



Strategic Plan 2020–2024

Including Work Plan for 2020

version 18 March 2020

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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 discussed by the NBIS Board (Styrgrupp) and decided by the NBIS Annual General Meeting (Stämman). The current version is covering the period from 2020 to 2024 and was approved by the Board on 18 March 2020 and by the Annual General Meeting on 15 April 2020. 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 2020 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 to form **a single point of contact** for all users needing bioinformatics support. NBIS thereby has all types of bioinformatics support 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, genetic variation, comparative genomics, phylogenomics, transcriptomics, proteomics, metabolomics, systems biology, single-cell biology, biostatistics, 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 for 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 is ending.
- 3. Effective use of resources** – The large scope of many projects puts an increased demand on the ability to analyse the data effectively, using considerable programming/scripting skills, needed to automate much of the analysis. This is outside of the competence of most biological/medical researchers.
- 4. 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.
- 5. Expertise** – For research groups already having skilled bioinformaticians NBIS can, due to our 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 2020–2024, we see new areas emerging, e.g. continued rapid development in single cell omics, advances in artificial intelligence (AI) techniques, and high-content imaging informatics. We see more projects combining imaging with genomics, proteomics and other omics techniques, and it is therefore logical to formalise our

collaborations with BioImage Informatics and AIDA Data Hub, which will establish important competence in the NBIS platform. We have therefore added a module 8 – Bioimage informatics – which will be in place fully by 2021.

NBIS will 2020–2024 continue our activities that were favourably reviewed by the Swedish Research Council (VR) recently: *“The NBIS approach is the only way forward to enable statisticians and informaticians to use optimal methods for data access and data analysis. These aspects are well documented in the proposal. The opportunity to support emerging new fields of application is of great interest.”*

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 even 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. Furthermore, NBIS provides advanced training for PhD students, post-docs, and PIs.

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 and infrastructure to facilitate bioinformatics analyses and having 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, SNIC, SUNET and SND. NBIS main user groups are at academic institutions, but NBIS will also interface with hospitals, governmental agencies, and commercial companies for mutual benefits. Internationally, NBIS constitutes the Swedish ELIXIR node and engages in Nordic and European collaborations.

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 SNIC and SUNET. These computational and storage needs are set up in close collaboration with SNIC/SUNET, in order to avoid duplication of efforts and to benefit from their competences.

Data management

As science in general, and life sciences in particular, is facing an unprecedented growth of data output due to technological advances in instrumentation, NBIS will strive to be positioned to be able to handle this so-called data tsunami for Swedish life science. This entails investing in and building up of know-how in data storage, data management and data publication, but also to convey this knowledge to Swedish life science researchers and to follow the development on a European and international level. NBIS will be able to assist and train Swedish scientists in all parts of the data management life cycle, from plan, collection and analysis to archiving and data publication.

For the long-term storage of high-throughput sequencing data (NGS data) and other types of data, NBIS collaborates with SUNET and SNIC. We see that this area develops fast and the needs are increasing considerably every year. NBIS will in its role as national infrastructure coordinate the Swedish bioinformatics-related storage needs, which will be especially important for small research groups, where NBIS can assure that they get access to storage. Large user groups (e.g. SciLifeLab) typically have their own staff handling storage issues and thereby have their own momentum, leaving mainly the coordination to be performed by NBIS in close contact with the respective user group.

Proper management of research data is becoming increasingly important. NBIS shall act to guide and facilitate for Swedish life science research to make the generated data **FAIR (Findable, Accessible, Interoperable and Reusable)** to maximise impact of public investment. NBIS follows the international development in this area and engages in relevant international collaborations, mainly in ELIXIR coordinated activities, such as the ELIXIR-CONVERGE project on data management.

NBIS provides infrastructure to the Swedish life science community for data management, publishing data, and submitting data to appropriate international repositories, many within ELIXIR. Since 2012, NBIS has the authorisation to mint so-called doi:s, Digital Object Identifiers, which are persistent pointers to data sets. NBIS now coordinates doi:s with SciLifeLab Data Centre, as we have a common Figshare instance. Persistent identifiers are a prerequisite for data publication, as they enable data sets to be found even if there is a change of storage location. Using doi:s, it is also possible to cite data sets, and to search for data using metadata queries.

