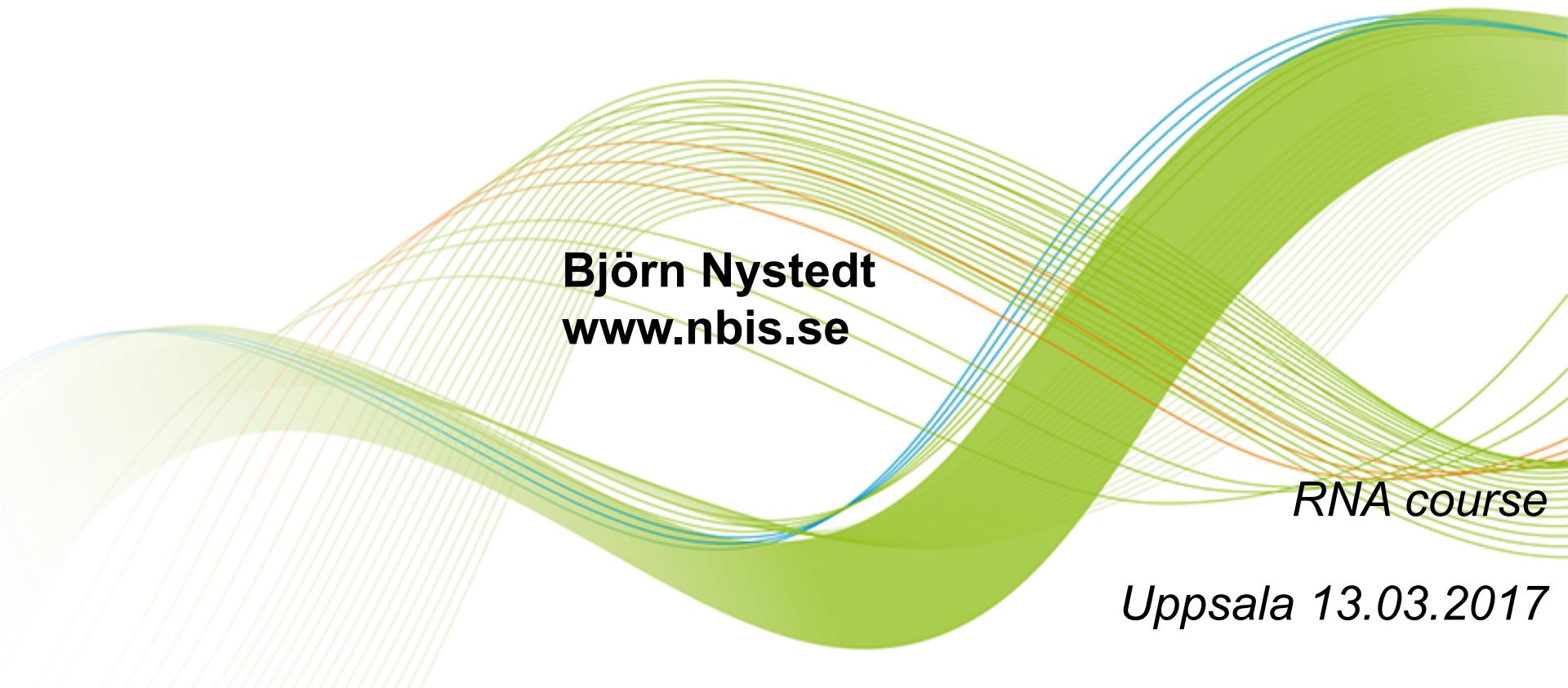


SciLifeLab Bioinformatics Platform

National Bioinformatics Infrastructure Sweden (NBIS)



