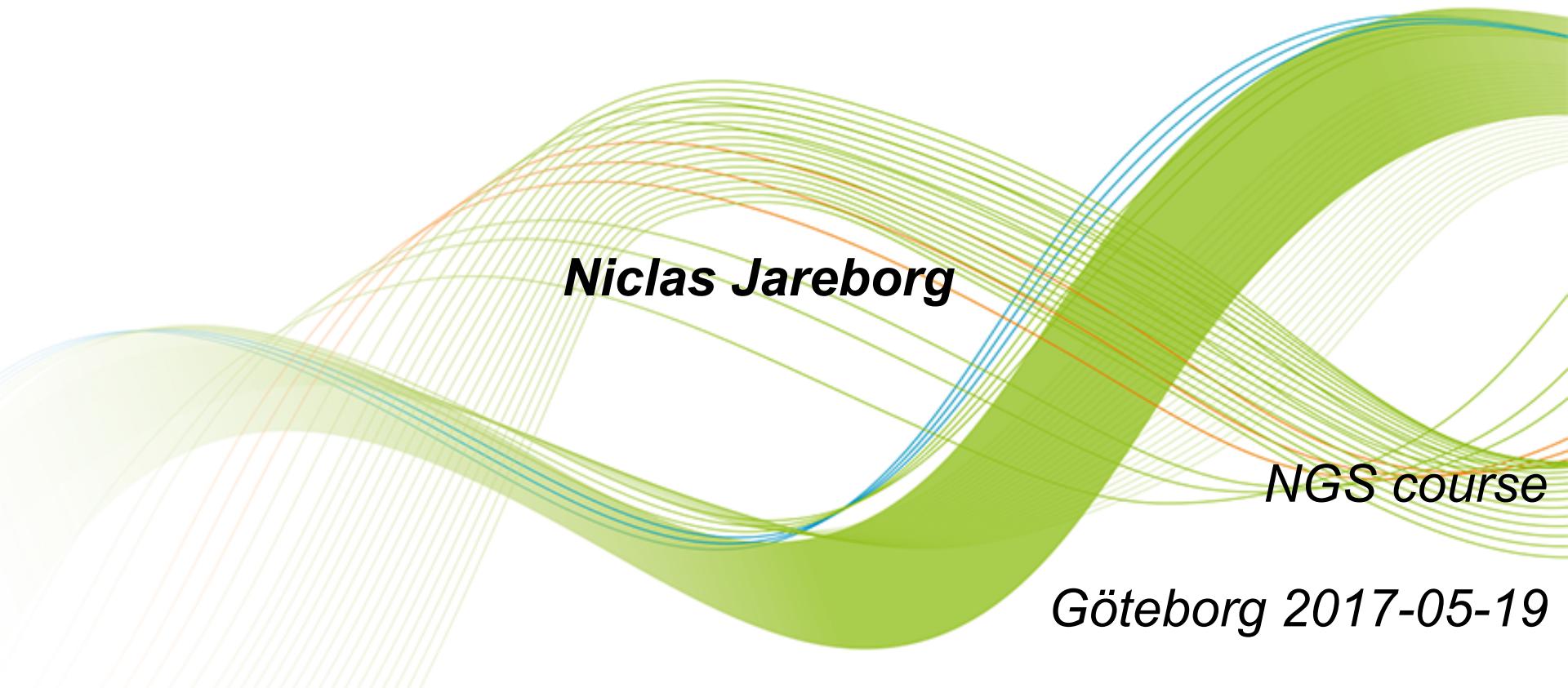


SciLifeLab Bioinformatics Platform

National Bioinformatics Infrastructure Sweden (NBIS)



