The SIB Swiss Institute of Bioinformatics was founded 20 years ago. Today, it represents the largest national bioinformatics network in Europe. In this special edition of our SIB Profile, we are looking back over 20 years of services to the life sciences and health community.

Since 1998, SIB’s activities have developed along two main axes: infrastructure for life scientists and the bioinformatics community. As we write, the Institute brings together 800 members, 67 research and service groups, 12 core facilities and over 150 resources, covering fields as diverse as proteomics, phylogeny and machine learning. Alongside these two main axes, SIB also has at heart a number of strategic activities: the sustainability of core data resources and personalized health, to name but two.

Defending the long-term sustainability of data resources is rooted in our Institute’s DNA. SIB is part of the Global Life Sciences Data Resources working group, whose mission is to develop a global infrastructure to support and ensure the sustainability of core data resources, which are of crucial importance for the life sciences. An SIB-led study to identify a candidate funding model for knowledgebases has also been completed and will contribute to informing the working group.

On the personalized health front, the Institute initiated the implementation of a national secure and interoperable infrastructure, as part of the Swiss Personalized Health Network (SPHEN). By the close of the year, the network had approved 15 projects – in fields as varied as oncology and inflammatory disease immunotherapy – in which SIB Group Leaders are well represented.

SIB is also involved in developing personalized health capacities in Swiss hospitals, and bringing bioinformaticians and clinical actors together to harmonize routine next-generation sequencing practices on a national scale. And along with the University Hospitals of Basel and Lausanne, SIB has just launched a Certificate of Advanced Studies in personalized molecular oncology for professionals who are faced with the new challenges posed by complex genomic data, for the benefit of patient care.

Over these years, SIB has come a very long way, with the critical support of many bodies: we would like to thank the State Secretariat for Education, Research and Innovation (SERI), the Federal Assembly, the Swiss National Science Foundation and all those in funding roles, as well as our partner institutions.

While there are still exciting challenges in front of us, we would like to express our heartfelt gratitude to all SIB members, without whom none of this could have happened, and whose expertise and dedication has brought Swiss bioinformatics to where it is today.

Two decades of Swiss bioinformatics are something to celebrate, and we hope you will join us for the festivities and actions planned throughout the year.

Foreword
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SIB in brief

OUR MISSION AND OBJECTIVES

SIB leads and coordinates the field of bioinformatics in Switzerland. Our data science experts join forces to advance biological and medical research and enhance health.

Infrastructure

SIB provides the national and international life science community with a state-of-the-art bioinformatics infrastructure, including resources, expertise and services.

DATABASES AND SOFTWARE TOOLS
SIB creates, maintains and disseminates a large portfolio of databases and software tools worldwide.

CORE FACILITIES AND COMPETENCE CENTRES
It offers easily accessible, in-depth expertise and support in bioinformatics through core facilities and competence centres.

PERSONALIZED HEALTH
It supports researchers and clinicians with know-how, resources and a secure data infrastructure dedicated to personalized health.

Community

SIB brings together world-class researchers based in Switzerland and delivers training in bioinformatics.

SCIENTIFIC COLLABORATION
SIB represents and promotes Swiss bioinformatics nationally and internationally, by bringing together bioinformatics research and service groups from Swiss universities and research institutes. SIB fosters scientific collaboration and innovation at the highest level of excellence.

TRAINING IN BIOINFORMATICS
It provides life scientists and clinicians with a large portfolio of courses and workshops. It fosters exchanges among bioinformatics and computational biology PhD students, and trains them using the most up-to-date methods necessary for their research.

Top
Life scientists rely on various types of infrastructure to perform their research: knowledge-based resources, computing power, software...

Bottom
Collaboration is at the heart of science. And it all starts with a strong community.
Bioinformatics: a definition

FIGHTING DISEASES. DISCOVERING SPECIES. UNDERSTANDING LIFE.

Life scientists and clinicians have always tried to assemble data and evidence to find the right answers to fundamental questions. In 2018, data are no longer lacking. But a different kind of problem has emerged. Nowadays, new technologies are producing data at an unprecedented rate. Indeed, so much data - and of so many kinds (see right-hand examples) - that they can no longer be interpreted by the human mind alone.

Enter bioinformatics.

Bioinformatics is the application of computer technology to the understanding and effective use of biological and clinical data. It is the discipline that stores, analyses and interprets the ’big data’ generated by life science experiments, or clinical data, using computer science. And dedicated data experts. This multidisciplinary field brings together biologists, computer scientists and mathematicians, as well as statisticians and physicists.

Computer-based approaches have become indispensable to science, by allowing researchers to advance their understanding of complex systems.

Bioinformatics encompasses:

DATA- AND KNOWLEDGEBASES for storing, retrieving and organizing biological information, including biocuration, to maximize the value of biological data;

SOFTWARE TOOLS for modelling, visualizing, analysing, interpreting and comparing biological data;

COMPUTING AND STORAGE INFRASTRUCTURE to process large amounts of data;

ANALYSIS of complex biological datasets or systems in the context of particular research projects;

RESEARCH in a wide variety of biological fields using computer- and data science and leading to applications in diverse areas, from agriculture to precision medicine. (SEE P.50-51)

What sort of data are we talking about? Bioinformatics is dealing with a broad spectrum of complex data types. Often all at once.

Sequence data
Expression data
Imagery data
Test data
And more...
Converting biological questions... 

Massive amount of data and data types: genetics, text, biochemical, imagery... 

Life sciences and health actors

...into answers with various applications

Basic research Medicine Ecology Agriculture

SIB Swiss Institute of Bioinformatics

Creating databases and software tools

Analysing complex datasets and conducting research

Providing computing and storage infrastructure

Tailoring treatment to cancer patients

Understanding how chameleons change colour

Predicting the spread of the bird flu
Some of the things bioinformatics has made possible since 1998

Though the beginnings of bioinformatics can be tracked back to the early 1950s, key biological and medical questions have been solved thanks to technological breakthroughs and bioinformatics advances in the past two decades.

Detecting susceptibility to a disease
Spotting our genetic Achilles’ heel

Individuals share genetic peculiarities, or variations, in their DNA sequence, which can predispose them to conditions such as diabetes.

Today, it is possible to selectively sequence all the protein-coding regions in a genome in one go, thanks to a technique known as WHOLE EXOME SEQUENCING (WES).

