

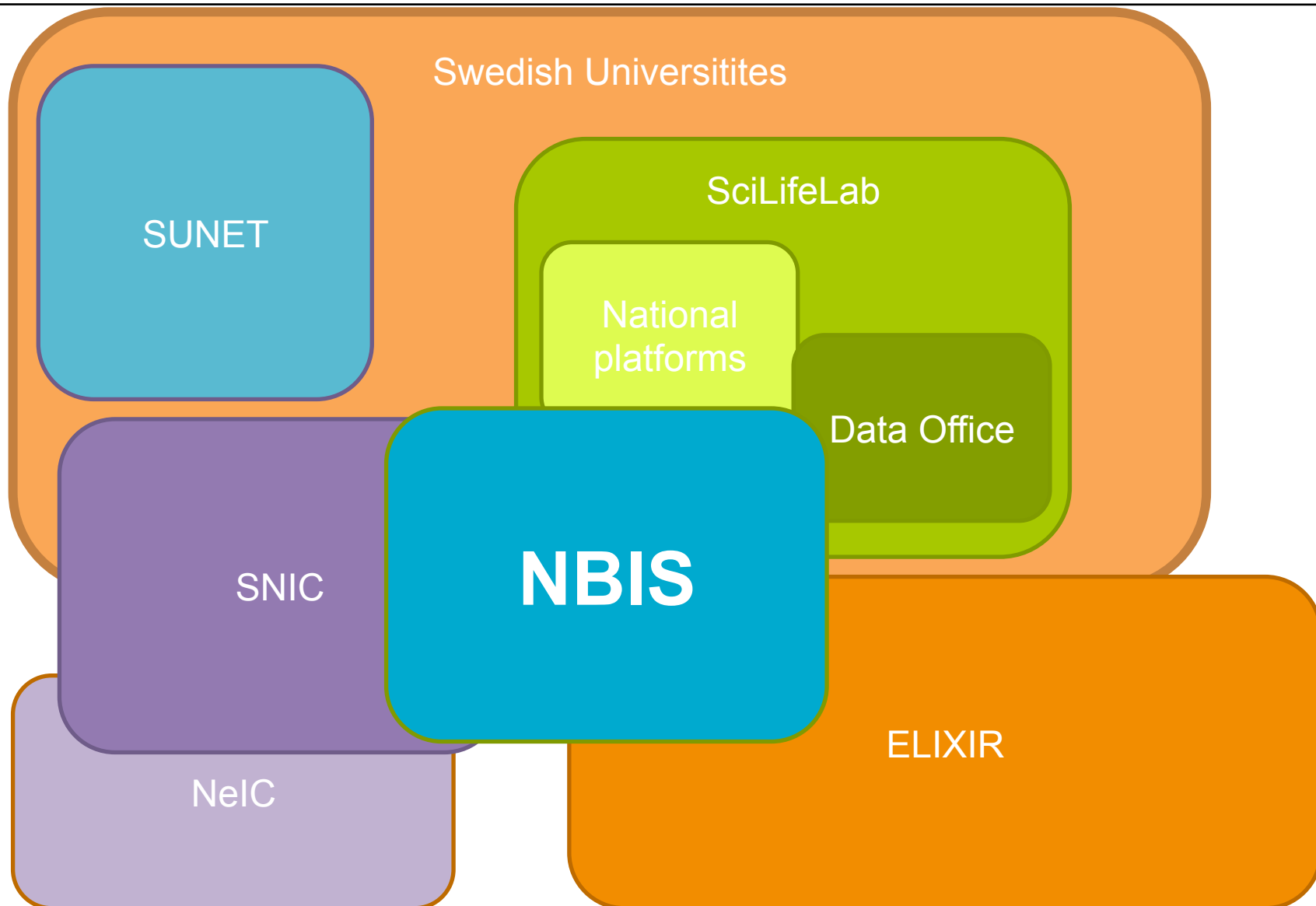
SciLifeLab Bioinformatics Platform

National Bioinformatics Infrastructure Sweden (NBIS)

