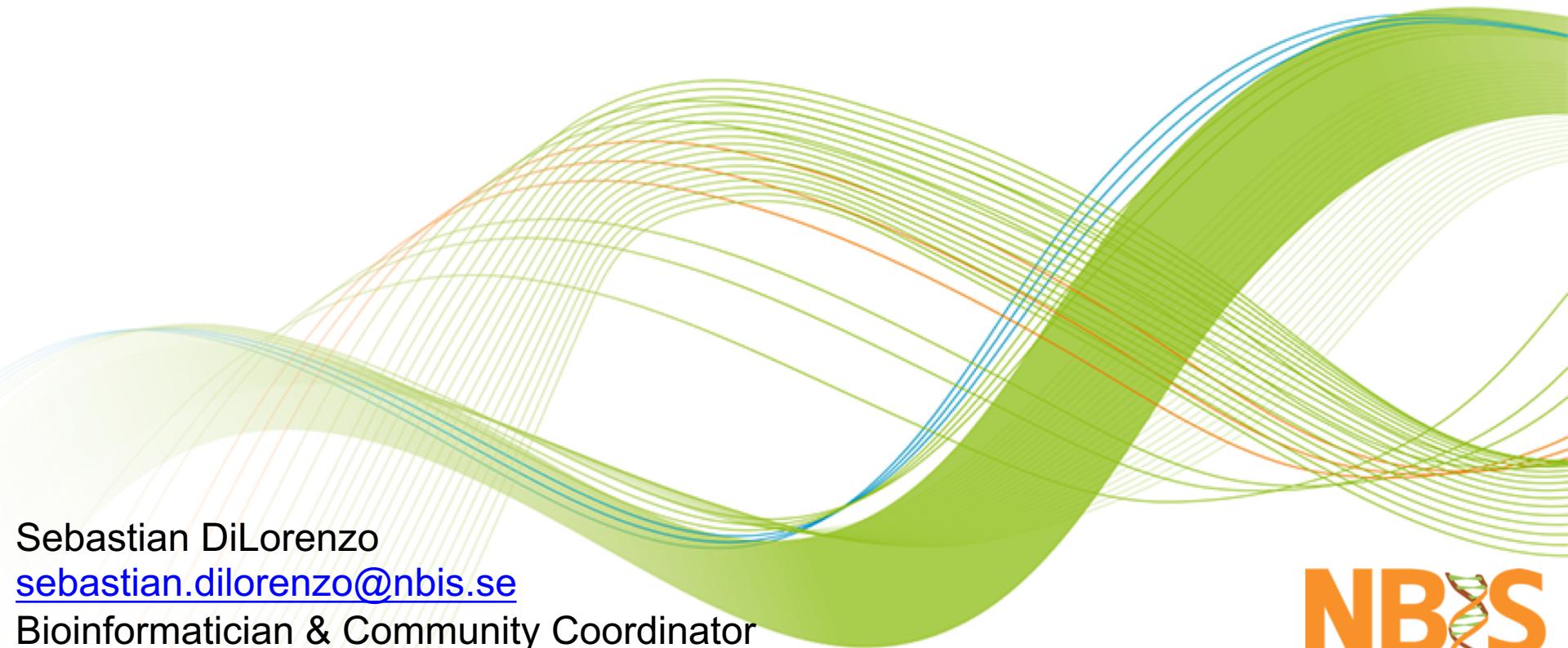




National Bioinformatics Infrastructure Sweden



Sebastian DiLorenzo

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Bioinformatician & Community Coordinator



SciLifeLab platforms

SciLifeLab

SciLifeLab national service

National
Genomics
Infrastructure

Clinical
Diagnostics

Single-cell
Biology

National
Bioinformatics
Infrastructure
Sweden

Bengt Persson



NBIS
NATIONAL BIOINFORMATICS
INFRASTRUCTURE SWEDEN



VR

SNIC



Computer
resources
free for
Swedish
researchers

NBIS - Support, Tools and Training

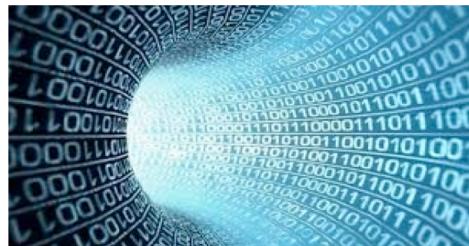
Future storage infrastructure



400 consultations



>1600 compute projects
>800 software and databases



Data publishing and open science
Secure sharing of sensitive data



Efficient tools and
workflows



30 training events
500 PhD/post-docs



200 research projects

SciLifeLab Bioinformatics (NBIS) and Genomics (NGI) were both top-ranked in the most recent VR evaluation.

