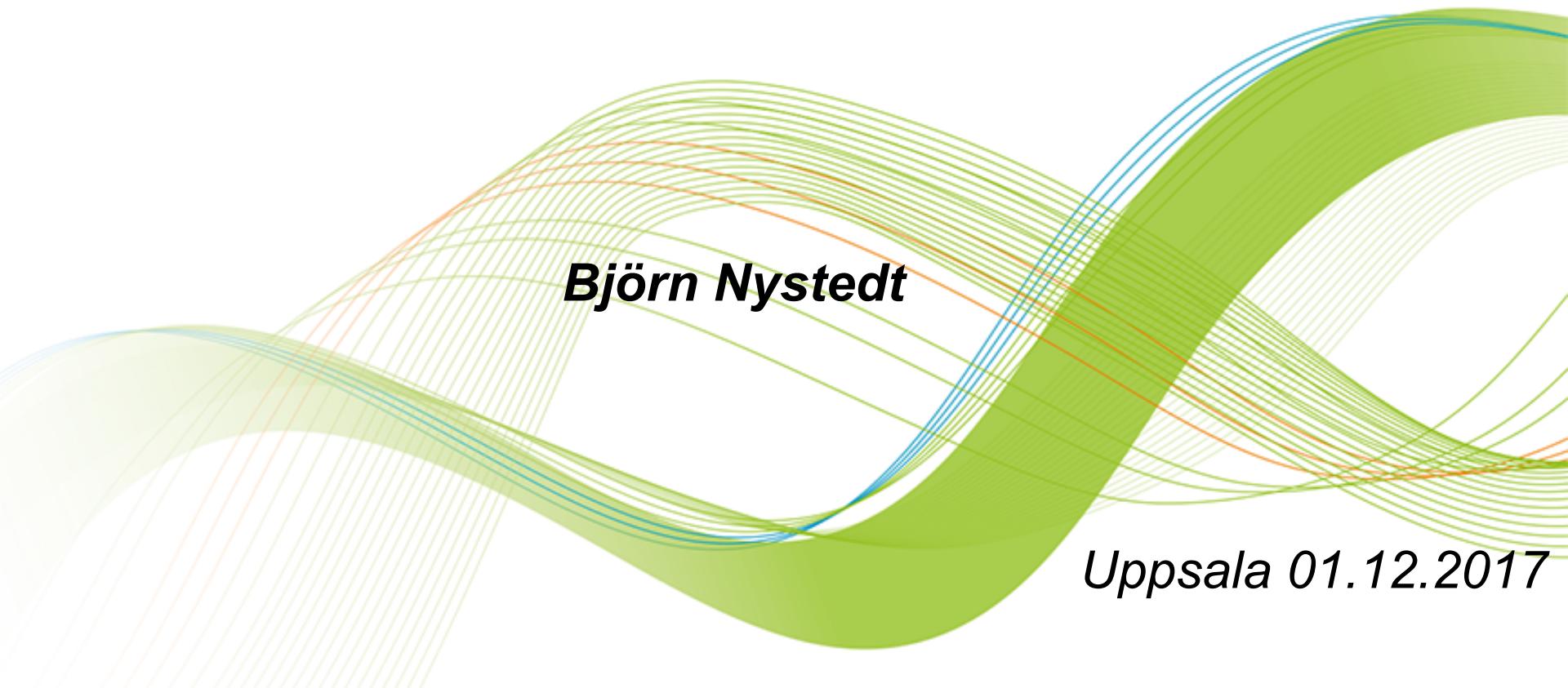

SciLifeLab Bioinformatics Platform

National Bioinformatics Infrastructure Sweden (NBIS)



Björn Nystedt

Uppsala 01.12.2017

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



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.”

