



Proteiner, pixlar och populationer: Arbetssätt och lärdomar från att stötta Svenska bidrag till globala biodata-resurser

NBIS - National Bioinformatics Infrastructure Sweden

Wolmar Nyberg Åkerström, NBIS, SciLifeLab 2025-12-16

SciLifeLab

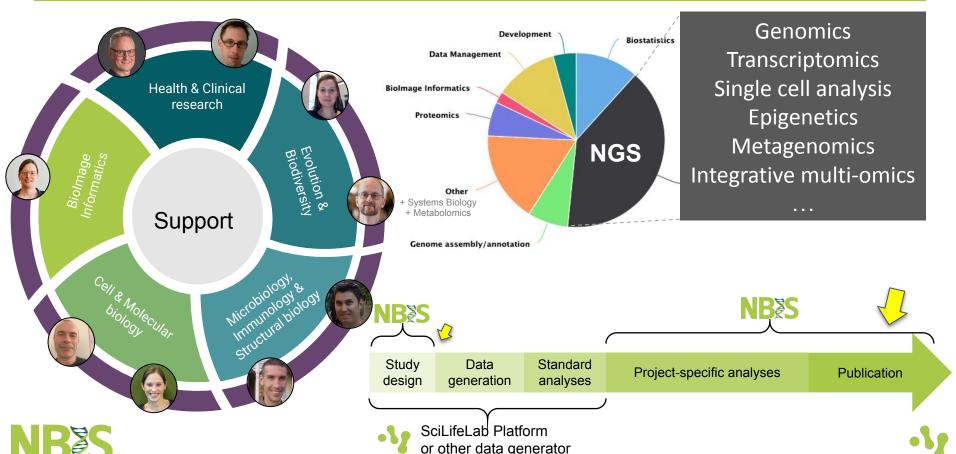


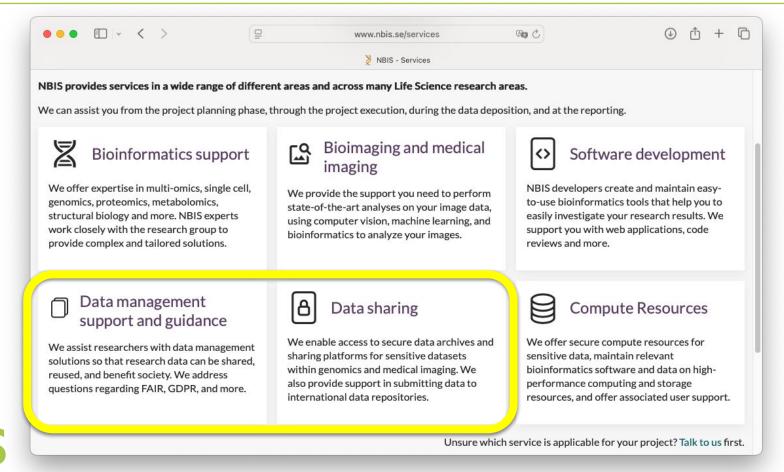


NBIS research area teams & subjects



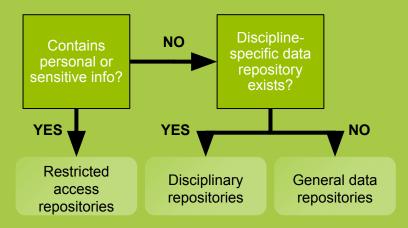
Support













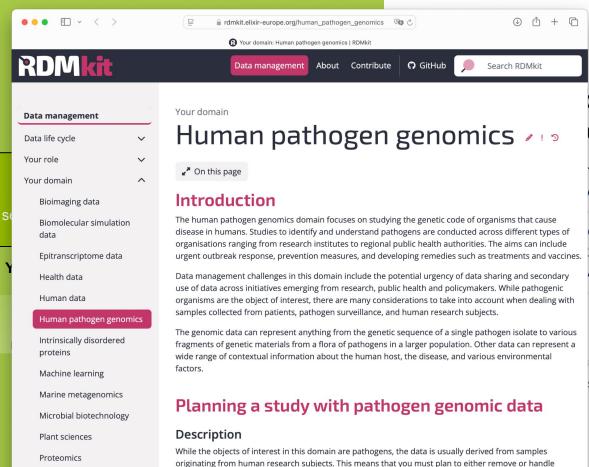


Data submission to public repositories

Making the data underlying your research publicly available to others is a fundamental part of a FAIR research process. When publicly available and appropriately described, data can be re-used by yourself as well as others. Domain-specific public repositories offer the most direct routes to making your data FAIR. This workshop will give you the why, where and how of data sharing via repository submission, including hands-on exercise. No prior knowledge is required in order to attend this workshop.