Using this technique, SIB Group Leader Jacques Fellay and his team identified a mutation which increases the severity of common colds among children. SEE P. 54

Improving the function of a protein’s function
Tell me your shape and I’ll tell you what you do

The structure of a protein determines how it functions and interacts with other proteins and molecules in order to perform its vital tasks. Twenty years ago, predicting a protein’s function was tricky, as it was difficult to change a protein’s sequence in order to find out how this would affect its structure and function.

Today, our knowledge on 3D PROTEIN STRUCTURES has grown dramatically to reach 90% coverage in some model organisms, such as the bacterium Escherichia coli.

SIB’s core resource SWISS-MODEL, a platform for generating 3D protein models, can be used by scientists to engineer proteins in order to improve vaccines. SEE P. 64

Tracing our origins
Making do with less to see more

ANCIENT DNA has a lot to say about how organisms evolved and populated the world. Over time, though, DNA undergoes damage, and becomes a story in which paragraphs are missing. This has long impeded researchers’ ability to read into our past.

Today, bioinformatics techniques account and to some extent, compensate for the less than ideal quality of the data. More knowledge can thus be extracted from less material.

SIB Group Leader Laurent Excoffier and his team took part in a study that shed light on the social and reproductive behaviour of our Paleolithic ancestors, over 30,000 years ago. SEE P. 60

Revealing the multitude
Many birds and one stone

Imagine our guts, a handful of soil, or a drop of blood. Everything is teeming with microscopic entities, including bacteria, viruses, fungi and more. How can you identify each one of them?

Today, the overall DNA present in a sample – taken for example from a patient infected by an unknown pathogen – can be retrieved thanks to METAGENOMICS, whether it represents a single or indeed several thousand species.

SIB Group Leader Jérôme Goudet and his team used metagenomics to document the fungal biodiversity of alpine soils. SEE P. 61

Unfolding the tree of life
Splitting paths

We have been building the tree of life for centuries, ever since scientists took an interest in how species – or populations – were related. For years, however, relationships were merely based on what could be seen with the naked eye or through the microscope.

Today, PHYLGENETIC TECHNIQUES are able to delve into the invisible, and compare genotypes and phenotypes in far greater detail, thus refining the tree of life, but also predicting the evolution of a disease.

SIB Group Leader Tanja Stadler developed a phylogenetic tool to model the dynamics of the Ebola virus during the 2014 outbreak in West Africa. SEE P. 62

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Activities for the layperson

Bioinformatics is at the heart of life science research. As it is involved in issues pertaining to health, ethics and society, explaining its role to the public is crucial. SIB has been involved in communicating bioinformatics for 20 years, through a range of fun activities and at various events.

In 2017, the SIB team met you at:

A STAGE PLAY TO UNDERSTAND OUR ORIGINS
Inspired by a research carried out by SIB Groups on the peopling of Australia, *Genome Odyssey* was produced at the Museum of Ethnography in Geneva.

SIB HACKATHONS
Children, developers and structural biologists used programming or augmented reality apps to tackle biological problems, as part of the 2017 Open Geneva Hackathons.

In 2018, meet the SIB team at:

IMAGES DE SCIENCE
Geneva, Museum of Science History, until 26 August
Science image exhibition in the grounds of the Museum, with two panels provided by SIB.

MYSTÈRES DE L’UNIL
University of Lausanne, 2-3 June
Discover the field of metagenomics with a workshop entitled *Bacteria, viruses, fungi and me…*

NUIT DE LA SCIENCE
Geneva, Museum of Science History & Campus Biotech, 7-8 July
What is the role of bioinformatics in drug design, personalized health and metagenomics?

PLANÈTE SANTÉ LIVE
Geneva, Palexpo, 4-7 October
Latest advances in clinical bioinformatics in the field of personalized health.

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In 2018, SIB celebrates its twentieth anniversary. In the following pages, discover some of the Institute’s highlights over the past two decades; learn about its role in federating a growing Swiss bioinformatics community; and find out more about its engagement to make open data sustainable.
CHAPTER 2 20 years of commitment Federating Swiss bioinformatics

A few questions to Mauro Dell’Ambrogio, State Secretary for Education, Research and Innovation.

Switzerland is a small country benefiting from an excellent international position on the scientific, innovation and education fronts. Small, but highly decentralized, it is also one of the few countries in the world with a coordinated bioinformatics infrastructure, provided by SIB to life scientists and clinicians with the support of the Swiss government for 20 years.

When did you first hear about bioinformatics?

Mauro Dell’Ambrogio. I was appointed State Secretary for Education and Research in 2008. Bioinformatics was a growing scientific field then and I quickly discovered SIB and its ambitions.

In particular, it was during my first official trips abroad to visit life science research institutions, that I noticed the ‘Swiss-Prot’ database being mentioned by different scientists, and in different parts of the world. For me, this was the best sign that this was an important initiative in Switzerland. The topic of bioinformatics has been with me for almost 10 years now.

Every four years, the Swiss government allocates significant funding to SIB. Can you tell us a bit more about the underlying process, and the key elements that have contributed to making the Institute eligible for funding over the past 20 years?

MDA First of all, the federal funding is primarily allocated to SIB for its function as a supporting institution for research in life science. When we prepare the fund allocation every four years, the applicants have to undergo a scientific evaluation. If the result is that something is indispensable for the development of a specific domain, it is of national importance and demonstrates a high level of quality, we can provide subsidiary funding in addition to support from the universities and cantons.

Another key element that contributes to SIB’s eligibility is its governance model: it has to be a non-commercial research institution, which is independent from the universities. By including both the providers and users of bioinformatics services among its researchers, the Institute ensures it can lead and coordinate the field.

So SIB is a good example of how we can reconcile competition and cooperation in research.

How are Switzerland positioned internationally in terms of its bioinformatics infrastructure?

MDA Switzerland has been a first-mover in the bioinformatics field internationally. The Swiss government has been financing bioinformatics infrastructure since 1998. In 2013, the European Research Infrastructure ELIXIR was founded, with reference to the SIB model. This and other developments in Europe have since strengthened our leading position in bioinformatics, not only as pioneers in the field, but also in research as a whole.

How do you expect the role played by bioinformatics to evolve over the next 20 years?

