

# National Bioinformatics Infrastructure Sweden

Strategic Plan 2025–2029

Including Work Plan for 2025

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## **Executive summary**

NBIS will during 2025–2029 continue our activities in providing excellence in bioinformatics support to researchers in Sweden, enabling world-class life science by offering support, infrastructure and advanced training. NBIS forms the Swedish node in ELIXIR (the European infrastructure for biological information). The organisational structure allows for changes in support needs over time as new techniques are developed and utilised. In 2023, NBIS celebrated 15 years of providing bioinformatics support.

NBIS is well connected to several national data-producing infrastructures, to avoid bottle-necks in data analysis and to ensure maximal impact of Swedish research data from a broad range of technology areas. NBIS constitutes the Bioinformatics platform at SciLifeLab, a national resource centre for molecular biosciences, and SciLifeLab funding to NBIS ensures good integration with data-generating services and training efforts at SciLifeLab, and adds improved flexibility for NBIS to meet new user demands and establish new service areas.

NBIS supports both research groups not having their own bioinformatics resources and large established research groups with their own bioinformaticians needing specialised expertise. As scientists develop greater competence in bioinformatics, NBIS will successively focus on more advanced expertise, but still provide broad support when needed.

A fundamental part of NBIS is the formation of a sustainable bioinformatics infrastructure for life sciences, consisting of access to tools and data. The NBIS infrastructure is typically constructed as domain-specific supporting layers utilising resources from e-infrastructure providers such as NAISS and SUNET.

With the new SciLifeLab initiative IDS (Integrative Data Services), NBIS will together with SciLifeLab Data Centre be central in the efforts to improve the data handling at all SciLifeLab platforms to benefit all users. NBIS will lead the planned SciLifeLab-wide project consultations to be held at the beginning of a new project. NBIS will also engage in the initial platform meetings with users at project start in order to provide bioinformatics competence and to facilitate planning of downstream bioinformatics analyses. NBIS will also be engaged in data management.

For data management, our vision is that Swedish life science researchers should apply good data management practices so that the research outputs produced are available to the global research community, and to society at large, according to the principles of Open Science, Reproducible Research, and FAIR. NBIS provides data management support, training in efficient data management practices and collaborates with relevant national and international stakeholders.

NBIS has since long been engaged in building infrastructure for sensitive human data. NBIS has established the Swedish node in the international infrastructure Federated European Genome-phenome Archive (FEGA), and NBIS participates in the implementation project GDI (Genomic Data Infrastructure) 2022–2027, where we as ELIXIR-SE together with ELIXIR-FI lead the infrastructure work. GDI and FEGA are instrumental to enable the European 1+ Million Genomes project (1+MG). Furthermore, NBIS is engaged in the EU IMI project Bigpicture 2021–2027 aiming at creation of an infrastructure for digital pathology. NBIS contributes with expertise in systems development and with bioimage informatics via AIDA Data Hub. From 2023, NBIS also engages in European Federation for Cancer Images (EUCAIM), especially regarding interoperability with GDI and Bigpicture. NBIS works closely with GMS (Genomic Medicine Sweden) in several of these human data projects.

Our systems development unit provides access to professional and usable software tools and makes them usable to the research community. With recent advances in computational resources, Deep Learning and Artificial Intelligence (AI) has become highly relevant for life sciences, and NBIS will provide systems development support to users creating AI-based tools. Further, NBIS has since long supported users in sharing data in a FAIR fashion, which is a prerequisite for data to be used in AI.

NBIS will continue to develop components in <u>Human Protein Atlas</u> (HPA), and maintain the <u>Metabolic Atlas</u> (MA).

NBIS support for computational resources ensures the life science community's needs for support and training on national e-infrastructures for compute and storage. We build and maintain a nationally available computational environment for bioinformatics, pushing the envelope on usability.

As a national infrastructure it is important for NBIS to be visible and connected to local bioinformatics activities, and NBIS therefore has assigned site coordinators, and we work in close collaboration with local resources, including recently established research data offices.

NBIS support for bioinformatics projects enables world-class life science research and maximises scientific and societal impact of collected data, and is one of our major activities. NBIS offers nation-wide support, providing expertise in multiple areas within bioinformatics. Resources are prioritised, giving high priority to scientific excellence. To cater for different user needs, NBIS provides well-defined support tracks, ranging from consultations via user fee track and peer review track to partner projects.

NBIS training offers high quality training programmes aimed at the Swedish Life Science research community that provide a standard to follow when handling and analysing bioinformatics data. The training follows up-to-date best practices for effective teaching, using Open and FAIR training materials. Moreover, our training is inclusive and meets the needs of the research community in terms of accessibility.

Most information about NBIS is spread via our website (<a href="https://nbis.se">https://nbis.se</a>) but we also conduct a number of other outreach activities that have proven important to inform the scientific community about the support that NBIS can provide, making bioinformatics easily accessible for life science researchers.

Internationally, NBIS forms the Swedish node in the <u>European infrastructure for biological information ELIXIR</u>. NBIS is providing the Human Protein Atlas as an ELIXIR service. We are also engaged in multiple EU projects related to ELIXIR. Furthermore, NBIS plans to continue our activities in Nordic collaborations.

#### Introduction

NBIS (National Bioinformatics Infrastructure Sweden) is a distributed national research infrastructure with support from Science for Life Laboratory (SciLifeLab), the Swedish Research Council, Knut and Alice Wallenberg Foundation, and Swedish universities. NBIS is hosted by Uppsala University. NBIS constitutes the SciLifeLab Bioinformatics Platform.

NBIS is formed by 11 partners: Uppsala universitet (hosting NBIS), Chalmers tekniska högskola AB, Göteborgs universitet, Karolinska Institutet, Kungl. Tekniska högskolan, Linköpings universitet, Lunds universitet, Naturhistoriska riksmuseet, Stockholms universitet, Sveriges lantbruksuniversitet, and Umeå universitet

The NBIS Strategic Plan serves to formulate the long-term general goals for NBIS. It is decided by the NBIS Board (Styrgrupp). The current version is covering the period from 2025 to 2029 and was approved by the NBIS Board on 20 May 2025. The strategic plan will be revised at least annually, considering input from NBIS partners, the International Advisory Board and the Reference Group. The plan describes the major activities foreseen within the areas of Infrastructure, Support, Outreach, Training, Organisation and International.

The detailed goals to be achieved during 2025 are highlighted in green boxes.

#### **Aim**

NBIS provides excellence in bioinformatics support to researchers in Sweden, enabling world-class life science by offering support, infrastructure and advanced training. NBIS constitutes the SciLifeLab Bioinformatics platform and forms the Swedish node in ELIXIR (the European infrastructure for biological information). The organisational structure allows for changes in support needs over time as new techniques are developed and utilised.

NBIS supports both research groups not having their own bioinformatics resources and large established research groups with their own bioinformaticians needing specialised expertise. As life scientists get successively more educated in bioinformatics, the scope of NBIS will be shifted towards a focus on more advanced expertise.

## **Vision and Mission**

Enable world-class life science research and maximise scientific and societal impact of collected data by:

- Providing expert knowledge, innovative data integration, advanced training, efficient data publication for open science, and access to high-performance data analysis methods
- Coordinating bioinformatics support within Sweden and making bioinformatics easily accessible for life science researchers
- Swiftly responding to changes in support needs as new techniques are developed and utilised
- Forming the Swedish ELIXIR node and participating in relevant international projects

## **Background**

NBIS (National Bioinformatics Infrastructure Sweden) with its extensive experience in large-scale data analysis is in a unique position to be a key driver for data-driven life science. NBIS was established in 2016 after fusion of four infrastructures (of which BILS dates back to 2008) to form a single point of contact for all users needing bioinformatics support. NBIS thereby provides support in a wide array of bioinformatics areas within one organisation, facilitating user contacts and enabling efficient service provision. NBIS provides excellence in bioinformatics support to researchers in Sweden, enabling world-class life science by offering expertise, infrastructure and training. The topics covered by our experts include genome assembly, genome annotation, evolutionary genomics, population genomics, comparative genomics, transcriptomics, proteomics, metabolomics, systems biology, single-cell biology, biostatistics, machine learning, systems development, data management, image analysis and multi-omics integration. Furthermore, NBIS forms the Swedish node in ELIXIR (the European infrastructure for biological information). The organisational structure allows for changes in support needs over time as new techniques are developed and utilised.

NBIS is a **distributed infrastructure** with staff placed at all major universities in Sweden, creating access points to facilitate contacts with our users. Nevertheless, all projects are **nationally prioritised** and allocated to achieve best possible efficiency and competence matching. All NBIS staff are placed in excellent scientific environments in order to keep up-to-date with front-line achievements in bioinformatics and to create a critical mass at each site.