Learning outcomes:

- · Know the benefits of data sharing
- Know how to find a suitable repository for different types of data
- Have experience of a repository submission



human data during your study.

Rare disease data

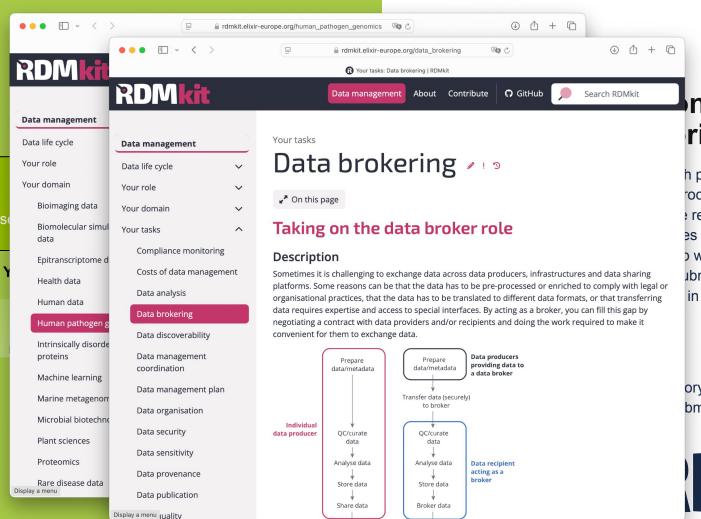


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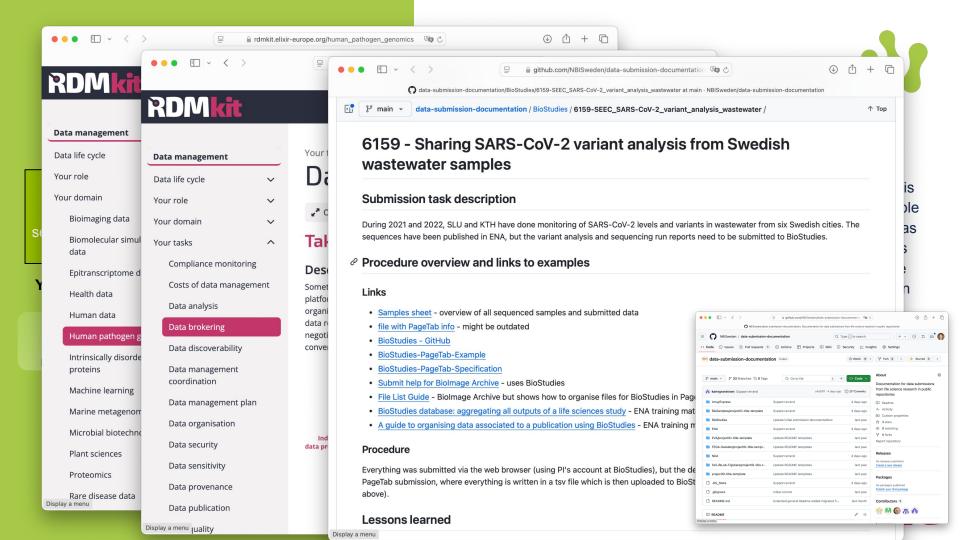