MDA Personalized health and big data will be among the main challenges for science in the next years. I think the experience and possibilities SIB offers will be of utmost importance, in particular in the field of personalized health, where we gave a central role to the Institute in the framework of the national “Swiss Personalized Health Network” initiative.

The growing needs for bioinformatics infrastructure provision

A sustained support of the federal government to SIB over the years, matching the growing need for bioinformatics infrastructure provision.

As you know, this year marks SIB’s 20th birthday. Would you like to express your wishes to the Institute, its employees and its members?

MDA Switzerland is among the top countries for research and innovation. However, it is a continuous challenge to remain at the top, and institutions like SIB play a key role in supporting our country’s leading position.

I would therefore like to thank all those at SIB who have helped to strengthen Switzerland’s standing as a location for top research for the past 20 years. Their work benefits society and the economy – today and tomorrow.

With this I wish you all every success for the future!

Watch this interview online on our Youtube channel: goo.gl/uR6WvB

“SIB is a good example of how we can reconcile competition and cooperation in research”
CHAPTER 2  20 years of commitment

Representing bioinformatics

IN SWITZERLAND...

A steady increase of the bioinformatics community

SIB Members, Groups and Employees over the years

Faces of Swiss bioinformatics

In addition to its history of collaboration with other national bioinformatics institutes, SIB also acts as the Swiss Node of ELIXIR, the organization for a sustainable infrastructure to support life science research across Europe.

Thanks to its pioneering role, SIB’s expertise is renowned outside Switzerland and the Institute is regularly invited to share its experience with and present its activities to international institutes and organizations. Recent visits from abroad have included delegates from Bahrain, China, Greece, Iran and South Africa.

All roads lead to bioinformatics

SIB Members come from a wide range of educational backgrounds

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Bioinformatics as a career

SIB Members’ current positions are diverse too

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Data collected from 91 SIB Members as part of an internal survey (2017)

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Cristina Casals Casas, PhD

UniprotKB/Swiss-Prot biocurator

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<th>Field</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Biology</td>
<td>40%</td>
</tr>
<tr>
<td>Other (engineering, bioinformatics, administrative…)</td>
<td>16%</td>
</tr>
<tr>
<td>Informatics</td>
<td>16%</td>
</tr>
<tr>
<td>Mathematics</td>
<td>9%</td>
</tr>
<tr>
<td>Physics</td>
<td>9%</td>
</tr>
<tr>
<td>Chemistry</td>
<td>6%</td>
</tr>
<tr>
<td>Mathematics</td>
<td>3%</td>
</tr>
</tbody>
</table>

Bioinformatics as a career

SIB Members’ current positions are diverse too

<table>
<thead>
<tr>
<th>Role</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bioinformatician, Computational Biologist</td>
<td>44%</td>
</tr>
<tr>
<td>Others (Biochemist, Structural Biologist, Mathematician…)</td>
<td>16%</td>
</tr>
<tr>
<td>N/A</td>
<td>11%</td>
</tr>
<tr>
<td>Biocurator</td>
<td>9%</td>
</tr>
<tr>
<td>Software Developer</td>
<td>3%</td>
</tr>
<tr>
<td>Development team Leader</td>
<td>2%</td>
</tr>
<tr>
<td>Administrative Staff</td>
<td>1%</td>
</tr>
<tr>
<td>Statistician</td>
<td>4%</td>
</tr>
</tbody>
</table>

Data collected from 91 SIB Members as part of an internal survey (2017)
Some examples of collaboration

Nationwide working groups
Under the lead of its Clinical Bioinformatics Group, SIB is fostering collaborative working in the health sector and in particular, it has set up two working groups on "mutmic mutation calling" and "microwave typing and characterization", which bring together bioinformaticians and clinical actors from all major SWISS HOSPITALS, with the aim of harmonizing routine next-generation sequencing (NGS) practices at the national level.

Innovation/hospitals

The GENEVA UNIVERSITY HOSPITALS (HUG) and SIB's Clinical Bioinformatics and Vital-IT Groups are jointly developing and upgrading OnosBenchTM, a sample-to-report platform used by the HUG Clinical Pathology Division for the management and analysis of NGS patient data in routine cancer diagnosis. The INSTITUTE OF MEDICAL GENETICS OF ZURICH has recently joined the partnership, to further collaborate on the development of a tool serving the specific needs of medical genetics.

Award-winning platform to make data discoverable

The SourceData platform, developed by EMBIO in collaboration with SIB's Vital-IT Group, provides an intuitive interface for researchers and publishers alike, enabling them to share figures and their underlying data in a machine-readable, searchable format. In addition, SourceData provides a public interface where scientists can efficiently find and re-use published results.

Converting ideas into applications
The SIB Legal and Technology Transfer Office ensures seamless collaboration between SIB Groups, academic institutions and the private sector, with over 100 contracts signed each year.

Some papers with high impact

The following SIB (co-)authored papers have been cited over 1,000 times since their publication date:

- STRING v9.1: Protein-protein interaction networks, integrated over the tree of life. By Szklarczyk D et al. NUCLEIC ACIDS RES. 2013
- STRING v9.0: Protein-protein interaction networks, with increased coverage and integration. By Franceschini A et al. NUCLEIC ACIDS RES. 2011
- Arlequin suite ver 3.5: a new series of programs to perform population genetics analyses under Linux and Windows. By Excoffier L and Lischer H. MOLEC ECOL. RESOUR. 2010
- Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. By Speedie J et al. NAT GENET. 2016
- Transcriptome-wide identification of RNA-binding protein and microRNA target sites by PAR-CLIP. By Hartman M et al. CELL 2010
- STRING v8 – a global view on proteins and their functional interactions in 650 organisms. By Jensen L et al. NUCLEIC ACIDS RES. 2009
- ExPaSy: The proteomics server for in-depth protein knowledge and analysis. By Gasteiger E et al. NUCLEIC ACIDS RES. 2003

* Source: European Bioinformatics Institute (EMBL-EBI) - citations are based on open citation data.
CHAPTER 2 20 years of commitment

... and bioinformatics skills development

One of SIB’s missions is to train the next generation of bioinformaticians and to ensure that life and health scientists make the best use of bioinformatics resources, many of which are developed by SIB Groups. The SIB Training team promotes and coordinates training in bioinformatics, both in Switzerland and internationally.