NBIS supports both research groups without bioinformatics resources and large established research groups with their own bioinformaticians needing specialised expertise. As life scientists are getting successively more educated in bioinformatics, the scope of NBIS is continuously shifting towards a focus on very advanced expertise. The advantages of an infrastructure like NBIS are:

- **1. Guarantee of excellence.** NBIS can provide a multitude of expertise, which is difficult to match by a locally employed bioinformatician.
- **2. Long-term stability.** NBIS is a sustainable resource in contrast to PhD students or post-docs who need to move on when the time-limited position ends.
- **3.** Effective use of resources. The large scope of many projects puts an increased demand on the ability to analyse data effectively, using considerable programming/scripting skills, to automate much of the analysis. This is outside the competence of many researchers.
- **4. Reproducibility.** NBIS is a key driver to implement open science and reproducible research as routine practice in Swedish life science, with related data and source code from supported projects deposited in public repositories (human sensitive data subject to controlled access). Since 2018, NBIS provides regular courses in reproducible research for PhD students and post-docs.
- **5. Critical mass.** It is hard to reach a critical mass of bioinformaticians in an individual research group and therefore not possible to get synergies from the collective learning.
- **6. Expertise.** For research groups already having skilled bioinformaticians NBIS can, due to our competence breadth, provide complementary expertise.

#### **Overview of NBIS activities**

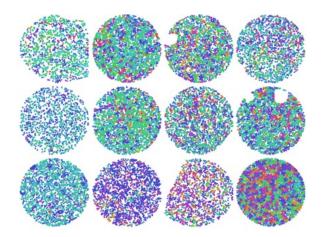
NBIS provides specialised competence having **experts** in multiple essential domains of large-scale analyses, but also has **generalists** able to integrate data from different omics areas. One of the strengths is our ability to assign **several experts from different domains** when required. NBIS keeps up-to-date with scientific developments, and for the period 2025–2029, we foresee several major developments:

- A continued need to meet new and rapidly transforming technology areas, such as singlecell and spatially resolved omics and new avenues in computational structural biology combining computational predictions with experimental data across a broad range of applications.
- Precision medicine, for which the formation of a European human genomics data infrastructure including analytical tools, access to medical imaging and AI (Artificial Intelligence)-based tools will be instrumental.
- Data-driven life science with needs of open data requiring professional data management and FAIRification of data, reuse of existing data profiting of reproducible science and advanced machine-learning methods. (FAIR=Findable, Accessible, Interoperable and Reusable).

We also see new areas emerging, listed below:

AI and deep learning. These methodologies will enter new aspects and opportunities
across almost all areas of life science, such as medical imaging, human genetics, ecology,
evolution, antibiotic resistance, infectious disease, and drug development. This
development will affect both future user support and our provision of tools for
bioinformatics analyses as new cross-border data science and life science research initiatives
emerge.

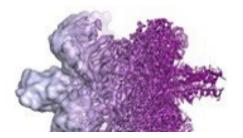
NBIS will contribute 0.5 FTE in the SciLifeLab AI action group (AIAG), as also is done by SciLifeLab Data Centre, supporting the SciLifeLab AI lead's work towards an AI strategy for SciLifeLab.



• Spatial omics. NBIS was early in adopting spatially resolved omics data, with the Spatial Transcriptomics (ST) and In-Situ Sequencing (ISS) technologies developed at SciLifeLab as prominent examples. Currently we see an extremely exciting development of combinations of high-content microscopy imaging, in-situ omics technologies, and single-

cell omics, with the potential to revolutionise our understanding of cellular and sub- cellular processes and to improve the accuracy of pathology and tissue-based diagnostics.

- **Diagnostic imaging.** Thanks to the incorporation of the AIDA Data Hub in 2021, NBIS is now successfully serving clinic-native precision medicine research in pathology and radiology imaging. Through support for compute and access to data from clinical sources, on both national and international scales, scientific advances with great potential for clinical and societal impact will be enabled.
- Structural biology. As radically improved methods for computational protein structure prediction opens new avenues for research, the demand for structural biology competence is expected to increase dramatically as many more researchers than today will start to apply it across a broad range of research fields.



- Archaeology, ancient DNA (aDNA) and paleoproteomics. NBIS is, since several years, engaged in aDNA studies, and recently also in paleoproteomics. With improved lab protocols and the recently established aDNA service at SciLifeLab, we expect these technologies to be utilised on museum specimens and in archaeology by many more users and at a much larger scale than today.
- **Drug development.** Bioinformatics, machine learning, and public data mining is expected to be increasingly important for the development of new drugs and treatments, e.g. by computational chemistry on massive on-demand compound libraries and antibody bioinformatics. NBIS has initiated contacts with the Chemical Biology Consortium Sweden (CBCS) and the SciLifeLab Drug Discovery and Development platform (DDD) for optimal synergistic operations and services.

NBIS interacts with the DDLS (Data-Driven Life Science) programme funded by KAW 2021–2032 and coordinated by SciLifeLab. Of special importance, our efforts to help users to FAIRify data and provision of important data resources in our function as the Swedish ELIXIR node will

be of value to enable the DDLS effort to be successful. NBIS will also provide support to the increased number of researchers in this area. In 2023, SciLifeLab as part of the DDLS programme established Data Science Nodes (DSNs) in the four areas of DDLS. For the users, it is important that the DSN activities are well coordinated with those of NBIS, and we will strive to achieve optimal coordination between NBIS and Data Centre in these matters.



NBIS will for the period 2025–2029 continue our activities that have been well received by the scientific community and favourably reviewed multiple times by the Swedish Research Council (VR): "The NBIS approach is the only way forward to enable statisticians and informaticians to use optimal methods for data access and data analysis." (from evaluation 2019 for the period 2021–2024), "NBIS is

an important infrastructure for bioinformatics, analysis and scientific insights into genomic data produced in research. It has a clear track record of wide scale users in Sweden distributed among different universities, and it is contributing to excellent scientific outputs." (from evaluation 2021 for equipment for human data), and "NBIS has played a vital role in providing bioinformatics support and education to the scientific community in Sweden within life science during many years and has an outermost important role to play ahead in the rapidly transforming life science landscape where data handling, bioinformatics and AI are becoming more and more central" (from evaluation 2023 for the period 2025–2028).

NBIS provides Sweden with the necessary means to allow for simultaneous exploitation of vast amounts of biological data coming from different research fields and derived at different scales, from the molecule to the organism, and to the population. It also allows for integration of these data with information from other disciplines, such as chemical, medical and environmental data. A national bioinformatics infrastructure enables advanced user support at a level that single research groups (or even single institutions) cannot reach. We provide specialised expertise in a number of areas, and our staff can simultaneously participate in multiple projects. NBIS thereby enables our users to benefit from the data-driven life science.

NBIS enables world-class life science research to maximise scientific and societal impact of publicly and privately funded research by providing expert knowledge, creative data integration, advanced training, efficient data publication and access to high-performance data analysis methods. NBIS coordinates bioinformatics support within Sweden and makes bioinformatics easily accessible for life science researchers.

## **Overall strategy**

In order to fulfil our vision and mission, NBIS provides a number of activities, as detailed in the following. This includes provision of expertise in **support** and **infrastructure** to facilitate bioinformatics analyses including access to necessary computational and storage resources. NBIS also provides routes for data publishing and engages in numerous advanced **training** activities.

NBIS coordinates its activities with other relevant infrastructures, such as other platforms at SciLifeLab, NAISS, SUNET and SND. NBIS main user groups are at academic institutions, but NBIS also interfaces with the healthcare sector, governmental agencies, and commercial companies for mutual benefits. Internationally, NBIS constitutes the Swedish ELIXIR node and engages in Nordic and European collaborations.

With the new SciLifeLab initiative IDS (Integrative Data Services), NBIS will together with SciLifeLab Data Centre be central in the efforts to improve the data handling at all SciLifeLab platforms to benefit all users. NBIS will lead the planned SciLifeLab-wide project consultations to be held at the beginning of a new project. NBIS will also engage in the initial platform meetings with users at project start in order to provide bioinformatics competence and to facilitate planning of downstream bioinformatics analyses. NBIS will also be engaged in data management.

## **Infrastructure**

A fundamental part of NBIS is the formation of a sustainable bioinformatics infrastructure for life sciences, consisting of access to tools and data. The NBIS infrastructure is typically constructed as domain-specific supporting layers utilising resources from e-infrastructure providers such as NAISS, SUNET and SciLifeLab Data Centre. These computational and storage needs are set up

in close collaboration with these actors, in order to avoid duplication of efforts and to benefit from their competences.

One example is Rshiny apps developed and maintained by NBIS for users, but hosted at hardware at SciLifeLab Data Centre. Another example is Nextcloud, which AIDA Data Hub uses to share anonymous research data with the world, but the back-end services are operated by SciLifeLab Data Centre.

#### **Data management**

The vision of NBIS data management unit is that Swedish life science researchers apply good data management practices so that the research outputs produced are available to the global research community, and to society at large, according to the principles of Open Science, Reproducible Research, and FAIR.

A detailed **NBIS Data management strategy** that outlines the scope and guiding strategic decisions underlying the operations of the Data Management was published in 2024 (<a href="https://doi.org/10.17044/scilifelab.27604476">https://doi.org/10.17044/scilifelab.27604476</a>). In the strategy, the following strategic Mission has been established to work towards the vision.

