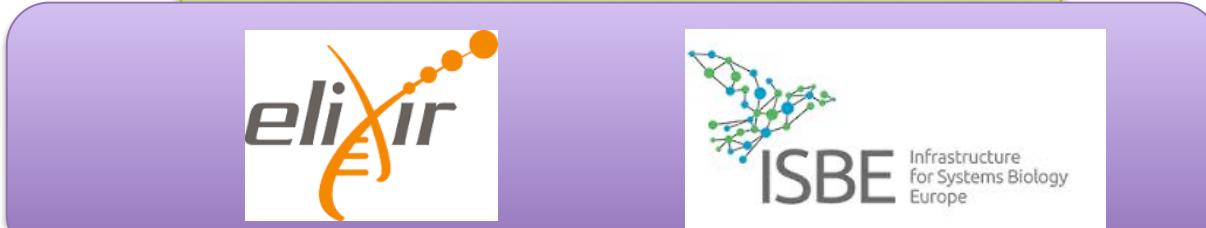


Our role

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.

NBIS activities

Support, tools and training



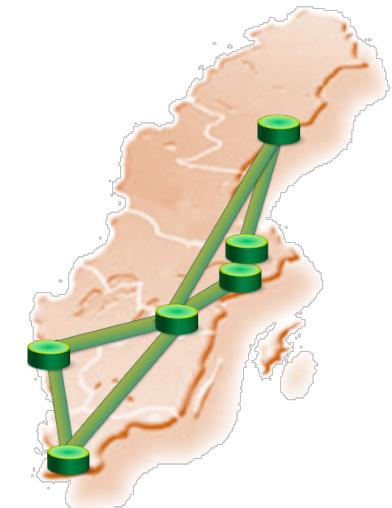


Custom-tailored support

www.nbis.se

www.scilifelab.se/platforms/bioinformatics/

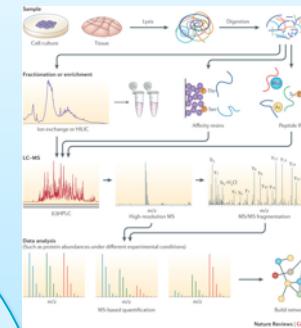
- Study design consultation (free)
www.nbis.se/support/supportform/index.php
+ drop-in sessions every week @ all 6 sites
- Support (User fee 800 kr/h)
www.nbis.se/support/supportform/index.php
- Long-term support and systems biology
(500h, free, scientific evaluation)
www.nbis.se/support/supportform/index.php?form=longterm