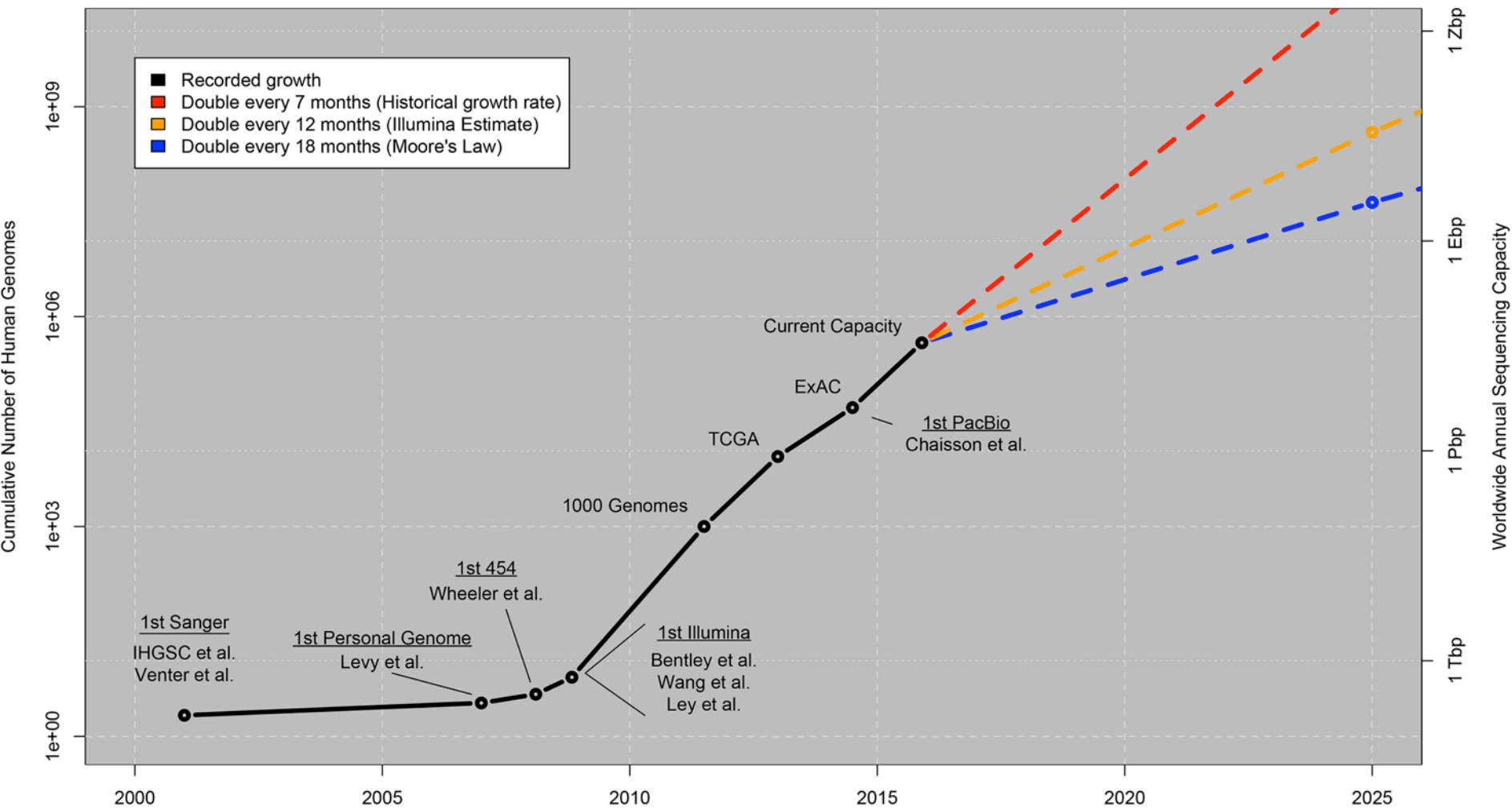


Scientific impact: 7/7 (“Outstanding”)

Overall score: 7/7 (“Outstanding”)

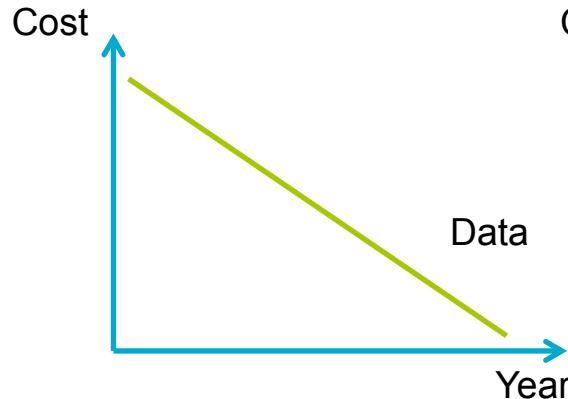
Data growth

Growth of DNA Sequencing

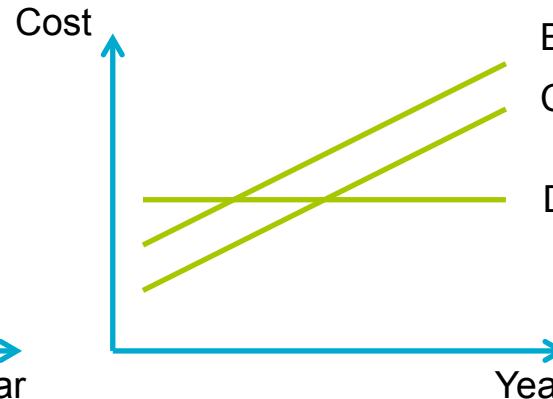


Stephens ZD, Lee SY, Faghri F, Campbell RH, Zhai C, et al. (2015) Big Data: Astronomical or Genomical?. PLoS Biol 13(7): e1002195. doi:10.1371/journal.pbio.1002195