#### Mission (Overall objectives and the approach to reach those objectives)

*Support*: Swedish life science researchers shall have access to data management support to be able to manage their research data according to international best practice. To enable this, NBIS Data Management shall:

- Develop **procedures** for providing efficient data management support to research projects throughout the research data lifecycle.
- Provide data management **planning** knowledge and resources/tools for research projects.
- Provide support and resources/tools for data submission to recommended community deposition databases.
- Provide support and resources/tools to enable reproducible research.



*Training*: Swedish Life Science researchers shall have access to training in efficient data management practices that can be applied in their daily work.

- Provide Data Management training for researchers.
- Provide guidelines and documentation for data management "best practises".
- Promote awareness of the importance of Open Science, Reproducible Research, and FAIR in the Swedish life science community.

A detailed data management training strategy is described in the published **NBIS DM team training Strategy** 2025 – 2028 (https://doi.org/10.17044/scilifelab.27606417.v2).

Collaboration: Data Management practices, solutions and training for Swedish Life Science shall be established in collaboration with NBIS internally, as well as with relevant national and international stakeholders

- Collaborate with **NBIS** staff to develop data management aspects of support procedures and policies.
- Contribute to Data Management activities and capacity building within ELIXIR.
- Collaborate with SciLifeLab Data Centre to develop data management policies, procedures and solutions to support researchers.
- Interact with **other national and international stakeholders** to harmonise data management policies, procedures and solutions to support researchers.

#### Work plan for 2025:

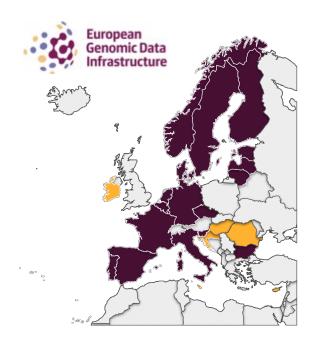
- Collaborate with the SciLifeLab Data Centre on activities to support the IDS
  initiative at SciLifeLab, where NBIS is primarily responsible for providing data
  management knowledge content, while Data Centre provides the infrastructure, with
  the exception of those cases where NBIS operates and develops infrastructure
  components, e.g. FEGA Sweden, GDI and 1+MG.
- Continue collaboration with the SciLifeLab Data Centre to provide Data management support to other prioritised SciLifeLab initiatives, such as DDLS, Pandemic preparedness, and Biodiversity.
- Continue to explore solutions for brokering data to ENA and other deposition databases together with SciLifeLab Data Centre and external international stakeholders (ELIXIR Data Platform, ELIXIR Nodes and EMBL-EBI).
- Contribute to the deliveries of the NBIS Technology Development Project on harmonising technical metadata from the Genomics units, in collaboration with the NGI and SciLifeLab Data Centre.
- Further develop the helpdesk function for data submissions to the Swedish node of the Federated EGA (FEGA Sweden), to support Swedish researchers in depositing sensitive data according to best-practice standards. This also includes assisting Swedish universities in establishing data access governance routines regarding access to sensitive data in FEGA Sweden for which the universities are data controllers.
- Contribute to the work in the European GDI and GoE projects regarding data management aspects.
- Continue to provide assistance and training to researchers in creating data management plans, including providing suitable templates and tools (together with SciLifeLab Data Centre).
- Continue providing efficient data management support to NBIS analysis support projects.
- Hold the Introduction to Data Management Practices course for researchers twice.
   Extend the data management training portfolio with contributions to the DDLS
   Research School (together with SciLifeLab Training Hub).
- Together with SciLifeLab Data Centre, arrange and perform Data Management outreach events at the SciLifeLab sites in Linköping and Göteborg.
- Lead and contribute to the ELIXIR Research Data Management Community.
- Lead and contribute to tasks in the work plans for ELIXIR Data and Interoperability platforms.
- Follow and engage in other European developments in Data management, such as in the EOSC Association.
- Contribute to the work in the Research Data Alliance (RDA) through leading roles in the Technical Advisory Board the Life Science Data Infrastructures IG, and running the TIGER grant to create an interactive web interface.

#### Human data

NBIS has since long been engaged in building infrastructure for sensitive human data. Driven by the European member state initiative 1+ Million Genomes project (1+MG), EU funding has been obtained – 2020–2023 with the B1MG coordination and support action and 2022–2027 with the implementation project Genomic Data Infrastructure (GDI) having a budget of 40 MEUR. The major part of GDI is the construction of the human genomics data infrastructure, work led by NBIS as ELIXIR-SE together with ELIXIR-FI. NBIS also engages in European Federation for Cancer Images (EUCAIM), especially regarding interoperability with GDI and Bigpicture. NBIS works closely with GMS (Genomic Medicine Sweden) in several of these human data projects.

NBIS plans to host the Swedish node in the 1+MG landscape, together with GMS. NBIS will host the Swedish front-end, 1+MG-SE, together with hosting of data originating from universities, while GMS will host data originating from the regions. The 1+MG-SE node will point to the GMS-hosted node for their data, enabling a unified Swedish front-end in 1+MG, even though data are stored at different sites.

For the 1+MG project, NBIS will work for the establishment of a Genomics EDIC (European Digital Infrastructure Consortium) as the legal body for the 1+MG.



Related to GDI, NBIS is part of a recent EU application for the Genome of Europe project (GoE), aiming at coordinated sequencing in 20+ European countries and storing data in GDI. NBIS leads the data management efforts in this project, while NGI leads the Swedish sequencing efforts.

To cater for the needs of publishing sensitive genome sequence data, NBIS has established the Swedish node in the international infrastructure Federated European Genome-phenome Archive (FEGA). We have established a help desk to provide data management support to users and expect a large workload in the coming years. The federated framework with national nodes, where metadata (data about data) are stored centrally, while datasets are stored locally and only accessed with proper agreements, is an elegant solution to allow for international data access within the current legal framework. There is a large unmet need for this type of service in Sweden, and with

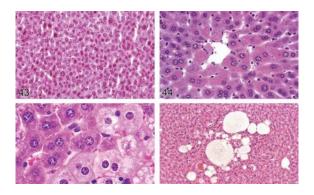
1+MG and the coming European Health Data Space (EHDS), the importance of these activities as well as the scope will increase. In order to pave the road for EHDS, NBIS is also engaging in the EU project TEHDAS2, Towards the European Health Data Space 2024–2027. TEHDAS2 is in Sweden coordinated by the E-health agency (E-hälsomyndigheten).



The data stored in this infrastructure must adhere to international standards that make data discoverable, retrievable, and re-usable for the research community. The FEGA and GDI ensure these standards and principles in a way that storage solutions at individual universities cannot.

NBIS collaborates with Genomic Medicine Sweden (GMS) on access to international databases, and we work together in 1+MG. NBIS participates in a Vinnova-funded GMS-led project on digital consents, of importance for the 1+MG project. NBIS is also part of another Vinnova-funded project 2024–2027 aiming at coordination between the Swedish EU-DIGITAL projects, including GDI and EUCAIM.

Furthermore, NBIS is engaged in <u>Bigpicture</u>, which is a 6 year 70 MEUR flagship EU-IMI project 2021–2027 with the aim to establish a Petabyte platform for European digital pathology AI. Joel Hedlund at the AIDA Data Hub is WP lead for the Bigpicture repository infrastructure development, and the systems development work is done by NBIS systems development unit together with staff at ELIXIR-FI.



In addition, our AIDA Data Hub unit is engaging in ASHA, which is a 4yr 73MSEK Vinnova systems demonstrator started in 2023, with the aim to construct interconnected spaces for primary and secondary use of standardized health data, engaging Swedish healthcare regions, EHR systems provider Cambio, and start-up company PredictMe. Here, AIDA Data Hub provides a secondary use health data space for ASHA on SDS 2.0, with a driver use case in PredictMe development of innovative epigenetic/multimodal AI tools for personalized medicine in long COVID and other systemic diseases.

NBIS is participating in the EU projects ELIXIR-STEERS for FAIR software management and node capacity building, EOSC-ENTRUST for secure computing environments on sensitive data, and ERDERA – European Rare Disease Research Alliance – where NBIS works on bioinformatics analyses and creates effective pipelines for these analyses. In collaboration with GMS, NBIS also

plans to enable execution of these pipelines on regional data in the NGP (National Genomic Portal), hosted by VGR (Region Västergötland).

NBIS will also engage in the recently started EU project CANDLE, aiming for setting up national cancer data nodes across Europe. NBIS will here work together with the Swedish National Board for Health and Welfare (Socialstyrelsen), which is coordinating Swedish cancer data.

#### Work plan for 2025:

- Continue our engagement in the EU projects GDI, EUCAIM, Bigpicture, GoE, TEHDAS2, ELIXIR-STEERS, EOSC-ENTRUST and ERDERA.
- Engage in the new EU project CANDLE.
- Continue hosting FEGA-SE and plan for hosting 1+MG-SE.
- In GDI, co-organise the fourth technical workshop in Paris in October 2025 together with the French partner.