Niclas Jareborg

NGS course

Lund 2017-10-27

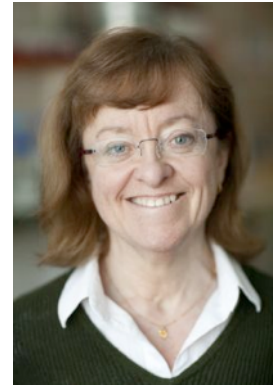


SciLifeLab

National service

The Swiss army knife for Swedish
Life Science researchers

Local scientific
center



Director: Olli Kallioniemi

Co-director: Lena Claesson-Welsh

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

Next
Generation
Diagnostics

Single-cell
omics

National
Bioinformatics
Infrastructure
Sweden

Bengt Persson



Merge of BILS, WABI and
more; complete 2016.
National, distributed

VR

SNIC

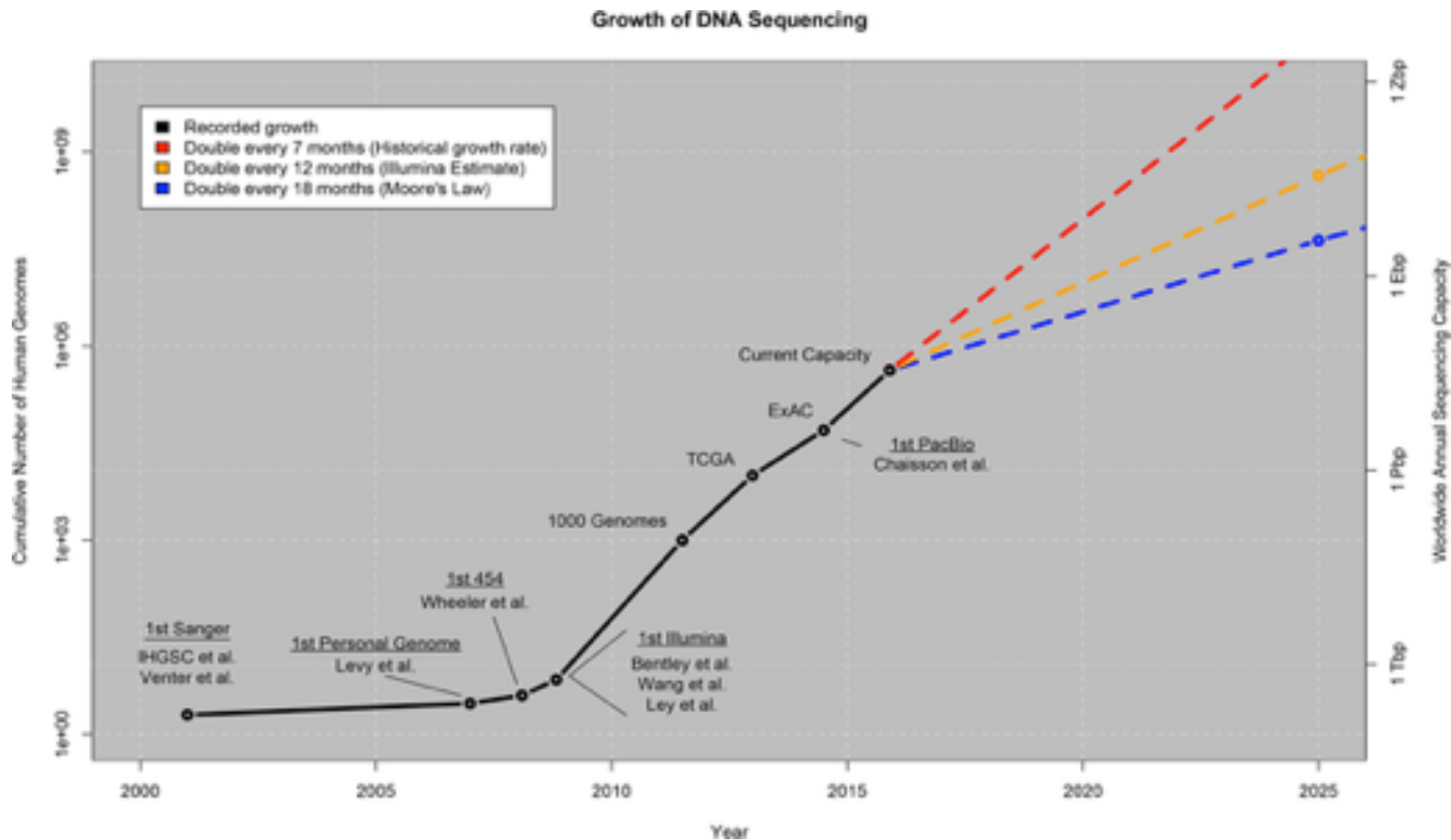


Computer
resources
free for
Swedish
researchers



Bioinformatics as infrastructure

Data growth



NBIS activities

Support, tools and training



Support



Tools



Training



- **Support and Infrastructure**

Wide competence in bioinformatics, Assembly/Annotation, SysDev

- **Long-term support (WABI)**

Large collaborative projects selected by scientific ranking

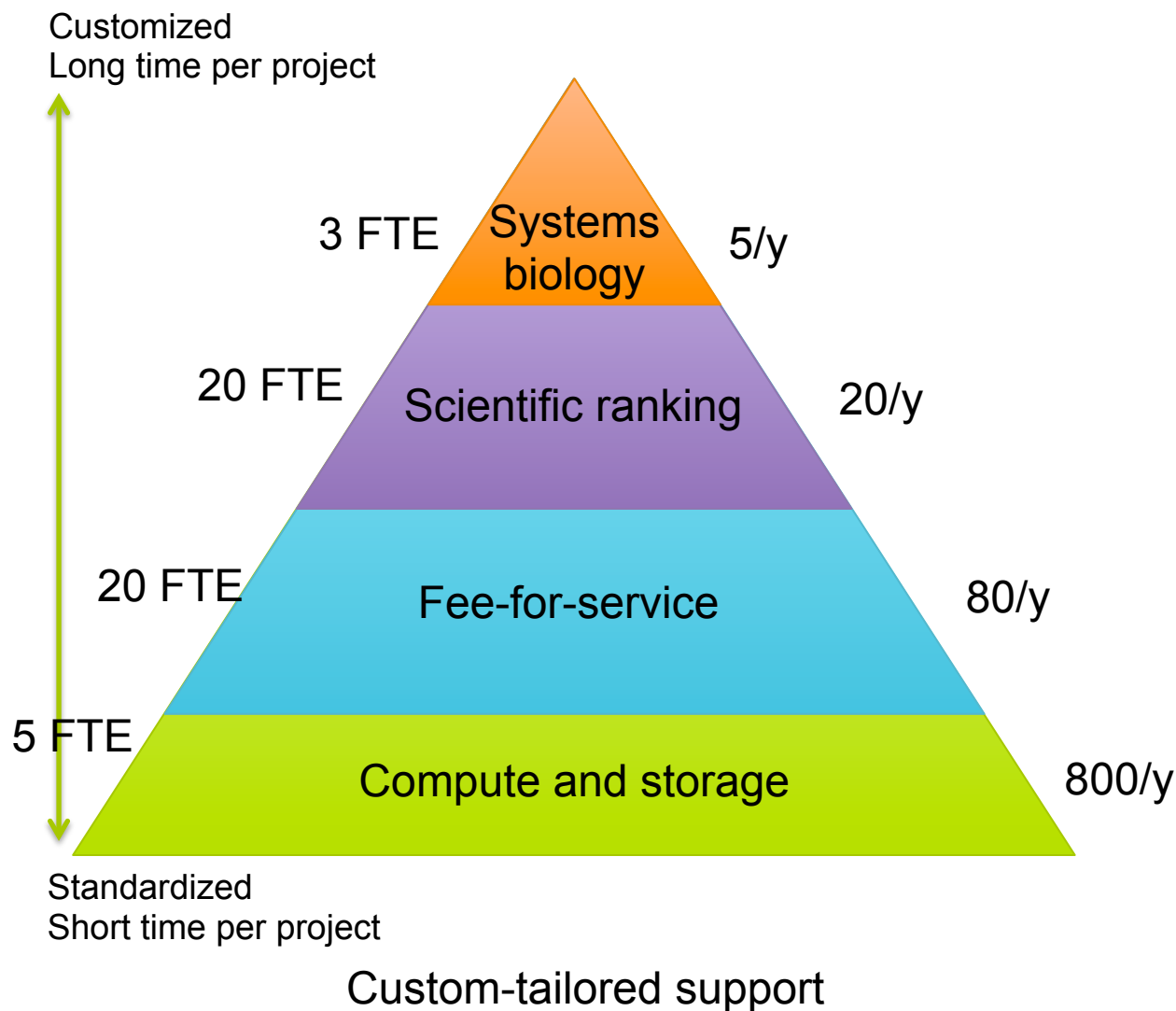
- **Systems biology**

Network analyses and Integrative bioinformatics

- **Compute and storage**

Computational and storage resources for bioinformatics, especially next-generation sequencing