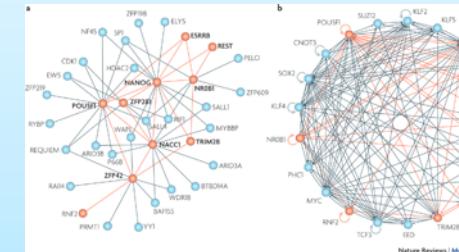
Bioinformatics support



Genomics



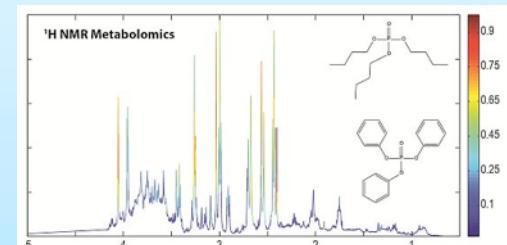
Proteomics



Systems biology



Biostatistics

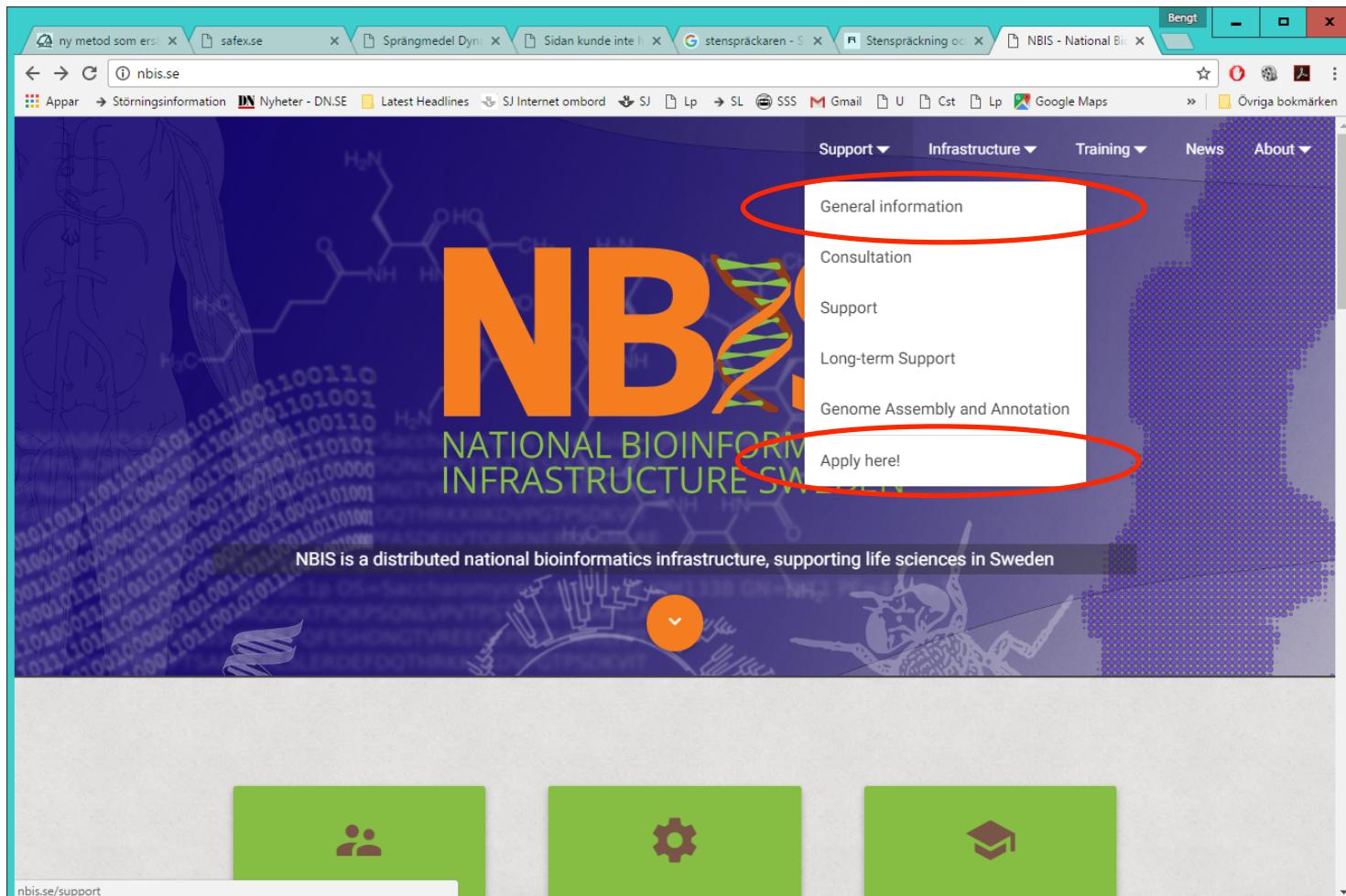


Metabolomics

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 the NBIS website homepage. A red circle highlights the "Support" dropdown menu, which is open to show options: General information, Consultation, Support, Long-term Support, and Genome Assembly and Annotation. Another red circle highlights the "Apply here!" button located at the bottom of the same menu.