Björn Nystedt
www.nbis.se

RNA course

Uppsala 13.03.2017

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

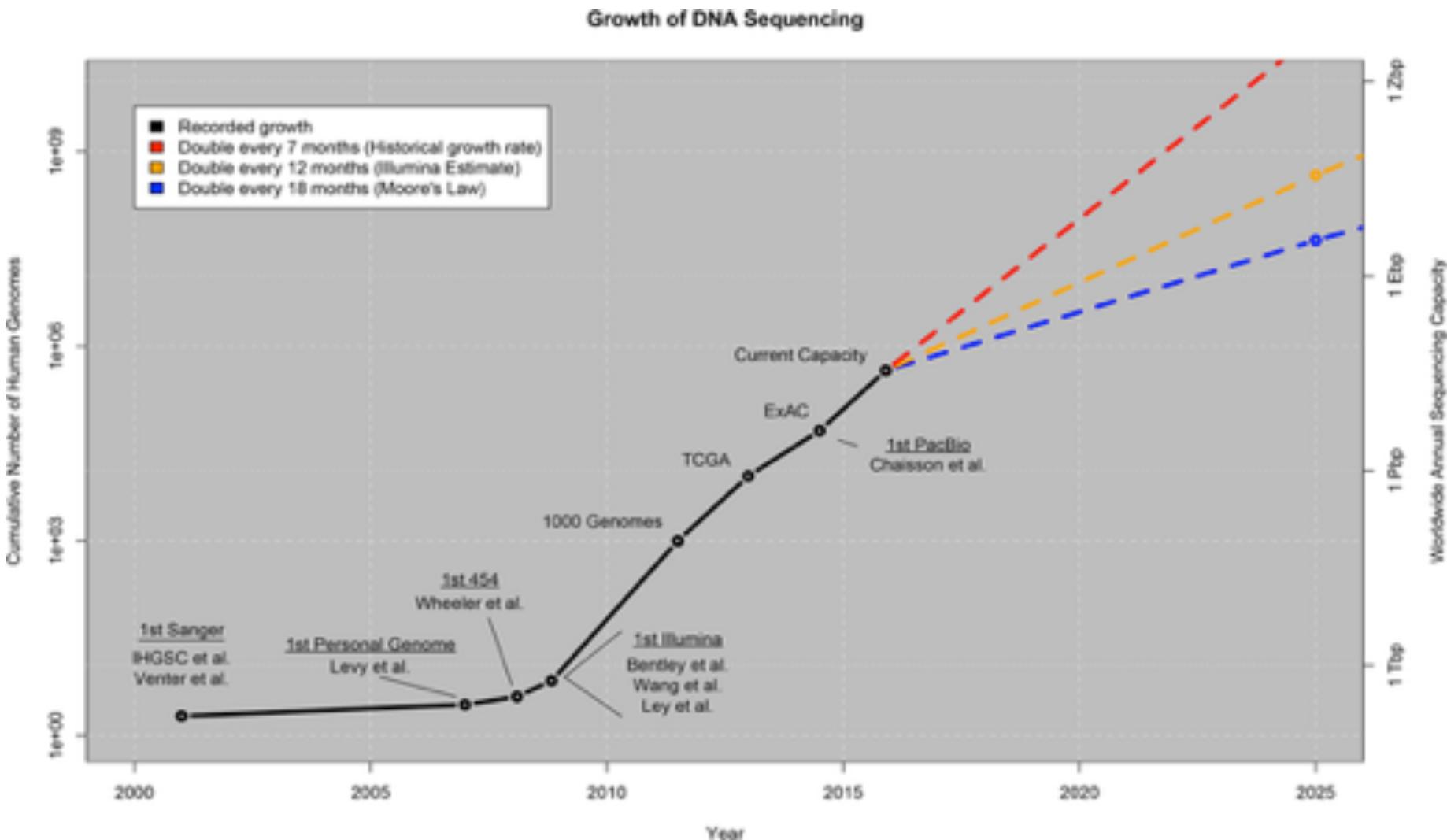
SNIC



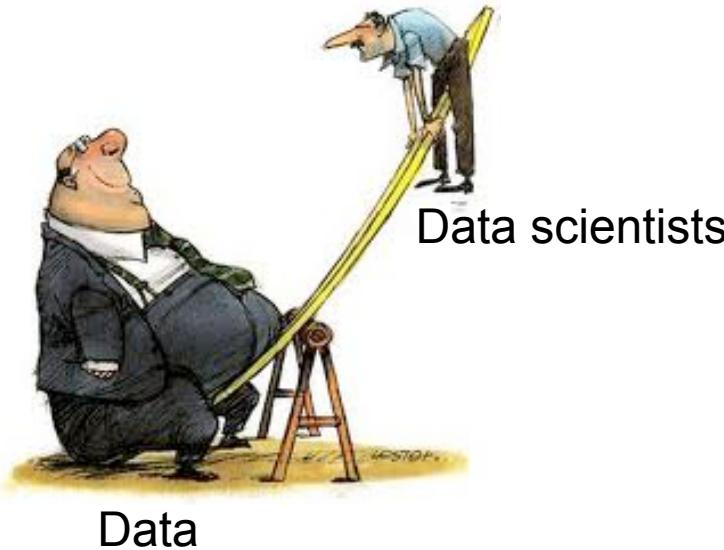
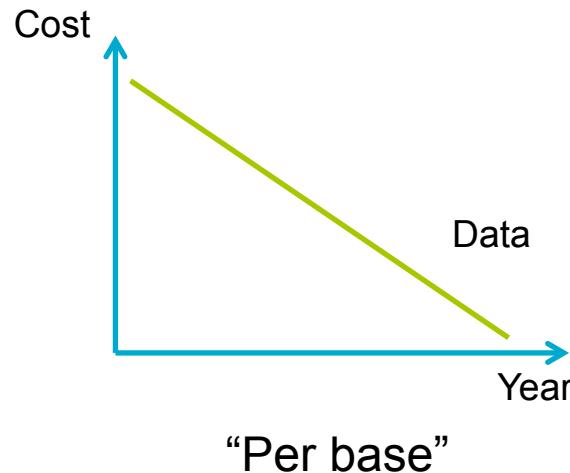
Computer
resources
free for
Swedish
researchers

Bioinformatics as infrastructure

Data growth



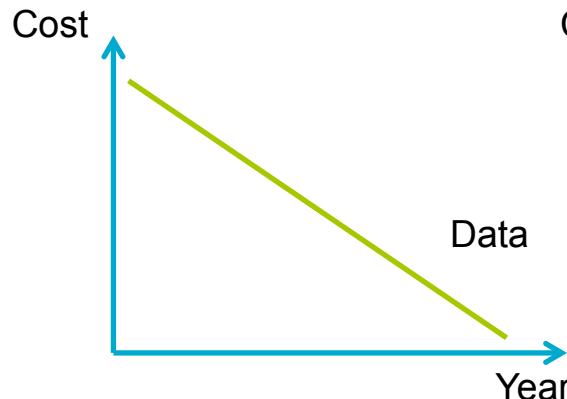
Production is cheap, analysis is not



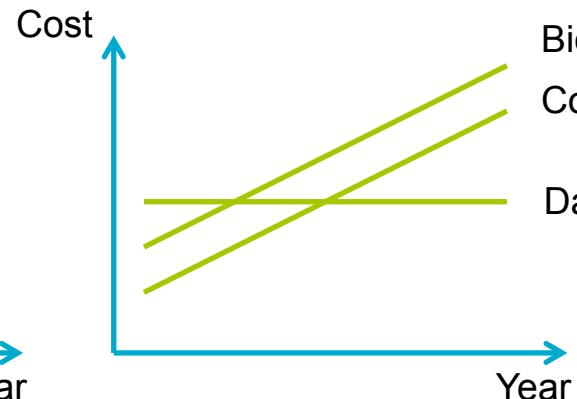
Our role

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.

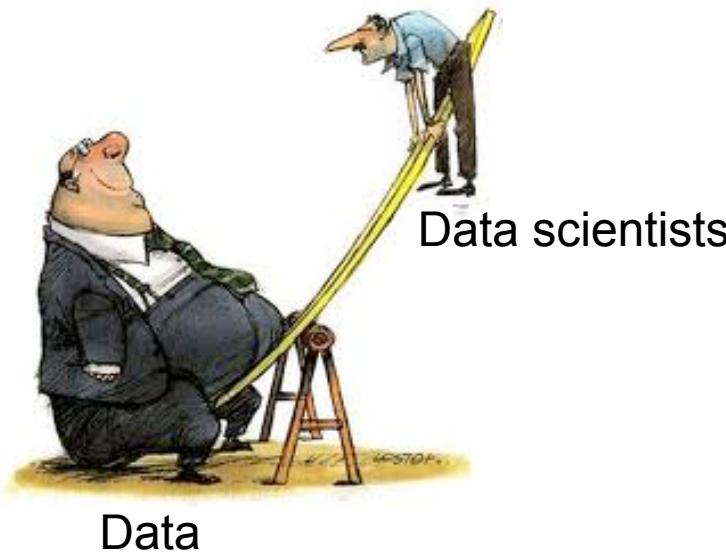
Production is cheap, analysis is not



“Per base”



“Per project”