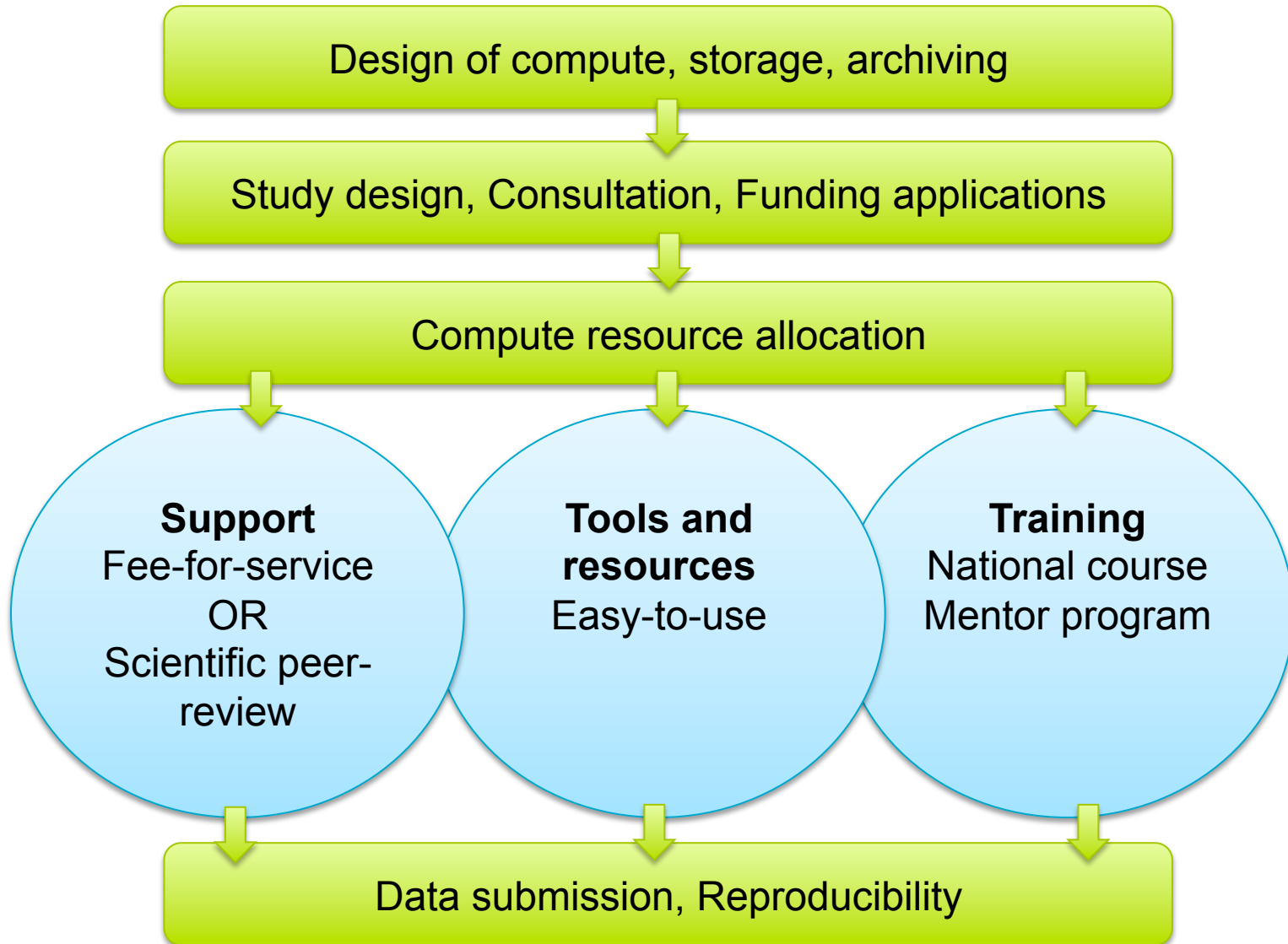
Training



5 FTE
Systems development



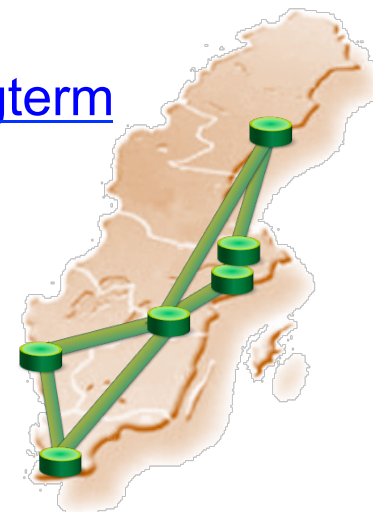
1 FTE
Data management





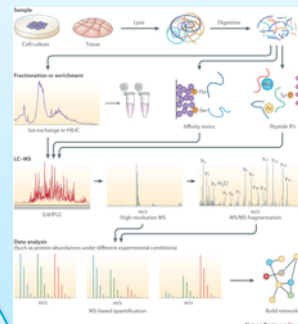
- Study design consultation (free)
www.nbis.se/support/supportform/index.php
+ drop-in sessions every week @ all 6 sites
- Short- and Medium-term support (User fee 800 SEK/h)
www.nbis.se/support/supportform/index.php
- Long-term support and systems biology
(500h, free, scientific evaluation)
www.nbis.se/support/supportform/index.php?form=longterm