Niclas Jareborg

NGS course

Göteborg 2017-05-19

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

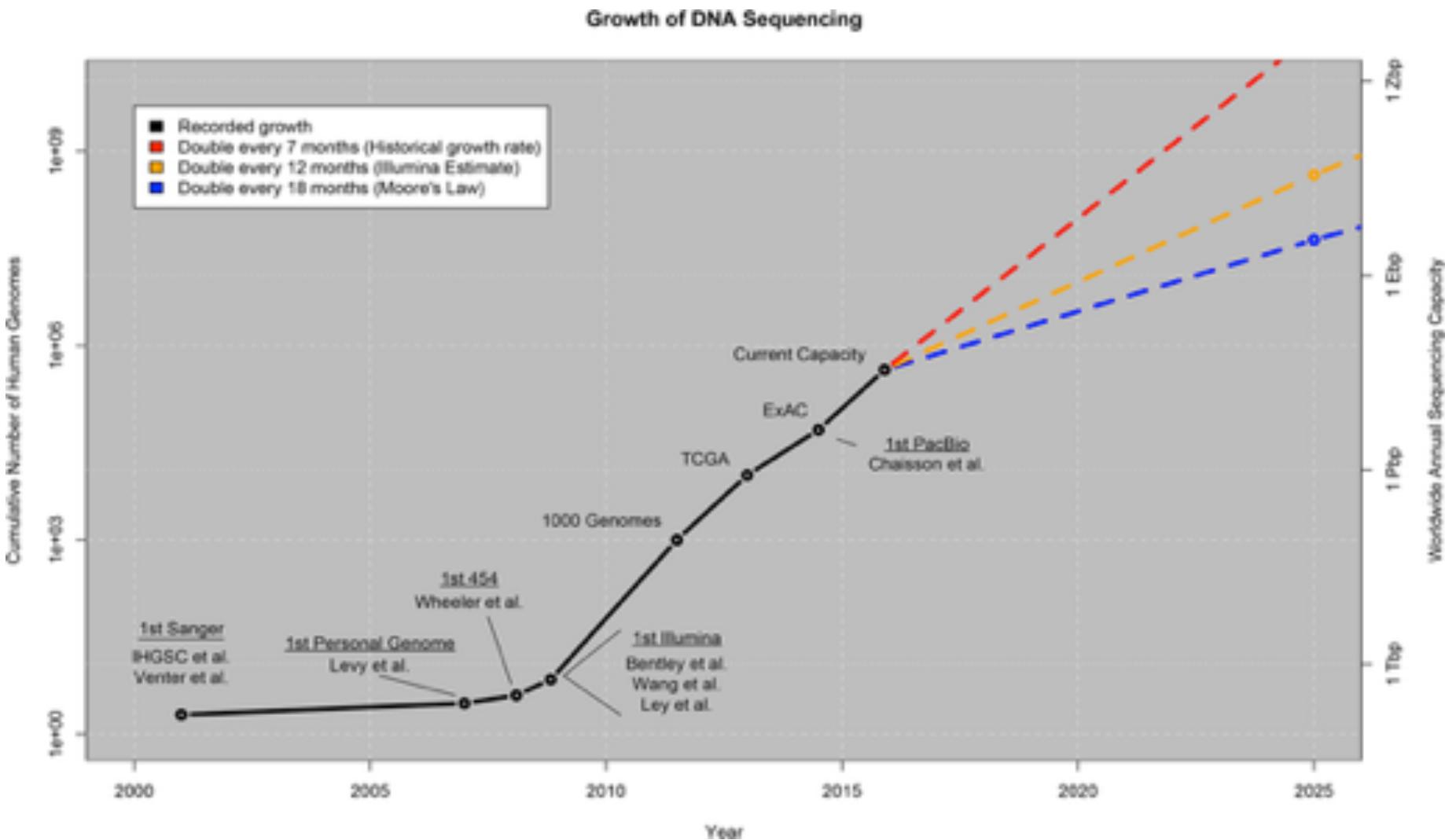


Computer
resources
free for
Swedish
researchers



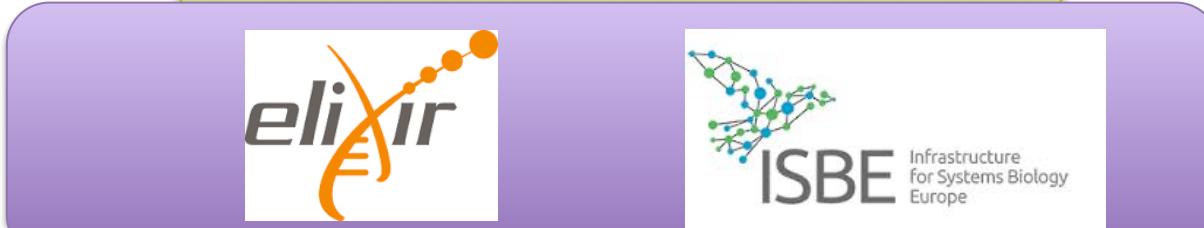
Bioinformatics as infrastructure

Data growth



NBIS activities

Support, tools and training



- **Support and Infrastructure**

Wide competence in bioinformatics, Assembly/Annotation, SysDev

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