NBIS, together with the SciLifeLab Data Centre, will continue to support researchers to make **Data Management Plans (DMPs)**. In this work, we coordinate our efforts with the Data Offices at the respective universities. To cater for the needs of publishing human genome data, NBIS is establishing the **local Swedish node (EGA-SE)** within ELIXIR to meet the large need for this service in Sweden, enabling participation in the European 1+ million genome project (1+MG). NBIS is also engaged in the 1+MG through leadership of their WG5 (Bengt Persson) together with the Finnish ELIXIR node (Tommi Nyrönen and Ilkka Lappalainen).

NBIS has systematic procedures to ensure that **all analyses from supported projects are reproducible** and that the related data and source code becomes deposited in **public repositories** (with human sensitive data subject to controlled access). NBIS provides **courses in reproducible research** for the life science community. Reproducible research enables others to build upon previous work in a sustainable way. All support provided by NBIS strives to be FAIR and reproducible.

Work plan for 2020:

- Provide assistance and training to researchers in creating data management plans, including provision of suitable template documents and/or web-based tools.
- Continue to coordinate the NBIS activities with those of the SciLifeLab Data Centre.
- Set up a Swedish local EGA node within the framework of ELIXIR-Excelerate project in collaboration with SUNET.
- Engage in the European 1+ Million Genome initiative.
- Continue our engagement in the Nordic NeIC-supported project Tryggve2 aiming at providing secure handling of sensitive data in a federated manner across countries.
- Follow and engage in the European developments in this area, including developments in ELIXIR efforts in interoperability, FAIRification of data, and activities in the RDA (Research Data Alliance).

Systems Development & Tools

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 team 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 team 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 (*e.g.* “AI Innovation in Sweden”) 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.

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NBIS will continue to develop components in the HPA ([Human Protein Atlas](#)) which now is an ELIXIR core data resource. We will also make the [Metabolic Atlas](#) (genome-scale metabolic models) integrated with other ELIXIR resources and hopefully also an ELIXIR core data resource in the future. Already in 2019, [curated metabolic maps were integrated in HPA](#).

Prioritisation of new 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 [coding guidelines](#) to aid in developing better software, and our staff also participates in the ELIXIR task Software [best practices](#). We maintain **public repositories** (GitHub) for codes and scripts that are developed by NBIS.

Computational resources to our users will predominantly be provided by SNIC, but other research e-infrastructures, such as EGI (European Grid Infrastructure), EOSC (European Open Science Cloud) and emerging computational services within ELIXIR will also be utilised.

Within the Systems Biology module, NBIS will implement tools for analysis of high-throughput data and visualisation of results, adhering to established standards in order to enable integrative data analysis, thus providing a comprehensive set of tools sufficient to perform the complete analysis of data.

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

Work plan for 2020:

- Maintain prediction servers and bioinformatics tools of importance for the Swedish life science community, of which several are expected to also be of interest for ELIXIR.
- Engage in the development of tools for genome analyses.
- Develop nextflow workflows together with NGI.
- 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.

Compute and Storage

High throughput biomedical science nowadays depends on high-performance computers for bioinformatics analysis. The hardware is maintained by SNIC but NBIS provides expertise needed to efficiently access the computational and storage resources. We expect a continuing high number of projects as well as increasing sizes due to decreasing costs for sequencing and the increased

capacity of NGI. In order to preserve the frontline position of Swedish bioscience, we need to stay in the forefront of e-Infrastructure resources and related expertise. With growing numbers of project, increasing sample and data sizes, we will develop plans for how to accommodate for this scenario in the best way. We will explore the benefits of cloud computing and data analytics frameworks such as MapReduce, Hadoop, and Spark. We will also investigate how scientists can improve the automation of bioinformatics analysis using scientific workflow tools. In addition, we will investigate more efficient methods for storage of biological data, including user interfaces for working with multiple, tiered solutions, enabling cost-efficient utilisation of available hardware.

Support for users is currently focused on the environment present at UPPMAX. By leveraging new software deployment methods, we will make this environment more portable and vastly improve our ability to support users also on other SNIC systems and elsewhere.

Rackham reaches end-of-life during this period. We expect that at least 1500 users and their 5+ PB of data will need administrative and/or technical help moving to a replacement system.

Increased support for automatic workflows and digital research environments. Workflows in e.g. Galaxy are growing in power and popularity, and are quickly becoming an important tool also for experts and non-experts alike.