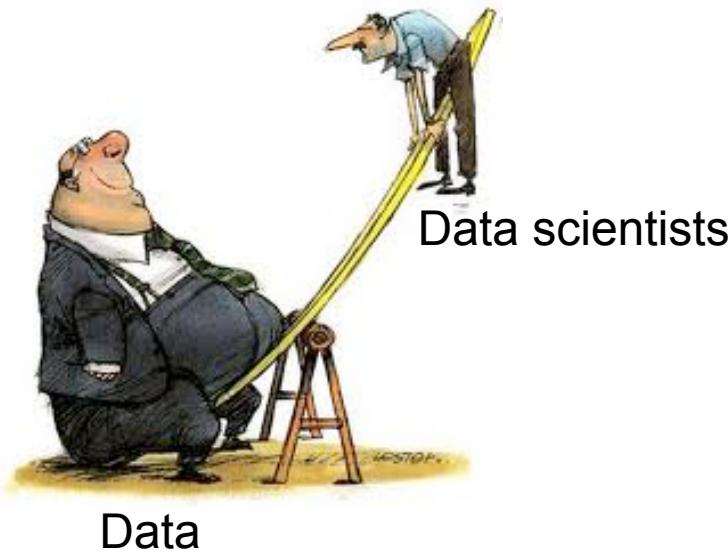
<http://127.0.0.1:8081/plosbiology/article?id=info:doi/10.1371/journal.pbio.1002195>



“Per base”



“Per project”

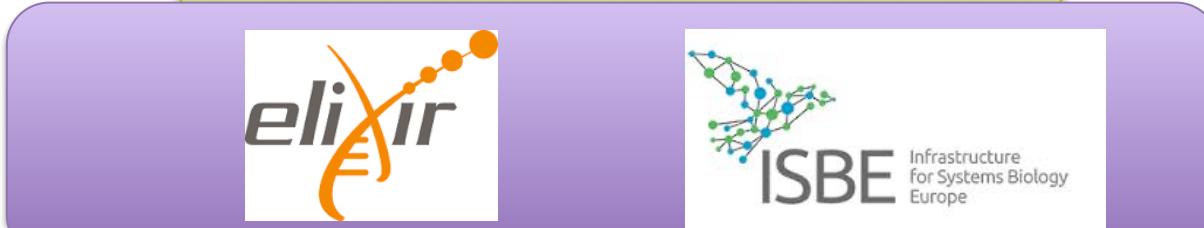


Our role

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



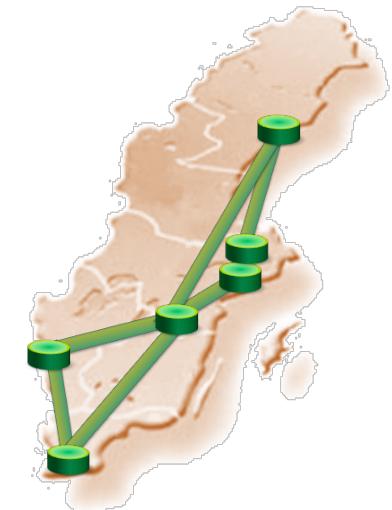


Custom-tailored support

www.nbis.se

www.scilifelab.se/platforms/bioinformatics/

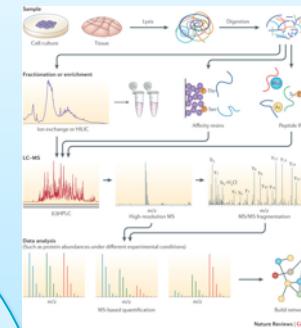
- Study design consultation (free)
www.nbis.se/support/supportform/index.php
+ drop-in sessions every week @ all 6 sites
- Support (User fee 800 kr/h)
www.nbis.se/support/supportform/index.php
- Long-term support and systems biology
(500h, free, scientific evaluation)
www.nbis.se/support/supportform/index.php?form=longterm