Development of training activities over time

In the early years, the courses organized by SIB consisted mostly of introductions to sequence analysis, protein structure, statistics, phylogeny or evolution. Over time, more advanced and technical topics joined the training portfolio, such as high-performance computing and programming (machine learning, Markov models, advanced R and scripting languages). The courses continued to evolve together with the sequencing techniques. Today, several courses focus on high-throughput data analysis (ChIP-seq, RNA-seq, GWAS and NGS).

PHD TRAINING NETWORK

The PhD Training Network was created as a community support for doctoral students carrying out their research in bioinformatics or computational biology in Switzerland. The Network aims to foster interactions and exchanges among SIB PhD students and train them in the most up-to-date methods necessary for their doctoral research. At the time of its creation in 2007, the Network had 19 PhD students in Lausanne, Basel and Geneva. Ten years later, it has seen close to 170 alumni defending their PhD thesis. Today, it has 240 members spread across eight Swiss universities.

TRAINING WITHOUT BORDERS

Benefiting from strong connections with the international and European bioinformatics training community, the SIB Training team regularly co-organizes and hosts workshops, such as the “Workshop in Education for Bioinformatics” for the International Society for Computational Biology, or the ELIXIR “Train the trainer workshop”, which is bringing the European training community to Switzerland.

What do the participants say?

96% consider that SIB courses are excellent or very good
94% consider that SIB courses have the right balance between theoretical and practical content
93% consider that SIB courses are relevant to their work

Some of the most taught skills

Best coding practices HPC Machine learning
ChIP-seq R software Metabolomics
Statistics Long read sequencing Protein databases
Experimental design Variant annotation Regular expressions
Next-generation sequencing RNA-Seq
UNIX Python Proteogenomics and immunopeptidomics
Computer-aided drug design Snakemake
Sensitive data management Perl 6

FIGURES FROM 2017

9 courses 193 participants
2 courses 98 participants
9 courses 193 participants

2007
2006
1998
Open access does not mean free of cost, especially for databases that need significant human resources. With the increasing demand for open data production, the question is a pressing one, and SIB has conducted a study to identify the most suitable funding model to ensure that open access to essential biological information can be maintained.

## What are core data resources for the life sciences?

Christina Durinx. Imagine a centralized encyclopaedia of the function of hundreds of thousands of proteins, made accessible to life scientists around the world. The database is manually curated by a team of SIB experts who facilitate data reuse and maximize the value of biological data. This is an example of what is referred to as a core data resource on the international scene. Such resources are of fundamental importance to the broad life science community and the long-term preservation of biological data. They provide complete collections of generic value to life science, and show high levels of usage, scientific quality and service.

What is the threat they are facing?

CD Today’s life science research cannot be conducted without data resources. The issue is that most open-access databases are currently financed by research grants and institutional funds. If a database applies for a research grant, it will compete against research projects. Moreover, such grants are cyclical, so they are not stable over time.

The situation is actually quite dramatic: a recent survey revealed that two-thirds of databases have one year or less of guaranteed funding.

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We should bear in mind that scientists have entrusted their research results to these databases. So the fact that their existence is so unstable could result in the loss of an immense wealth of biological information, and the associated investments.

What is the study about and what is the outcome?

CD Under the Infrastructure model, funding agencies would set aside a fixed percentage of their research grants. The amount would subsequently be re-distributed to core data resources according to well-defined selection criteria (see above). Based on existing data, we have been able to estimate that less than 1% of the total amount dedicated to research grants in the life sciences would be sufficient to cover the costs of the core data resources worldwide.

Of course, this model requires major changes in how funds are now distributed and necessitates collaboration at an international level.

### Why is SIB preoccupied with this issue?

CD Caring for sustainability really is in our Institute’s DNA and raison d’être. SIB was founded following a funding crisis, to ensure the long-term sustainability of the Swiss-Prot database, which is now part of the UniProt consortium. Twenty years after SIB’s creation, the topic of the long-term sustainability of its knowledgebases is still a hot topic.

SIB is working closely with ELIXIR, an intergovernmental organization that is building a sustainable infrastructure for biological information across Europe. This study was done in the context of the ELIXIR Data Platform and benefited from ELIXIR’s support. SIB also plays an active role in an international coalition that is working on a mechanism to provide global support for core data resources. Joining forces globally is the key to the long-term sustainability of knowledge preservation.

CD Making open data sustainable: the example of UniProtKB/Swiss-Prot

While the importance of knowledgebases is increasingly recognized, the question of their sustainability is frequently raised. How can a tight number of curators keep up with the rapid growth in biomedical literature, i.e. currently over one million papers published a year? A study led by SIB’s Swiss-Prot Group reveals that in fact, no more than 2-3% of all publications indexed in PubMed each year appear to be relevant for UniProt curation. The sheer volume of biomedical papers published each year is therefore a poor way to evaluate biocuration effort.

These results demonstrate, for one thing, that expert curation in UniProt can keep up with the increasing number of biomedical publications. They also underline the fact that, now more than ever, curators play a crucial role in shedding light on biomedical knowledge, to support researchers in identifying the most relevant and substantiated evidence.

**Expert curation is sustainable:** UniProtKB/Swiss-Prot as a case study

By Poux S et al. Bioinformatics, 2017

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**References**

- Funding knowledgebases: Towards a sustainable funding model for the UniProt use case
- By Gabriela C et al., F1000Research 2018, 6(ELOXIR) 2021
- [Watch the interview online on our Youtube channel](https://www.youtube.com/watch?v=Geo-gI0w2ZV)

---

**Watch this interview online**

- **UniProtKB/Swiss-Prot** is a well-known and widely used protein information database in the knowledgebase. It is the most widely used protein information database in the world. The database is manually curated by a team of SIB experts who facilitate data reuse and maximize the value of biological data. This is an example of what is referred to as a core data resource on the international scene. Such resources are of fundamental importance to the broad life science community and the long-term preservation of biological data. They provide complete collections of generic value to life science, and show high levels of usage, scientific quality and service.

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Two decades of SIB

1986

**BIRTH OF SWISS-PROT**
Amos Bairoch, then a PhD student at the University of Geneva, creates what has since become the world reference resource for protein knowledge, UniProtKB/Swiss-Prot. At that time, the various versions are distributed on magnetic tapes!

1993

**CREATION OF THE FIRST LIFE SCIENCE WEB PORTAL**
To exchange information within the community, Ron Appel launches the first life science server, which is one of the top 50 first websites in the world. It is accessed 7,295 times even during its first month of existence, and over 2 million times per month a few years later.