#### **AIDA Data Hub**

The vision of AIDA Data Hub for 2030 is to be a global role model for effective and sustainable e-infrastructure services for clinical research and innovation in data driven precision health. The mission is to accelerate creation of clinical and patient value through development and provisioning of cost-effective, best-in-class e-infrastructure services beyond stakeholder capabilities. Our strategy is to enhance the productivity of research and innovation beyond what is possible elsewhere, through improved sensitive data services, increasing data availability, and strengthening the community. We strengthen prospects for sustainable services through active stakeholder dialogue and development of effective business models.

Our AIDA Data Hub (AIDA DH) unit is an e-infrastructure for research and clinical innovation in data driven precision health, with a base in multi-modal diagnostics and medical imaging AI, hosted by the Linköping University Center for Medical Image Science and Visualization (CMIV). AIDA DH provides secure compute and storage resources, data and sharing services, and advanced support and training in legal and ethical policy, AI development, and system design, to researchers, industry, and caregivers of national significance, in collaboration with national and international communities in areas of common interest. The unit provides services based on user fees to allow sustainable operations and development, with discounts offered when possible to maximise high-impact research, and where fee waivers are offered to co-funding communities and data sharing parties to incentivise building the data commons and increasing data availability for research.

Engagements include AIDA, NAISS, ASHA (VINNOVA systems demonstrator), Bigpicture (EU project, where AIDA DH is work package lead and infrastructure development is done together with the NBIS systems development unit), BrainChild, CMIV, EUCAIM (EU project for cancer image data), MT4H Focus area Datadriven Hälsa, Vård och Omsorg, Precision medicine centre network, SCAPIS, Strategy for region common healthcare data management.

AIDA DH Sensitive Data Services for AI training on sensitive personal medical data are primarily directed to leading edge researchers in medical imaging AI. It is used to provide trusted research environments for the AIDA community, and is also used to facilitate sharing of sensitive data for research, including all SCAPIS core CT examinations.



AIDA DH Data Science Platform (DSP) was

launched early 2025 as a secure data science platform for research and clinical innovation in data driven precision health, co-located with national/European flagship compute systems such as NAISS Tetralith and KAW Berzelius and the upcoming NAISS Arrhenius and AI Factory Mimer. It supports advanced data usage patterns like long term primary storage, and lets users collaborate, annotate, share, federate, and train AI.

AIDA DH also operates the AIDA Dataset register that facilitates FAIR sharing of high-quality datasets for OpenScience in medical imaging AI research, providing citable SND DataCite DOIs and search engine optimised landing pages, helping national research groups share data nationally and globally. Data sharing is carried out in collaboration with the SciLifeLab Data Centre, and transfers are often carried out using the AIDA Nextcloud service, operated in collaboration with the SciLifeLab Data Centre.

The AIDA DH unit offers advanced support and training in medical imaging diagnostics AI research and innovation. Areas include data sharing and policy, system design for sensitive data, and advanced research support in AI development. NBIS user fees are applied to the support, but with the option to include fee waivers for co-funding communities.

Training events cover technical topics at advanced AI development workshops, and GDPR and data management topics at regular AIDA AI courses for clinicians, aiming to increase availability of clinical AI training data for research.

#### Work plan for 2025:

AIDA DH unit will continue its engagements as described above, and continue its strategy implementation with work in four strategic areas:

- Increased data availability. Support development and adoption of open standards for health data; Support extraction and enrichment of prioritised clinical data for research; Promote publication and facilitate access to high quality datasets.
- Data Science Platform. Offer computable long term primary storage close to flagship compute; Enable striking the balance between security and usability as appropriate for each customer; Support advanced data usage patterns including collection, collaboration, visualisation, annotation, AI training, and federated analysis.
- Strengthen community. Provide advanced support and training in Sensitive data, AI development, and System design. Form partnerships to increase researcher access to infrastructure capabilities and for development in areas of common interest.
- Sustainable services. Map stakeholders and partnerships; Create engagement plan; Develop business models for effective engagement.

#### **Systems Development & Tools**

The mission of the systems development unit is to provide secure, usable and maintainable software solutions for all our users' needs.

Access to professional and usable software tools is a cornerstone in current day research projects. This is particularly the case in fast-changing, data-intensive fields like next generation sequencing and proteomics. Easy-to-use tools that enable researchers to carry out more of the data analysis themselves will also help to alleviate the demands for bioinformatics support in the form of consultancy.

While the development of new methods and algorithms is typically carried out in research projects, making these tools usable to the research community and keeping them maintained is often neglected due to lack of resources, expertise or incentives.

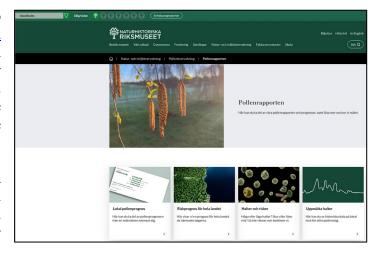
The NBIS systems development unit creates user interfaces and provides support in deploying tools so that they can be used by the entire life science community, and not just by bioinformaticians. The combination of bioinformatics experts, who can appraise the scientific value and usefulness of tools, with developers who have the capability of making tools stable and accessible, allows for development projects driven by user needs.

The development efforts will entail creating user interfaces (e.g. web interfaces), providing assistance in programming best practices (documentation, source code management, bug tracking), and deployment. The development unit will also assist in internal development projects, e.g. for integrating data services with other initiatives.

Bioinformatics has since long used machine learning for sequence pattern recognition and structure prediction. With recent advances in computational resources, these techniques have improved considerably and are summarised under terms such as Deep Learning and Artificial Intelligence (AI). NBIS will participate in national AI initiatives and benefit from the cross-disciplinary research on for example feature selection/extraction and pattern recognition, which are highly relevant for advancing NBIS capability on Big Data analysis. We will provide systems development support to users creating AI-based tools, and we will, together with our eInfrastructure partners, provide the computational requirements necessary for new AI-based tools. Further, NBIS has since long supported users in sharing data in a FAIR fashion, which is a prerequisite for data to be used in AI.

NBIS will continue to develop components in the <u>Human Protein</u> Atlas (HPA) which since long is an ELIXIR Core Data Resource and now also a Global Core Biodata Resource. We will continue to maintain the <u>Metabolic Atlas</u> (MA; genome-scale metabolic models).

NBIS will continue our collaboration with the Swedish Museum of Natural History (NRM) to support the pollen prognosis system (PLUPP) and the new



NRM website. The functionality of the API has been enhanced during the previous year, enabling users to incorporate real-time pollen forecasts into a wide range of applications. The advanced pollen prognosis is currently being incorporated into the newly launched NRM website, which includes new ways of representing current and historic data via plots. NBIS and NRM have now signed a 5-year support contract to ensure the smooth functionality of the API.

Prioritisation of major systems development projects proposed by staff or the user community are done regularly (typically every 2–3 months) by the management, according to the general principles:

- Highest prioritised are international agreements and national services of large interest.
- Next level are projects of large impact for Swedish users and of central importance for NBIS functions.
- Medium-prioritised are individual support projects for systems development that are treated as other support cases, including charging user fees.

Methods and software developed within NBIS are made publicly available through Open Access publication and Open Source licensing of software. NBIS has developed and published <u>coding guidelines</u> to aid in developing better software, and our staff also participates in the ELIXIR task Software <u>best practices</u>. We maintain **public repositories** (GitHub) for codes and scripts that are developed by NBIS.

Computational resources to our users will predominantly be provided by NAISS. For infrastructure services, SUNET will be used for FEGA-SE encryption, and EGI (European Grid Infrastructure) will be used for prediction servers. In the future, we expect that emerging computational services within ELIXIR and EOSC (European Open Science Cloud) to be utilised.

NBIS continuously enters relevant Swedish tools in the ELIXIR Tools Registry.

The systems development unit contributes to work in ELIXIR-STEERS on software management.

#### Work plan for 2025:

- Continue with the Human Protein Atlas as ELIXIR service contributed by Sweden.
- Maintain prediction servers and bioinformatics tools of importance for the Swedish life science community, of which several are also of interest for ELIXIR.
- Engage in the development of tools for genome analyses.
- Develop Nextflow workflows for data processing and analysis, partly together with other SciLifeLab platforms.
- The management makes regular prioritisations on new systems development projects.
- Enter relevant Swedish tools in the ELIXIR Tools Registry.
- Participate in European collaborations on systems development to enable efficient bioinformatics tools for large-scale analyses and management of data both human and non-human.
- Continue supporting the new pollen prognosis system PLUPP and the newly released NRM website.

#### Support for computational resources (SCoRe)

The vision of this unit is to ensure the life science community's needs for support and training on national e-infrastructures for compute and storage. We build and maintain a nationally available computational environment for bioinformatics, pushing the envelope on usability.

High throughput life science depends on high-performance computers for data processing and bioinformatics analysis. The computer systems (hardware) are provided to researchers primarily by NAISS. However, software provisioning for the community has been an ongoing challenge, not only because of the wide variety of software and the productivity of the community that develops

software for genomics and life science research, but also due to the changing landscape of how software is developed, delivered, and used. We provide support to researchers in the domains of biological sequencing, biological imaging, drug screening, and other life sciences with expertise related to and support on high-performance computational and storage resources, as well as cloud resources when that is appropriate.