www.scilifelab.se/platforms/bioinformatics/
www.nbis.se

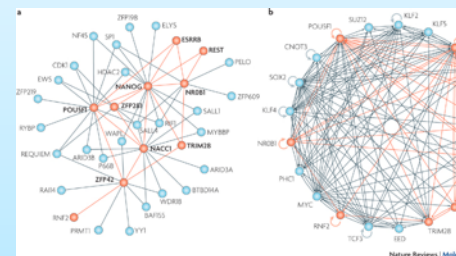




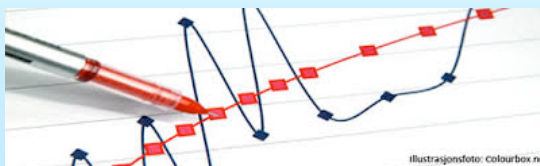
Genomics



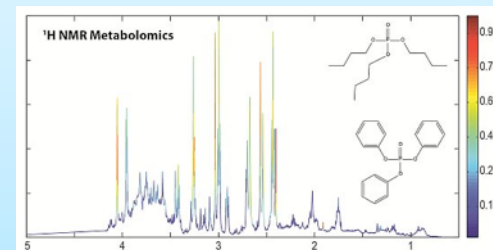
Proteomics



Systems biology



Biostatistics

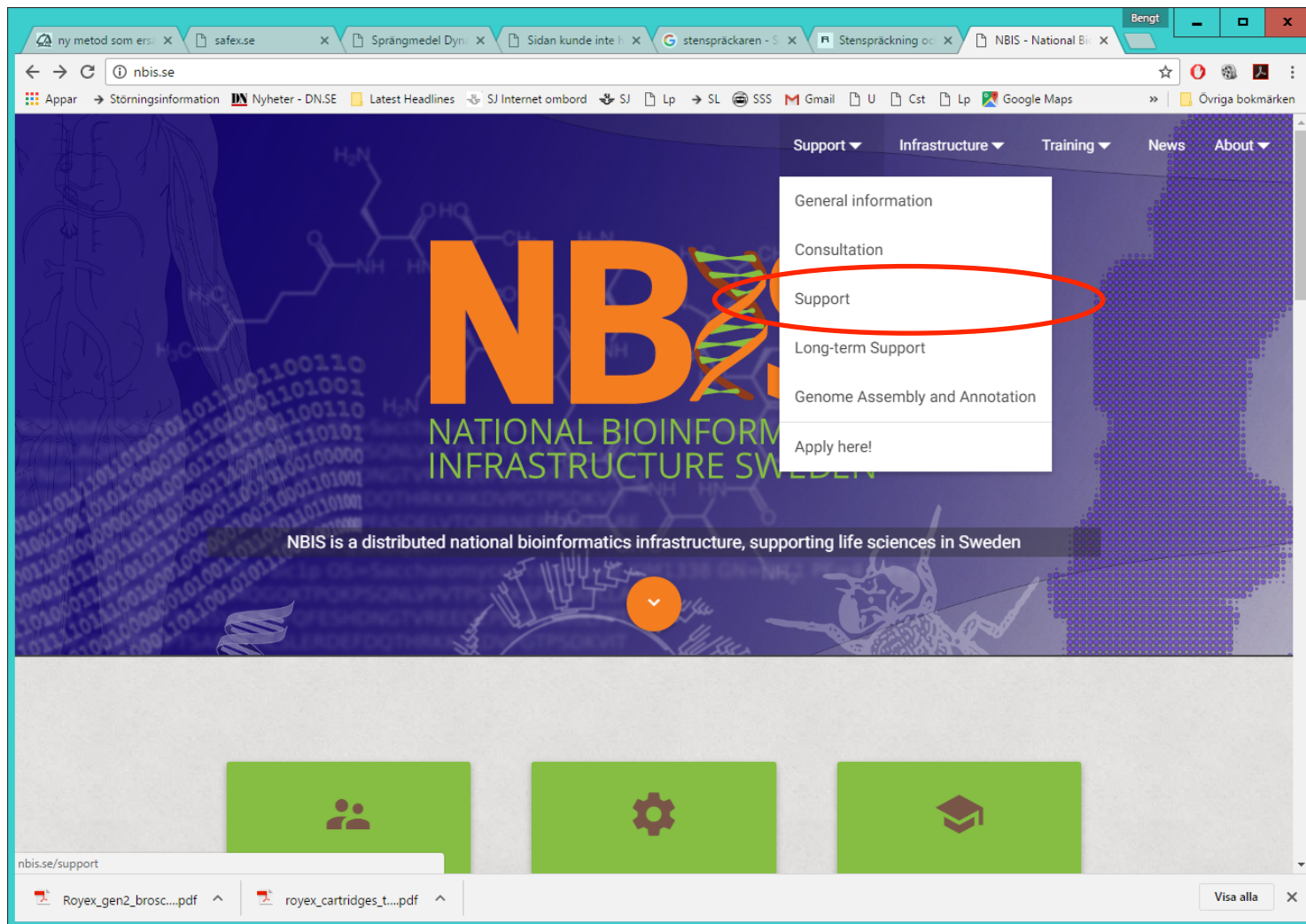


Metabolomics

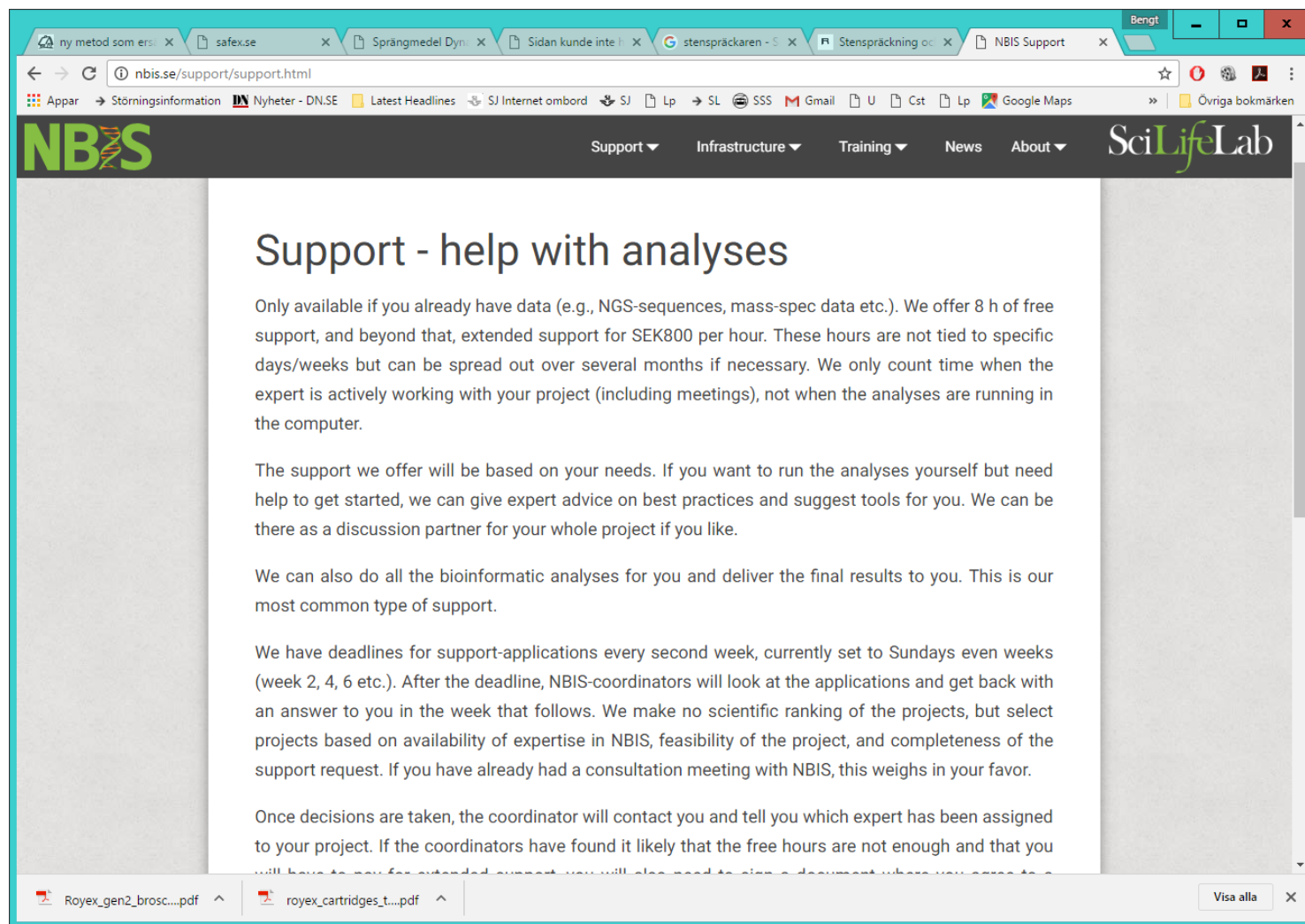
2 tracks!