Large collaborative projects selected by scientific ranking

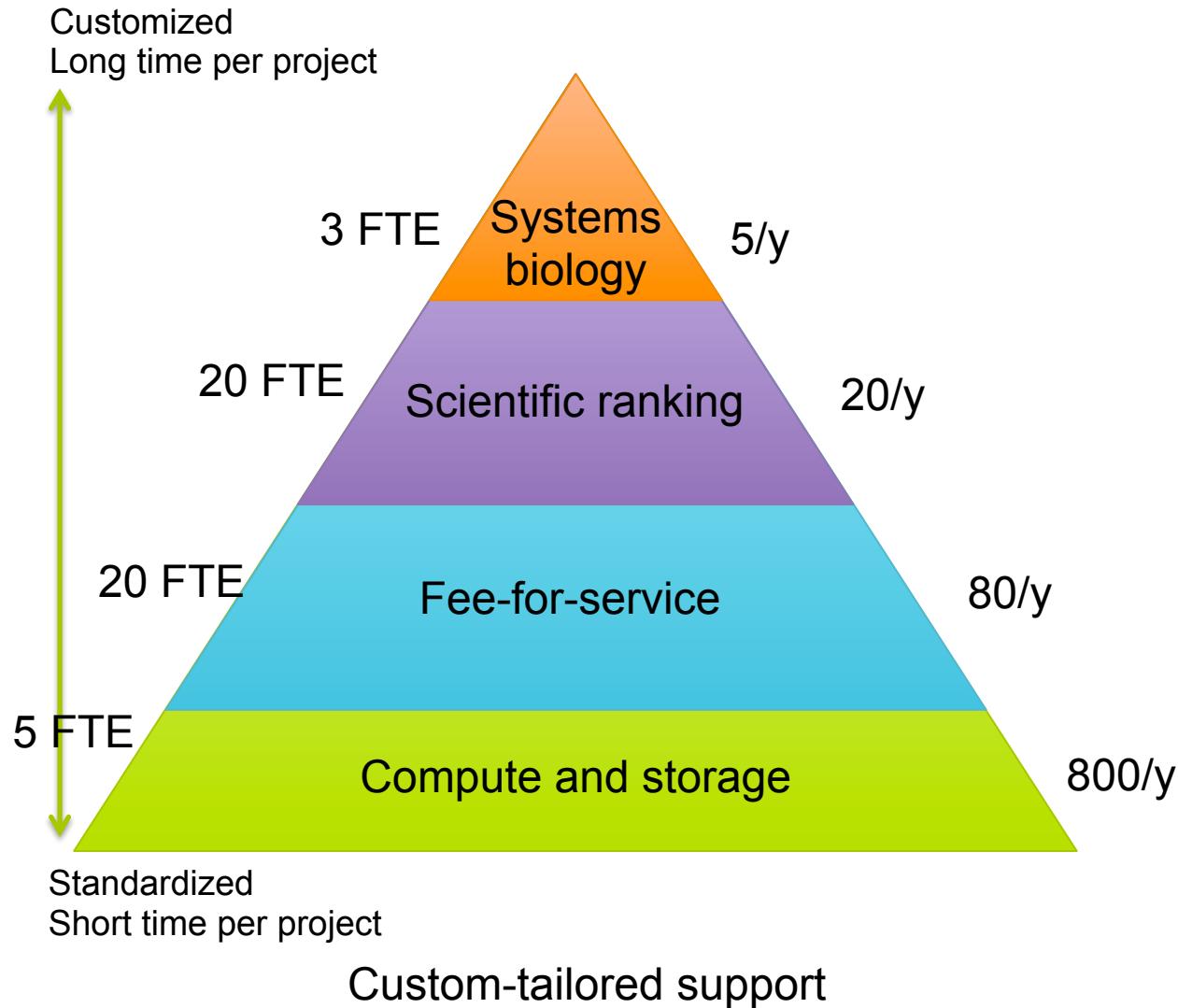
- **Systems biology**

Network analyses and Integrative bioinformatics

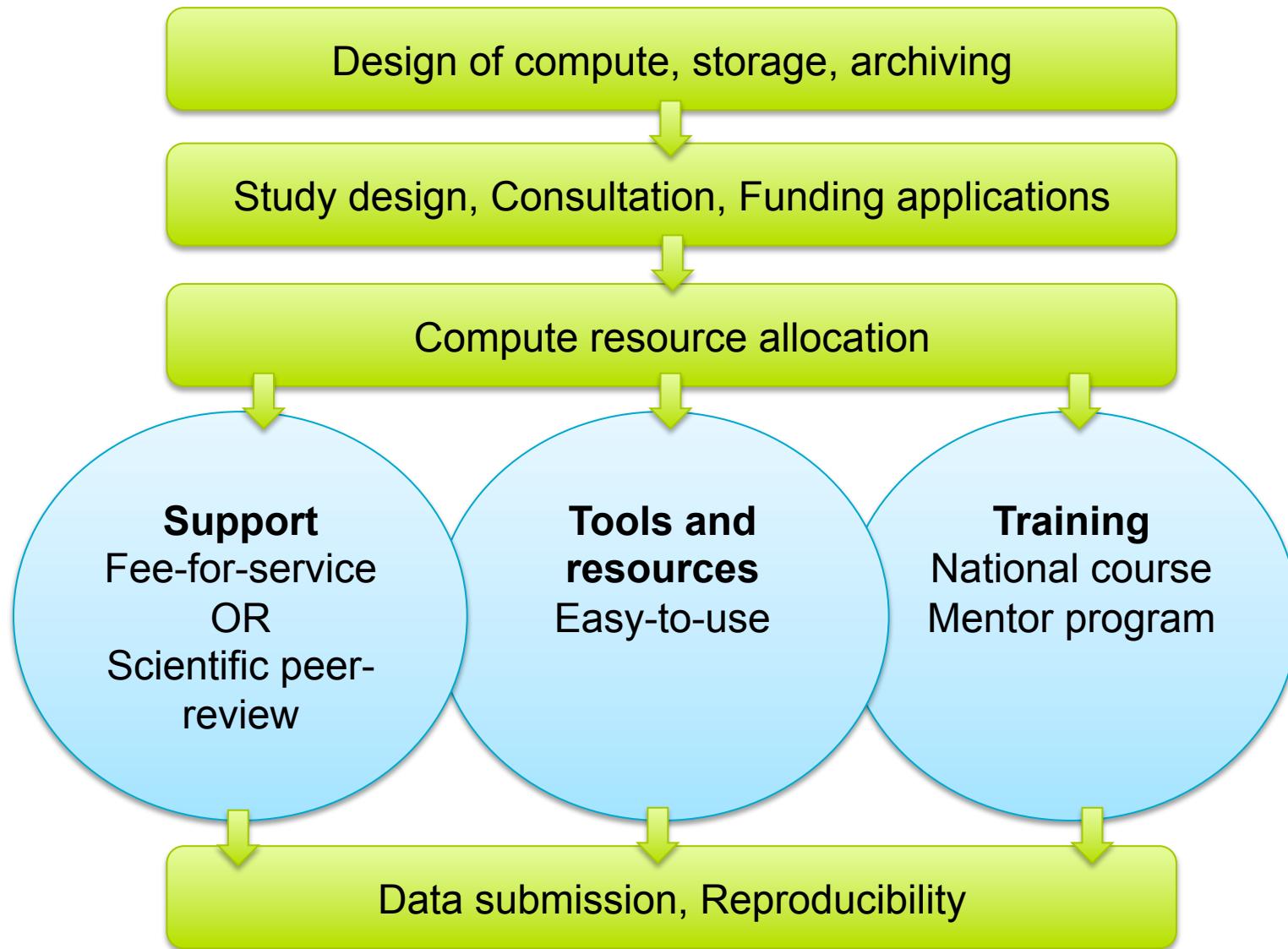
- **Compute and storage**

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





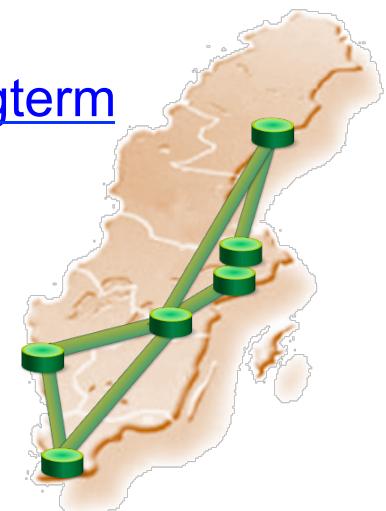
User benefits





- 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

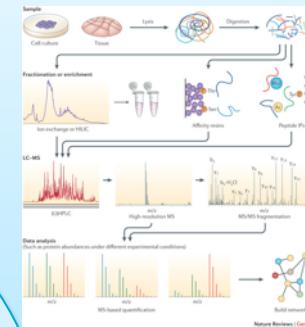
www.scilifelab.se/platforms/bioinformatics/
www.nbis.se