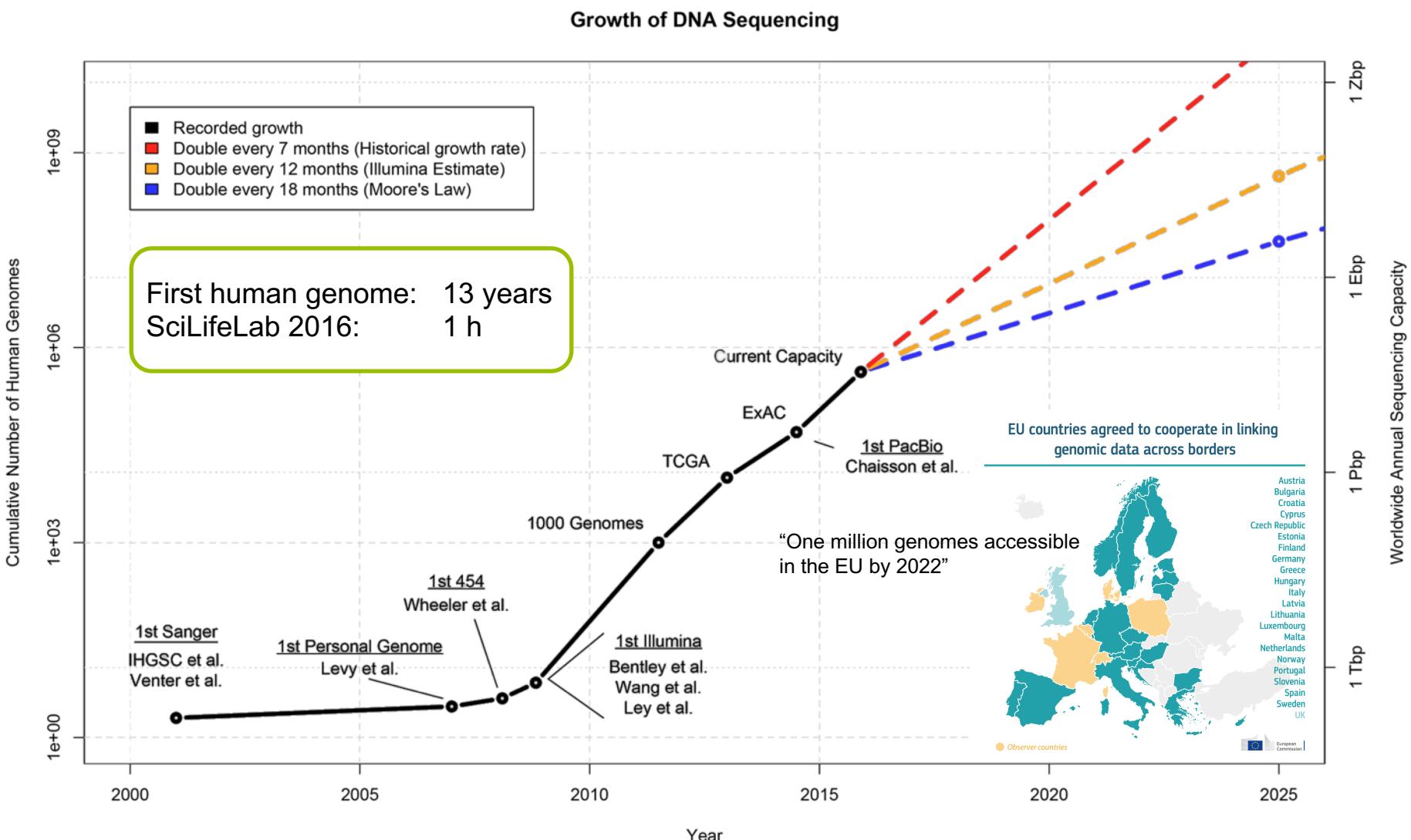
“NBIS is probably the largest genuinely national and fully established bioinformatics infrastructure in Europe.”