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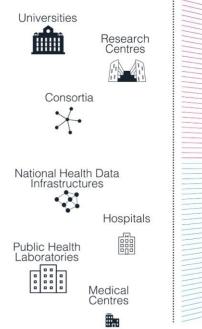
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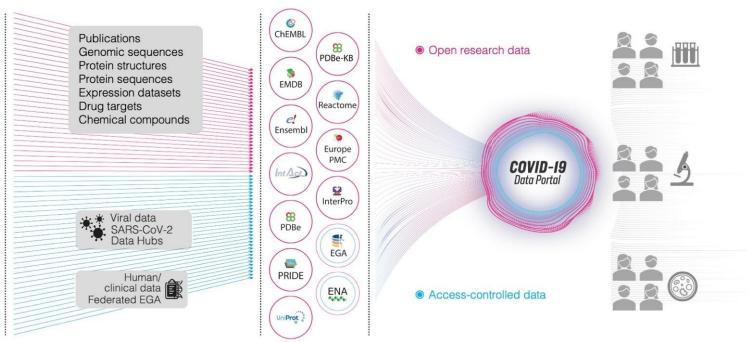
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Connected data resources

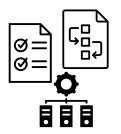








Data and metadata in context (I)











Study & data design

Sampling & specimen collection

Sample preparation

Sample analysis & data generation

Data processing to prepare inputs for analysis

Data analysis

Communicating results

Procedures

data protection, ethics permit, infrastructure, standards, protocols, data dictionaries, data access, ...

Biosamples and instruments

populations (statistical) and inclusion criteria, physical processing steps, working storage conditions, long-term storage location, sample quality assessment, sample annotations, reagents, instruments, kits, ...

Data and computational workflows

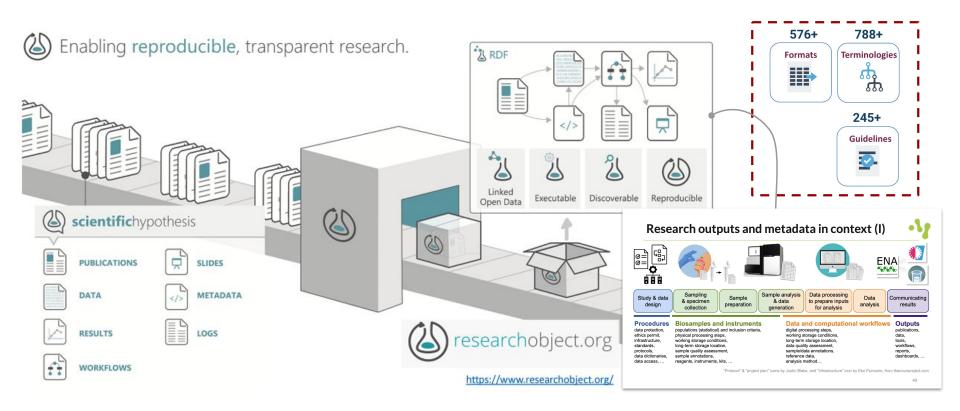
digital processing steps, working storage conditions, long-term storage location, data quality assessment, sample/data annotations, reference data, analysis method...

Outputs

publications, data, tools, workflows, reports, dashboards, ...



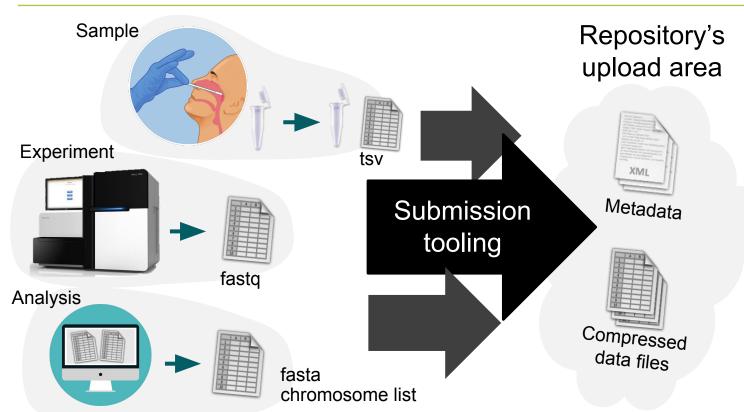
Data and metadata in context (II)