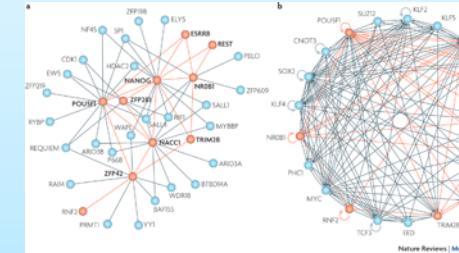
Bioinformatics support



Genomics



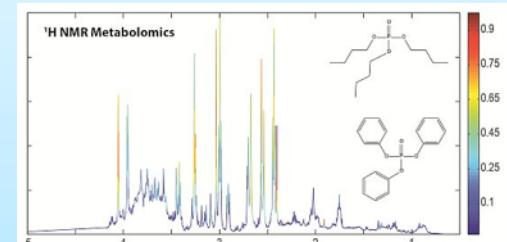
Proteomics



Systems biology



Biostatistics

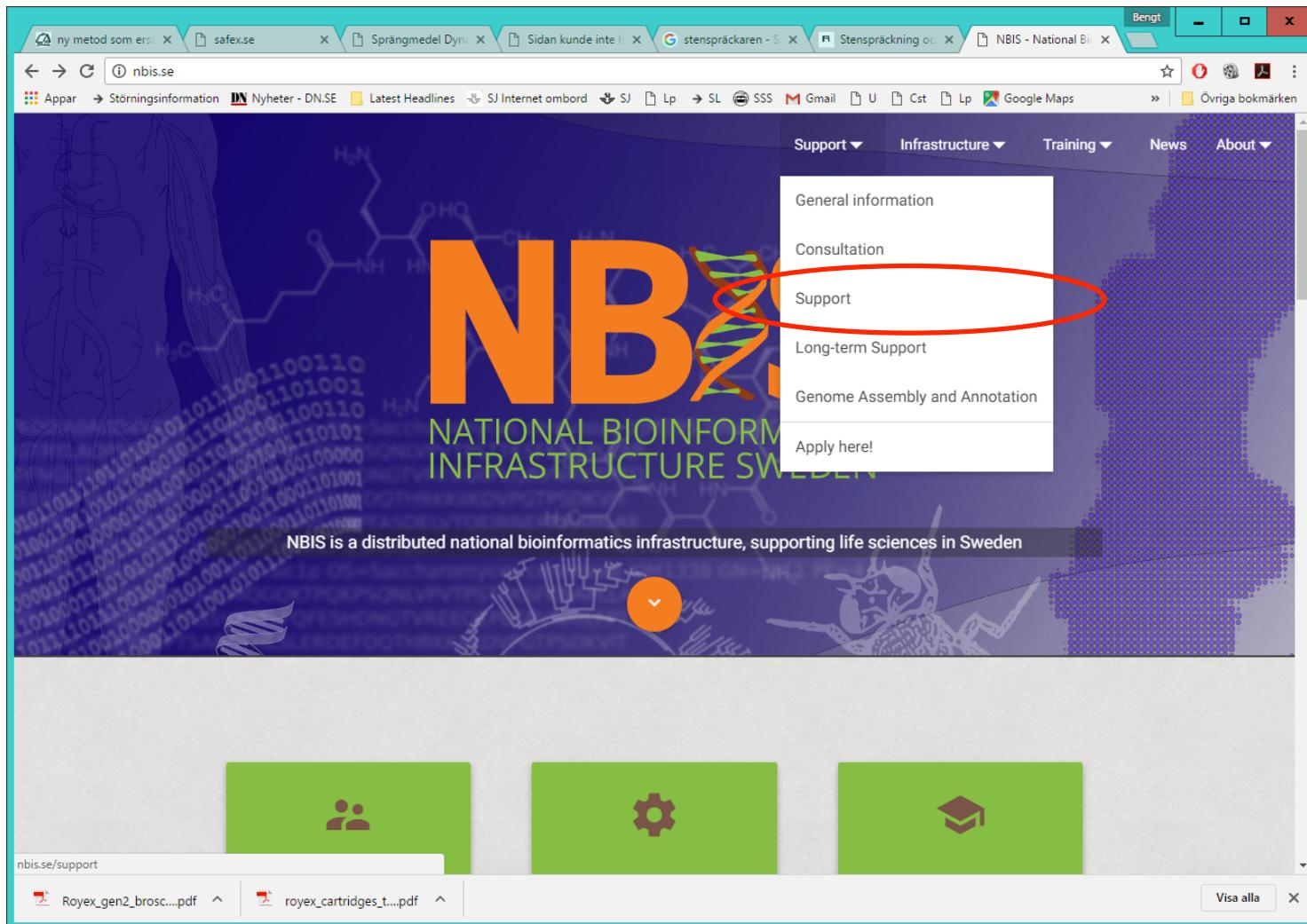


Metabolomics

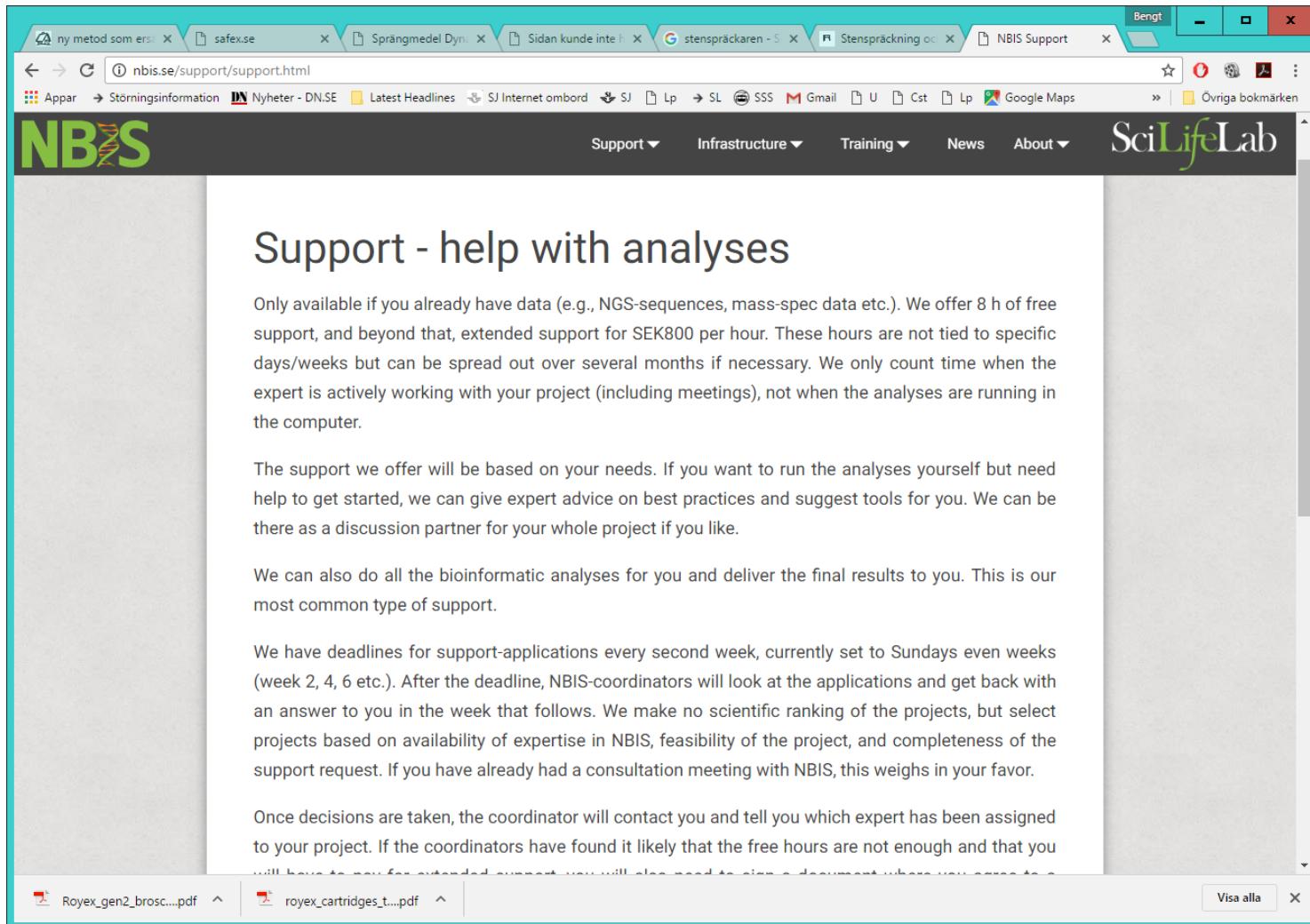
2 tracks!

- 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 URL nbis.se in the address bar. The page features a large orange 'NBIS' logo with a DNA helix integrated into the letter 'B'. Below the logo, the text 'NATIONAL BIOINFORMATICS INFRASTRUCTURE SWEDEN' is visible. A banner at the bottom states: 'NBIS is a distributed national bioinformatics infrastructure, supporting life sciences in Sweden'. On the right side of the header, there is a navigation menu with dropdowns for 'Support', 'Infrastructure', 'Training', 'News', and 'About'. A red oval highlights the 'Support' dropdown menu, which is open and displays the following options: 'General information', 'Consultation', 'Support' (which is circled in red), 'Long-term Support', 'Genome Assembly and Annotation', and 'Apply here!'. At the bottom of the page, there are three green buttons with icons: a person icon, a gear icon, and a graduation cap icon. The status bar at the bottom of the browser shows two PDF files are being viewed: 'Royex_gen2_brosc....pdf' and 'royex_cartridges_t....pdf'. A 'Visa alla' (View all) button is also present.



The screenshot shows a web browser window with multiple tabs open at the top. The main content area displays the NBIS support page, which includes sections about support for analyses, deadlines, and contact information. At the bottom, there are links to PDF files for Royex_gen2_brosc...pdf and royex_cartridges_t...pdf.

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.

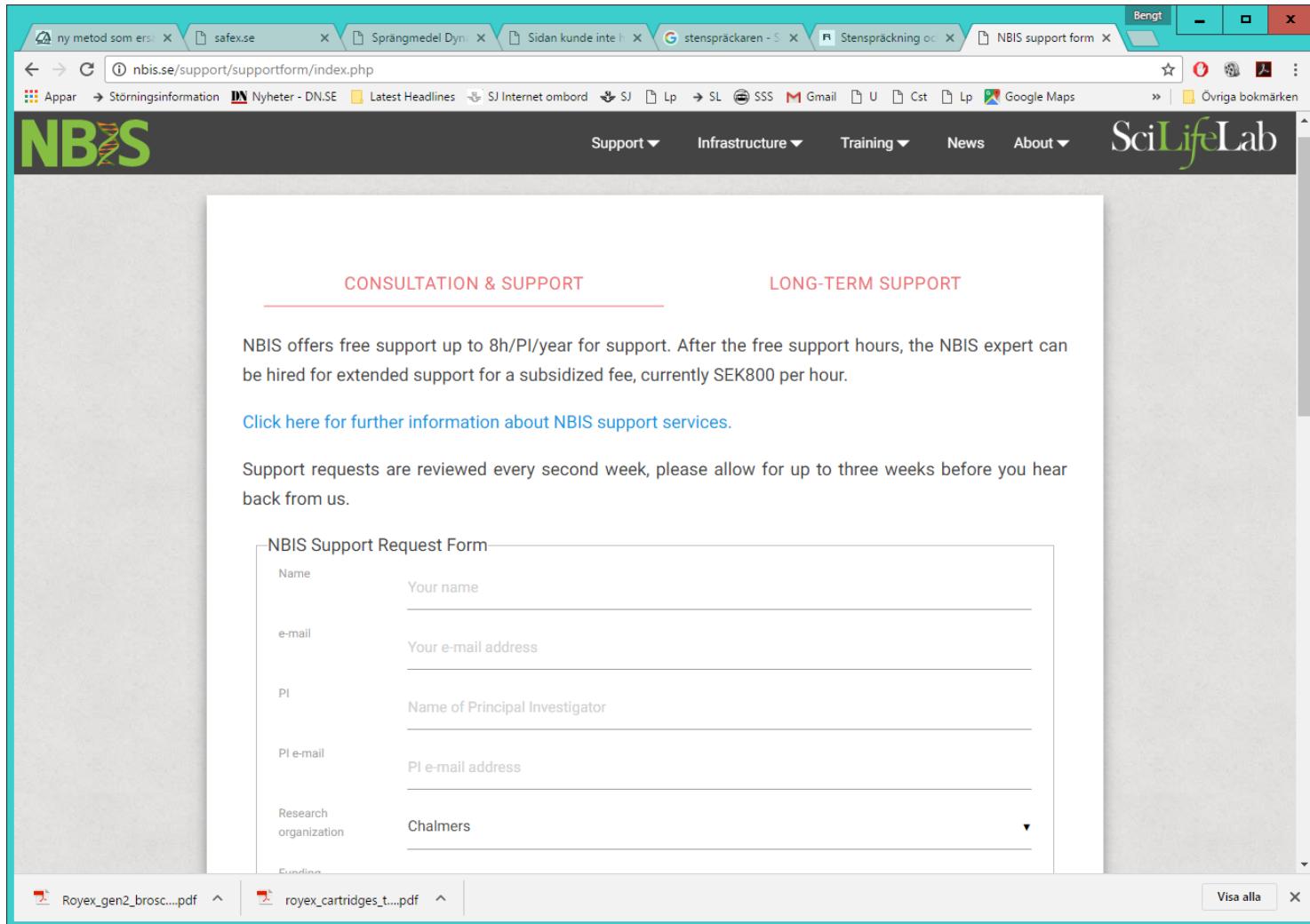
The support we offer will be based on your needs. If you want to run the analyses yourself but need help to get started, we can give expert advice on best practices and suggest tools for you. We can be there as a discussion partner for your whole project if you like.