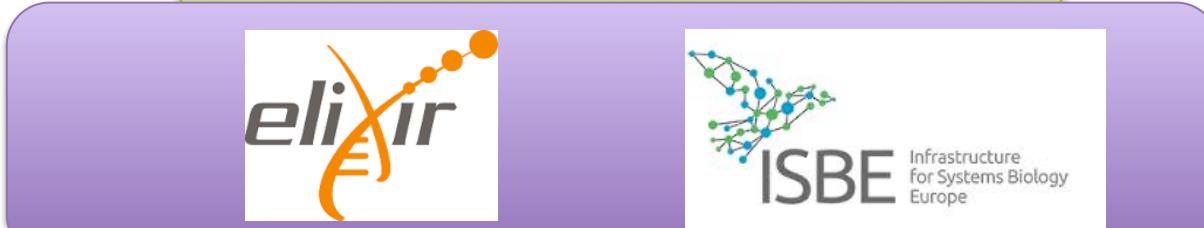


Our role

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



4 facilities, ~60 FTEs

- **Support and Infrastructure**

Wide competence in bioinformatics, Assembly/Annotation, SysDev

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

Large collaborative projects selected by scientific ranking

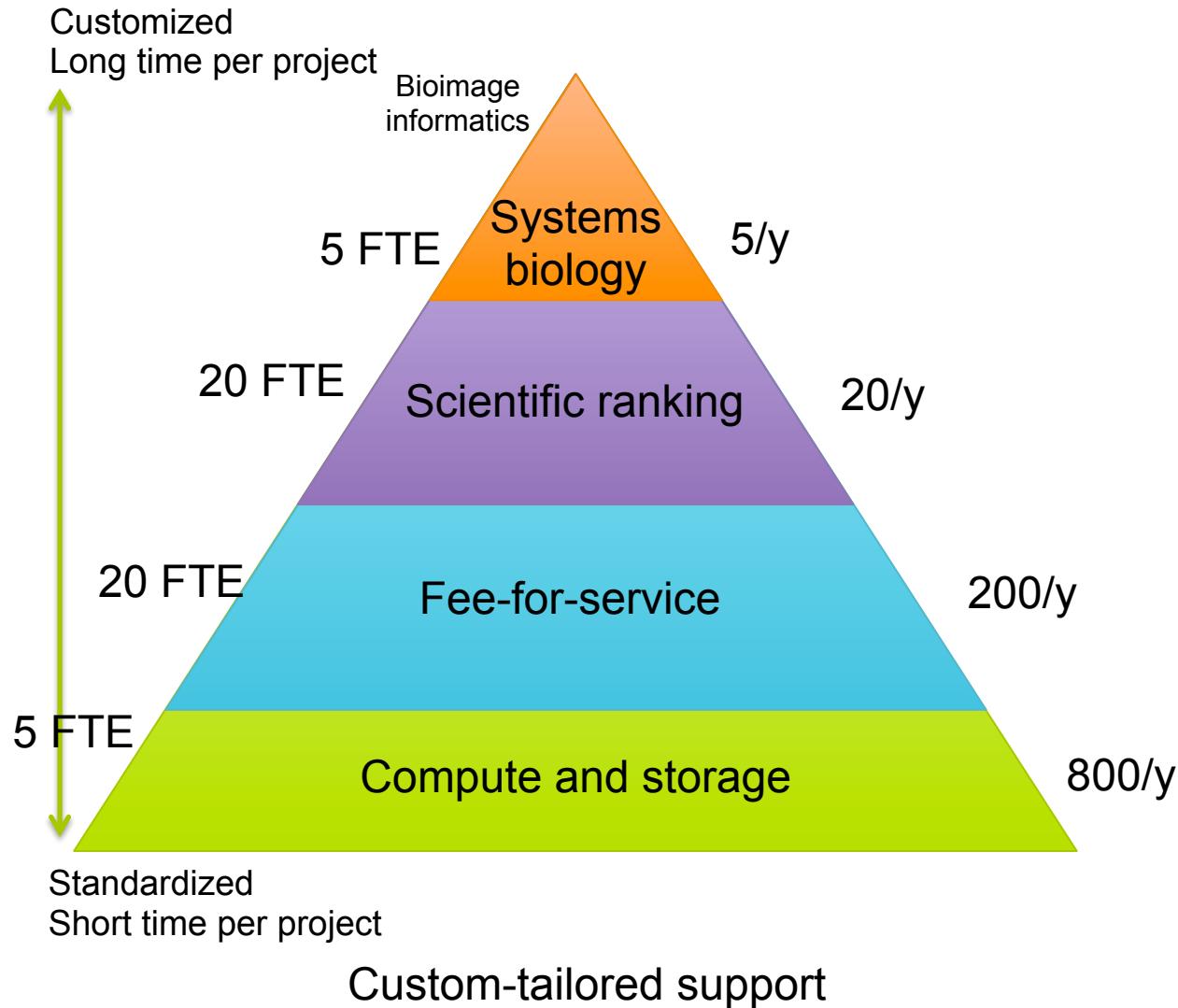
- **Systems biology**

Network analyses and Integrative bioinformatics

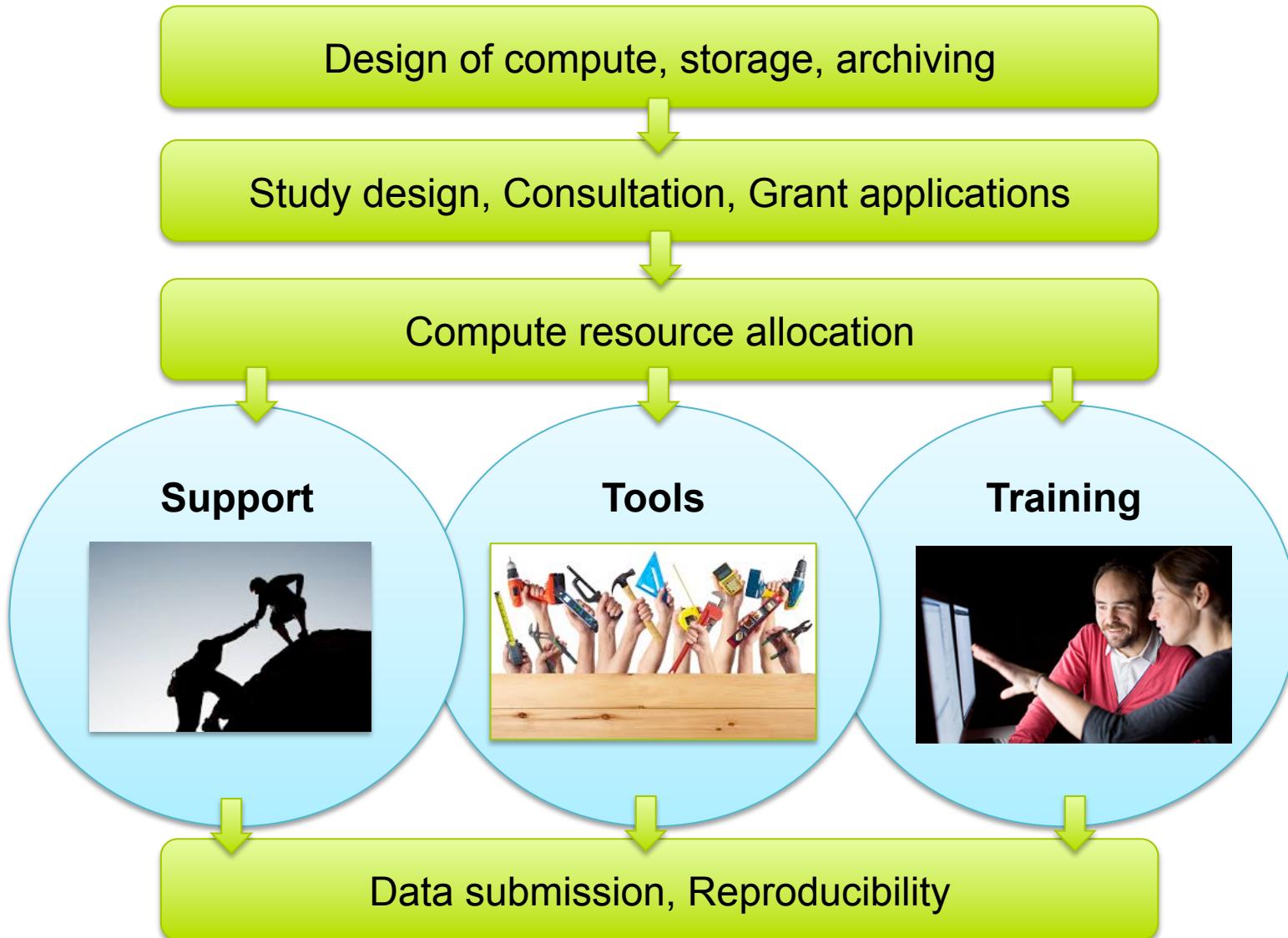
- **Compute and storage**