Depositing data to a repository







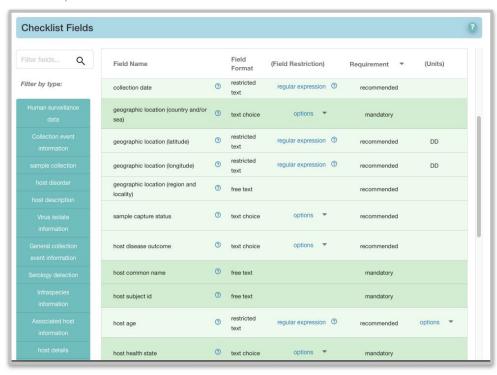






Data repository conventions example

https://www.ebi.ac.uk/ena/browser/view/ERC000033



https://ena-docs.readthedocs.io/en/latest/submit/fileprep/reads.html

Fastq format

We recommend that read data is either submitted in BAM or CRAM format. However, single and paired reads are accepted as Fastq files that meet the following the requirements:

- · Quality scores must be in Phred scale.
- Both ASCII and space delimitered decimal encoding of quality scores are supported. We will automatically detect the Phred quality offset of either 33 or 64.
- · No technical reads (e.g. adapters, linkers, barcodes, primers) are allowed.
- Single reads must be submitted using a single Fastq file and can be submitted with or without read names.
- Paired reads must be submitted using two Fastq files.
- . The first line for each read must start with '@'.
- . The base calls and quality scores must be separated by a line starting with '+'.
- Paired read names must either use Casava 1.8 read names (regular expression:
 \@(.+)(+\\\\\)([0-9]+): [\m): [0-9]*[02468] (\\$|:.*\\$) or must end with |/1 or |/2 optionally

```
followed by a space and a comment.
```

- Read names must not exceed a length of 256 characters.
- · The Fastq files must be compressed using gzip or bzip2.
- The regular expression for bases is "^([ACGTNactgn.]*?)\$"

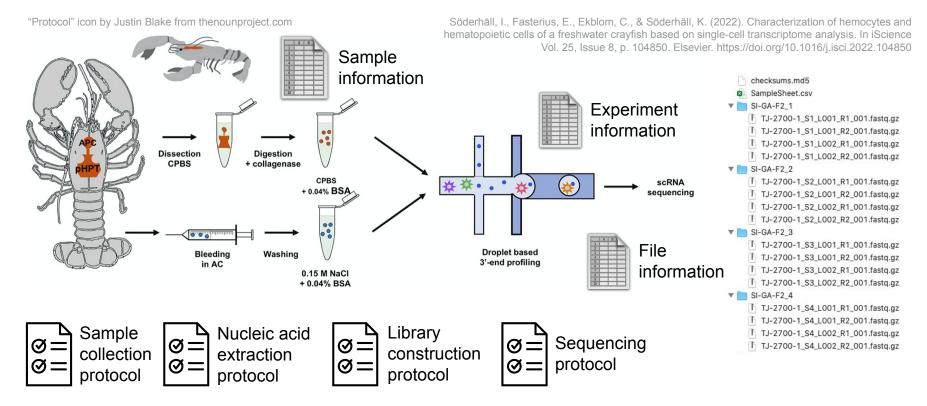
Example of Fastq file containing paired reads (prior to Casava 1.8):

```
@read_name/1
GATTIFGGGTTCAAAGCAGTATCGATCAAATAGTAAATCCATTTGTTCAACTCACAGTTT
+
!'**((((****+))%%++)(%%).1***-+*''))**55CCF>>>>>CCCCCCC65
@read_name/2
GATTIFGGGGTTCAAAGCAGTATCGATCAAATAGTAAATCCATTTGTTCAACTCACAGTTT
+
!''**((((****))%%++)(%%).1***-+*''))**55CCF>>>>>CCCCCCC65
...
```





Single-cell RNA sequencing example







Sample information as (meta)data

ENA Checklist: ERC00001 – ENA default sample checklist

OLS / Experimental Factor Ontology

EFO

Population	Organism	Pacifastacus leniusculus
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Genotype wild type genotype
Geographical location Sweden, Lake Erken