The Wallenberg Artificial Intelligence, Autonomous Systems and Software Program (WASP) has invested in a large-scale computer system for artificial intelligence/machine learning (AI/ML) methods, to be taken into production starting late 2020. We will support our community with AI/ML needs on that resource.

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 2020:

- Provide user support within compute and storage.
- Follow usage patterns and facilitate efficient utilisation of the computational and storage resources.
- Coordinate with NGI.
- Follow the development in the compute and storage area and make suitable pilot studies

Interactions with other research infrastructures

NBIS constitutes the Bioinformatics Platform at Science for Life Laboratory, which is a national infrastructure for high-throughput techniques. NBIS is increasing the interactions with other SciLifeLab platforms as their users' needs for bioinformatics support become more pronounced. NBIS already has **regular meetings** with NGI (National Genomics Infrastructure) and DD (Diagnostics Development). Starting 2020, we have **joint staff** with CBCS (Chemical Biology

Consortium Sweden) in chemoinformatics, and **embedded NBIS staff** in other SciLifeLab platforms, as NBIS is piloting within proteomics facilities. Further details are provided below.

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

NBIS, in close collaboration with data-producing infrastructures, *e.g.* DD, 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.

On the data publishing side, NBIS has ongoing collaboration 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.

On the international side, NBIS follows the work in EOSC-Life, coordinated by ELIXIR, and in which all European BMS infrastructures are partners. NBIS is also actively engaged in RDA (Research Data Alliance). Furthermore, NBIS follows the work in GA4GH (Global Alliance for Genome and Health).

Work plan for 2020:

- Regular meetings with SNIC and SUNET to coordinate computational and storage and issues.
- Regular meetings with the SciLifeLab facilities NGI, DD and Proteomics.
- When motivated, coordinate NBIS activities with relevant platforms at SciLifeLab and relevant national infrastructures.
- Participate in relevant European and international infrastructure meetings.

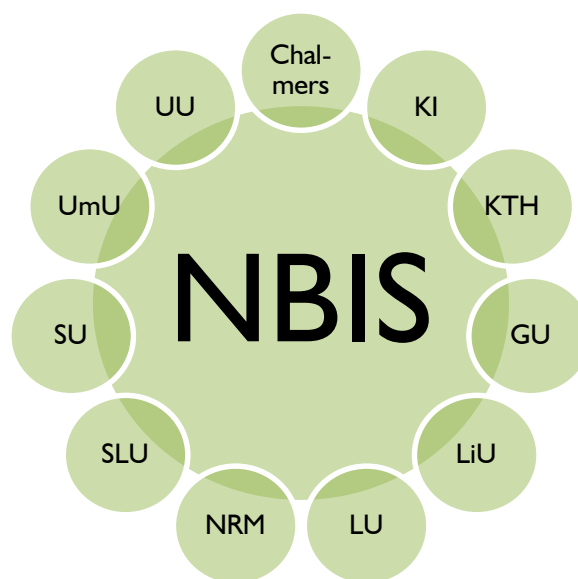
Increased local interactions

In order to optimise coordination between the national infrastructure NBIS and local bioinformatics activities, *e.g.* core facilities, we will assign site coordinators 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. Bioinformatics is entering more and more research projects, and we expect local support facilities to be more involved in support needs. Already now, NBIS has regular drop-in sessions in Gothenburg together with the local core facility.

We will, piloting in 2020, start by at three sites assign a person as **NBIS site coordinator**, typically 20 %, responsible for keeping contacts between the national activities of NBIS and these local activities. This is important for knowledge transfer, where new techniques taken up 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.

Since bioinformatics is increasingly entering more and more research projects, we expect that local support facilities will successively take a larger part of the total support needs. We also expect a trend where common and standardised analyses are done at multiple local sites, where new state-of-the-art techniques are provided by NBIS. Furthermore, the local facilities will be tailored to meet local needs that are better catered for by a local facility than the NBIS which is providing the national perspective. These NBIS site coordinators will be instrumental in guiding 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 community coordinator that is part of the management group.

NBIS can affiliate locally funded bioinformaticians giving them access to NBIS knowledge exchange meetings, provide valuable contacts with NBIS staff performing similar work and thereby enrich their network.