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





User benefits

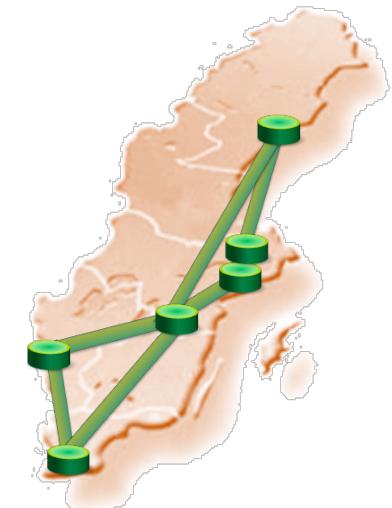




Custom-tailored support

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

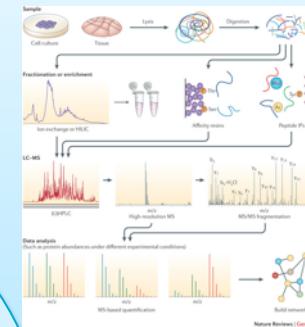
- Study design consultation (free)
www.nbis.se/support/supportform/index.php
+ drop-in sessions every week @ 6 sites
- Short- and Medium-term support (User fee 800 kr/h)
www.nbis.se/support/supportform/index.php
- Long-term support and systems biology
(500h, free, scientific evaluation)
www.nbis.se/support/supportform/index.php?form=longterm