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

How to get support nbis.se



The screenshot shows a web browser window with the nbis.se website. The browser's address bar displays 'nbis.se'. The website's header features a navigation menu with links for 'Support', 'Infrastructure', 'Training', 'News', and 'About'. The 'Support' dropdown menu is open, showing options: 'General information', 'Consultation', 'Support' (highlighted with a red circle), 'Long-term Support', 'Genome Assembly and Annotation', and 'Apply here!'. The main content area has a large 'NBIS' logo and the text 'NATIONAL BIOINFORMATICS INFRASTRUCTURE SWEDEN'. Below this, a banner states 'NBIS is a distributed national bioinformatics infrastructure, supporting life sciences in Sweden'. At the bottom, there are three green buttons with icons representing people, a gear, and a graduation cap. The browser's taskbar at the bottom shows two open PDF files: 'Royex_gen2_brosc....pdf' and 'royex_cartridges_t....pdf'.



ny metod som ers... X safex.se X Sprängmedel Dyn... X Sidan kunde inte... X stenspräckaren -... X Stenspräckning oc... X NBIS Support X Bengt

nbis.se/support/support.html

Appar → Störningsinformation → Nyheter - DN.SE → Latest Headlines → SJ Internet ombord → SJ → Lp → SL → SSS → Gmail → U → Cst → Lp → Google Maps → Övriga bokmärken

NBIS Support ▾ Infrastructure ▾ Training ▾ News About ▾ SciLifeLab

Support - help with analyses

Only available if you already have data (e.g., NGS-sequences, mass-spec data etc.). We offer 8 h of free support, and beyond that, extended support for SEK800 per hour. These hours are not tied to specific days/weeks but can be spread out over several months if necessary. We only count time when the expert is actively working with your project (including meetings), not when the analyses are running in the computer.

The support we offer will be based on your needs. If you want to run the analyses yourself but need help to get started, we can give expert advice on best practices and suggest tools for you. We can be there as a discussion partner for your whole project if you like.

We can also do all the bioinformatic analyses for you and deliver the final results to you. This is our most common type of support.

We have deadlines for support-applications every second week, currently set to Sundays even weeks (week 2, 4, 6 etc.). After the deadline, NBIS-coordinators will look at the applications and get back with an answer to you in the week that follows. We make no scientific ranking of the projects, but select projects based on availability of expertise in NBIS, feasibility of the project, and completeness of the support request. If you have already had a consultation meeting with NBIS, this weighs in your favor.

Once decisions are taken, the coordinator will contact you and tell you which expert has been assigned to your project. If the coordinators have found it likely that the free hours are not enough and that you will have to pay for extended support, you will also need to sign a document where you agree to...

Royex_gen2_brosc...pdf ^ royex_cartridges_t...pdf ^ Visa alla X

ny metod som ers... X safex.se X Sprängmedel Dyn... X Sidan kunde inte... X stenspräckaren - S... X Stenspräckning oc... X NBIS support form X Bengt

nbis.se/support/supportform/index.php

Appar → Störningsinformation Nyheter - DN.SE Latest Headlines SJ Internet ombord SJ Lp → SL SSS Gmail U Cst Lp Google Maps Övriga bokmärken

NBIS Support ▾ Infrastructure ▾ Training ▾ News About ▾ SciLifeLab

CONSULTATION & SUPPORT **LONG-TERM SUPPORT**

NBIS offers free support up to 8h/PI/year for support. After the free support hours, the NBIS expert can be hired for extended support for a subsidized fee, currently SEK800 per hour.

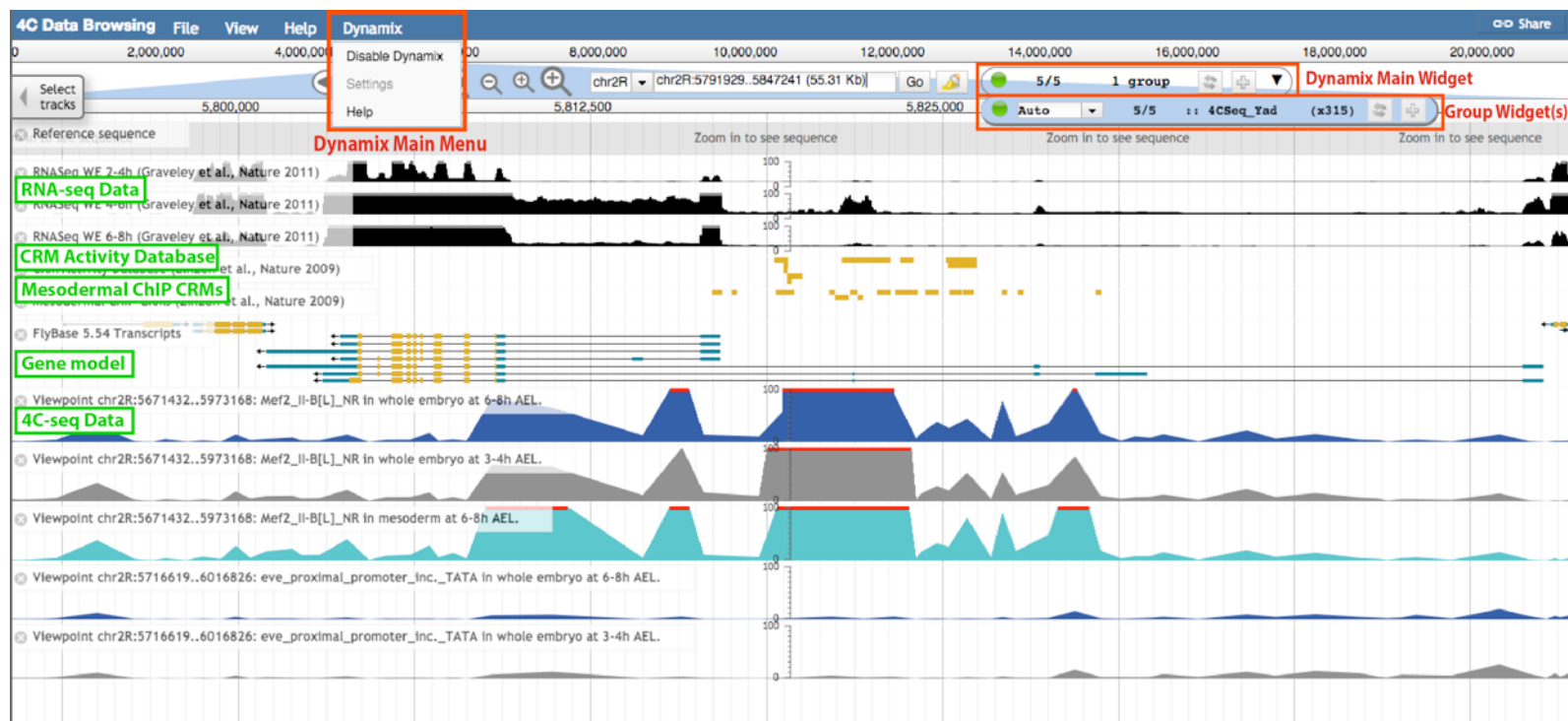
[Click here for further information about NBIS support services.](#)