“[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.”

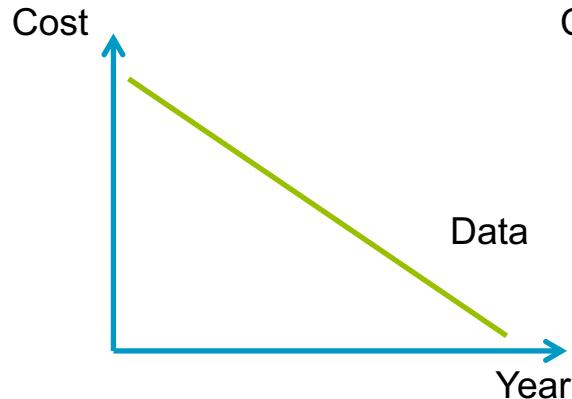


Scientific impact: 7/7 (“Outstanding”)
Overall score: 7/7 (“Outstanding”)

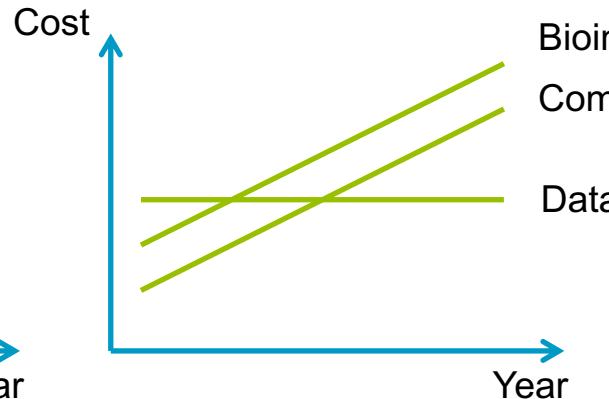
Data growth



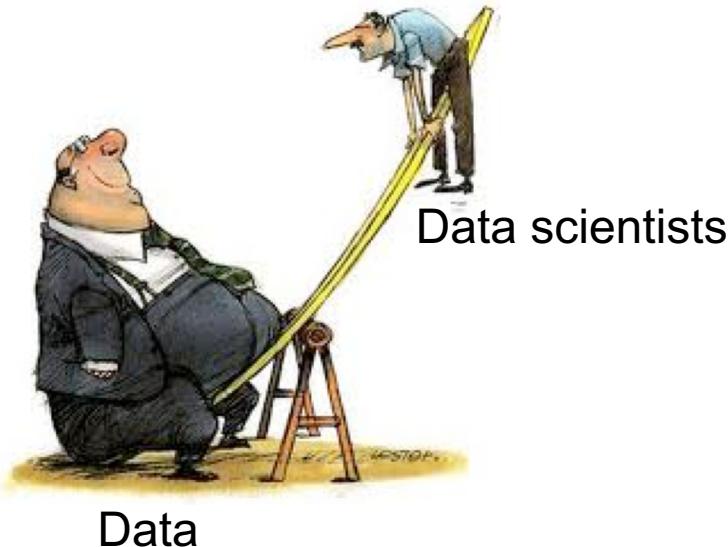
Production is cheap, analysis is not



“Per base”