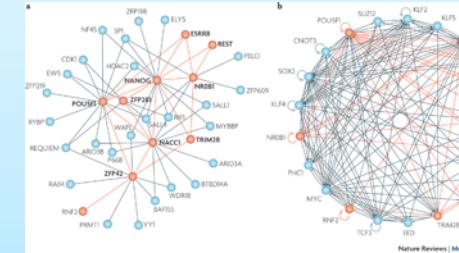
Bioinformatics support



Genomics



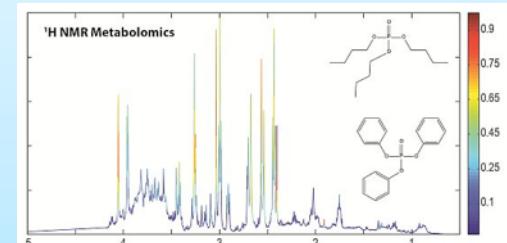
Proteomics



Systems biology



Biostatistics

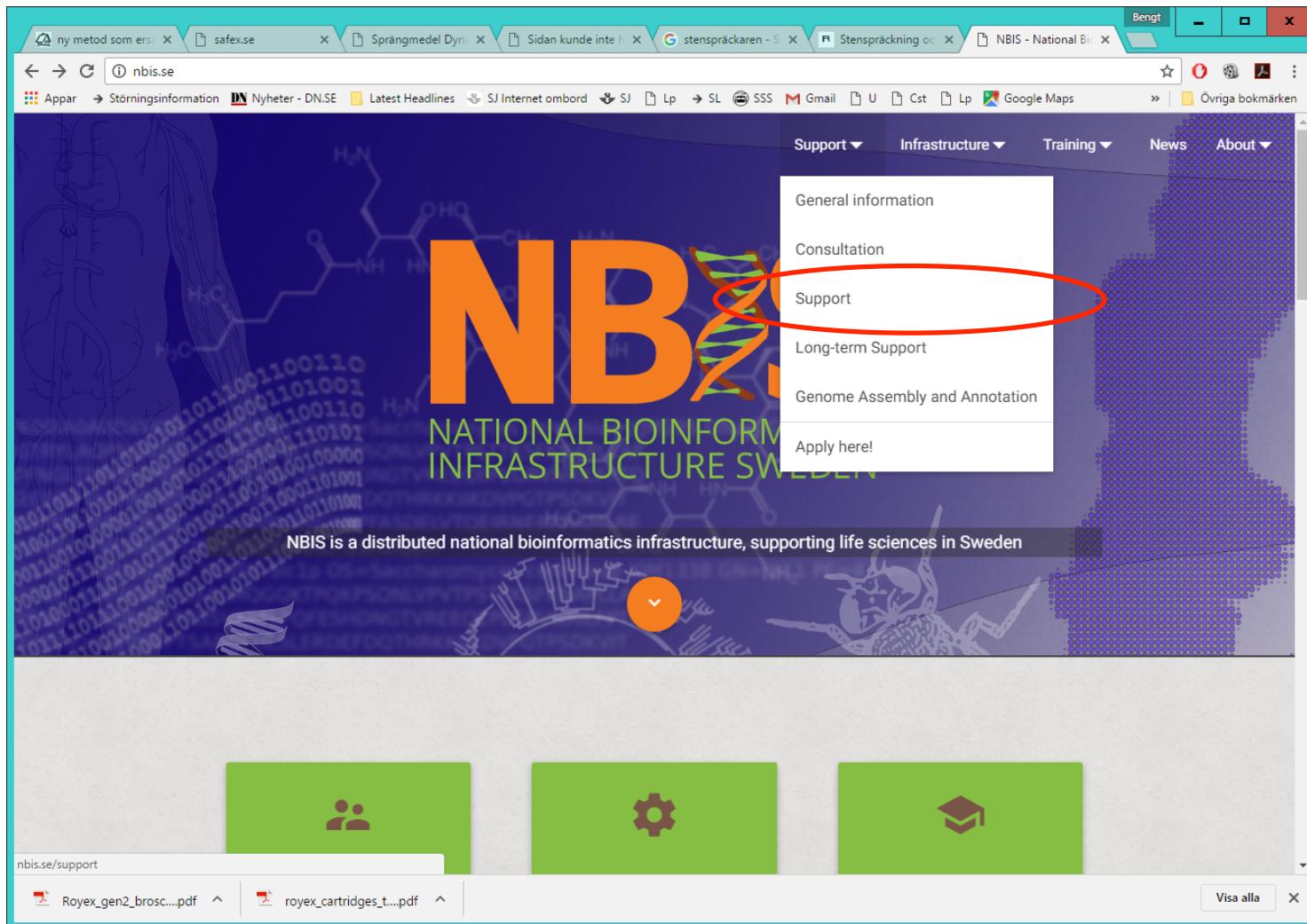


Metabolomics

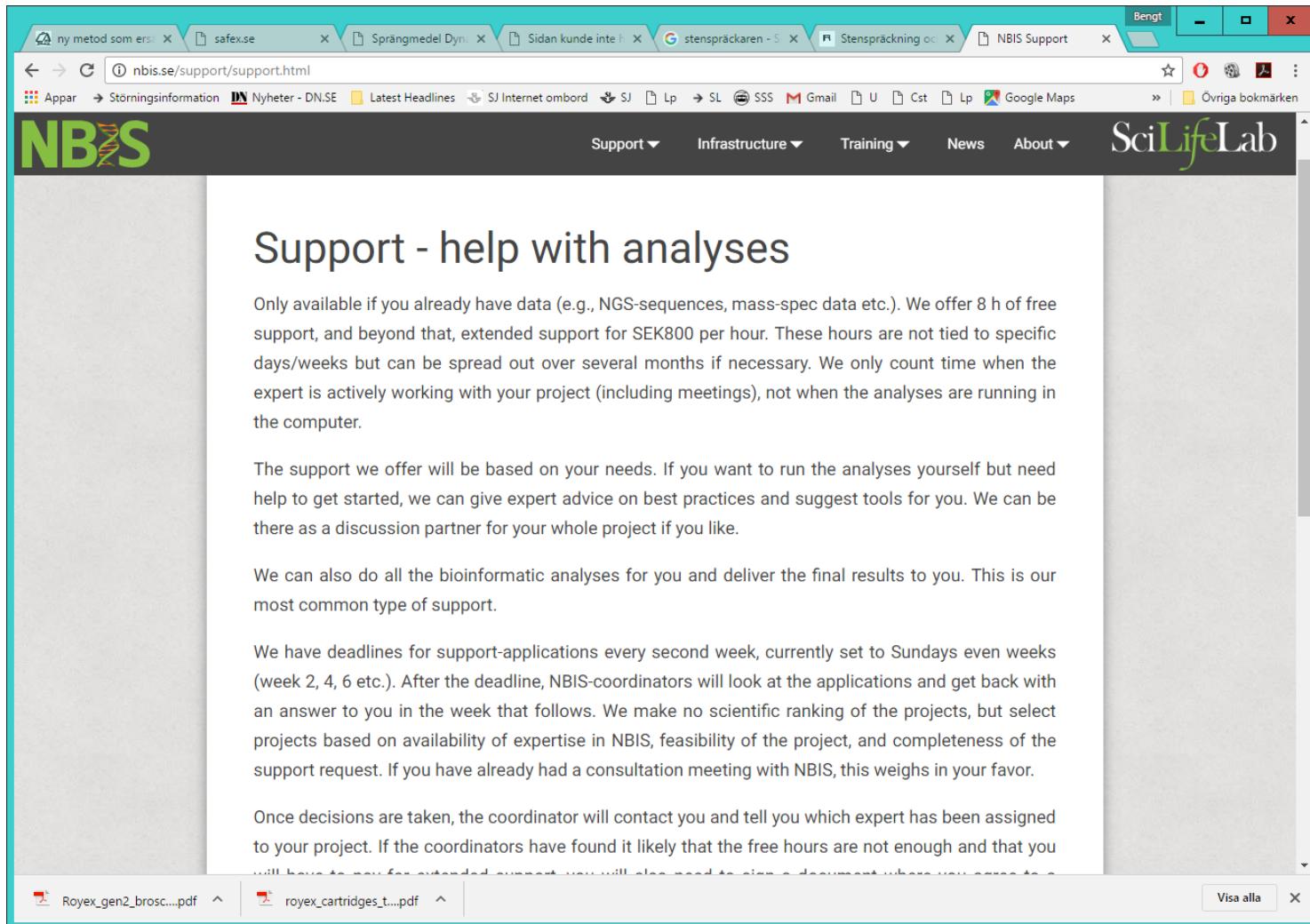
2 tracks!

- Fee-for-service (800kr/h)
Rapid turnaround
- Scientific ranking (free)
“Long-term Support”
3 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 has a dark blue background featuring a DNA helix and various scientific illustrations. A navigation bar at the top includes links for Support, Infrastructure, Training, News, and About. A dropdown menu is open under the Support link, listing options: General information, Consultation, Support (which is circled in red), Long-term Support, Genome Assembly and Annotation, and Apply here!. Below the menu, there are three green buttons with icons: a person icon, a gear icon, and a graduation cap icon. At the bottom of the page, there are two PDF files listed in a sidebar: "Royex_gen2_brosc....pdf" and "royex_cartridges_t....pdf".



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

CONSULTATION

NBIS offers bioinformatics consultations as a free service.