Support requests are reviewed every second week, please allow for up to three weeks before you hear back from us.

NBIS Support Request Form

Name	Your name
e-mail	Your e-mail address
PI	Name of Principal Investigator
PI e-mail	PI e-mail address
Research organization	Chalmers ▾

Royex_gen2_brosc....pdf ^ royex_cartridges_t....pdf ^ Visa alla X



- 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

Henrik Lantz



Cost effective with high quality!

4 FTE, joint effort by Long-term Support and Systems Biology

Projects apply in the regular Long-term Support calls

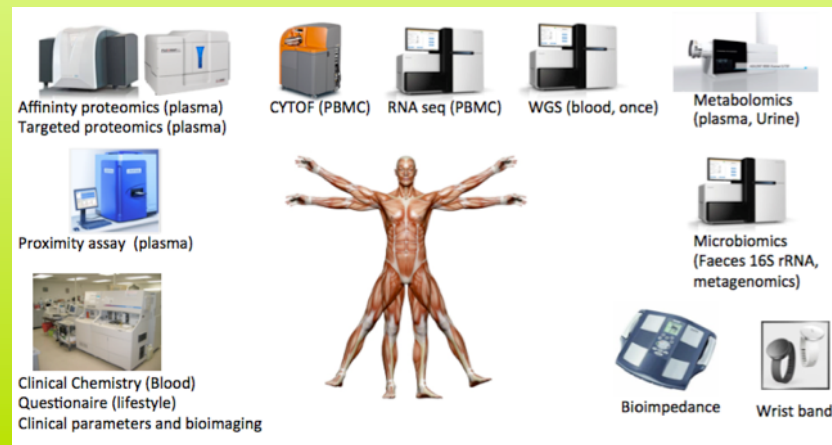
Combine data from SciLifeLab platforms

- 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
- Mosler
- 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 SYSDEV BETS BOARD http://bit.ly/nbis_bets_board

NBIS should enable world-class life science by providing expert knowledge, creative data integration, advanced training, efficient data publication and data and analysis methods.

NOW						NEXT						
We have done the experimenting, we have learned, and we're ready to build it for the world. It is fully funded, plans are clear and we can predict with reasonable confidence what impact the Bet may have.						We're experimenting with different ways to create value with an opportunity we've identified so we can build the right thing, the right way, at the right time.						We believe that we're investigating
Rank	Bet/Project	Ext	PO	PL	PM	Rank	Bet/Project	Ext	PO	PL	PM	
1	Local EGA	x	NJ?	?	6?	1	Tools and guides collection		PE	?	<1	Impute GW
2	SweFreq	x	Adam A	NJ	<1	2						Permutatio
3	ePouta integration pilot	x	Antti P	JH?	1	3						Developme
4	microMosler			NJ?	JH	4						Improve bi
5	WGS structural variation WF			BN	PO	5						Single-cell
6	Falcon on milou			HL	JB	6						Chip-seq p
7	Pouta Blueprints			OS	JH							CWL for N
8	web-servers with EGI cloud vo.NBIS.se			MB	NS							OpenCB
9	Human Metabolic Atlas (HMA)			TS	LH							Local Ense
10	SSC HPC2N region dynamic resource allocation			MB	NS							NBIS Tools
11	Database for SciLife/AZ proj	x	BPn?	JV?	4?							IgDiscover

Ongoing dev projects not resourced by the dev team

	WGS somatic variant calling WF			BN	(SJ)	0
	Haloplex variant calling pipeline			PE	ML	0
	WhatsHap: Genomic phasing tool			PE	MM	0

Open prioritization and background descriptions

Features Business Explore Pricing This organisation Search Sign in or Sign up

NBIS NBIS -- National Bioinformatics Infrastructure Sweden
Sweden https://nbis.se

Repositories People

Pinned repositories

MrBayes

MrBayes is a program for Bayesian inference and model choice across a wide range of phylogenetic and evolutionary models. MrBayes uses Markov chain Monte Carlo (MCMC) methods to estimate the posterior...

11 5

wgs-structural

Whole Genome Sequencing Structural Variation Pipelines

1 3 5

sweFreq

1 3

ENA-submission

Tool for easing the submission of data to the ENA

1 1

LocalEGA

NBIS repository for the Local EGA code

1 1

developmental

Small scripts and one document (README.md) describing guidelines for the development of software within NBIS.