The SCoRe staff faces researchers in their day-to-day work from the time they start planning their data analysis to the point where papers are published and data are moved out. This makes the unit an effective interface between NBIS and those parts of the research community not receiving other forms of support, allowing the collection of user



needs in e.g. software or hardware resources or technical training. SCoRe is involved in strategic planning as well as support and educational efforts to bridge between NBIS, SciLifeLab and NAISS.

With the growing power and utility of machine learning methods, we see a trend towards more projects that use AI tools alongside more traditional tools. SCoRe is meeting this challenge by working more closely with other support providers, as well as by working to increase internal competence in this area.

The majority of bioinformatics users are now working on systems built without their particular needs in mind. This makes it challenging for seasoned researchers and new students alike. SCoRe strives to improve usability for all life scientists on current and future systems. This work takes us beyond the Swedish borders and we work closely with international infrastructures to make the Swedish systems, services, policies and standards both align and spearhead the international research community.

We are making progress in connecting all relevant support activities on nationally and regionally available systems where life science is being conducted. We aim to further increase the number of people present in the life science support community to make the end user experience as intuitive, smooth and similar on every such system as possible. This means including non-NBIS staff under the umbrella of SCoRe.

It is expected that long-term storage systems of various kinds will continue to spring up locally and nationally to meet the need for archival and publication of research data. We will increase our level of support towards the adoption of good practices with regards to long-term data storage and publication according to FAIR principles and legal requirements.

#### Work plan for 2025:

- Continue providing user support within compute and storage and interact with NAISS, UPPMAX, PDC, and other actors.
- Follow usage patterns and facilitate efficient utilisation of the computational and storage resources.
- Coordinate with NGI, Data Centre, and other relevant SciLifeLab units.
- Follow the development in the compute and storage area and make suitable pilot studies.

#### Interactions with other research infrastructures

NBIS is increasing the interactions with other SciLifeLab platforms and capabilities as the needs of users' of other platforms for bioinformatics support becomes more pronounced. NBIS has **dedicated platform contacts** to all SciLifeLab platforms. NBIS has **regular meetings** with NGI (National Genomics Infrastructure), Clinical Genomics, and SciLifeLab Data Centre. NGI also participates in the weekly virtual drop-in sessions that NBIS hosts where users can ask bioinformatics or sequencing related questions. We have **joint staff** with CBCS (Chemical Biology Consortium Sweden) and the Cryo-EM infrastructure, and during 2024 we plan to expand with joint staff also at the Metabolomics platform. Furthermore, we have **directed collaborations** with several SciLifeLab platforms, *e.g.* NBIS is providing image- and data analysis support for the Single Cell and Spatial Biology Platform.

Since 2024, NBIS has been looking into using part of the SciLifeLab funding for collaborative projects, TDPs, with other platforms. We will also provide bioinformatics support for the TDPs that the Precision Medicine Capability are initiating.

NBIS, in close collaboration with data-producing infrastructures, e.g. Clinical Genomics, will provide expertise in systems development and access to ELIXIR-related databases and tools enabling improved and cost-efficient health care. We collaborate with Genomic Medicine Sweden (GMS), mainly on data and knowledge discoverability and sharing, in GA4GH Beacon and federated human genomic data technologies. We also coordinate our efforts in 1+MG, where we both are active. Furthermore, NBIS participates in an GMS-led Vinnova project to facilitate for patients to give their consent for secondary use of genomics data.

NBIS has ongoing data publishing collaborations with the SciLifeLab Data Centre, SUNET and SND, Swedish National Data Service. Furthermore, NBIS follows the progress with MAX-IV and ESS for potential new user communities.

NBIS has regular contacts with NAISS in order to get optimal provision of computational and storage resources. Also coordination with the application experts in bioinformatics will be important.

On the international side, NBIS follows the work in EOSC. UU as host university of NBIS is one of the founders of EOSC. NBIS is also actively engaged in RDA (Research Data Alliance). Furthermore, NBIS is working in GA4GH (Global Alliance for Genome and Health) with development of the htsget product for GDI.

NBIS has since 2022 regular meetings with SBDI – Swedish Biodiversity Data Infrastructure – for the benefit of all users in these scientific areas.

#### Work plan for 2025:

- Regular meetings with NAISS and SUNET to coordinate computational and storage and issues.
- Regular meetings with the SciLifeLab platforms Genomics, Clinical Genomics, Clinical Proteomics, Single Cell and Spatial Biology, and Metabolomics.
- When motivated, coordinate NBIS activities with relevant platforms at SciLifeLab and relevant national infrastructures.
- Start with joint staff at the Metabolomics platform.
- Participate in relevant European and international infrastructure meetings.
- Continue our engagements in several ELIXIR Focus Groups.
- Participate in the EU project and develop assembly standards and methods to be used in ERGA.
- Continue our engagements in EOSC.

#### **Local interactions**

As a national infrastructure it is important for NBIS to be visible and connected to local bioinformatics activities. To this end, NBIS has assigned site coordinators to drive, participate in and/or facilitate local activities. This is important for knowledge transfer, where new techniques acquired by NBIS should be spread also to the local sites. Similarly, NBIS will faster become aware of needs from local facilities that need national support. The site coordinators are already well connected bioinformaticians on site with the mandate to use 20% of their time for local bioinformatics interactions.

NBIS works in close collaboration with local resources, e.g. the Core Facilities at University of Gothenburg and Sahlgrenska Academy, and is active in the local SciLifeLab sites. NBIS is also involved in managing the local bioinformatics networks in Gothenburg (GOTBIN) and Lund (LUBI). The local interactions are important to guide users to the right level of support, so that the future landscape of Swedish bioinformatics support is optimally shaped with respect to user satisfaction and resource usage. All NBIS site coordinators will be under the responsibility of the outreach coordinator that is part of the management group. Furthermore, the NBIS reference group, with representatives from all 11 partners, will help to communicate new areas of interest for NBIS as they emerge locally and contribute to all partners staying updated on future life science trends.

NBIS can affiliate locally funded bioinformaticians and bioinformaticians at other SciLifeLab platforms giving them access to NBIS knowledge exchange meetings, dedicated slack channels for interactions with NBIS staff, internal training events and the opportunity to participate as experts at NBIS bioinformatics drop-ins. This will provide valuable contacts with NBIS staff performing similar work and thereby enrich their network.

The 11 partner universities have in recent years established supporting functions for data management, in many cases as Data Access Units in the SND network. These research data office functions are important stakeholders to collaborate with to ensure that the data management support services, offered by NBIS, can aid the life science researchers at these sites taking the local

considerations into account, as well as harmonizing the data management support and guidance nationally. As the local support functions for data management are still in many cases understaffed and need to cater for the complete range of scientific domains at the different institutions, NBIS works in close collaboration with the data offices to offload a lot of the burden for the local data management support for the life science domain.

The BioImage Informatics unit has since long close contact with all major Swedish local Microscopy Facilities.

AIDA Data Hub participates in the SciLifeLab Linköping site infrastructure group and has many local interactions and collaborations through its projects. In 2025 the AIDA Hub launched its Data Science Platform (DSP), which will be used locally in several activities by LiU and Region Östergötland (RÖ) for secure environments to enable collaborations for research and clinical innovation in data driven precision health. Such activities include research projects at LiU CMIV, RÖ Centra for Information driven Precision health (CIP) and RÖ precision medicine laboratory. The servers for the AIDA Data Hub Sensitive Data Services (SDS) are hosted in the same RÖ secure computer rooms that also host the hospital electronic health record production servers. The DSP servers are instead hosted in the same NSC/NAISS secure computer rooms where the upcoming national/European flagship systems Arrhenius and Mimer systems will be built.

#### Work plan for 2025:

Continued and increased engagement in, or startup of, local bioinformatics groups.

## **Support**

The vision of NBIS support unit is to enable world-class life science research and maximise scientific and societal impact of collected data. Our mission is to help catalyse the Life Science transformation towards large-scale molecular research by ensuring that excellent research projects have access to advanced (applied) bioinformatics competence, and by broad community training.

NBIS supports excellence in research. One of our major activities is support, where NBIS staff helps researchers with bioinformatics tasks in various projects. The time spent in each project varies from short (weeks) to long (months). The topics for NBIS experts are decided by the NBIS Board and Management, following suggestions from open NBIS calls, NBIS partners, the International Advisory board, the Reference Group, evaluations, or the Board itself. In order to be flexible and to test the needs for new topics, the Board can decide upon launching short-term (1–2 years) project-type activities. This will enable NBIS to provide expertise in the areas needed by the life science researchers.

Currently, NBIS provides expertise in many areas within bioinformatics: genome assembly, genome annotation, genetic variation, comparative genomics, phylogenomics, transcriptomics, proteomics, metabolomics, systems biology, single-cell biology, biostatistics, machine learning/AI, image analysis and multi-omics integration.

The NBIS support unit is thematically divided into five teams (cf. organisation on page 29):

- Health and Clinical Research
- Cell and Molecular Biology
- Evolution and Biodiversity

- Microbiology, Immunology and Structural Biology
- BioImage Informatics

NBIS has users from **all Swedish major universities** and predominantly from the faculties of medicine, science, technology, and pharmacology. NBIS handles all support projects at the **national level** and assigns the **best expert** for each project, regardless of geographic location. Furthermore, NBIS has the possibility to assign **multiple experts, when needed** for providing expertise in multiple areas or for provision of long-term redundancy.