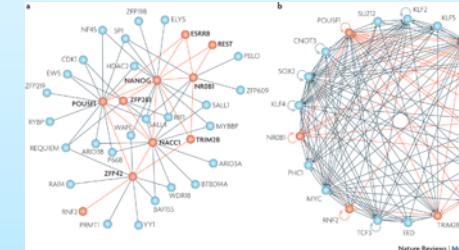
Bioinformatics support



Genomics



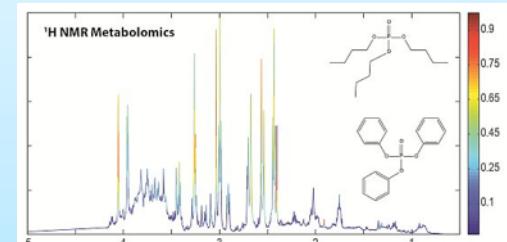
Proteomics



Systems biology



Biostatistics

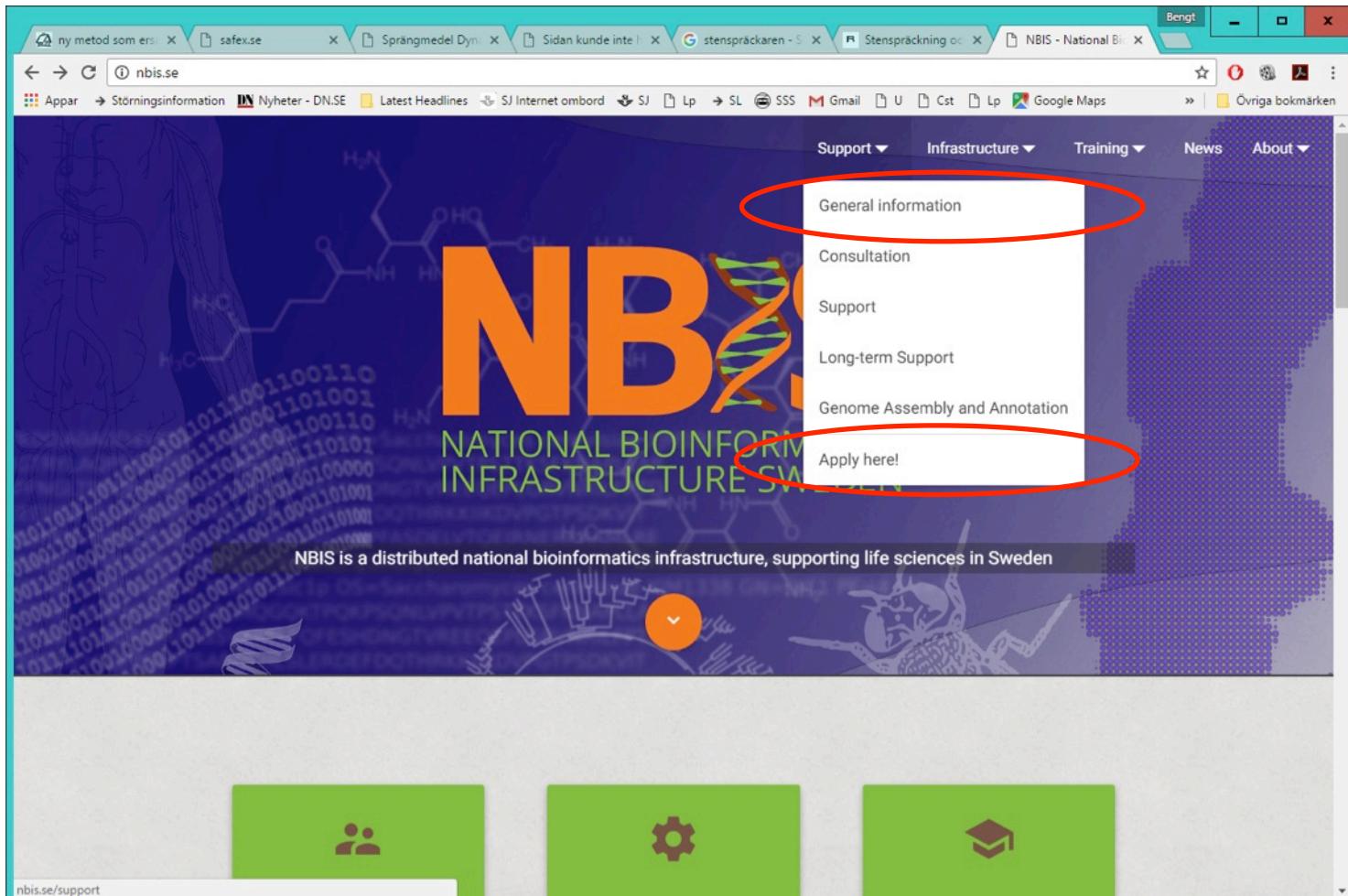


Metabolomics

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



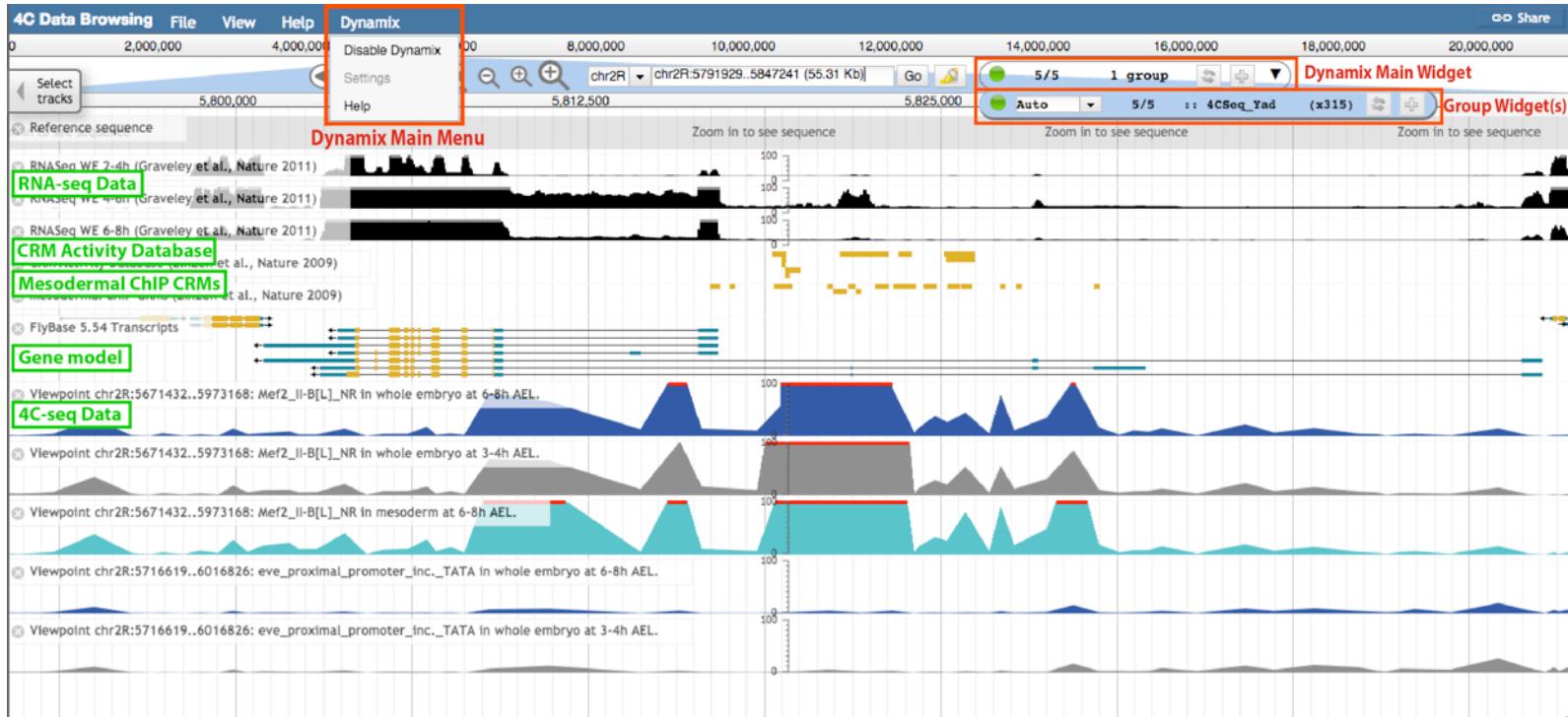
The screenshot shows the NBIS website homepage. A red circle highlights the "Support" dropdown menu, which is open to reveal several options: "General information", "Consultation", "Support", "Long-term Support", and "Genome Assembly and Annotation". Below these options, another red circle highlights a white button labeled "Apply here!". The background of the page features a blue banner with the NBIS logo and text about being a distributed national bioinformatics infrastructure supporting life sciences in Sweden.