1996

**A FUNDING CRISIS LEADING TO SIB’S CREATION IN 1998**
The Swiss-Prot knowledge-base, which by then has around 100,000 users, is threatened with extinction due to a lack of financial support. Thousands of support messages from the international scientific community flow in. As a result, the Swiss authorities recommend that a stable long-term funding mechanism be sought to ensure the survival of Swiss-Prot and bioinformatics, in the form of a publicly funded foundation.

1998

**CREATION OF GENEBIÖ**
Created to facilitate fund matching between industries and the Swiss government, GeneBIÖ, SIB’s commercial entity, was first focused on protein identification and characterization as well as proteome imaging.

2018

**A WEB SERIES, A MOBILE GAME AND A BOOK…**
To mark our 20th anniversary in a surprising way, different projects were launched to promote bioinformatics within and outside the research community during the year:
- Share the everyday life and challenges of a bioinformatician in a 7-episode humorous web series;
- With the mobile game “Genome Jumper”, explore our genetic variability and why we have curly or straight hair, or lack the capability to digest milk;
- Discover the colourful and surprising side of SIB’s Group Leaders, captured by Swiss photographer Nicolas Righetti, while learning about the history of bioinformatics in a science and art book.

Stay tuned for the release of these projects on www.sib.swiss/20years

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**20 YEARS OF SIB!**
A year of festivities and projects to celebrate 20 years of commitment to life sciences and health.

**CERTIFICATE OF ADVANCED STUDY IN PERSONALIZED MOLECULAR ONCOLOGY**
First of its kind in Switzerland, the CAS is launched jointly by the University Hospitals of Basel and Lausanne and SIB.

**NEW PARTNER**
**SWISS INSTITUTE OF ALLERGY AND ASTHMA RESEARCH (SIAR), DAVOS**

---

**CREATION OF THE SIB SWISS INSTITUTE OF BIOINFORMATICS**
A team of visionary Swiss scientists (Ron Appel, Amos Bairoch, Philipp Bucher, Victor Jongeneel and Manuel Peitsch) unite to create one sole bioinformatics organization. It includes five founding groups from the Universities of Geneva and Lausanne, the Geneva University Hospitals (HUG), the Ludwig Institute for Cancer Research, the Institut Suisse de Recherche Expérimentale sur le Cancer (ISREC) and Glaxo Wellcome Experimental Research.*

**Number of SIB (co-)authored publications since 1998**
5 groups
20 members
6 institutional partners

**Key articles**
- Lischer H in Mol Biol Evol 2005
- Jensen L et al. in Nucleic Acids Res. 2009
- Speliotes E et al. in Nat Genet. 2009
- Landgraf P et al. in Nature 2004
- Bucher, Martin, et al. in Nucleic Acids Res. 2007

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**Number of SIB Members**
20

---

* The HUG remained an institutional partner of SIB until 2006, before joining again in 2016; ISREC remained until 2008 when it was integrated into EPFL and Glaxo Wellcome Experimental Research until 2002.

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**SIB Profs’ 2019**

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**SIB Profs’ 2019**
CHAPTER 3

Activities 2017

SIB’s activities revolve around five pillars:

I. DATABASES AND SOFTWARE TOOLS
II. CORE FACILITIES AND COMPETENCE CENTRES
III. PERSONALIZED HEALTH
Community
IV. TRAINING IN BIOINFORMATICS
V. SCIENTIFIC COLLABORATION

Discover our latest developments and achievements on each of these fronts in the following pages.
Providing life scientists with a complete bioinformatics infrastructure

I. DATABASES AND SOFTWARE TOOLS

Most of the resources developed by SIB Groups are available in open access via the SIB bioinformatics resource portal ExPASy (www.expasy.org) and cover a broad range of domains. Here are a few examples:

<table>
<thead>
<tr>
<th>DOMAIN</th>
<th>SUB-DOMAIN</th>
<th>DATABASE</th>
<th>SOFTWARE TOOL</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genes and genomes</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sequence alignment</td>
<td></td>
<td>LAALIGN, Newick Utilities, T-Coffee</td>
<td></td>
</tr>
<tr>
<td>Similarity search</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Characterization/ annotation</td>
<td></td>
<td>BLAST, BLAST+, LAALIGN, Phyloglob</td>
<td></td>
</tr>
<tr>
<td>Transcriptsomics</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Protein sequence and identification</td>
<td></td>
<td>exPASy, UniProtKB/Swiss-Prot, VirusZone</td>
<td></td>
</tr>
<tr>
<td>Protein functionality and 2-DE data</td>
<td></td>
<td>SWISS-2D PAGE, WORLD-2D PAGE Repository</td>
<td></td>
</tr>
<tr>
<td>Protein characterization and function</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Families, domains and motifs</td>
<td></td>
<td>MyTree, PROSITE</td>
<td></td>
</tr>
<tr>
<td>Post-translational modification</td>
<td></td>
<td>MyDomains, pfTools, PRATT</td>
<td></td>
</tr>
<tr>
<td>Protein-protein interaction</td>
<td></td>
<td>STRING, UniProtKB/Swiss-Prot</td>
<td></td>
</tr>
<tr>
<td>Similarity search/ alignment</td>
<td></td>
<td>BLAST, ClusterW, MyHits</td>
<td></td>
</tr>
<tr>
<td>Imaging</td>
<td></td>
<td>ImageMaster / Melanie, M_VERBOSE</td>
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<tr>
<td>Proteins and proteomes</td>
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<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Evolution and phylogeny</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Structural biology</td>
<td></td>
<td>SWISS-MODEL Repository, SwissSideChain, SwissBioIsostere</td>
<td></td>
</tr>
<tr>
<td>Systems biology</td>
<td></td>
<td></td>
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</tr>
</tbody>
</table>

SIB core resources

A stringent process to ensure the highest level of excellence

SIB’s External Scientific Advisory Board (SAB) is in charge of providing recommendations on the selection of SIB’s core resources every four years. Assessing competitive criteria, including the scientific impact and return-on-investment, along with SIB’s global portfolio management and strategic orientation, SAB members select best-in-class resources and provide guidelines for their continuous development. Once validated by the Board of Directors, the SIB management team then assists resource managers in implementing their action plan and exchanging best practices.