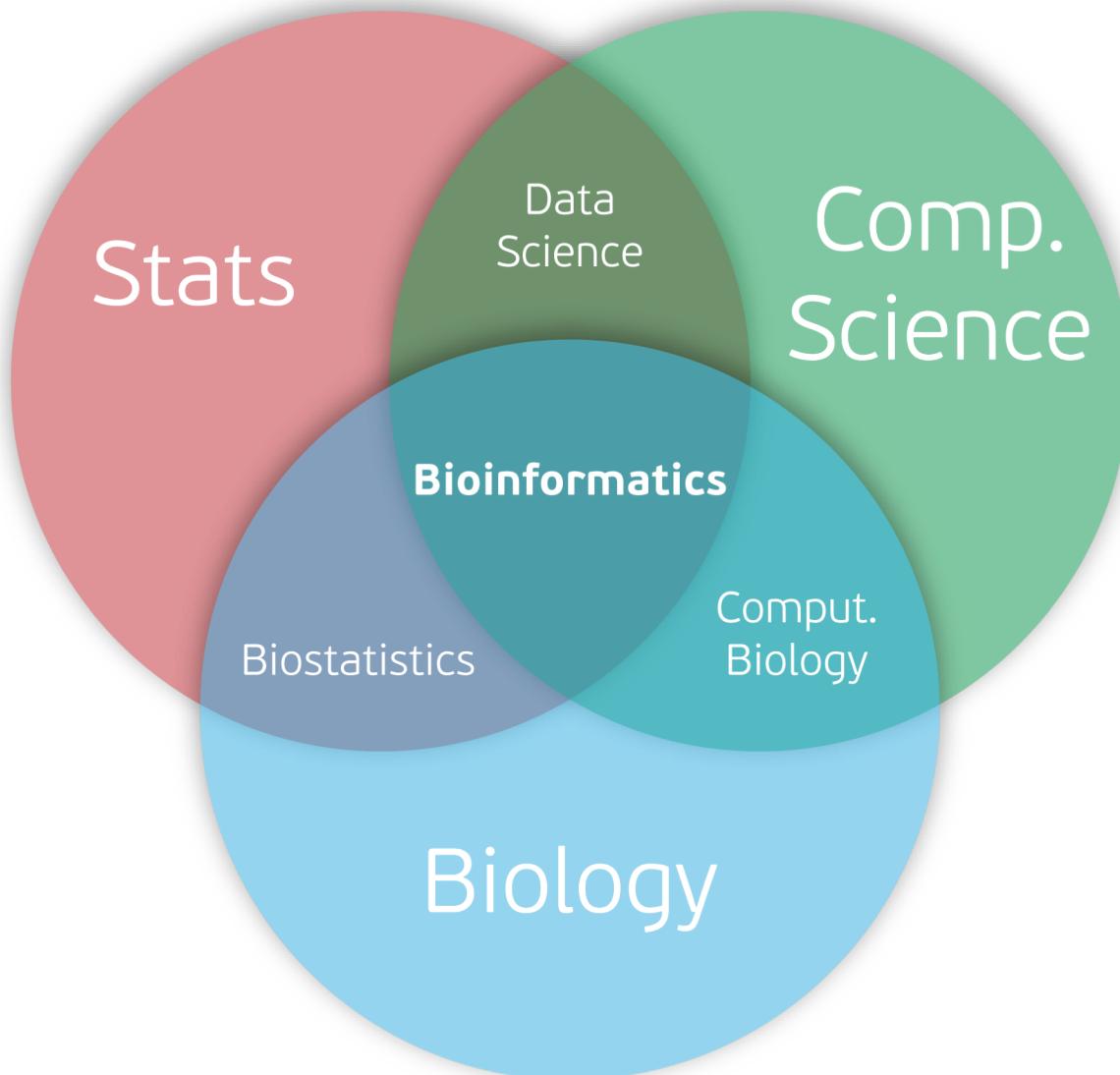
“Per project”