[Click here for further information about the NBIS consultation service.](#)

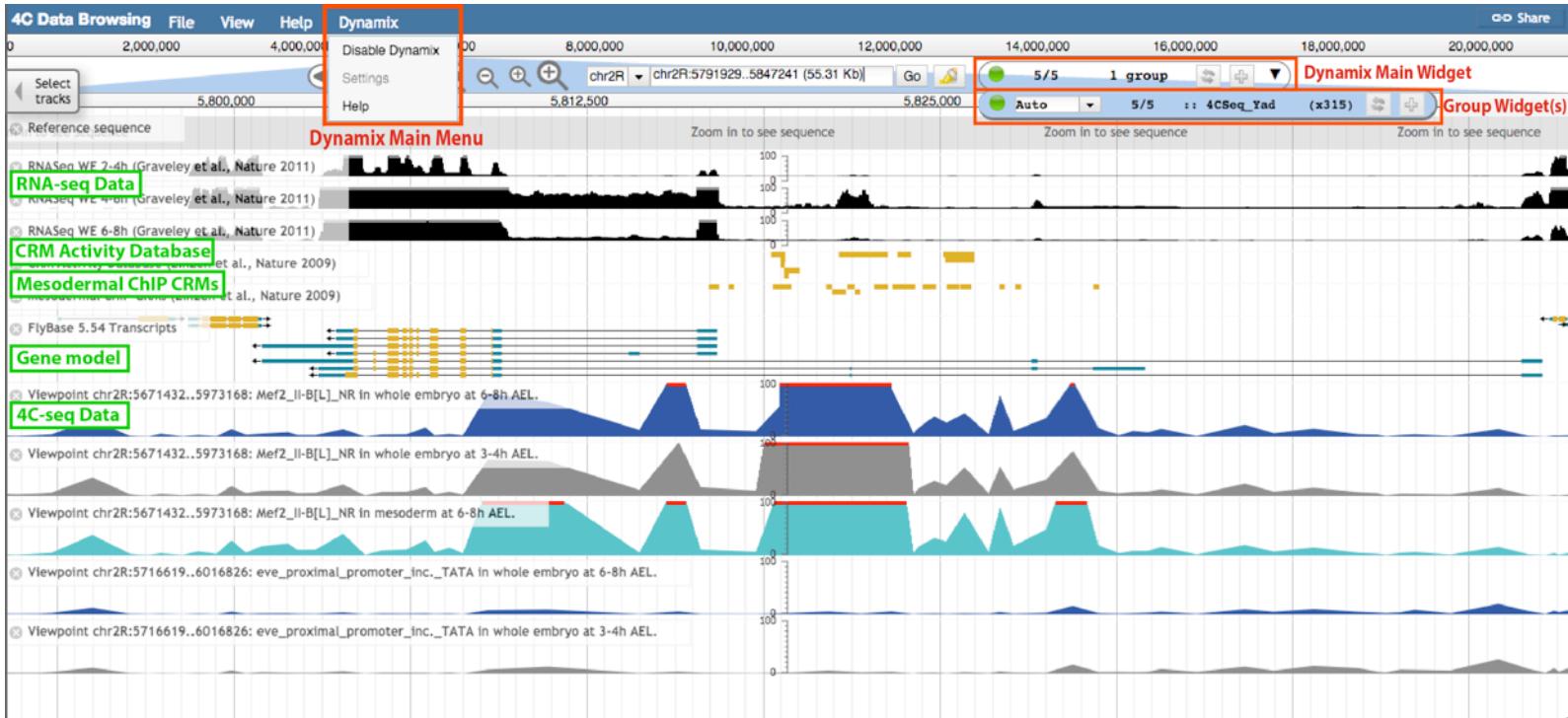
Support requests are reviewed every second week, please allow for up to three weeks before you hear back from us.

By applying for NBIS Support you also agree to our [user agreement](#).

NBIS Consultation 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	ALF
Project Title	Please include descriptive keywords.
Subject	NGS
Description	Brief project description.
Starting time	ASAP