Support ▾ Infrastructure ▾ Training ▾ News About ▾

General information

Consultation

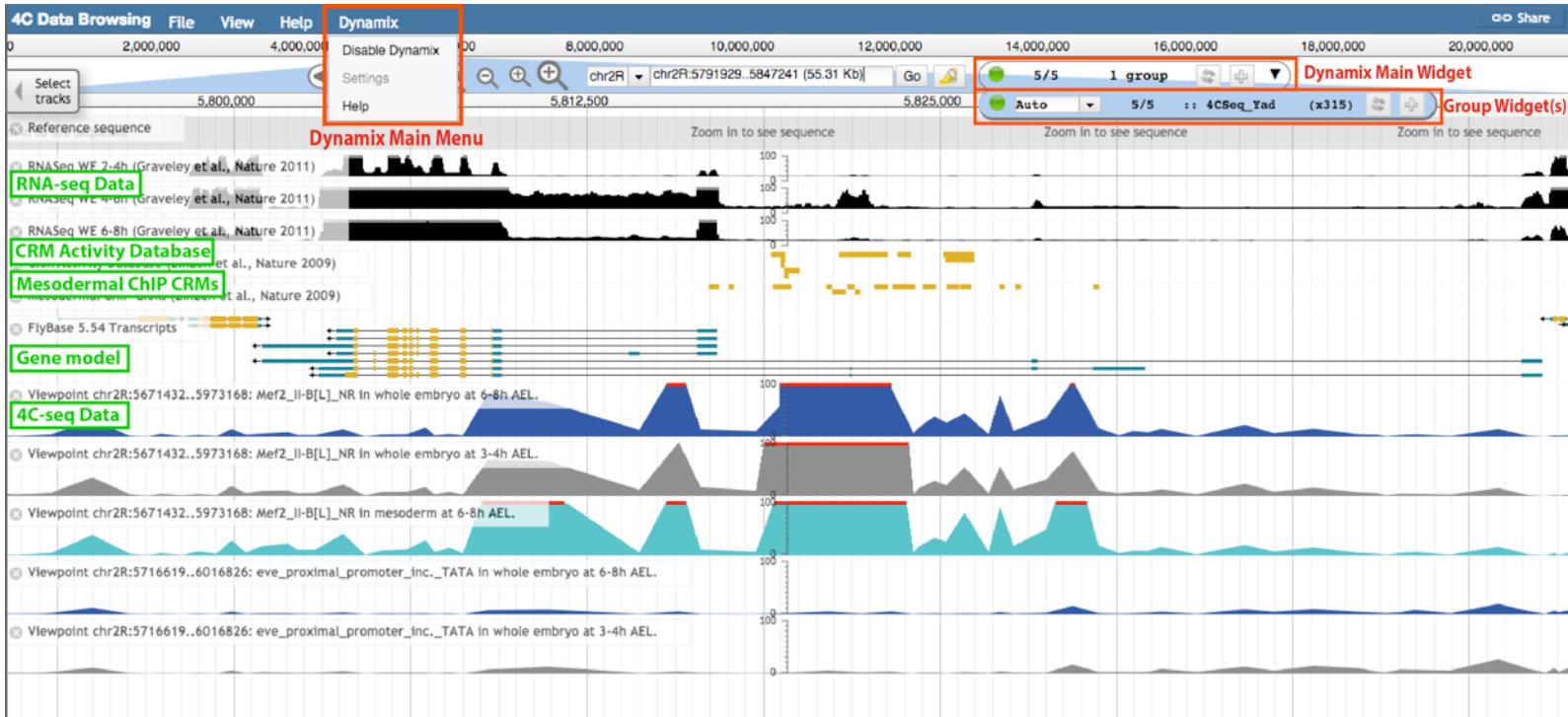
Support

Long-term Support

Genome Assembly and Annotation

Apply here!