Support constitutes about half of NBIS activities, and our users are from all Swedish major universities. NBIS provides bioinformatics support in the form of different services, ranging in commitment from short meetings to extensive collaborations.

NBIS offers bioinformatics consultations services provided for free, where our staff does not perform any work on the users' data. In one-to-one project consultations, we discuss research projects with users; the consultation time is limited to 3 h per project. The consultation focuses on project planning and many consultations result in a support project later on. In addition to these one-to-one meetings, we arrange weekly online bioinformatics drop-in sessions. These are coordinated nationally to optimal expert matching to the user's questions. At some sites they are complemented with on-site drop-ins. These informal events allow researchers to get feedback and guidance on experimental design, choice of analysis methods, software etc. They also give the opportunity to learn more about NBIS services and how to apply for support. Our user survey shows them to be highly appreciated by the community (average grade 9.5/10; 370 answers).

We will continue the major activity of **bioinformatics hands-on support service**, where NBIS staff work actively in the projects for shorter or longer time, allowing research groups access to cutting-edge expertise that would otherwise be difficult to obtain. We provide three hands-on support tracks.

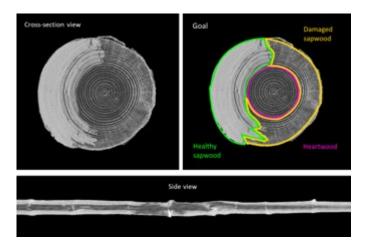
In the **User Fee Support track**, the focus is on short and medium-sized projects (typically 40–500 h) under a user-fee based model (currently 800 SEK/h = 80 EUR/h). In the last few years, we have seen an increase in the time requested, reflecting more complex data and often of different types. Projects are accepted continuously with the aspiration of having short waiting times, with most analyses starting within a few weeks from signed contract. We make a technical evaluation of each project to assure its feasibility and that we have the specific competence within NBIS.

The **Partner Project track** is intended for projects with a large bioinformatics component, where NBIS can enter as a project partner based on cost coverage by the research project. This track is intended for projects requiring NBIS support of at least 12 person months over 2–5 years (e.g. 0.5 FTE over 2 years). Since 2019, the number of partner projects has increased, and we foresee further increases during the next years.

The **Peer Review Support (WABI) track** provides extensive support to a limited number of scientifically outstanding projects that involve very large data sets and/or require extended, creative and customised analyses to accurately answer the scientific questions. The primary funding is from Knut and Alice Wallenberg Foundation (KAW) and no user fee is charged. Supported projects are selected in a rigorous scientific peer-review process in open national calls three times annually. Knowledge transfer is a key aspect of the support model, and dedicated researchers working handson alongside the support staff is required. The DDLS programme includes additional funding to the WABI Support track from 2022 onwards, with an instruction to broaden the range of expertise to include support for Cryo-EM projects.

In order to facilitate contacts with NBIS experts, we have staff at all major university sites. They constitute easy-to-find local entry points into the NBIS infrastructure. These local contact points will have good knowledge about the NBIS staff at other sites. NBIS staff has a national responsibility and should serve the users' needs regardless of their affiliation.

The **BioImage Informatics** team develops, implements and adjusts image analysis tools for quantitative analysis of primarily microscopy data. Much focus is on large scale analysis based on machine learning and deep learning/AI. We work with shorter and longer projects and offer research support via the BIIF nodes in Uppsala and Stockholm, and collaborate closely with the application experts at the AIDA Data hub at LiU. The unit is also involved in training in the form of organising courses and workshops in digital image processing and analysis with life science applications. More recent training has included AI and deep learning, as well as workflows for spatial omics. Since 2023, the AIDA Data Hub also offers advanced AI support (see above).



#### **Prioritisation**

As a national infrastructure NBIS aims at providing bioinformatics support in all projects where our competence is asked for. However, when the available resources are not sufficient to match the total needs, a prioritisation has to be made according to the principles listed below.

User Fee Support accepts projects continuously and prioritises according to the principles listed below. New support projects are assigned to the appropriate staff member(s) by a project coordinator. Managers and project coordinators follow up that each project proceeds according to plan.

Peer Review (WABI) Support has application rounds 3 times per year, where projects are scientifically ranked by the Proposals Evaluation Committee, an independent committee of scientists from Swedish universities. The managers prioritise the projects in agreement with this ranking, additionally taking technical feasibility into account.

Below are the current prioritisation principles, as decided by the NBIS Board and supported by the International Advisory Board. The prioritisation is done by the NBIS managers, based upon information from the NBIS staff.

- Technical feasibility
- Availability of data

- Projects that are judged excellent by VR or our external prioritisation committee are prioritised.
- Projects where the NBIS staff has appropriate competence are prioritised.
- Projects where the NBIS efforts make a large impact are prioritised.

In order to more efficiently be able to help more users, NBIS will provide guidance so that the users become able to do part of the bioinformatics analyses on their own. In line with this, NBIS staff also devotes part of their time to training activities. Furthermore, NBIS maintains a useful infrastructure, including tools and data handling, available for the users (cf. Infrastructure, above).

#### New services from 2024/2025

Since 2024/2025, two new capacities are integrated into NBIS – LU-fold and PReSTO. **LU-fold**, initiated by Assoc. Prof. Gemma Atkinson, enabling a fantastic opportunity to rapidly level up the structure prediction capacity and new complementary skills at NBIS and at the same time add an already growing community of users. **PReSTO** addresses that the structural biology community in the future heavily relies on supercomputer resources. PReSTO is an already mature framework, offering a unique opportunity to make this transformation possible in a short matter of time, and will give Swedish and SciLifeLab users a competitive advantage by providing an easy-to-use interface and a broad repertoire of preinstalled plug-n-play software.

### **Biodiversity**

NBIS is heavily involved in projects on biodiversity and non-model organisms. Through emerging initiatives this is expected to increase even further in the next few years.

**Support and training.** The Evolution and Biodiversity team supports projects in diverse areas, including genome assembly, comparative genomics, and populations genomics, but also expanding subjects such as ancient DNA where we have very strong competence. New types of data are emerging, for example distribution data and remote sensing data, and we have with recent recruitments adapted to meet the analysis needs for these subjects, including machine learning expertise. An advanced population genomics course funded by DDLS was run for the first time in 2023 with very positive feedback, and will now be run annually.

National collaborations. A VR-funded infrastructure project on the creation of reference genomes for Swedish species of conservation or commercial interest was wrapped up in 2024 with very positive results and a promise of extended collaborations with external stakeholders in the future. NBIS staff is leading the recently established Data Science Node in Evolution and Biodiversity (DSN-EB) funded by the DDLS programme and will through this initiative in collaboration with SciLifeLab Data Centre support the research community with the creation of services of interest by the community. We have close connection also with the SciLifeLab capability in Planetary Biology and use this connection to further gauge the needs and interests of the Swedish research community.

International collaborations. The Earth Biogenome Project (EBP) has an over-arching aim of creating reference genomes for all existing eukaryote species. In collaboration with the European Reference Genome Atlas (ERGA) and the Biodiversity Genomics Europe-project (BGE), NBIS is assembling and annotating genomes to the EBP standard to support European biodiversity research. We are also heavily involved in the creation of pipelines to facilitate increased speed and

decreased costs of reference genomes at the European level. We also have a driving role in the ELIXIR Biodiversity community with responsibility especially for networking of European biodiversity focused organisations.



#### Work plan for 2025:

- Continue providing support according to the guidelines above.
- As part of the SciLifeLab IDS effort:
  - o engage in the SciLifeLab-wide initial project consultations.
  - o engage in the initial project consultations held by the respective SciLifeLab platforms.
  - o engage in data management questions by the platforms
  - o provide competence in pipeline development
- The Board has set the academic user fee for NBIS support to 800 SEK per hour.
- The Board has set the Partner Project user fee to 49 kSEK/month+social fees. The OH surcharge is 35%.
- Encourage researchers to include bioinformatics costs in project grant proposals. NBIS will assist in estimating these costs.
- NBIS continuously follows trends in life science research in order to be prepared for emerging technologies and new bioinformatics approaches.
- Continue providing integrated support with the SciLifeLab Spatial Omics facility, with focus on combining multiple imaging modalities and informatics.
- Expand our metabolomics support in collaboration with SciLifeLab Metabolomics platform.
- Deliver reference genomes to Earth Biogenome Project quality in the Biodiversity Genome Europe project.
- Increased connection and support to non-academic external stakeholders, including several Länstyrelser and other governmental organisations.

## **Training**

The vision of NBIS training unit is to offer high quality training programmes aimed at the Swedish Life Science research community that provide a standard to follow when handling and analysing bioinformatics data. The training follows up-to-date best practices for effective teaching, using Open and FAIR training materials. Moreover, our training is inclusive and meets the needs of the research community in terms of accessibility.