Work plan for 2020:

- Assign a community coordinator to be main contact between NBIS Management and Site coordinators and to administer NBIS affiliates.
- Assign site coordinators at 3 sites during 2020.

Support

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 (days) to long (weeks/months). The topics for NBIS experts are decided by the NBIS Board, 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, and multi-omics integration.

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. On the lighter end of the spectrum, we arrange **bioinformatics drop-in sessions** at all sites in Sweden, providing 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 these. In conjunction with large grant applications, we arrange extra drop-in sessions with focus on these applications. We will arrange **~250 drop-in sessions** annually, which are much appreciated by the community. We also offer **bioinformatics consultations**, where we discuss research projects with users. This service is provided for free, but the NBIS staff do not perform any work on the users' data. Many consultations result in a support project later on.

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 **Short- and Medium-term 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). During 2019, the number of partner projects has increased, and we foresee further increases during the next years.

The **Long-term Support 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 hands-on alongside the support staff is required.

In order to facilitate contacts with NBIS experts, we have staff at all major university towns. 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.

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.

Short- and Medium-term Support has application rounds at least every 2 weeks, where projects are prioritised 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.

Long-term 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 according to technical feasibility in agreement with this ranking.

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 which 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).

Work plan for 2020:

- Continue providing support according to the guidelines above.
- Increase staff in order to match increased needs.
- Perform pilot activities as decided by the Board after the open call in 2019.
- For 2020, the Board has set the academic user fee for NBIS support to 800 SEK per hour.
- For 2020, the Board has set the Partner Project user fee to 42 kSEK/month + LBK + 35% OH.
- 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.

Training

NBIS staff is involved in a wide range of training activities targeted towards the Swedish Life Science community, both in our own workshops and as invited teachers/speakers in courses arranged by others. Training offered by NBIS ranges from participation in advanced bioinformatics courses, graduate student courses and similar to individual training of researchers in order to teach them new bioinformatics tools and to help them utilise bioinformatics tools more efficiently. 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 of training, both at an introductory level and at an advanced level. NBIS will also sponsor and co-organise workshops. At our web site we will announce available courses, facilitating users to take courses at different universities.

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. We are continuously evaluating the demand for training activities, and we are keeping our courses state-of-the-art. NBIS is encouraging our experts to professionally develop in pedagogics and cognitive science. Currently, the demand for NBIS courses is larger than our resources permit. NBIS has since 2015 run the **Bioinformatics Advisory Mentorship** Programme, a mentor programme where PhD students (typically 15–20 new per year) get a senior NBIS expert as a personal advisor for up to two years of their PhD studies.

NBIS workshops and training events are spread over all SciLifeLab sites in Sweden with the aim to reach as many national life scientists as possible. In 2018, NBIS launched a 10 days international summer school in R – RauhR – at Campus Gotland in Visby. This course was very successful and will be run annually to attract international participants as well as national students. NBIS staff also organise and teach at international post-graduate courses, e.g. single-cell omics (with SIB, GOBLET and EMBO), molecular evolution ([Evomics, Czech Republic](#)), and advanced R programming ([RauhR](#)).

NBIS engages in the **ELIXIR Training Programme** for development of new software engineers, biocurators and other professionals needed to operate the bioinformatics infrastructures. NBIS will participate in ELIXIR advanced courses, e.g. single cell transcriptomics, also 2021–2024. NBIS has successfully integrated the **eLearning platform** offered by ELIXIR-Slovenia as part of our training activities. Furthermore, hackathons, train-the-trainer events, instructor training, and software carpentry workshops are regularly arranged with ELIXIR.

Training is also one important area of future collaborations between NBIS and industry, where we can enable companies to attend NBIS training events.

Work plan for 2020:

- During 2020, we will arrange several training events on at least the following topics:
 - next generation sequencing data analysis
 - statistics
 - genome annotation
 - data management
 - programming courses, *e.g.* in Python and R
 - RaukR course in Visby
 - Single-cell transcriptomics
- During 2020, we will in the areas where we see increased demand, set-up new or modify existing training events to meet the needs of our users.
- Apply an e-learning approach to courses where applicable.
- Provision of “course packages” to increase the efficiency of our training activities.
- Continue with our National Advisory Mentor Programme in Bioinformatics.
- Continue our engagement in the National Research School in Medical Bioinformatics.
- Provision of information on bioinformatics courses available in Sweden and within ELIXIR.
- Provide self-study material and lecture notes in relevant bioinformatics courses on the NBIS website.
- Collaborate with other SciLifeLab platforms when relevant for advanced courses and seminar series.
- Collaborate with other ELIXIR nodes for advanced courses and training events.