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

Support form



The screenshot shows a web browser window with the NBIS support form page open. The page has a header with the NBIS logo and a SciLifeLab logo. It features two main sections: 'CONSULTATION & SUPPORT' and 'LONG-TERM SUPPORT'. Under 'CONSULTATION & SUPPORT', it says: '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.' A link 'Click here for further information about NBIS support services.' is provided. Under 'LONG-TERM SUPPORT', it says: 'Support requests are reviewed every second week, please allow for up to three weeks before you hear back from us.' Below this is a 'NBIS Support Request Form' with fields for Name, e-mail, PI, PI e-mail, Research organization, and Funding. At the bottom, there are two PDF files listed: 'Royex_gen2_brosc...pdf' and 'royex_cartridges_t...pdf'.

CONSULTATION & 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.

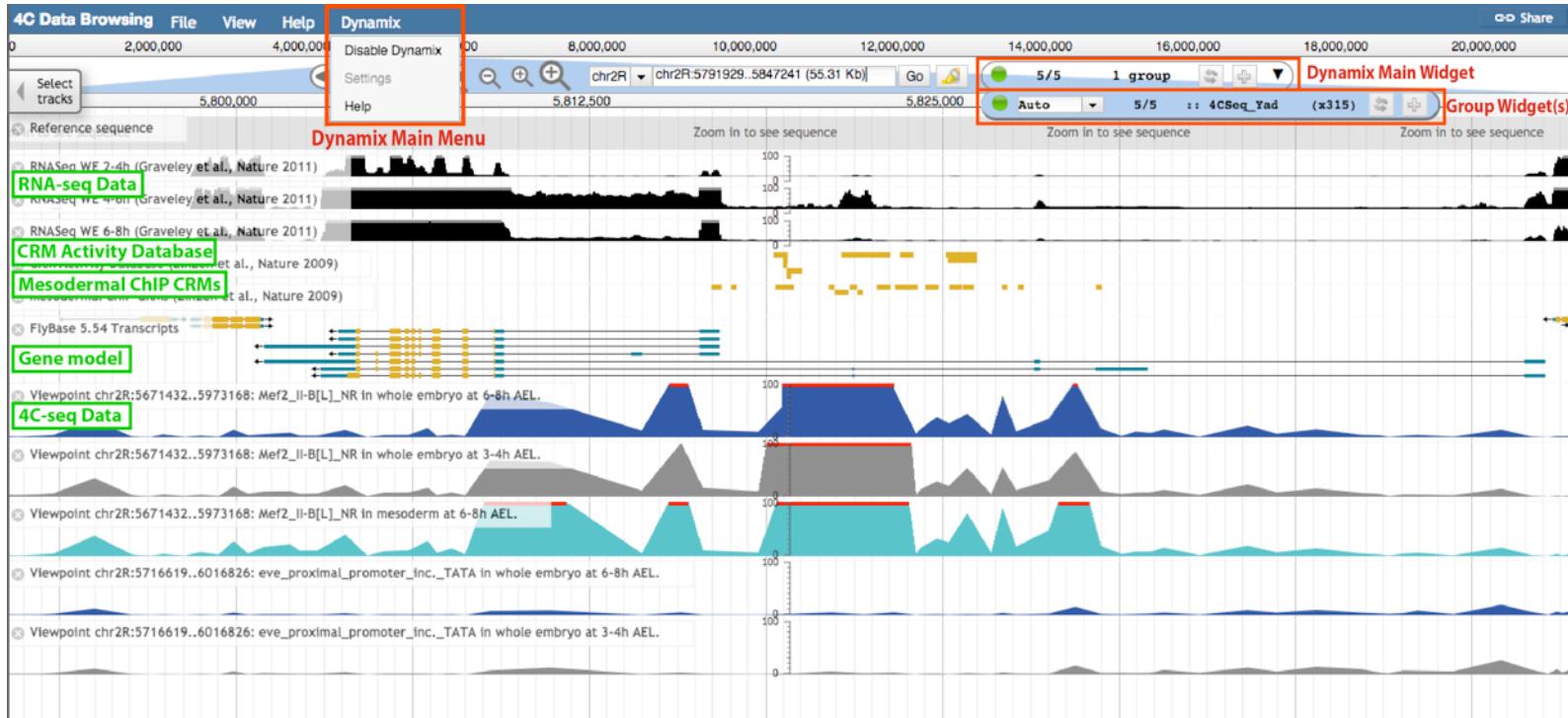
[Click here for further information about NBIS support services.](#)

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
Funding	

Royex_gen2_brosc...pdf royex_cartridges_t...pdf Visa alla



- 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!