In 2017, SIB core resources were used by about six million users worldwide

Source: Google Analytics

<table>
<thead>
<tr>
<th>Protocols &amp; proteomes</th>
<th>STRING</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>UniProtKB/Swiss-Prot</strong></td>
<td>Protein knowledgebase</td>
</tr>
<tr>
<td><strong>neXtProt</strong></td>
<td>Human protein knowledgebase</td>
</tr>
<tr>
<td></td>
<td>Glycomics@ExPASy</td>
</tr>
<tr>
<td></td>
<td>SWISS-MODEL</td>
</tr>
</tbody>
</table>

**UniProtKB/Swiss-Prot**

**TYPE**: Knowledgebase with manual expert curation

**DESCRIPTION**: Hundreds of thousands of protein descriptions, including function, domain structure, subcellular location, post-translational modifications and functionally characterized variants.

**HIGHLIGHT**: UniProtKB/Swiss-Prot is the expert curated part of UniProt, the most widely used protein information resource in the world, with over six million page views per month. Selected as an ELIXIR core resource.

**SEE P. 59**

**neXtProt**

**HIGHLIGHT**: Currently includes 9,643,763 proteins from 2,031 organisms and is the most used resource of its kind. Selected as an ELIXIR core resource.

**SEE P. 59**

**STRING**

**TYPE**: Knowledgebase

**DESCRIPTION**: Resource for known and predicted protein-protein interactions, including direct (physical) and indirect (functional) associations derived from various sources, such as genomic context, high-throughput experiments, co-expression and the literature.

**HIGHLIGHT**: Currently includes 8,149,013 proteins from 1,808 organisms and is the most used resource of its kind.

**SEE P. 59**

**Glycomics@ExPASy**

**HIGHLIGHT**: Currently includes 9,643,763 proteins from 2,031 organisms and is the most used resource of its kind. Selected as an ELIXIR core resource.

**SEE P. 59**

**SWISS-MODEL**

**HIGHLIGHT**: Currently includes 8,149,013 proteins from 1,808 organisms and is the most used resource of its kind. Selected as an ELIXIR core resource.

**SEE P. 59**

In 2017, SIB core resources were used by about six million users worldwide

Source: Google Analytics
SwissLipids NEW
A knowledge resource for lipids

**TYPE** Knowledgebase  
**DESCRIPTION** Information about known lipids, including knowledge of lipid structures, metabolism, interactions, and subcellular and tissue localization, providing a framework for the integration of lipid and lipidomic data with biological knowledge and models.  
**HIGHLIGHT** Contains information on over 400,000 lipid structures from over 180 major lipid classes.  
SEE P. 59

**SwissLipids** NEW
**DESCRIPTION** Web-based computer-aided drug design tools, from molecular docking (SwissDock) to pharmacokinetics and drug likeness (SwissADME), through virtual screening (SwissSimilarity) and target prediction of small molecules (SwissTargetPrediction).  
**HIGHLIGHT** Comprehensive and integrated web-based drug design environment.  
SEE P. 63

**EPD**
**DESCRIPTION** Web portal for regulatory genomics, including genome-wide annotations of regulatory sites and motifs, the webserver ISMARA for automated inference of regulatory networks and CRUNCH for automated analysis of ChIP-seq data.  
**HIGHLIGHT** User can upload raw microarray, RNA-seq or ChIP-seq data to automatically infer the core regulatory networks acting in the system of interest.  
SEE P. 57

**OrthoDB**
The hierarchical catalogue of orthologs

**TYPE** Automated phylogenomic database and software tool  
**DESCRIPTION** Comprehensive online catalogue of orthologs, including functional and evolutionary gene annotations and enabling the inference of putative gene functions.  
**HIGHLIGHT** Largest orthology resource, enabling users to generate publication-quality comparative genomics charts, and to upload, analyse and interactively explore their own pre-publication data.  
SEE P. 58

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SEE P. 58
II. CORE FACILITIES AND COMPETENCE CENTRES

Swiss life scientists with computational needs can count on a tight mesh of expert hubs in Switzerland whether they need access to high-performance computing resources, software development services, cutting-edge multidisciplinary bioinformatics expertise, specialized training – or all these at once.

Competence centres for life scientists

Vital-IT, an SIB-staffed group, started off as a small-scale high-performance computing centre in 2007. Now covering Western Switzerland and comprising a multidisciplinary team of scientists and technical staff, the group is involved in a wide diversity of projects on a national and international scale.

What is the added value of competence centres such as Vital-IT for life scientists? Ioannis Xenarios. Scientists have several options for keeping up with the increase in the amount and diversity of data generated today.

Some groups – originally ‘experimentalists at heart’ – have, for example, been able to successfully expand their competencies by hiring computational biologists, bioinformaticians or IT staff internally. Those who do not have the ‘luxury’ of having a bioinformatician in their own premises can turn to competence centres, which can address a diverse range of needs. In addition to taking the computational workload off the shoulders of the research groups, competence centres such as Vital-IT also provide software, storage and long-term data maintenance.

If these tasks were to be performed by the individual groups themselves, it would multiply the costs for their respective institutions. Competence centres thus offer an economy of scale and a coordinated environment, where expertise is shared amongst their members.

What sort of activities do you typically conduct? Ioannis Xenarios. Depending on the needs of the groups, we provide three types of activities. If the need is purely related to computing power or storage, we provide the necessary IT infrastructure. For groups that do not have the means to actually hire a bioinformatician, we also provide up to a dozen different types of expertise, according to the project. Interdisciplinarity is the key word here.

Finally, for scientists and groups that are not quite there yet in terms of generating their own data, but know that there are data out there and want to ‘probe’ the field, we help them to identify whether the investment and move towards a computational approach is worth it. This is therefore more a consulting and strategic counselling type of activity.

Since the beginning of 2017, the Swiss National Science Foundation (SNSF) allows researchers to include the costs of infrastructure usage for their projects in their grant application. What does this move represent, in your view? Ioannis Xenarios. Overall, the decision of the SNSF – and nearly all European funding agencies – is emphasizing the fact that, without provision for access to computational infrastructure and bioinformatics expertise, life scientists would not be able to function in the 21st century. However, the road to making all generated data compliant with Findable, Accessible, Interoperable and Re-usable (FAIR) principles has a cost that is often underestimated.