In the ongoing transformation of biology and medicine into large-scale data driven research, advanced training is a key factor to ensure Sweden's scientific competitiveness. NBIS has the mission to provide advanced bioinformatics courses and NBIS staff is involved in a wide range of training activities targeted towards the Swedish Life Science community. Here, NBIS offers a course catalogue with ~20 courses in advanced bioinformatics and data science topics as well as collaborative efforts with other European institutes in state-of-the-art methodology and research topics. In addition, NBIS offers commissioned training with approximately 3 training initiatives per year as part of the Bioinformatics Support. NBIS experts are also regularly co-organising training events and invited as teachers/speakers in events arranged by others.

Part of the bioinformatics and data management support, offered by NBIS in the various projects we undertake, is the individual training of researchers in order to teach them new tools and practices to help them utilise these tools more efficiently in their own projects. The training activities are also an efficient way to increase the flow of projects through the NBIS organisation by helping scientists to be able to perform parts of the bioinformatics analyses themselves.

There is an increasing need for training, both at an introductory level and at an advanced level, and this is seen throughout the international arena. NBIS also sponsors and co-organises different workshops. NBIS training events are announced via NBIS website, SciLifeLab website and the ELIXIR TeSS website in addition to announcements via social media and other bioinformatics fora. The aim is for the courses to be of a national spread, facilitating users at different universities to take our courses. NBIS is growing continuously and this is leveraged by progressively expanding the community in order to engage, educate and inspire as many individuals as possible and to reach beyond the academic realm.

Most courses are provided on-site but we also give courses online. The online format is in line with the openness and FAIRness of training material NBIS intends to uphold as one of its missions. In addition to courses, NBIS runs the highly appreciated and successful Bioinformatics Advisory Mentorship Programme, a mentor programme where PhD students (typically 15–20 new per year) get a senior NBIS expert as an advisor for up to two years of their PhD studies.

We are continuously evaluating the demand for training activities, and we are making sure to keep our content and topics state-of-the-art. We are also evaluating the training formats to ensure we provide training according to the demand. NBIS is encouraging our experts to professionally develop in pedagogics and cognitive science as we see the demand for NBIS expertise to be shared with the community.

The NBIS Training unit will continue to work closely with the Training Hub at SciLifeLab to further develop training infrastructure and to facilitate the use of open source tools, training-related services and open educational resources established by ELIXIR. We will utilise the SciLifeLab Training Hub to increase collaboration, co-delivery of courses and knowledge exchange with other SciLifeLab platforms and national stakeholders.

NBIS engages in the **ELIXIR Training Programme** to build and provide training infrastructure and best practices to enable delivery of courses by each ELIXIR node. Since 2021, ELIXIR-SE has the role as one of three Executive Committee members for the ELIXIR Training platform and hence has the role to strategically be part of developing training for the member nodes. Since the last years, ELIXIR training has successfully increased the training engagement across all nodes and with this has been able to successfully integrate the infrastructure and outcomes from the joint node efforts into the NBIS training work. NBIS is leading the multi-node service *The ELIXIR Training Metric Database (TMD)*, which can be used by all ELIXIR nodes for monitoring training quality and impact.

#### Work plan for 2025:

- During 2025, we will continue arranging several training events on relevant topics.
- Continue with our National Advisory Mentor Programme in Bioinformatics.
- All NBIS material and courses are Openly licensed and freely accessible.
- Arrange workshops for NBIS trainers where they can make training material FAIR.
- Increase our collaborations with other SciLifeLab platforms when relevant for advanced courses, symposia and seminar series.
- Continue to collaborate closely with SciLifeLab Training Hub.
- Take an active role in the DDLS Research school by providing the course *Principles* and tools for FAIR research Practices.
- Continue to collaborate with other ELIXIR nodes for advanced courses and training events.
- Continue to take a leading role for tasks in the ELIXIR Training Platform Work Programme.
- Update training quality and impact measurements in accordance with the updated ELIXIR TMD framework.
- Establish a comprehensive course catalogue on the NBIS website including clear learning objectives and prerequisites for each course.

### Information and Outreach

NBIS has a recent and improved web site (<a href="https://nbis.se">https://nbis.se</a>) for providing up-to-date information both to our users and for internal purposes. The SciLifeLab websites information regarding NBIS and its units have also been updated recently, in collaboration with the communications office of SciLifeLab. NBIS also has a project management system facilitating tracking of projects and allowing for NBIS staff to easily share data and information with their customers.

Outreach activities have proven important to inform the scientific community about the support that NBIS can provide, making bioinformatics easily accessible for life science researchers. These consist of:

- Annual symposium and user meeting.
- Presentations at different universities, providing the possibility to meet NBIS staff representing our wide variety of competences.
- Additional presentations at relevant symposia and conferences.
- National weekly Bioinformatics Drop-in sessions via Zoom enabling face-to-face contacts between researchers and NBIS experts, which many times is the first contact in a support case.

- Weekly local drop-in sessions on-site in Stockholm and Lund.
- Open thematic seminar series aimed at artificial intelligence and integrative omics (AI & IO) and at data management (together with Data Centre).
- A dedicated outreach effort aimed at Industry.

NBIS has an outreach group that creates, updates and maintains outreach materials for presentations, graphical assets and informational content.

The outreach group will strive to create meaningful and effective channels of information distribution to recipients that are interested in our services and courses. NBIS has put our X account on hiatus but has an active LinkedIn accounts for additional interactions and information dispersal and are working with SciLifeLab communications office to aid in getting relevant information to the users.

NBIS will continue to arrange seminars in Almedalen, as we have been doing since 2023. For 2025, we plan a seminar on European human data efforts in GDI, EUCAIM and Bigpicture.

#### Work plan for 2025:

- Weekly drop-in sessions.
- Assign additional site coordinators and increase interactions with SciLifeLab Node site coordinators.
- Create efficient site-side information distribution channels.
- Increase presence in local bioinformatics communities.
- Arrange a seminar in Almedalen.

## Collaboration with industry

NBIS has expanded our efforts towards increasing contacts with life science companies and offering them access to NBIS-provided tools, databases, expertise, and training. To spearhead this effort, NBIS has since 2024 assigned a dedicated coordinator for industry outreach. The objective is to identify and establish mutually beneficial collaborations that enhance bioinformatics support/research capabilities and drive innovation. NBIS will build on and draw experience from existing collaborative projects with industry. Furthermore, NBIS will provide dedicated places for industry at relevant courses in our advanced training programme.

In our efforts towards increasing NBIS engagement with life science enterprises and industry, we will foster close collaborations with actors that focus on facilitating interactions between academia and industry. We will also leverage the experience of ELIXIR partners and contribute to their efforts geared towards fostering interactions with innovative enterprises within life sciences sector.

AIDA Data Hub has extensive collaborations with industry, for example through our engagements with Bigpicture and the AIDA innovation arena. As a notable example, AIDA Data Hub provided GPU resources and data sharing services to a SpearPoint Analytics led AIDA project engaging national and international caregivers and academia, aiming to produce an AI tool for outcome-based grading of prostate cancer biopsies. This project was highlighted as a SciLifeLab industry case, and as a success story on the NBIS website.

In parallel to ASHA, AIDA Data Hub engages in a 2 MSEK MedTech4Health funded focus area collaboration with East Sweden Medtech (ESMT), to develop business models for scaling out the ASHA use of SDS 2.0 to further businesses and efforts, such as in the pharmacological sector.

#### Work plan for 2025:

- Evaluate NBIS's past and ongoing interactions with industry to identify new opportunities and potential challenges.
- Establish new and maintain current contacts with actors or organisations that focus
  on facilitating academia-industry interactions, including SciLifeLab External Relations
  Office, university innovation centres and business incubators.
- Provide support to companies at a full-cost rate when appropriate expertise and capacity are available.
- Extend invitations to scientists from companies to participate in NBIS training activities, with dedicated slots in selected courses.
- Invite scientists from companies to NBIS annual symposium and user meeting.
- Actively participate in the work of Industry and Innovation Focus Group within ELIXIR. Provide support to companies at a full-cost rate when we have appropriate expertise and capacity.

## **Organisation**

#### **NBIS** organisational chart AGM (Annual General Meeting) Uppsala University Board International Advisory Board National Reference Group Director сто Economy Community coord. Administration **ELIXIR** Support Training Swedish Data Human Systems Evolution Support and Data 1+MG-SE EGA-SE manage **ELIXIR** Informatic Biodiversity Biology Hub Outreach node ment ment resources Managers Managers Managers Managers Manager Proj. coord. Proj. coord. Proj. coord. Staff partly in other units Support units also contribute to infrastructure Infrastructure units also contribute to support ELIXIR-SE activities in many units. Director is also ELIXIR-SE Head of Node

The organisational governance is regulated in the NBIS consortium agreement, updated in 2025. The organisational scheme is depicted above. Organisationally, *Support* is treated as one unit, consisting of five teams, indicated by the five light blue boxes.

The Board is nominated by the Annual General Meeting (Partsråd) and assigned by Uppsala University after consultation with VR and SciLifeLab. The Board meets 3–4 times per year, often via teleconference. The SciLifeLab infrastructure director and Reference group chair participate in the Board meetings.