3

Search repositories... Type: All Language: All

LEGA-ingestion

Local EGA ingestion microservice

Python Updated 20 minutes ago

sweFreq

Python 3 1 Updated 2 hours ago

homo-horreur

Python 1 Updated 4 hours ago

Top languages

Python Java Shell Perl HTML

Most used topics

bioinformatics bioinformatics-pipeline

People

4

Public code repositories on GitHub

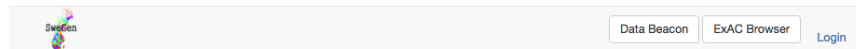
SweFreq – Swedish frequency database



- **Data dissemination – increasing the impact of data generated by SciLifeLab facilities**
- Currently: ***SweGen WGS Reference Variant Frequency Dataset***
 - 950 twin registry + 50 Northern Sweden
 - Deep coverage WGS (30X)
- ***More datasets to come***

swefreq.nbis.se

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



Swedish Frequency Database

The SweGen dataset contains whole-genome sequences for 1000 individuals sampled from the Swedish population. A detailed description of the project can be found here [\[add link to BioRxiv article later\]](#). This server hosts the variant frequency data generated within the SweGen project. This data can be used as a reference for the genetics research community and clinical genetics laboratories. Individual positions in the genome can be viewed using either the ExAC Browser or the Data Beacon by clicking the links above.

To access the variant frequency file you need to register.

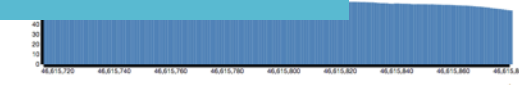


Region: 22 / 46615715 / 46615880

Coverage summary

Coverage metrics: Average: 30.000000, Individuals: 1000, Metrics: mean: 3

Genome browser



Genes

ENSG00000218891

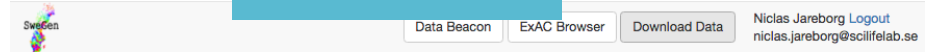
Variants

Alt: Missense + LoF | Include filtered (non-PASS) variants | Invert (highlight new variants)

Support table to CSV

Variant	Chrom	Position	Consequence	Filter	Annotation	Page	AltAlle Count	AltAlle Number	Number of Homozygotes	AltAlle Frequency
22-46615732 A/G (461387874)	22	46615732	p.Arg1702u	PASS	missense	1	1000	0	0.000000	0.000000
22-46615802 A/G (461414302)	22	46615802	p.Lys1090u	PASS	missense	1	1000	0	0.000000	0.000000
22-46615880 T/C (461500234)	22	46615880	p.Val2274u	PASS	missense	7	1000	0	0.000021	0.000021

File download

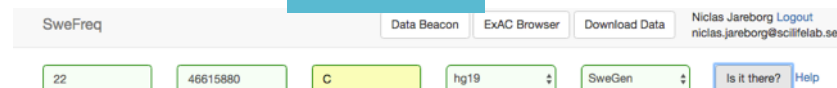


SweFreq Agreement

In case the agreement is short we can write it right here. Otherwise I can make a link to it. Some more text, bla bla Lorem ipsum dolor sit amet, eu ius graeco perfecto, velit delicatissimi an est, rebum persequeris mel no. Stet everti sea ne. Utamur laoreet interpretaris vix et, ex qui solum abhorreant. Vis detracto liberavisse te, sea odio possim instructor te, vidit scripta ex his. I hereby consent to the agreement: ☐

Download

Beacon



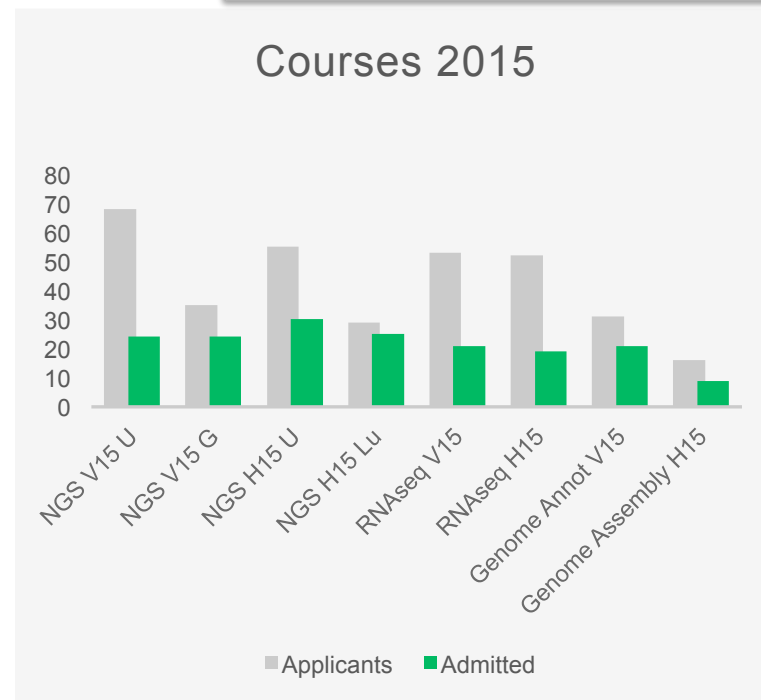
Yes

```
{ "query": { "position": "46541534", "dataset": "SweGen", "reference": "dummy", "chromosome": "22", "allele": "G" }, "beacon": "nbis-beacon", "response": { "observed": 0, "exists": true } }
```



- Bioinformatics Drop-In
 - Weekly at all sites – initial consultations
- 20-odd courses every year
 - Introduction to Bioinformatics using NGS
 - Introduction to Linux
 - Perl programming
 - Introduction to genome annotation
 - Introduction to multivariate analysis
 - RNA-seq
 - Advanced workshop on NGS data analysis
 - Advanced functional genomics
 - Advanced bioinformatics
- Additional local activities
- Bioinformatics Advisory programme
 - Mentorship in bioinformatics

Gender balance:
54% female / 46% male



From spring 2017, we plan to double our training efforts to match the increased demands from the scientific community

www.scilifelab.se/education/courses/
www.nbis.se/training/events.html

The Swedish Bioinformatics Advisory Program

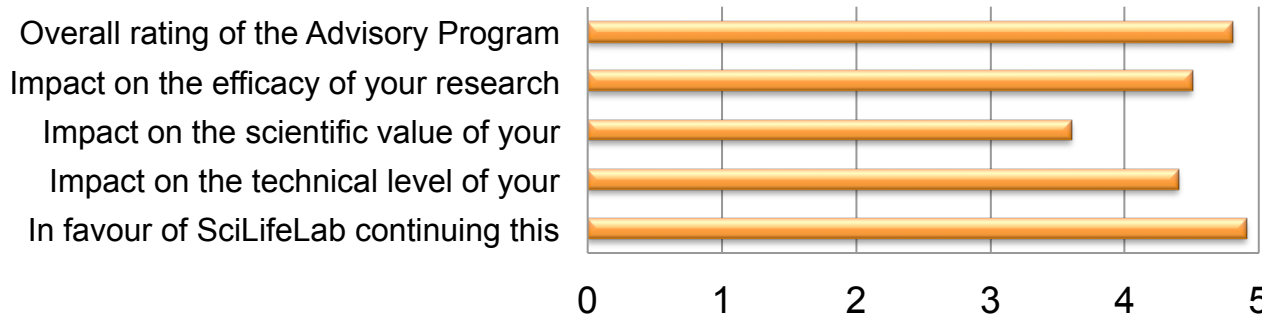
PhD students get a senior bioinformatician as a personal advisor during 2 years of their PhD. Monthly project meetings + two grand meetings per year to aid networking and knowledge transfer.