When writing this part of their grant application, life scientists should, for example, include not only the data generation aspects, but also the data and resource maintenance aspects beyond the end of the project, as these are likely to exceed the duration of their project grant by a long way. This is even more important in the light of the reproducible science arena, where this cost is rarely taken into account.

While the SNSF’s decision is a first step in the right direction, competence centres will play a crucial role in sensitizing and training scientists with whom they collaborate to the importance of data management – a role Vital-IT has been playing for the last 15 years.
A national network to support the computational needs of life scientists

A 2017 snapshot

Twelve core facilities and competence centres are distributed in all the major Swiss academic institutions, and coordinated by SIB.

GL = Group leader(s)
SI = Supporting institution(s)
IU = Infrastructure usage, when applicable
PB = Petabytes of storage capacity
CP = Core processing units
L = Location(s)

Vital-IT

GL Ioannis Xenarios
SI SIB, EPFL, University of Lausanne, University of Geneva, University of Bern, University of Fribourg
IU 1,200 AU
8 PB
9,900 CP
L Lausanne, Geneva, Bern, Fribourg
SEE P. 71

Bioinformatics 2 Core Facility (BCF)

GL Mauro Delorenzi and Frédéric Schütz
SI SIB, University of Lausanne
L Lausanne
SEE P. 68

Bioinformatics 3 Unravelling Group (BUGFri)

GL Laurent Falquet
SI University of Fribourg
L Fribourg
SEE P. 69

FMI 4 Computational Biology Group (FMICBG)

GL Michael Stadler
SI Friedrich Miescher Institute
IU 170 AU
1 PB
144 CP
L Basel
SEE P. 69

DBM 5 Bioinformatics Core Facility

GL Robert Ivanek
SI University of Basel, University Hospital Basel
L Basel
SEE P. 69

Centre for 6 Scientific Computing (sciCORE)

GL Torsten Schwede and Thierry Sengstag
SI SIB, University of Basel
IU 750 AU
8 PB
6,500 CP
L Basel
SEE P. 70

Interfaculty 7 Bioinformatics Unit (IBU)

GL Rémy Bruggmann
SI University of Bern
IU 150 AU
2 PB
600 CP
L Bern
SEE P. 70

Bioinformatics 8 Core Unit (BCU)

GL Luciano Cascione
SI Institute of Oncology Research
IU 20-40 AU
0.05 PB
80 CP
L Bellinzona
SEE P. 69

Scientific 9 Information Services (SIS)

GL Bernd Rinn
SI ETH Zurich
IU 2,600 AU
5.5 PB
67,000 CP
L Zurich, Basel
SEE P. 71

Service and Support for Science IT (S3IT) facility

GL Marcel Riedi
SI University of Zurich
IU 1,000 AU
7 PB
7,000 CP
L Zurich
SEE P. 70

NEXUS 10 Personalized Health Technologies

GL Daniel Stalkev
SI ETH Zurich
IU 50 AU
0.2 PB
192 CP
L Zurich
SEE P. 71

FGCZ Genome Informatics (FGCZ-GI)

GL Hubert Rehrauer
SI ETH Zurich, University of Zurich
IU 1,081 AU
0.3 PB
480 CP
L Zurich
SEE P. 69

Interfaculty 11 Bioinformatics Unit (IBU)

GL Rémy Bruggmann
SI University of Bern
IU 100 AU
2 PB
600 CP
L Bern
SEE P. 68

Centre for 12 Scientific Computing (sciCORE)

GL Torsten Schwede and Thierry Sengstag
SI SIB, University of Basel
IU 750 AU
8 PB
6,500 CP
L Basel
SEE P. 70

Bioinformatics 13 Core Unit (BCU)

GL Luciano Cascione
SI Institute of Oncology Research
IU 20-40 AU
0.05 PB
80 CP
L Bellinzona
SEE P. 69
III. PERSONALIZED HEALTH

SIB is playing a leading role in the Swiss personalized health landscape, by sharing its expertise with hospitals and by building the nationwide infrastructure that will enable patients’ data to be used for research.

The group provides expertise and support for the organization, analysis and interpretation of patient-related data (e.g. omics data), converting them into clinically useful information for health professionals.

It notably contributes to:

- Establishing consensus and good practices for diagnostic omics analyses across Swiss hospitals;
- Building partnerships with Swiss hospitals to develop, implement and sustain state-of-the-art diagnostic tools;
- Providing clinical bioinformatics training across Switzerland;
- Facilitating interactions between SIB’s research groups and the medical realm for clinical research projects.

2017 highlights

**WORKING GROUPS**

**THE SOMATIC MUTATION CALLING** working group has started to benchmark Swiss hospitals next-generation sequencing (NGS) practices and define common guidelines for the analysis and interpretation of genetic variants in cancer diagnosis.

The group is also a partner of SIB in the Swiss Variant Interpretation Platform, part of the BioMedIT project (SPHN), which aims to provide a central resource for clinicians and researchers to annotate and interpret clinically verified variants in cancer (see next page).

**THE MICROBE TYPING AND CHARACTERIZATION** working group has also started benchmarking NGS practices in the diagnostic of infectious diseases, in both virology and bacteriology.

**PARTNER HOSPITALS**

At the Geneva University Hospitals, OncoBench™ is now running in version 2, with new features and major improvements.

The Institute of Medical Genetics of Zurich has also entered the partnership to jointly develop a tool for the management and analysis of medical genetics data.

**CLINICAL RESEARCH COLLABORATIONS**

The group has been involved in several clinical projects, focusing on research and/or the development of resources of clinical utility. In particular, it is a partner in a multi-site NRP72 project led by the University Hospital of Basel, on the development of a Swiss surveillance database for molecular epidemiology of multi-drug resistant pathogens.

Launch of a Certificate of Advanced Studies

SIB has partnered with the University Hospitals of Basel and Lausanne to launch a unique, multi-disciplinary and multi-site Certificate of Advanced Studies (CAS) on personalized molecular oncology. The first of its kind in Switzerland, this programme aims to train the next generation of professionals from various backgrounds in this rapidly evolving field. www.pmo.unibas.ch

In 2017, SIB Clinical Bioinformatics group was also involved in several outreach-related events, to foster discussions and exchange ideas on personalized health with non-specialists: Expand Your Horizons, Café Scientifique and Leenaards SantéPerso.
SIB and the Swiss Personalized Health Network initiative (SPHN)

SIB PERSONALIZED HEALTH INFORMATICS GROUP (PHI)

The group contributes to the SPHN initiative in a number of ways. As the group in charge of the Data Coordination Centre (DCC), it establishes standards for the interoperability of health-related data. These standards will allow for efficient nationwide collaborative research across hospitals and academic research institutions. The group is also coordinating a network of bioinformatics core facilities at Swiss universities, to provide secure IT infrastructure and support personalized health research projects (BioMedIT project directly managed by SIB, see below).