Information and Outreach

NBIS has a web site (<https://nbis.se>) for providing up-to-date information both to our users and for internal purposes. 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 and to increase the number of NBIS customers. 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 various symposia and conferences.
- Weekly Bioinformatics Drop-in sessions enabling face-to-face contacts between researchers and NBIS experts, which many times is the first contact in a support case. These sessions are arranged at all sites (Uppsala, Stockholm, Göteborg, Linköping, Lund and Umeå).
- Training, cf. below.

Presentation material, e.g. flyers, roll-up, has been produced and will be continuously updated. NBIS also has Twitter and LinkedIn accounts for additional interactions.

NBIS aims to increase the collaboration with other platforms within SciLifeLab. This work for increasing contact surfaces for bioinformaticians within the SciLifeLab community started in 2018 with a newly launched Seminar series, *BiG Talks!* (Bioinformatics and Genomics seminar series arranged in collaboration between NBIS, NGI and CG). The seminar series is financially supported by SciLifeLab and talks are given by internationally well-renowned speakers within the fields of Bioinformatics and Genomics. The seminar series aims to increase the interaction between bioinformaticians and researchers within the SciLifeLab community. The BiG Talks are broadcasted in order to reach a wider audience with possibilities for participants to ask questions and comment in real time. The idea is that the seminar series will rotate at different SciLifeLab sites during the four times per year these take place.

Work plan for 2020:

- Continue to keep the NBIS web site up-to-date and informative – both for our users and for our staff.
- Weekly Drop-in sessions at the sites where NBIS personnel are located.
- Give presentations at the six sites (Umeå, Uppsala, Stockholm, Linköping, Göteborg and Lund).
- Annual symposium and user meeting will be held in 2020.
- Run the *BiG Talks!* seminar series during 2020.
- Continuous updates of information material, both web and print.

Collaboration with industry

NBIS will continue our efforts to increase contacts with life science companies to enable them access to NBIS-provided tools, databases, expertise, and training. Last year, we arranged an ELIXIR SME event in Stockholm with ~80 participants.

Work plan for 2020:

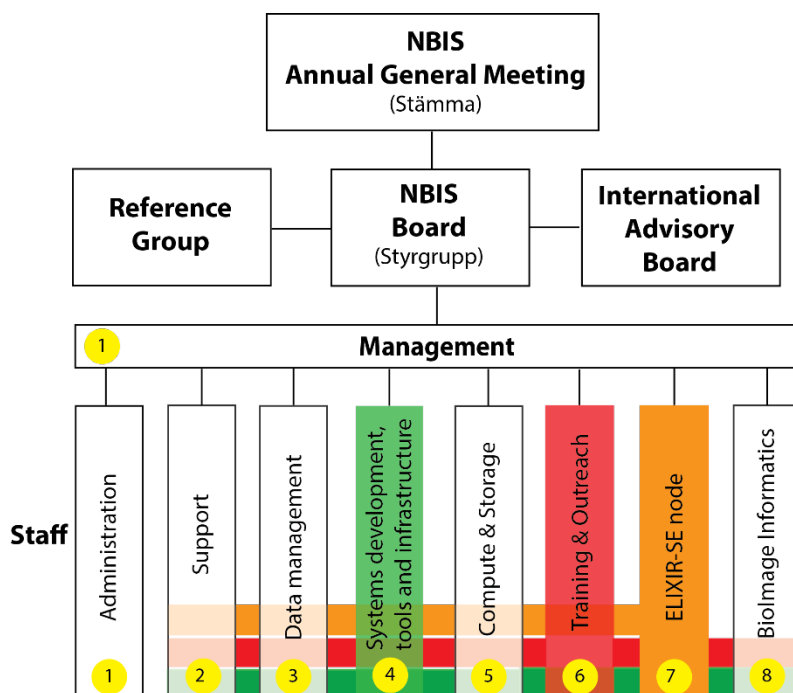
- Provide support to companies at a full-cost rate when we have appropriate expertise and capacity.
- Invite scientists at companies to NBIS training activities, to the annual symposium and user meeting, and to NBIS site visits.