Genome assembly and annotation



- 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

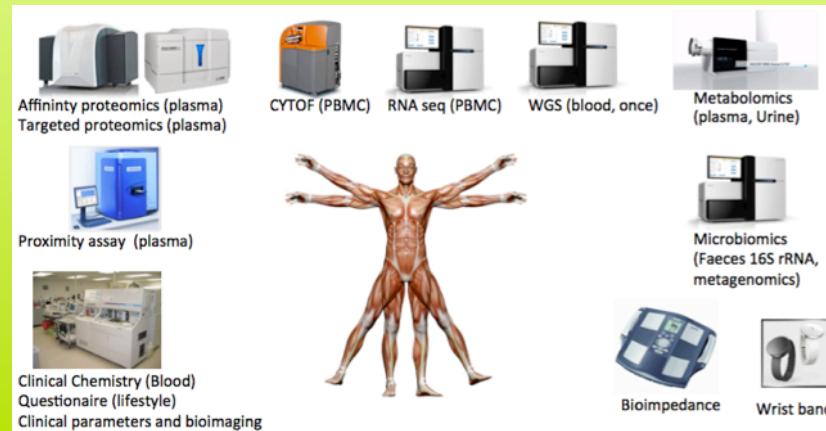
Combine data from SciLifeLab platforms

- 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





Tools and infrastructure

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

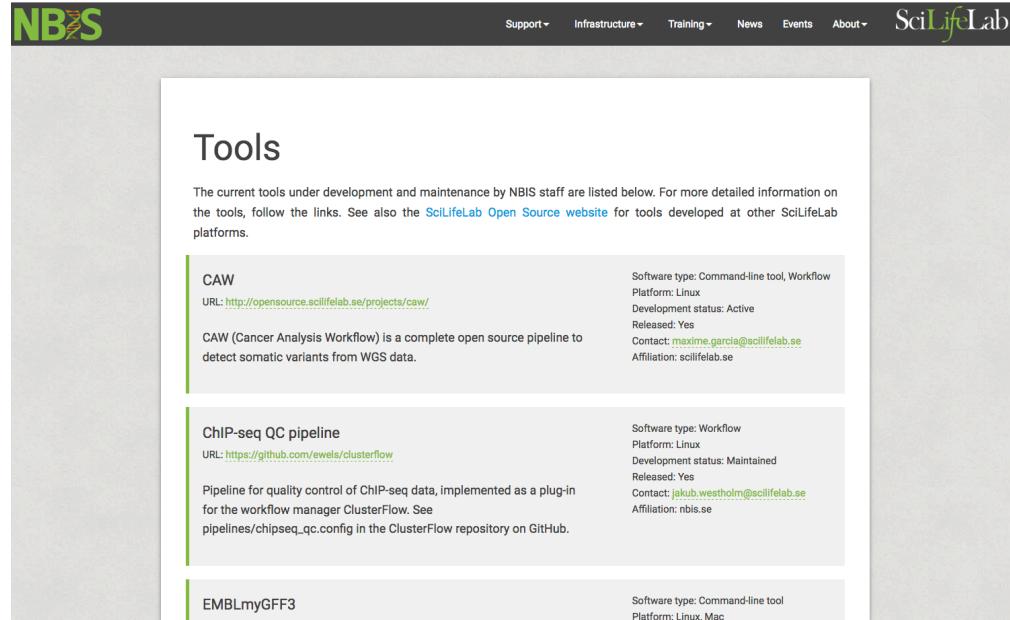
- MrBayes
- Structure prediction web services

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



The screenshot shows the NBIS Tools page. At the top, there is a navigation bar with links for Support, Infrastructure, Training, News, Events, About, and the SciLifeLab logo. Below the navigation bar, the page title is "Tools". A sub-header states: "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." Three tool entries are listed:

- CAW**
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**
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: 1000 Swedish genomes

SciLifeLab

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 ↗
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 hide labels

46,615,750 46,615,800 46,615,850 46,615,900 46,615,950 46,616,000

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/

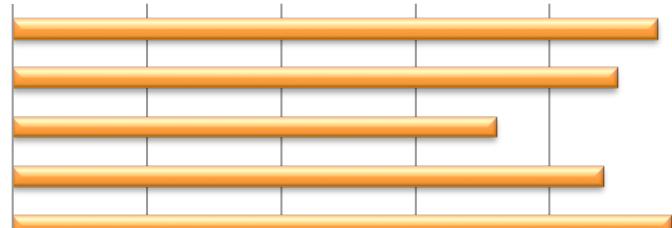
Last call (2017/2018): 111 applicants for 15 places

Next call opens Nov 2017!

The Swedish Bioinformatics Advisory Program

Student evaluation, June 2015

Overall rating of the Advisory Program



Impact on the efficacy of your research

Impact on the scientific value of your

Impact on the technical level of your

In favour of SciLifeLab continuing this



Also of interest to you

- 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

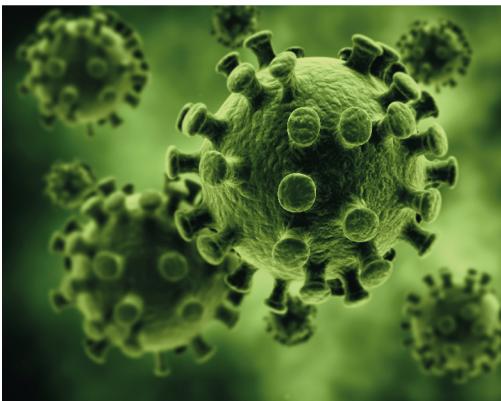
Science & SciLifeLab Prize

Science & SciLifeLab
PRIIZE
FOR YOUNG SCIENTISTS

Join the Symposium, Dec 11, 2017!

<https://scienceprize.ipages.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

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INFRASTRUCTURE SWEDEN