Individual laboratory aquarium since Sep 2020 Growth condition

Sampling date
Developmental stage
Body weight 020-11-06

adult

35

male

Specimen Organism part hematopoietic system

Sample Cell type hemocyte

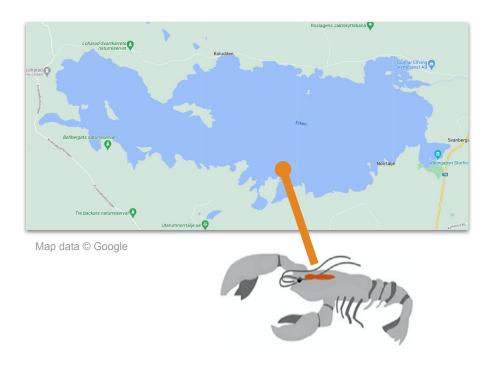




Sample collection protocols

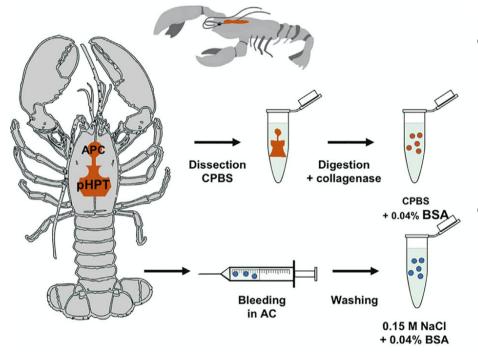
- Obtain freshwater crayfish (adult males) from lake Erken, Sweden (59.8 N 18.6 E)
- Maintain in the crayfish facility in running tap water,10-12°C, 12:12 light:dark cycle...
- Feed once a week

Map data © Google. Crayfish derived from Illustration in Söderhäll et al. (2022).









- Dissect and digest into single cells by incubation in 300 µl of 0.1% collagenase ... at room temperature for 20 min on a rotating plate ... then filtered through a 40 mm cell strainer
 - Pool isolated cells from four animals for scRNA-seq





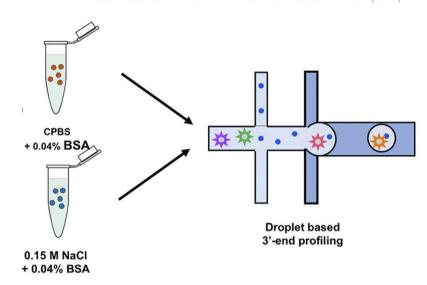
Library construction protocol

- Prepare sequencing libraries using Chromium Single Cell 3' reagent kit v3 (cat# 1000075/1000073/120262, 10xGenomics)
- According to the manufacturer's protocol

CG000183
Single Cell 3' Reagent Kit
User Guide, v3 chemistry,
10xGenomics



Protocol illustration derived from Illustration in Söderhäll et al. (2022).







Sequencing protocol

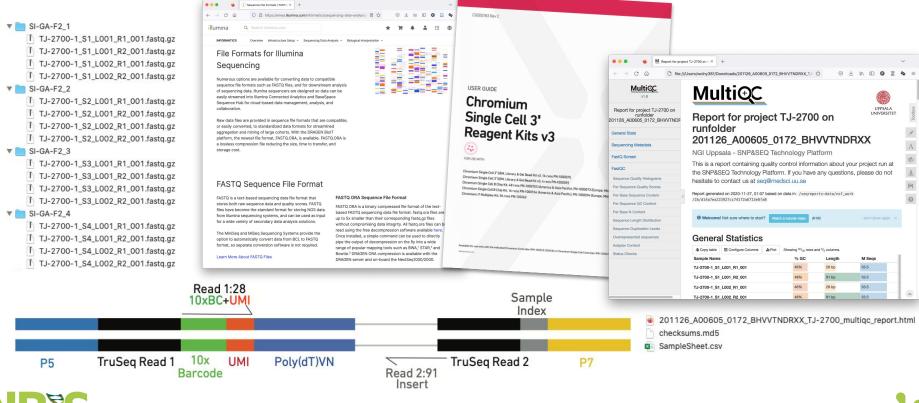


- 28+8+0+91 bp read length
- NovaSeq 6000 system
- SP flowcell
- v1 sequencing chemistry
- Include a sequencing library for the phage PhiX as 1% spike-in in the sequencing run