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

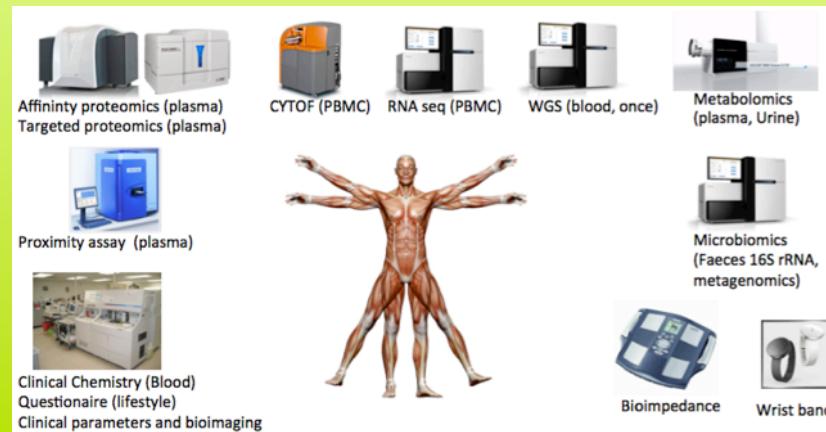
Combine data from SciLifeLab platforms

- 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

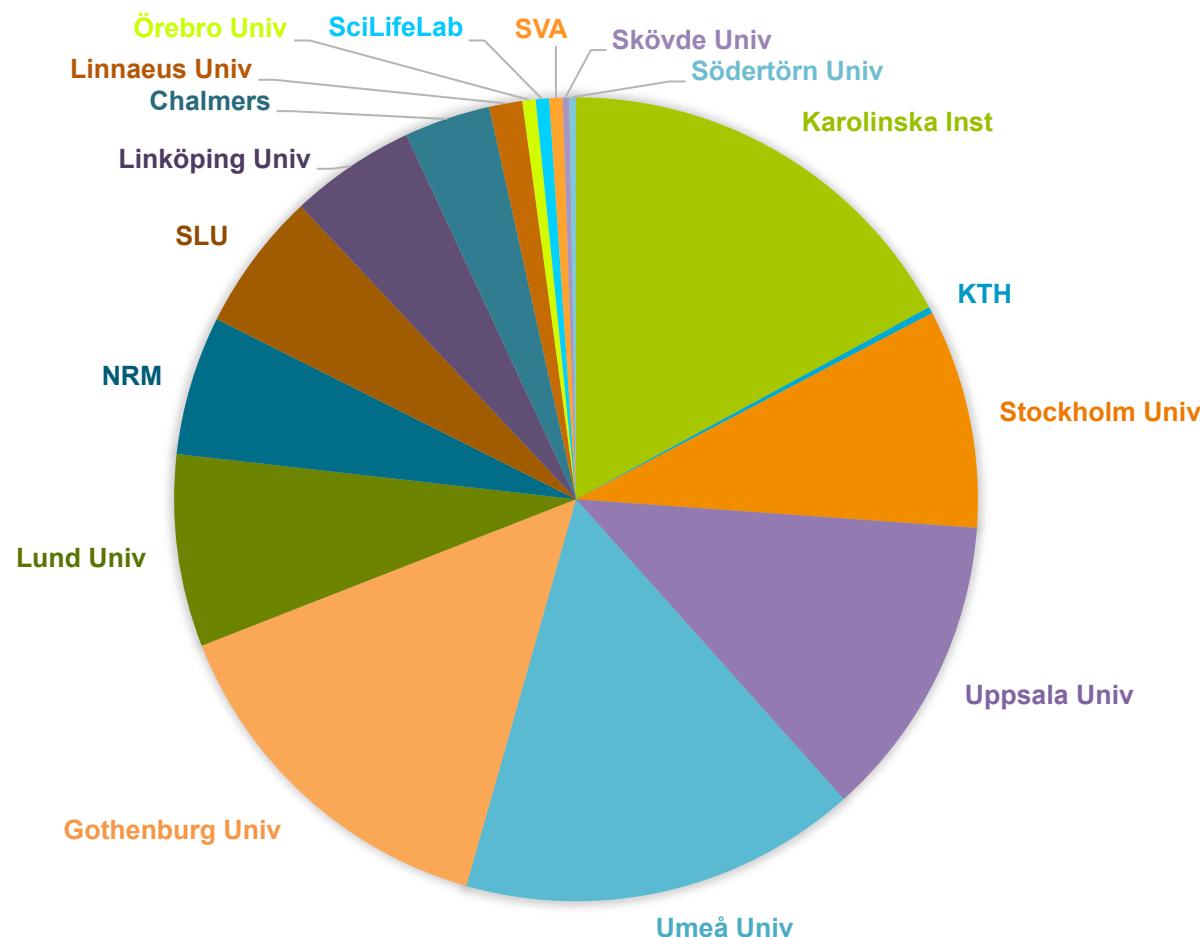
Support track for integrative projects

First call Feb 2016; First few projects initiated

Involves extensive integration of data



Geographical Distribution of Projects 2015





Tools and infrastructure

<https://docs.google.com/spreadsheets/d/1PrehKn2eb0ymfaFtCfvbLrOSKtpTL3qLcWZ2YwoXOIU/edit#gid=0>

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						Prioritization date: 2016-09-02					
NOW						NEXT					
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	
2	SweFreq	x	Adam A	NJ	<1	2					
3	ePouta integration pilot	x	Antti P	JH?	1	3					
4	microMosler		NJ?	JH	1?	4					
5	WGS structural variation WF		BN	PO	1	5					
6	Falcon on milou		HL	JB	<1	6					
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	BPn?	JV?	4?						
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

Tools and development projects
needs to be much more visible!
Work in progress....

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.

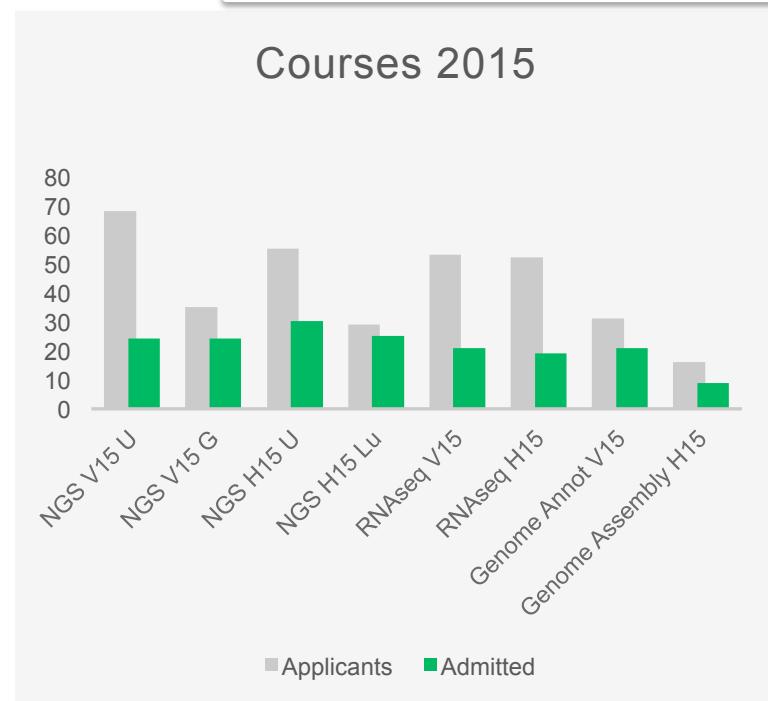
chr22:46,615,730-46,616,030 301 bp
46,615,750 46,615,800 46,615,850 46,615,900 46,615,950 46,616,000
hide labels - +

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



- 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.

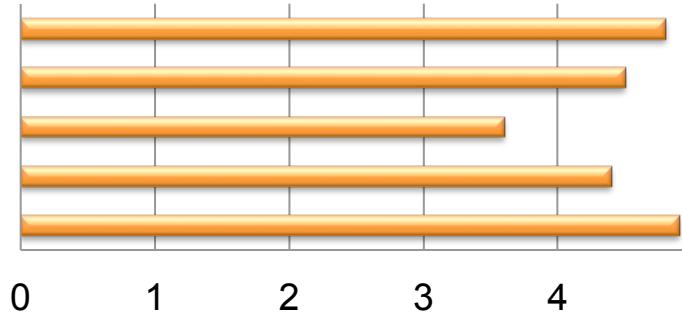
www.scilifelab.se/education/mentorship/the-swedish-bioinformatics-advisory-program/