For the coordination and leading the daily management of the infrastructure, a director is appointed by the Board. This is a 50% position, typically a professor in bioinformatics or other relevant field. The director has the executive responsibility for running the infrastructure, delegated from the Board. As the NBIS activities has grown, NBIS has since 2020 also a co-director in order to facilitate the running of the infrastructure. In addition, a technical manager (CTO; 100%) is appointed leading and coordinating the technical management of the infrastructure.

The activities of NBIS are divided into organisational units and teams, as depicted in the figure above. The managers together with the director form the management team that meet using video conference system at least monthly to coordinate activities and discuss operational matters. Important questions of policy, strategy and economy are put forward to the Board for decision. When time-wise suitable these meetings will be physical.

In order to further professionalise our services, especially for the European collaborations FEGA and 1+MG/GDI, we aim to implement components from FitSM (Federated IT Support Management), which provides an open source system for effective support management. During 2025, we plan to start the implementation both on FEGA and on some of our national services.

### **Electronic Meetings and Retreats**

In a distributed infrastructure like NBIS, it is important that all staff members are aware of the special competences of their colleagues at other sites. In order to achieve this, we use a text-based chat system (Slack) where we have set up many channels for discussing specific topics. Here, questions can be asked at any time and colleagues monitor their channels of interest as time allows. We also have a weekly short video meeting to inform about current NBIS activities of relevance to many in the organisation. The weekly meetings also provide opportunities for staff members to ask general questions, exchange ideas and socialise. In addition, we have annual retreats for all staff to increase interactions and give ample time for long-term planning and strategy discussions, bringing up new ideas, and develop the activities.

### **Topical meetings**

NBIS organises internal topical meetings, focused on a particular bioinformatics sub-discipline, *e.g.* Next Generation Sequencing, proteomics, metabolomics, AI & IO, large-scale data management, training. The purpose of these meetings is to facilitate internal networking and knowledge transfer. The topical meetings will allow for more in-depth discussions on new papers or methods, and of current NBIS projects. In order to minimise travelling time, the topical meetings will predominately be held on-line. At these meetings, relevant NBIS-affiliated persons are invited.

#### Professional development of staff

In order to assure a continuous competence development of the NBIS staff, they should ideally have their basis in a bioinformatics research environment, giving them opportunities to keep up with progress at the research frontiers and attending lectures and seminars. Furthermore, they should be given time for own education and development, e.g. when involving in support tasks needing additional competence. The time available for competence development is up to 20% (including the above mentioned own education associated with support tasks). As bioinformatics is a rapidly evolving discipline and new areas emerge, over time NBIS staff might move between different areas, depending on user needs and their own interests. NBIS is engaged in creating career development for staff scientists.

#### Work plan for 2025:

- Continue with chat-based knowledge exchange and weekly meetings.
- Continue and further develop topical meetings.
- Organise at least one internal course for competence building within NBIS.
- Arrange two retreats.
- Participate in SciLifeLab working group on career development for staff scientists.
- Start implementing components from FitSM.

#### **Affiliated NBIS persons**

In order to increase the national bioinformatics networking, NBIS enables affiliation of bioinformaticians at other SciLifeLab platforms and at core facilities. This will give them access to NBIS topical meetings, provide valuable contacts with NBIS staff performing similar work and thereby increase their network. NBIS affiliations are decided by the NBIS management. At time of writing, there are 19 NBIS affiliates.

#### Work plan for 2025:

- Continue with affiliation of relevant bioinformaticians to NBIS.
- Invite affiliated NBIS persons to knowledge exchange meetings.

## **Funding**

The major funding sources of national NBIS activities are the Science for Life Laboratory, the Knut and Alice Wallenberg Foundation and the Swedish Research Council (VR). NBIS also has financial support from the participating universities. In addition, users are contributing with user fees. These combined funding streams enable NBIS to grow successively as the demands for bioinformatics support increase. When suitable, NBIS will participate in national and international grant applications, predominantly in the infrastructure area, and NBIS currently participates in several EU projects.

#### Work plan for 2025:

- Increase user fee contributions.
- Continue expansion to meet the increased demands.
- Participate in relevant grant applications.

#### International

#### **ELIXIR** node

ELIXIR is the European infrastructure for biological information with currently 23 countries and more to join next years. Sweden is one of the founding members, and NBIS constitutes the Swedish node. Sweden is very active, e.g. with Human Protein Atlas (HPA), an ELIXIR Core Data Resource, reflecting its fundamental importance to the life-science community and long-term data preservation; with participation in multiple EU projects, especially on human data, data management, and biodiversity. NBIS is also engaged in multiple ELIXIR data management, training and capacity building efforts.



In the European 1+ Million Genomes initiative (1+MG), 27 countries have agreed to provide a cross-border federated network of national genome collections associated with relevant data for advancing data-driven health. NBIS is active in building and establishing components of the technical framework, e.g. the Federated EGA for secure storage of sensitive genome data, and the GA4GH Beacon for discoverability.

Since 2013, Sweden contributes with the Human Protein Atlas (HPA) and its integration into the ELIXIR landscape. HPA was declared ELIXIR Core Data Resource in 2018. Sweden engages in multiple ELIXIR-related activities, including the federated EGA, the Data Management Network, advanced training, and the 1+MG project.

To assure optimal coordination between ELIXIR user communities and Swedish researchers, NBIS will assign community liaisons for relevant user communities. This is already initiated for the ELIXIR communities Proteomics, Metabolomics, and Structure. Additional communities will be formed.

#### **Nordic collaboration**

NBIS aims at strengthening the Nordic collaborations with Norway, Denmark and Finland on computing, storage, training and on ELIXIR node activities. Since 2011, we have regular meetings between the Nordic ELIXIR heads of nodes. Travel costs for these meetings are kindly supported by a NordForsk grant. During 2013–2024, NeIC (Nordic eInfrastructure Collaboration) has supported the successful projects Tryggve, Tryggve2 and Heilsa Tryggvedottir. For the future, NBIS plans to particiapte in Nordic collaborations when suitable.

NBIS together with Data Centre plan for a meeting with ELIXIR-FI (CSC) during 2025.

#### Work plan for 2025:

- Continue the work on integration of Human Protein Atlas into the ELIXIR landscape and provide seamless integration with other important data sources.
- Contribute to ELIXIR 2024–2028 programme.
- Participate in relevant ELIXIR implementation studies and other ELIXIR-related activities.
- Continue our work on 1+MG, Federated EGA, GDI and related projects.
- Engage in relevant ELIXIR-related EU-projects.
- Establish contacts between new ELIXIR user communities and relevant groups in Sweden
- Collaborate in relevant areas with the Nordic ELIXIR nodes.
- Continue with our annual Sweden–Finland meetings.
- Engage in relevant international initiatives (e.g. EOSC, GA4GH, CINECA, RDA) that can benefit Swedish life science.

## Appendix I - Abbreviations

1+MG – European 1+ Million Genome Initiative

AI – Artificial Intelligence

AIDA – Analytic Imaging Diagnostics Arena

B1MG – Beyond 1 Million Genomes

BILS - Bioinformatics Infrastructure for Life Sciences

CINECA – Common Infrastructure for National Cohorts in Europe, Canada and Africa (an international project led by EBI)

DDLS - Data-Driven Life Sciences

EBI – European Bioinformatics Institute

ECDS – Environment and Climate Data Sweden

EGA – European Genome-phenome Archive

ELIXIR - European Infrastructure for Biological Information

EMBL – European Molecular Biology Laboratory

ESS – European Spallation Source

EUCAIM - European infrastructure for cancer image data

FEGA – Federated European Genome-phenome Archive

FTE – Full time equivalent

GA4GH - Global Alliance for Genome and Health

GDI – Genomic Data Infrastructure

GU - Göteborgs Universitet, University of Gothenburg

HPA - Human Protein Atlas

IAB - International Advisory Board

IDS – Integrative Data Services

IO – Integrative Omics

KI – Karolinska Institutet, Stockholm

KTH - Kungliga Tekniska Högskolan, Royal Institute of Technology, Stockholm

LiU – Linköping University

LU – Lund University

NAISS – National Academic Infrastructure for Supercomputing in Sweden

NBIS – National Bioinformatics Infrastructure Sweden

NeIC – Nordic eScience Infrastructure Collaboration

NGI – National Genomics Infrastructure

NGS – Next Generation Sequencing

NRM – Naturhistoriska Riksmuseet, Swedish Museum of Natural History, Stockholm

NSC – National Supercomputer Centre at Linköping University

PDC - PDC Centre for High Performance Computing at KTH

PI – Primary investigator

RDA – Research Data Alliance

SCAPIS - Swedish cardiopulmonary bioimaging study

SciLifeLab – Science for Life Laboratory

SLU – Sveriges Lantbruksuniversitet, Swedish University for Agricultural Sciences

SND – Svensk Nationell Datatjänst

SU – Stockholm University

SUNET - Swedish University Network

UmU – Umeå University

UPPMAX – Uppsala Multidisciplinary Center for Advanced Computational Science

UU – Uppsala University

VR – Vetenskapsrådet, Swedish Research Council

WABI – Wallenberg Advanced Bioinformatics Infrastructure