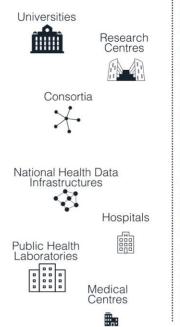
Files Delivered from Sequencing

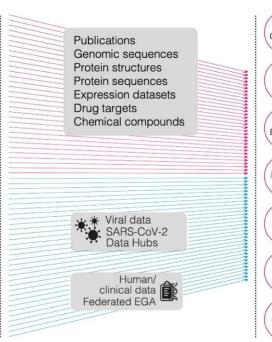




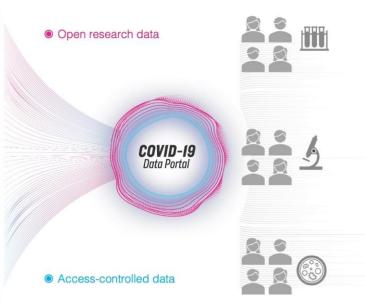


Connected data resources



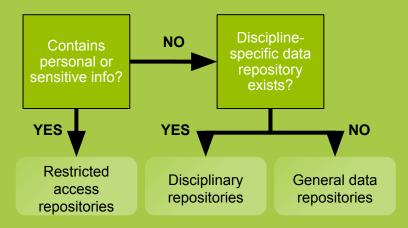














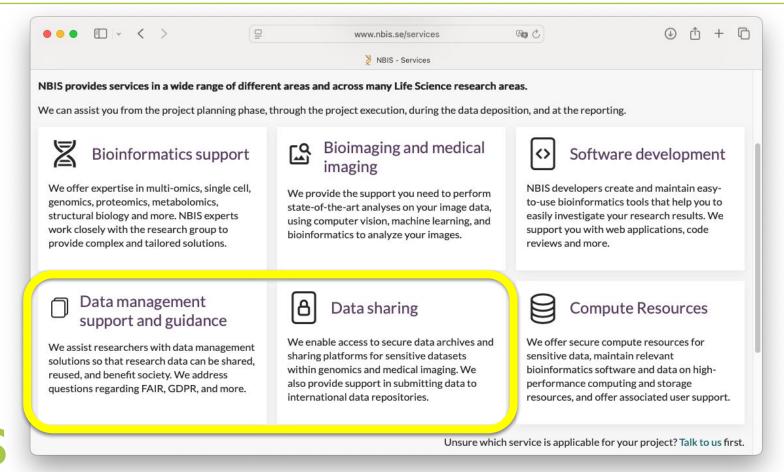


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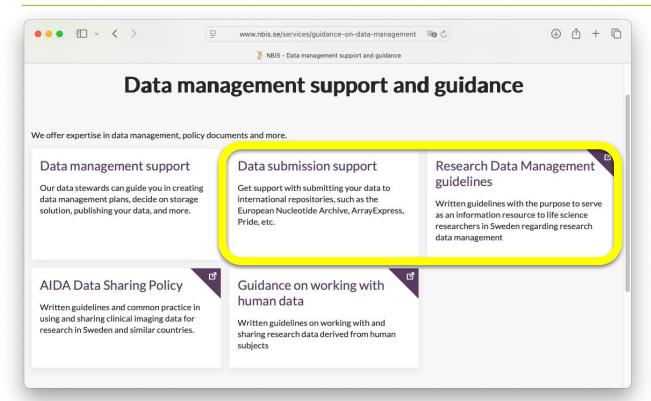
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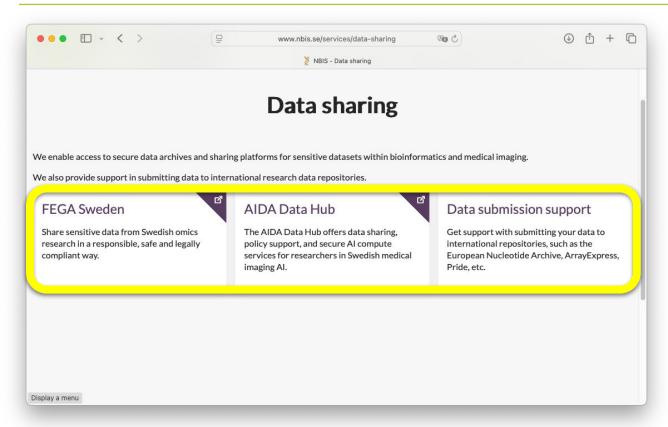


We assist researchers with data management solutions so that research data can be shared, reused, and benefit society. We address questions regarding FAIR, GDPR, and more.











Data sharing

We enable access to secure data archives and sharing platforms for sensitive datasets within genomics and medical imaging. We also provide support in submitting data to international data repositories.