Support ▾

- 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

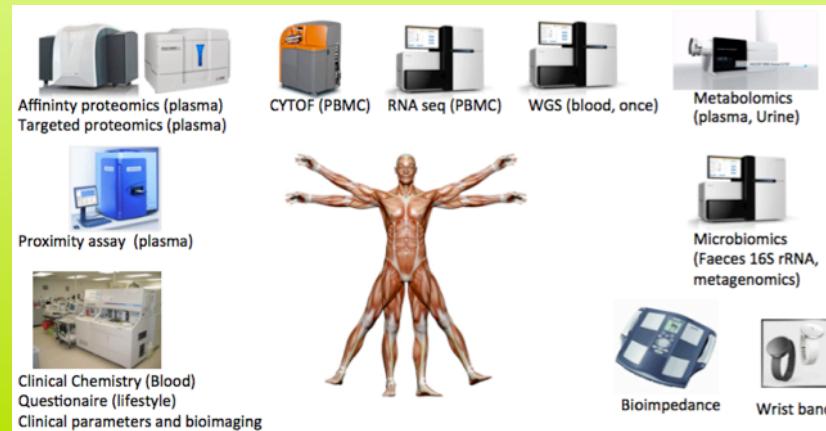
Combine data from SciLifeLab platforms

- 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

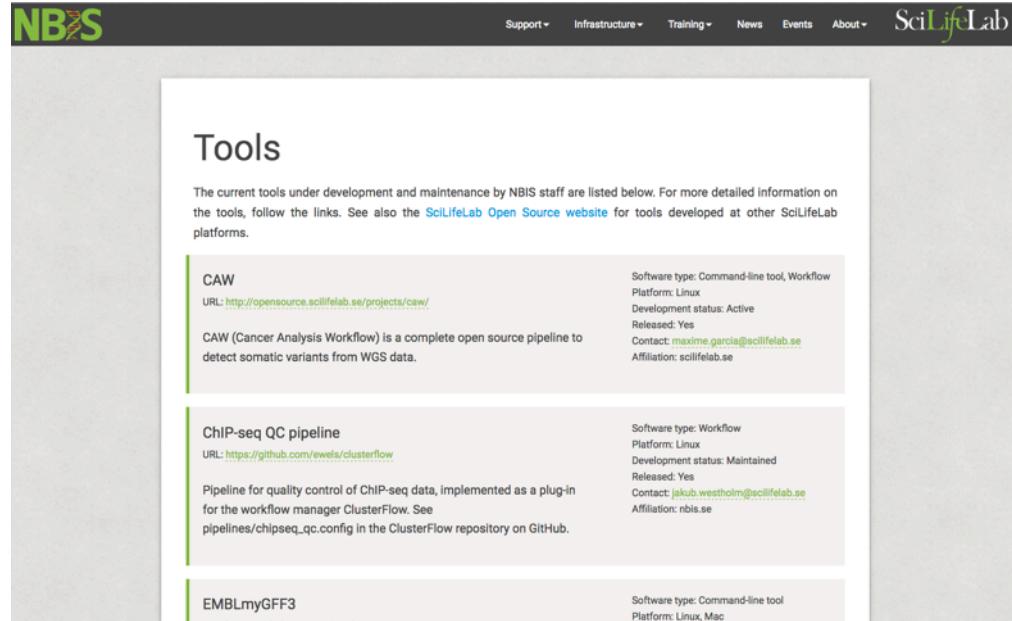
- MrBayes
- Structure prediction web services

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 SciLifeLab. 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/>
<http://opensource.scilifelab.se/>

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/

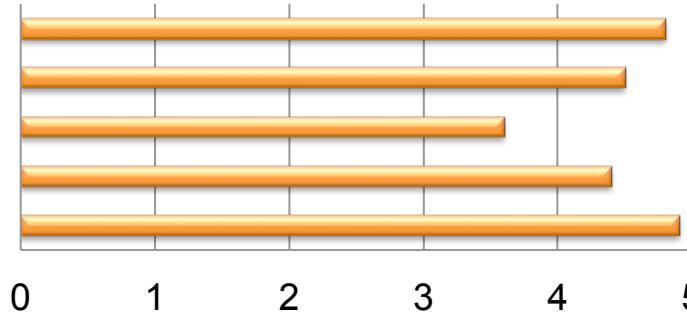
Last call (2017/2018): 111 applicants for 15 places

Deadline Dec 12, 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



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

SciLifeLab



NBiS
NATIONAL BIOINFORMATICS
INFRASTRUCTURE SWEDEN