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

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

The Swedish Bioinformatics Advisory Program

Student evaluation, June 2015



Elixir

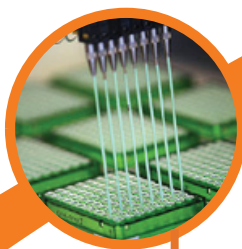
Why ELIXIR?

- Creating a robust infrastructure for biological information is a **bigger** task than any individual organisation or nation can take on alone
- These are issues of such complexity that no single institution or country can tackle alone
- Biology has by far the largest research community:
 - ~3 million life science researchers in Europe
 - >7 million web hits a day at EMBL-EBI alone

*ELIXIR connects national
bioinformatics centres and
EMBL-EBI into a sustainable
European infrastructure for
biological research data*



environment



bioindustries



agriculture



medicine

*ELIXIR underpins
life science research
– across academia
and industry*



NBIS coordinates the Swedish node in ELIXIR – the European infrastructure for biological information

- **Data**

Sustain core data resources

- **Tools**

Services & connectors to drive access and exploitation

- **Compute**

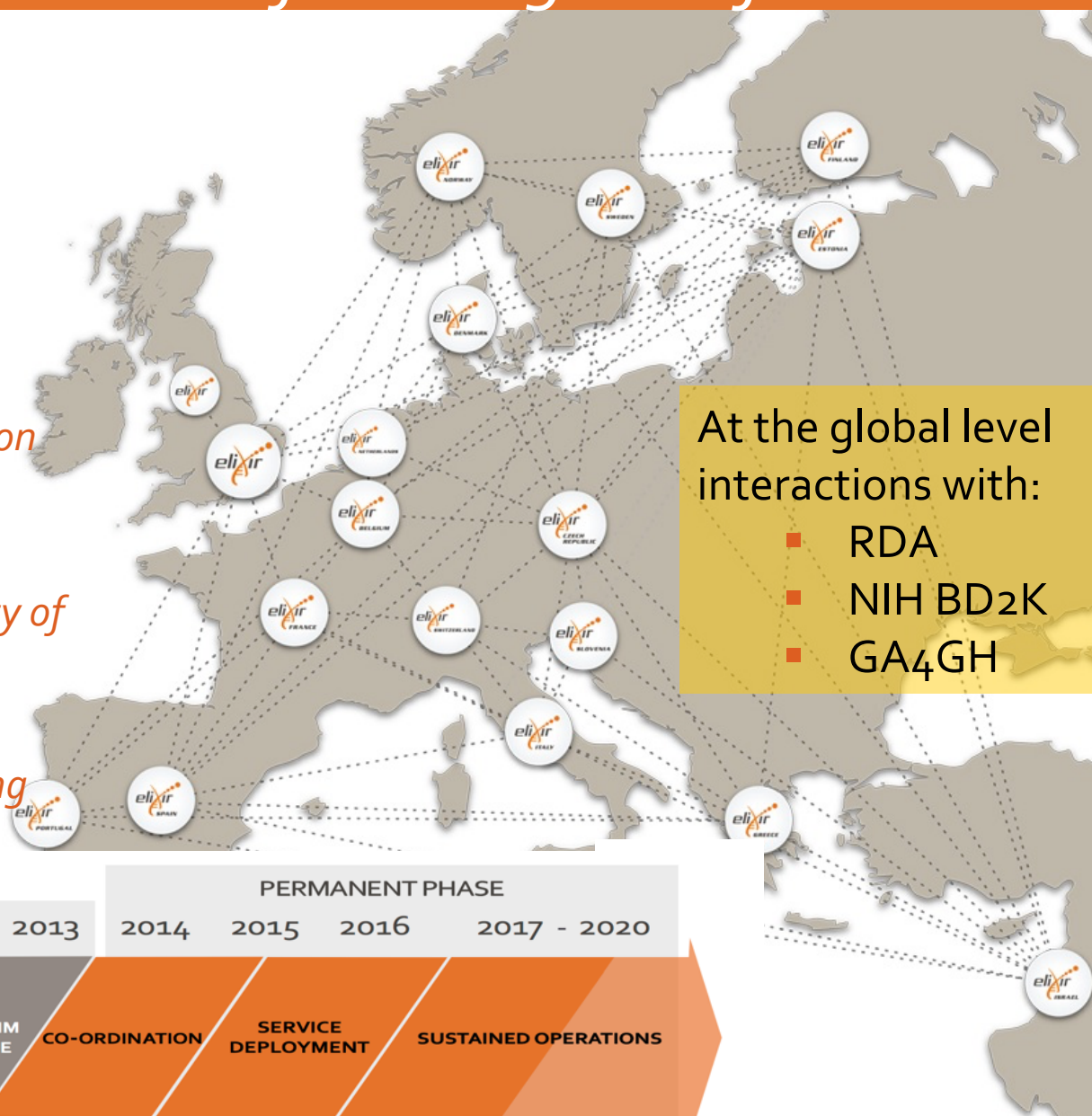
Access, Exchange & Compute on sensitive data

- **Standards**

Integration and interoperability of data and services.

- **Training**

Professional skills for managing and exploiting data



The screenshot shows the nbis.se website in a web browser. The browser's address bar displays 'nbis.se'. The website's header includes a navigation menu with links for Support, Infrastructure, Training, News, and About. The 'Support' menu is expanded, showing options: General information, Consultation, Support, Long-term Support, Genome Assembly and Annotation, and Apply here!. The main content area features a large orange 'NBIS' logo with a DNA double helix, followed by the text 'NATIONAL BIOINFORMATICS INFRASTRUCTURE SWEDEN'. Below this, a banner states: 'NBIS is a distributed national bioinformatics infrastructure, supporting life sciences in Sweden'. At the bottom of the website, there are two green buttons with icons: one with a group of people and another with a gear. The browser's taskbar at the bottom shows two open PDF files: 'Royex_gen2_brosc...pdf' and 'royex_cartridges_t...pdf'.