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

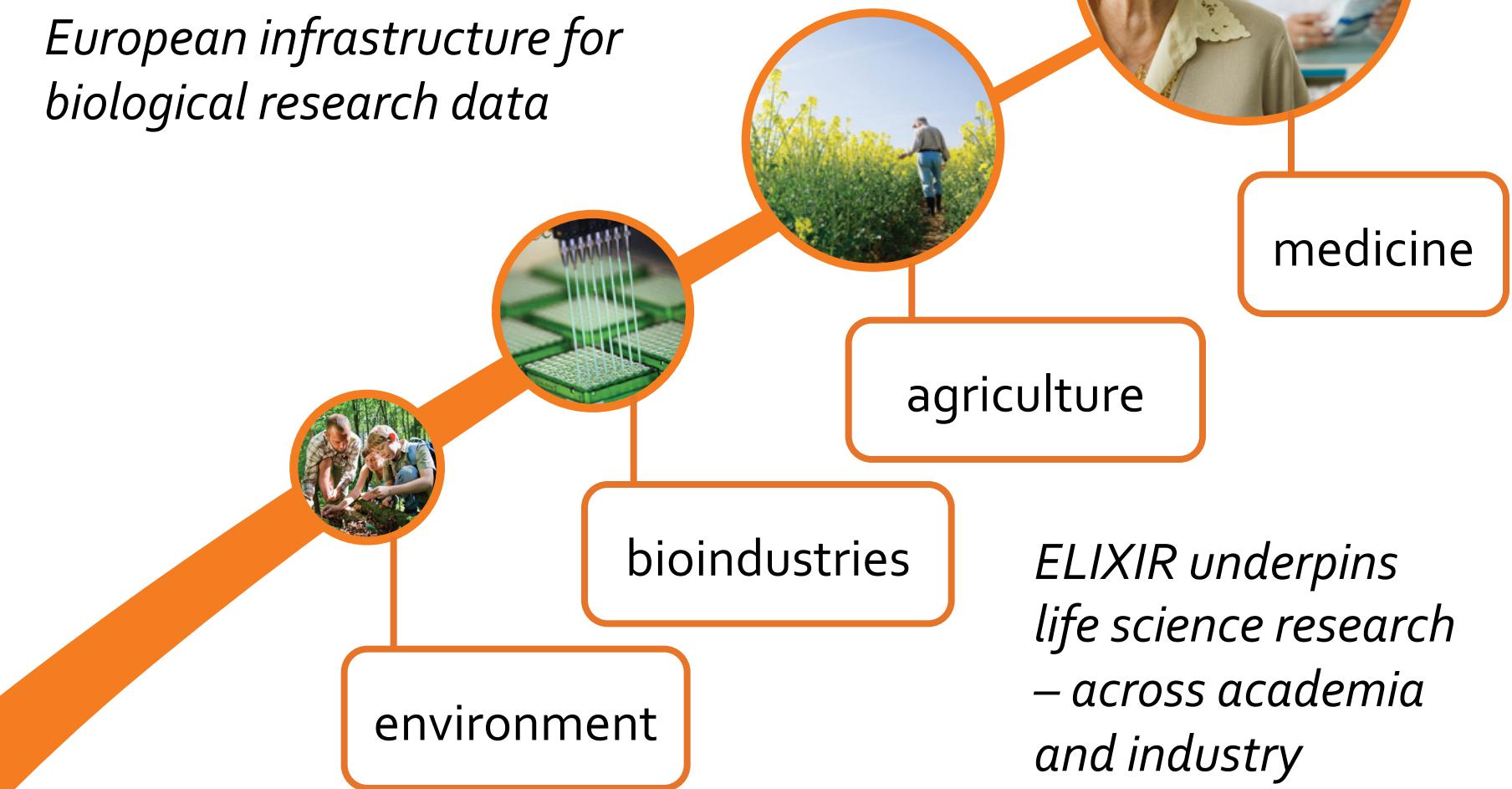


Elixir

Why ELIXIR?

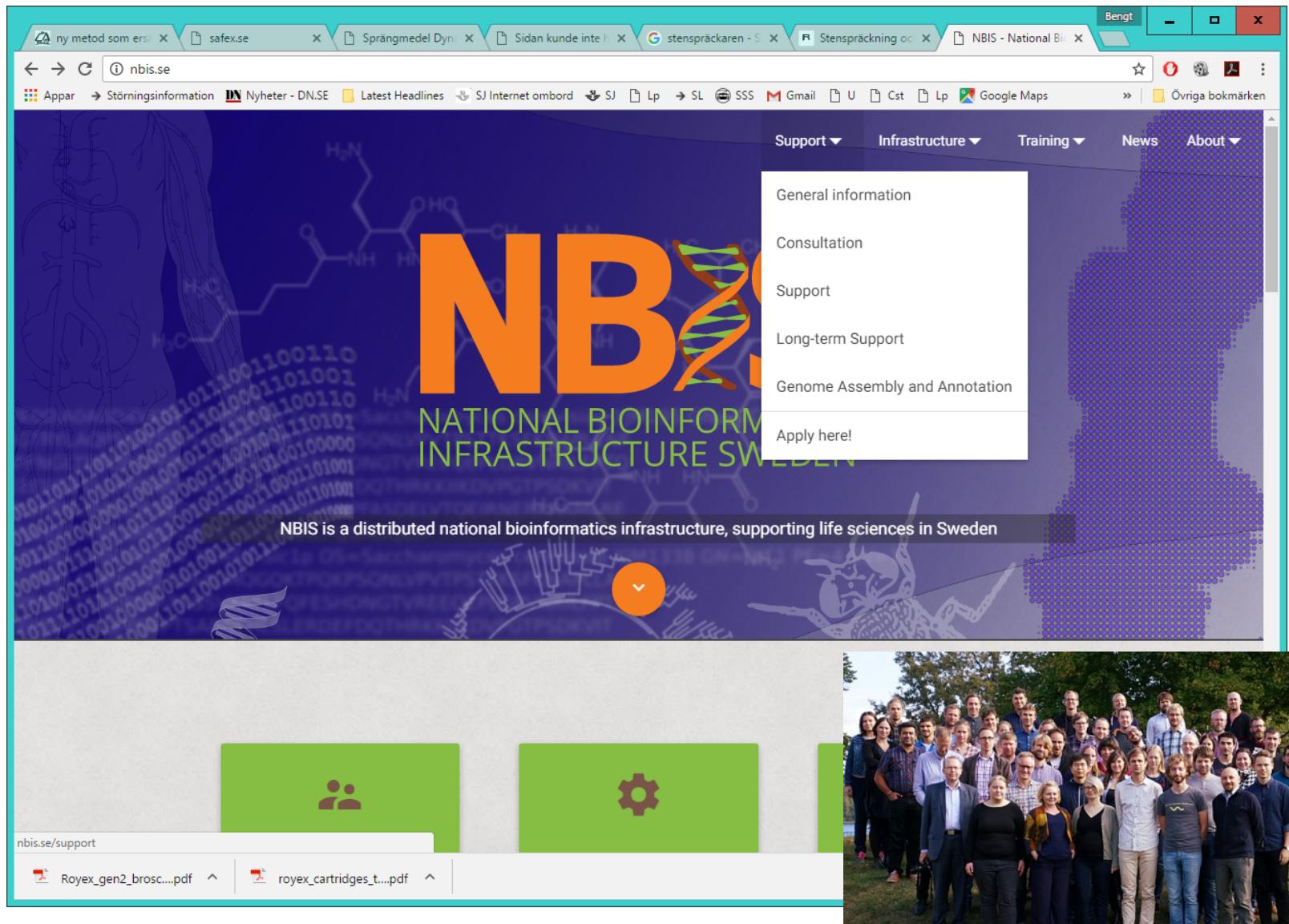
- Creating a robust infrastructure for biological information is a **bigger** task than any individual organisation or nation can take on alone
- These are issues of such complexity that no single institution or country can tackle alone
- Biology has by far the largest research community:
 - ~3 million 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*





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 footer contains links to "nbis.se/support" and PDF files for "Royex_gen2_brosc....pdf" and "royex_cartridges_t....pdf". To the right of the footer is a large 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

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

nbis.se/support Royex_gen2_brosc....pdf royex_cartridges_t....pdf