Henrik Lantz



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

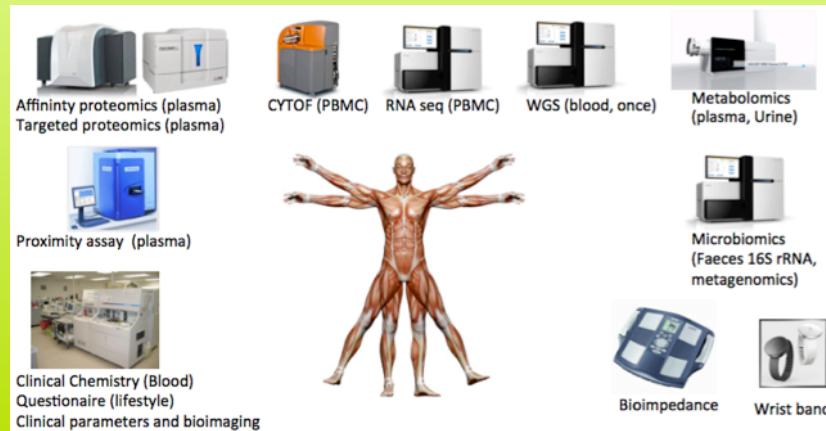
Combine data from SciLifeLab platforms

- 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

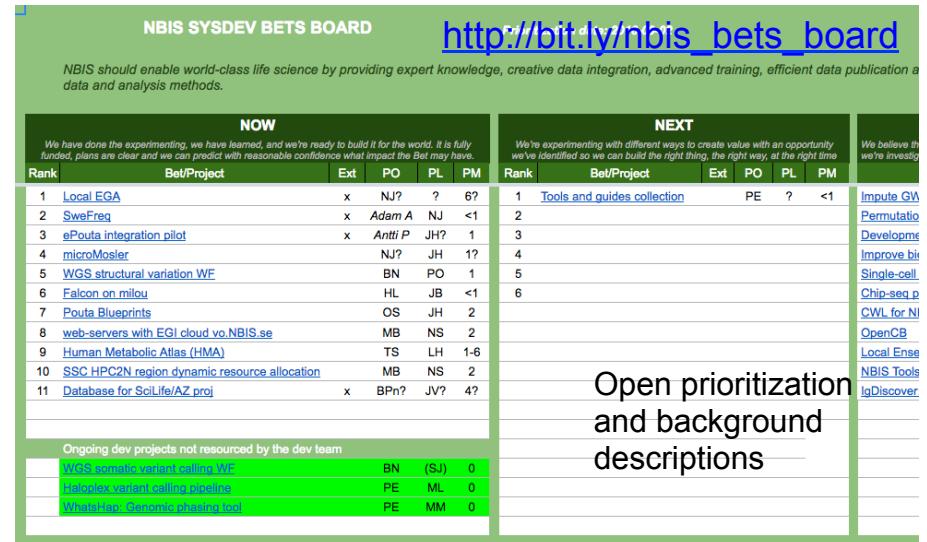
- MrBayes
- Structure prediction web services

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



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 analysis methods.

NOW

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.

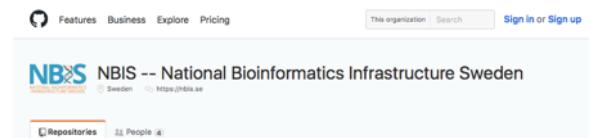
Rank	Bet/Project	Ext	PO	PL	PM
1	Local EGA	x	NJ?	6?	
2	SweFreq	x	Adam A	NJ	<1
3	ePouta integration pilot	x	Antti P	JH?	1
4	microMosler		NJ?	JH	1?
5	WGS structural variation WF		BN	PO	1
6	Falcon on milou		HL	JB	<1
7	Pouta Blueprints		OS	JH	2
8	web-servers with EGI cloud vo.NBIS.se		MB	NS	2
9	Human Metabolic Atlas (HMA)		TS	LH	1-6
10	SSC HPC2N region dynamic resource allocation		MB	NS	2
11	Database for SciLife/AZ proj	x	BPh?	JV?	4?

NEXT

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

Rank	Bet/Project	Ext	PO	PL	PM
1	Tools and guides collection	PE	?	<1	Impute GW
2					Permutatio
3					Developme
4					Improve bi
5					Single-cell
6					Chip-seq p
					CWL for NI
					OpenCB
					Local Ense
					NBIS Tools
					IgDiscover

Open prioritization and background descriptions

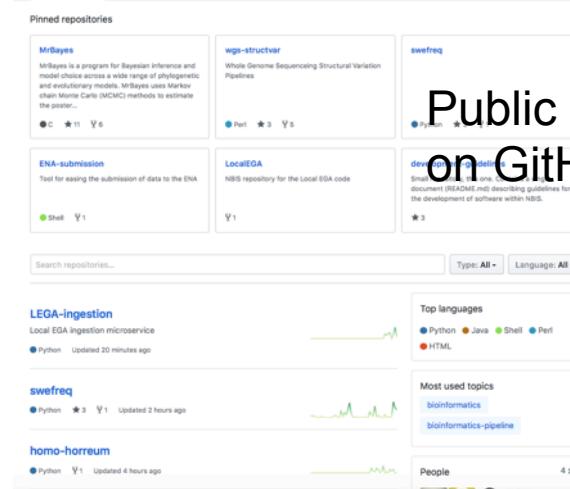


NBIS -- National Bioinformatics Infrastructure Sweden

Repositories People (4)

Pinned repositories

MrBayes	wgs-structvar	swefreq
ENA-submission	LocalEGA	development-tools



Search repositories... Type: All Language: All

Top languages Python Java Shell Perl HTML

Most used topics bioinformatics bioinformatics-pipeline

People

LEG4-ingestion Local EGA ingestion microservice Python Updated 20 minutes ago

swefreq Python Updated 2 hours ago

homo-horreum Python Updated 4 hours ago

Public code repositories on GitHub

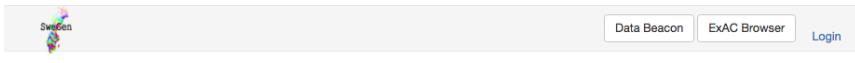
SweFreq – Swedish frequency database



- 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



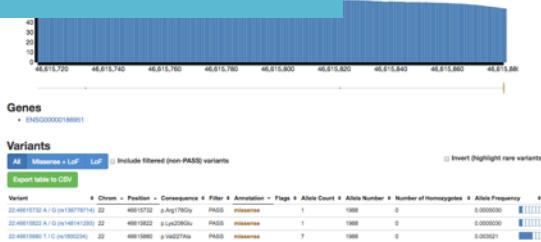
Swedish Frequency Database

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.



Genome browser



Variant	Chrom	Position	Consequence	Filter	Annotation	Flags	Alele Count	Alele Number	Number of Homozygotes	Alele Frequency
22:46615715 A/G (n=387174)	22	46615715	p.Arg189Gln	PASS	missense		1	1000	0	0.000000
22:46615822 A/G (n=1614120)	22	46615822	p.Lys237Ser	PASS	missense		1	1000	0	0.000000
22:46615880 T/C (n=190324)	22	46615880	p.Val227Ile	PASS	missense		7	1000	0	0.000217




SweFreq Agreement

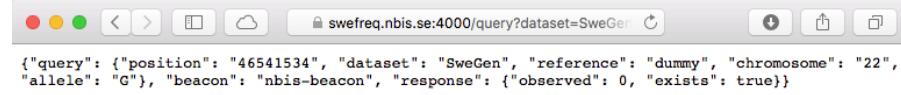
In case the agreement is short we can write it right here. Otherwise I can make a link to it.
 Some more text, bla bla Lorem ipsum dolor sit amet, eu ius graeco perfecto, velit delicatissimi an est,
 rebum persequitur mel no. Stet everti sea ne. Utam laoreet interpretaris vix et, ex qui solum
 abhorreant.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

[Download](#)

Beacon



Yes

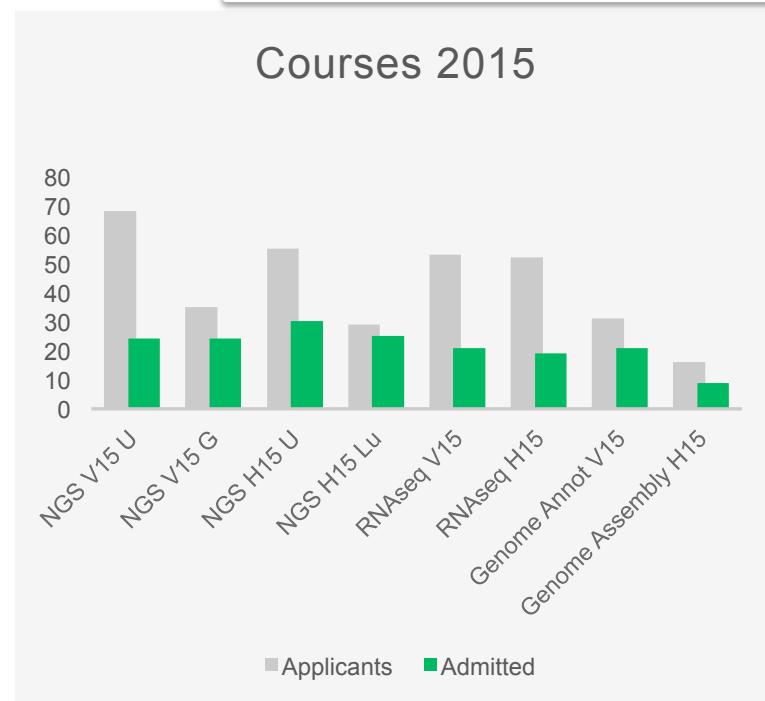


```
{"query": {"position": "46615154", "dataset": "SweGen", "reference": "dummy", "chromosome": "22", "allele": "G"}, "beacon": "nbis-beacon", "response": {"observed": 0, "exists": true}}
```



- 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.