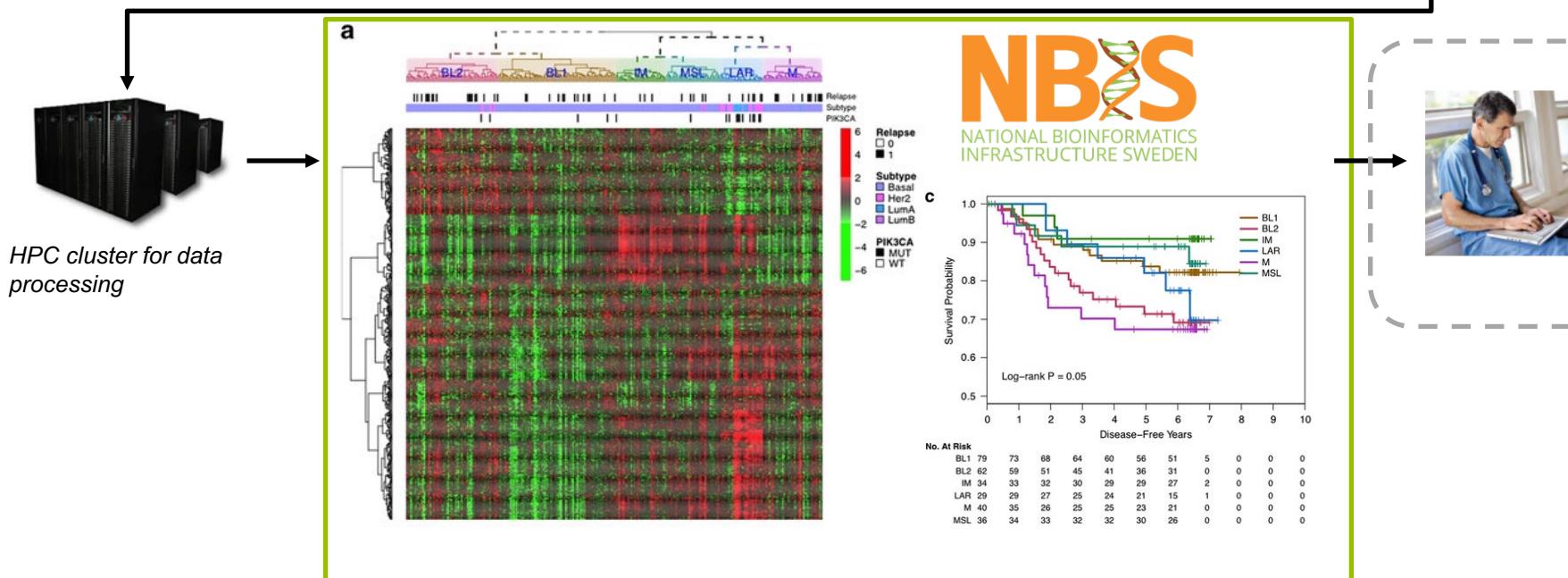
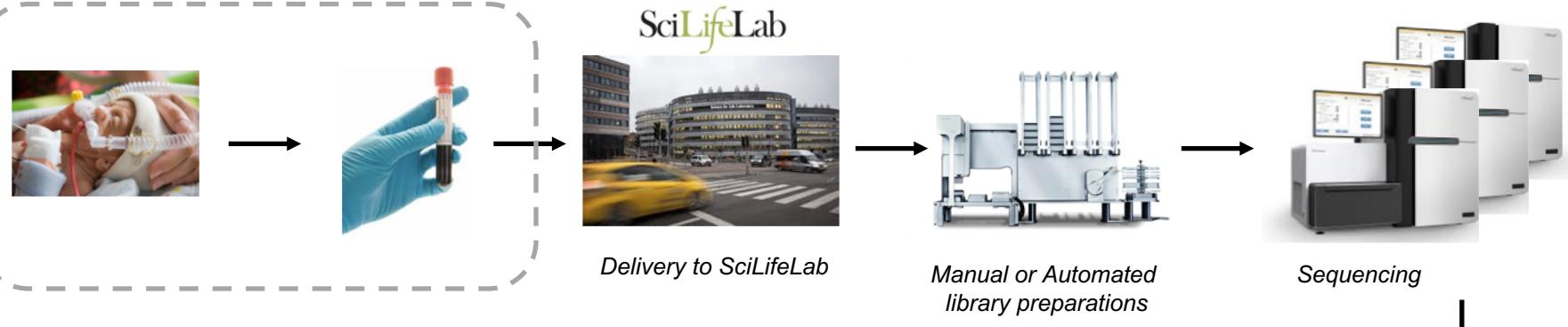
NBIS role

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.

What is bioinformatics?