Organisation

The organisational governance is regulated in the NBIS consortium agreement, updated in 2019.

The organisational scheme as envisioned in the applications to VR in 2015, 2017 and 2019 is depicted to the right.

The Board is nominated by the Annual General Meeting 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. An International Advisory Board is established since 2012.



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. 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, as depicted in the figure above (the numbers refer to modules in the VR application). Major interactions between the units are visualised as horizontal bars, but additional interactions also exist. 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.

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 have a weekly text-based chat, where current NBIS activities are discussed in an informal manner. An automatic e-mail reminder is sent to all staff, containing information about open support requests. The NBIS chats also provide opportunities for staff members to ask general questions, exchange ideas and socialise.

In addition, we have annual retreat(s) 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, 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. Some NBIS staff might want to spend this development time within a research project. 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 2020:

- Continue with weekly chats.
- 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.

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 knowledge exchange meetings, provide valuable contacts with NBIS staff performing similar work and thereby increase their network. NBIS affiliations are decided by the NBIS management.

Work plan for 2020:

- 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.

Work plan for 2020:

- 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 22 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), now an ELIXIR Core Data Resource, reflecting its fundamental importance to the life-science community and long-term data preservation;

ELIXIR-EXCELERATE 2015–2019, with NBIS efforts in data management, sensitive data (Federated EGA), genome annotation, single cell transcriptomics, data interoperability, and advanced training; ELIXIR-CONVERGE 2020–2022, where we will lead European efforts in data management; and the B1MG application for 2020–2022 to provide coordination support for the European 1+ Million Genomes Initiative.

In the **European 1+ Million Genomes initiative (1+MG)**, 20+ countries will 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. Efforts will be made 2020 and onwards to get Metabolic Atlas to become an ELIXIR Core Data Resource in the future. Sweden will engage in multiple ELIXIR-related activities, including the federated EGA, the ELIXIR-CONVERGE project, RDCConnect, 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. Since 2013, NeIC (Nordic eInfrastructure Collaboration) engages in supporting biomedical sciences, initiated by letters of interest from NBIS together with the Nordic ELIXIR nodes. The project Tryggve aims at constructing a federated solution across the Nordic countries for secure data handling and analysis, allowing for exchange of data when the ethical permits so allows. The first phase was very successful, and a second phase (Tryggve2) is granted 2017–2020. We have recently applied, also included Estonia, for a NeIC Heilsa project 2021–2023.

Work plan for 2020:

- 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 2019–2023 programme.
- Participate in ELIXIR-CONVERGE project activities according to the plans in the application.
- Participate in relevant ELIXIR implementation studies and other ELIXIR-related activities.
- Continue our work on Federated EGA.
- Engage in ELIXIR-related human data activities, including 1+MG, RDCConnect,
- Establish contacts between new ELIXIR user communities and relevant groups in Sweden
- Collaborate in relevant areas with the Nordic ELIXIR nodes.
- Plan for joint advanced training activities with the Nordic ELIXIR nodes.
- Continue our activities in Tryggve2.
- Engage and collaborate with NeIC concerning their activities on the biomedical arena.
- 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
CG – SciLifeLab facility Clinical Genomics
CINECA – Common Infrastructure for National Cohorts in Europe, Canada and Africa (an international project led by EBI)
DD – Diagnostic Development
EBI – European Bioinformatics Institute
ECDS – Environment and Climate Data Sweden
ELIXIR – European Infrastructure for Biological Information
EMBL – European Molecular Biology Laboratory
ESS – European Spallation Source
FTE – Full time equivalent
GA4GH – Global Alliance for Genome and Health
GU – Göteborgs Universitet, University of Gothenburg
HPA – Human Protein Atlas
IAB – International Advisory Board
iRODS – Integrated Rule Oriented Data System
KI – Karolinska Institutet, Stockholm
KTH – Kungliga Tekniska Högskolan, Royal Institute of Technology, Stockholm
LiU – Linköping University
LU – Lund University
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
SciLifeLab – Science for Life Laboratory
SLU – Sveriges Lantbruksuniversitet, Swedish University for Agricultural Sciences
SND – Svensk Nationell Datatjänst
SNIC – Swedish National Infrastructure for Computing
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