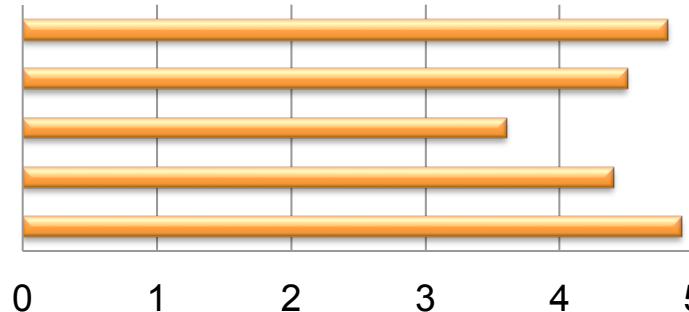
www.scilifelab.se/education/mentorship/the-swedish-bioinformatics-advisory-program/

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

The Swedish Bioinformatics Advisory Program

Student evaluation, June 2015

- 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

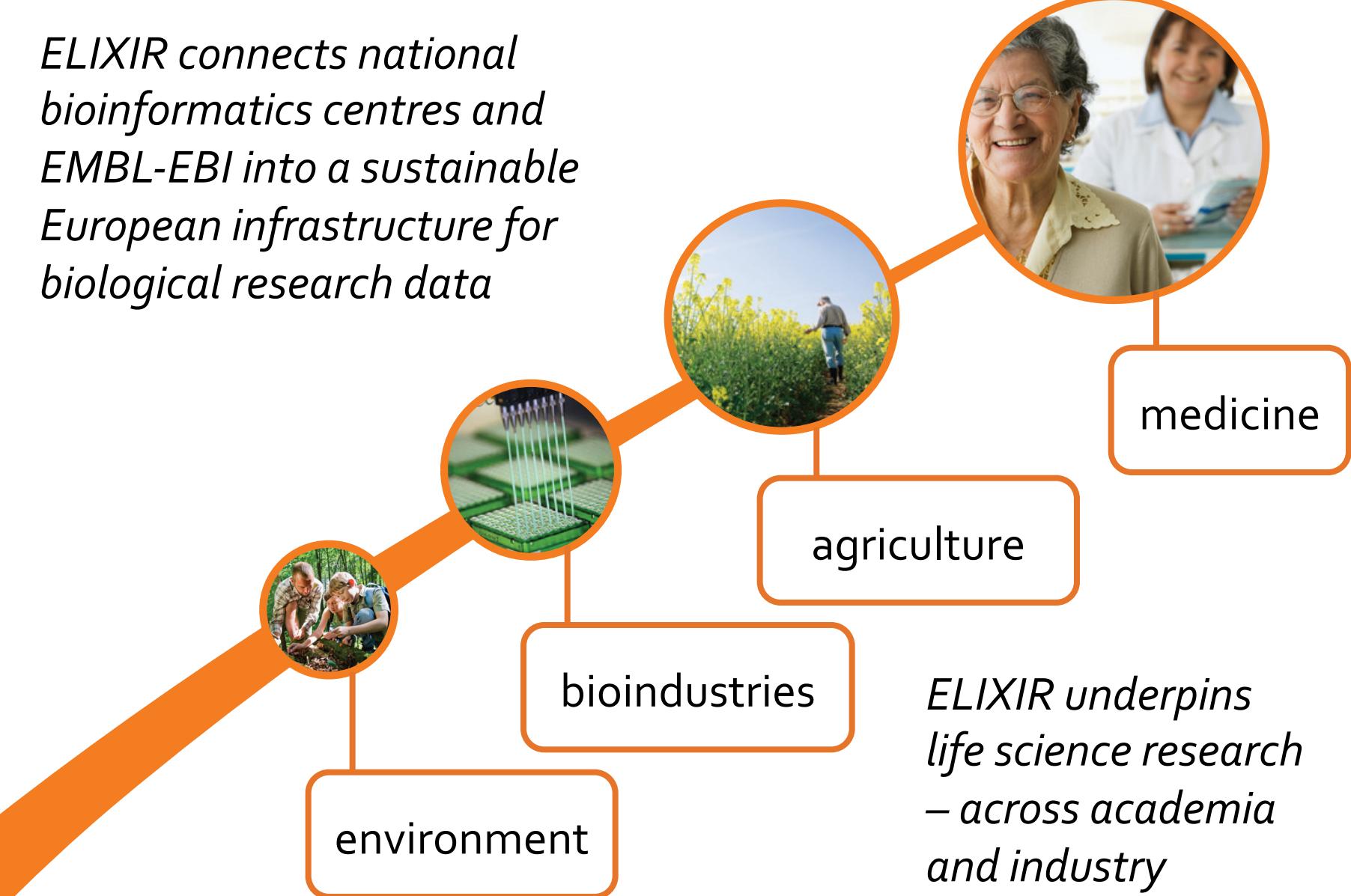


Elixir

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*



*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*

-

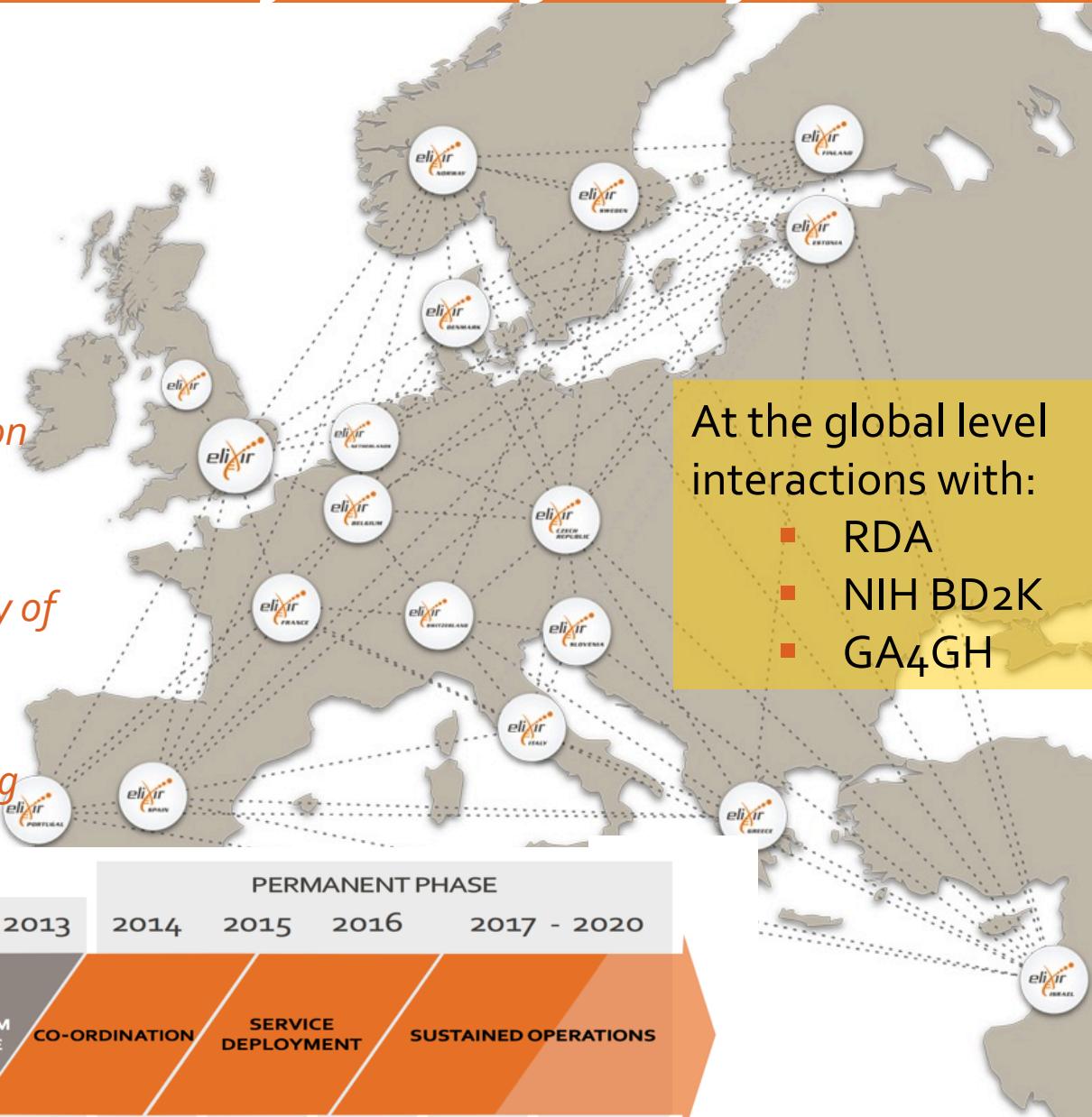
Standards

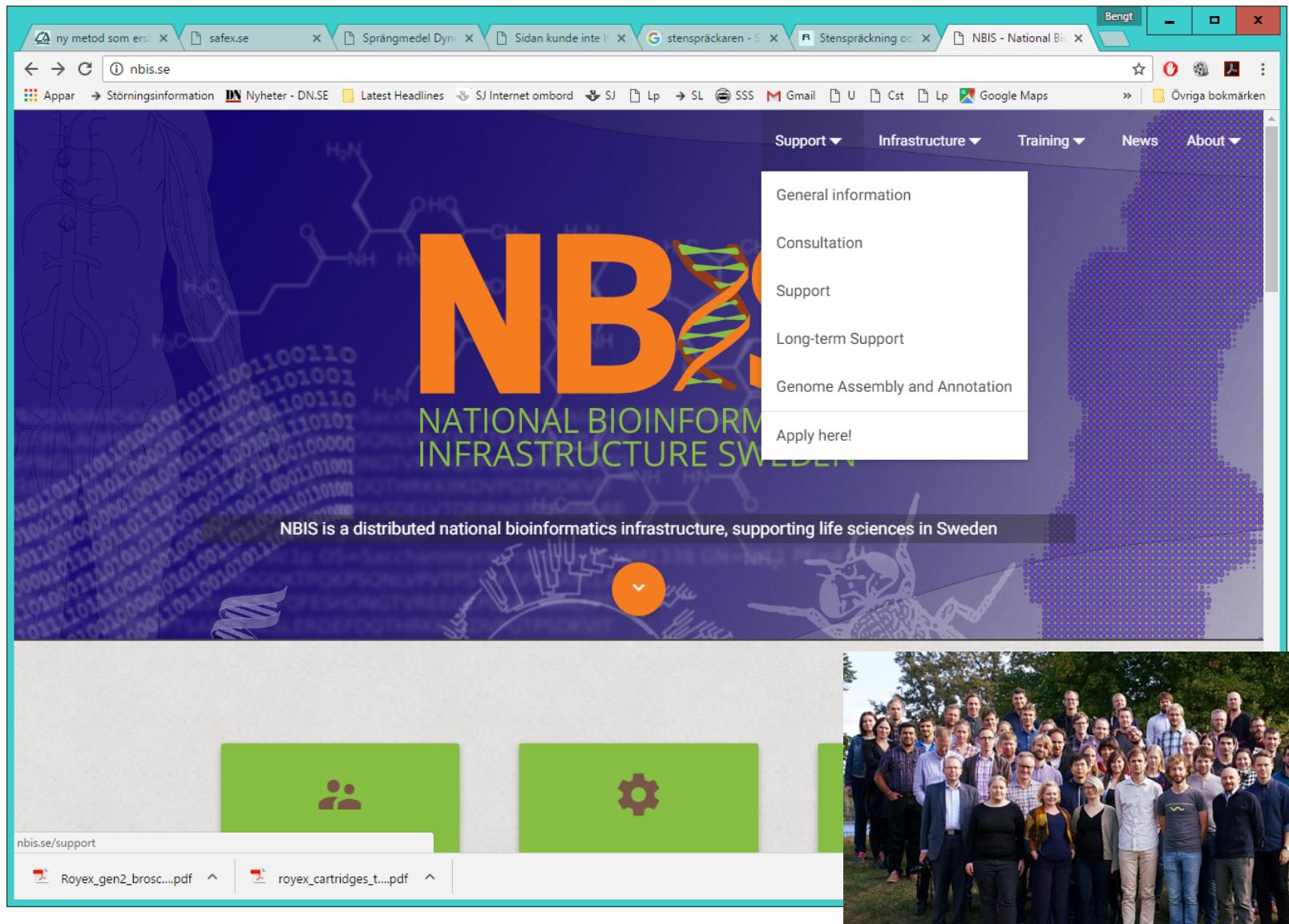
*Integration and interoperability of
data and services.*

-

Training

*Professional skills for managing
and exploiting data*





The screenshot shows the NBIS website homepage. The main header features the NBIS logo and the text "We're here for you! nbis.se". Below the header is a large banner with a blue background, featuring a DNA helix and chemical structures. The banner text reads: "NBIS is a distributed national bioinformatics infrastructure, supporting life sciences in Sweden". A support menu is open on the right side of the banner, listing options such as "General information", "Consultation", "Support", "Long-term Support", "Genome Assembly and Annotation", and "Apply here!". At the bottom of the page, there are two green buttons with icons: one for users and one for gears. The browser's address bar shows "nbis.se/support". A sidebar on the left contains links to "nbis.se", "Safex.se", "Språngmedel Dyn...", "Sidan kunde inte h...", "stenspräckaren - S...", "Stenspräckning oc...", "NBIS - National Bi...", and "Bengt". The bottom right corner of the screenshot shows a group photo of the NBIS team.

ny metod som ers... safex.se Språngmedel Dyn... Sidan kunde inte h... stenspräckaren - S... Stenspräckning oc... NBIS - National Bi... Bengt

nbis.se

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

Support Infrastructure Training News About

General information

Consultation

Support

Long-term Support

Genome Assembly and Annotation

Apply here!

NBIS NATIONAL BIOINFORMATICS INFRASTRUCTURE SWEDEN

NBIS is a distributed national bioinformatics infrastructure, supporting life sciences in Sweden

nbis.se/support

Royex_gen2_brosc...pdf royex_cartridges_t...pdf