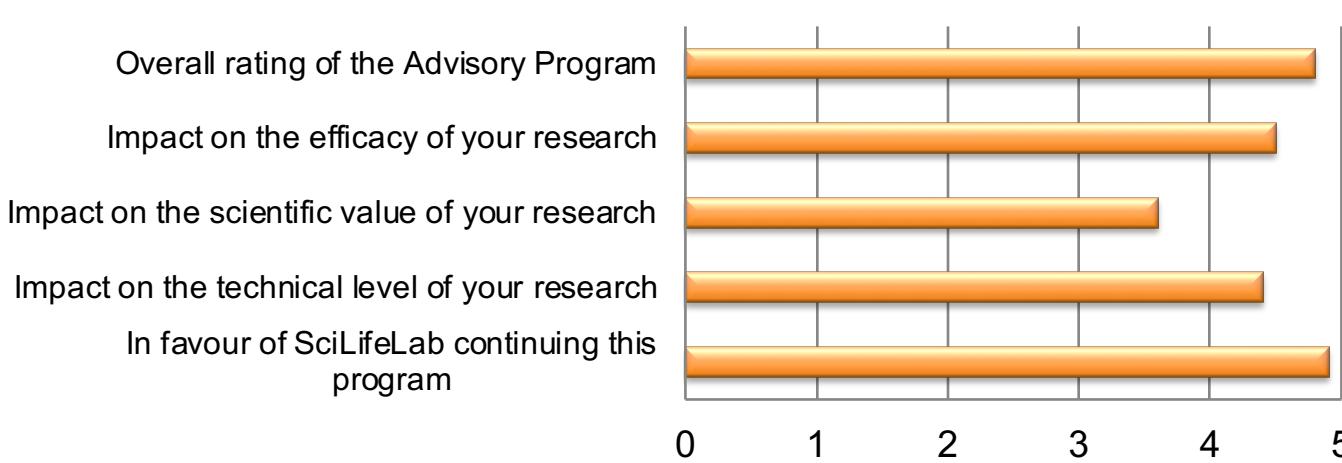
Example support project



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/

The Swedish Bioinformatics Advisory Program Student evaluation



Advanced R for Bioinformatics Summer School

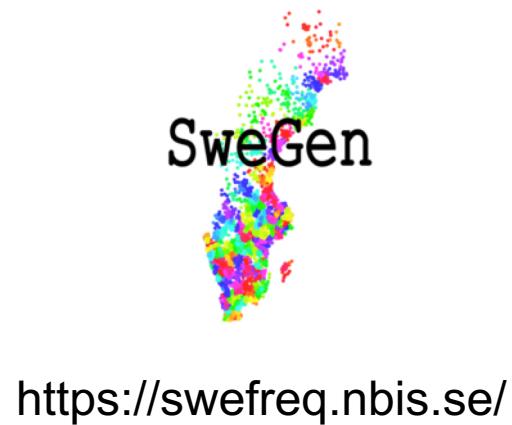
- R programming
- Some focus on bioinformatics related topics
- International



SweGen Variant Frequency Database

- 950 twin registry + 50 Northern Sweden
- WGS (30X)
- ExAC browser interface
- Data Beacon
- Full SNP frequency table download

Variant: 22:46615880 T / C



<https://swefreq.nbis.se/>

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 ⓘ
intron • PPARA - ENST00000434345
non coding transcript exon • PPARA - ENST00000493286

Population Frequencies

Population	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
SweGen	7	2000	0	0.0035
Total	7	2000	0	0.0035

Note: This list may not include additional transcripts in the same gene that the variant does not overlap.

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 ⌂ +

The visualization shows a genomic track for the region chr22:46,615,730-46,616,030, which is 301 bp long. The x-axis is labeled with genomic coordinates: 46,615,750, 46,615,800, 46,615,850, 46,615,900, 46,615,950, and 46,616,000. Below the track, there are several colored bars representing different sequencing lanes or samples. A 'hide labels' button and a zoom-in icon are located at the top right of the visualization area.

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



About European Portal Support Services

Submit data

en | sv

Genomics & transcriptomics Protein data Imaging data Compound and target data Health data

Accelerating research through data sharing

The Swedish COVID-19 Data Portal provides information, guidelines, tools and services to support researchers in utilising Swedish and European infrastructures for data sharing, in particular the [European COVID-19 Data Portal](#).

För information på svenska, [klicka här](#)

Data Types

Click to expand a data type and see available resources.

- Genomics & transcriptomics ▾
- Protein data ▾
- Imaging data ▾
- Compound and target data ▾
- Health data ▾

Support Services

General support services for Swedish COVID-19 researchers.

- Bioinformatics
- General data repository
- Resources
- Text and literature
- Submit data

Contact the Swedish COVID-19 Data Portal: datacentre@scilifelab.se

For bioinformatics support, please contact: <https://nbis.se/support/>

DATA TYPES

[Genomics & transcriptomics](#)
[Protein data](#)
[Imaging data](#)
[Compound and target data](#)
[Health data](#)

SUPPORT SERVICES

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SciLifeLab



<https://covid19dataportal.se/>

Thanks for listening!



NBIS
NATIONAL BIOINFORMATICS
INFRASTRUCTURE SWEDEN