INFORMATICS GROUP (PHI)

TAGS: interoperability; personalized health; data management; standardization; precision therapy; information security

FIRST INFRASTRUCTURE STEP FOR BIOMEDIT

In November 2017, the green light was given to launch the first step of the infrastructure implementation under the leadership of SIB Clinical Bioinformatics, together with the Vital-IT and SIS (ETH Zurich) Groups. SIB hospitals and SIB have decided to join forces to develop a common infrastructure allowing hospitals to share and unify the clinical interpretation of the genetic variants identified in their patients all over Switzerland.

WORKSHOP ON INFORMATION SECURITY AND DATA PROTECTION

The 2-days workshop took place in December and gathered about 40 SPHN stakeholders from Swiss university hospitals and research institutions, including specialists in IT security, data protection, legal and governance aspects.

A “driver” project for oncology treatment and research

SIB PI OLIVIER MICHELIN (IN ASSOCIATION WITH SIB’S JACQUES FELLAY, PATRICK RUCH AND SYLVAIN PRADERVAND)

This “driver” project aims to adapt the clinical information flow in order to capture essential data, which can then be used for treatment and research. It will be collaborating with the “Multidisciplinary Multicentre Molecular and Cellular Cancer Consortium” project.

A “driver” project for inflammatory disease immunotherapy

SIB PI MANFRED CLAASSEN

This Swiss-wide SPHN/PHRT* driver project aims to identify cell population biomarkers and therapeutic targets in inflammatory disease immunotherapy.

In addition to the involvement of the PHI Group, several other SIB Groups are playing an active role in the SPHN initiative via project calls and the implementation of BioMedIT.

The results of SPHN’s first project call were issued in December 2017. Of the 16 projects selected, five have SIB Group Leaders as co-applicants or co-Principal Investigators and three are led by SIB Group Leaders. These SIB-led projects include:

An “infrastructure development” project on cancer variants annotation

SIB PI DANIEL STEKHOVEN AND PATRICK RUCH

The Swiss Variant Interpretation Platform for Oncology (SVIP-O) aims to provide a central resource for clinicians and researchers to annotate and interpret clinically verified variants in cancer. The database and the governance mechanisms have been launched in the context of BioMedIT (see ‘First infrastructure step for BioMedIT’).

Creating a strong bioinformatics community

IV. TRAINING IN BIOINFORMATICS

23 SIB groups engaged in teaching activities

1,191 training days

81 experts and trainers

53 courses and workshops

94 training days

Stronger governance for training

The governance of the group was strengthened in 2017 to determine its strategy in terms of course content and audience reach.

Two SIB Group Leaders, Mark Robinson and Marc Robinson-Rechavi, have been elected by their peer Group Leaders to represent them on the SIB Training Committee. They have joined the four Group Leaders whose groups have long been involved in training activities: Romy Bruggmann, Mauro Delorenzi, Torsten Schwede and Ioannis Xenarios.

Moreover, three external scientists have joined the Committee to represent the academic and industry-based life scientists: Pierre Farmer (Novartis), Florian Steiner (UNIGE) and Christoph Handschin (Biozentrum).

Marc Robinson-Rechavi has agreed to serve as chair of the Committee and works closely with Patricia Palagi, Team Leader of the Training Group.

2017 highlights

In 2017, the network celebrated its 10th anniversary with a special conference for its 240 members in Bio/Bienne. Other events in 2017 included the autumn school in “Machine Learning Applied to Systems Biology”, jointly organized with SystemsX.ch, and the course on “Best practices in programming”, which were both very well attended.

Who took part in SIB courses in 2017?

PhD students 43%

Postdocs 26%

Principal Investigators 4%

Laboratory technicians 11%

Other scientists 14.5%

Other 5%

SIB PhD Training Network

In 2017, the network celebrated its 10th anniversary with a special conference for its 240 members in Bio/Bienne. Other events in 2017 included the autumn school in “Machine Learning Applied to Systems Biology”, jointly organized with SystemsX.ch, and the course on “Best practices in programming”, which were both very well attended.

Reaching out to new training audiences

SIB hosted a half-day workshop to present its computational biology resources to the Biology17 conference’s participants and is repeating this positive experience in 2018 (see page 47).

Scientists from the industry are also in need of quality training in bioinformatics. Reaching out to them is part of the new training strategy and action plan.

Who took part in SIB courses in 2017?

Other scientists 14.5%

PhD students 43%

Postdocs 26%

Principal Investigators 4%

Laboratory technicians 11%

Other 5%

The SIB Training portfolio is constantly evolving to meet the scientific community’s needs: find the full list of courses at www.sib.swiss/training

Creating a strong community

Creating a strong community
V. SCIENTIFIC COLLABORATION

SIB is a unique ecosystem, in which Swiss bioinformatics service providers rub shoulders with users – researchers working at the interface between experimental and computational biology – on a daily basis. It thus ensures that the two types of scientists understand each other and collaborate optimally. This is an essential ingredient in SIB’s core mission, which is to provide cutting-edge bioinformatics infrastructure in line with the fast-paced evolution of life science.

Key scientific events and research outcomes in 2017

At SIB, fostering scientific collaboration means creating opportunities for our scientists to exchange know-how, as well as promoting their work. Some of the actions implemented in 2017 are listed below, together with a digest of some of the research results released during the year.

14.12.2017
2,000+ follower on Twitter

Congratulations to the winners of the SIB Awards and [BC]2 Best Posters Awards 2017

SIB International Young Bioinformatics Award – CHF 10,000
SEBASTIAN WASEZAK, EMBL (Germany), for his outstanding work in computational and statistical biology. Following a PhD at EPFL in Bart Deplancke’s Lab, he is now a postdoc at EMBL in the group led by Jan Korbel.

SIB Bioinformatics Resource Innovation Award – CHF 10,000
The drug discovery resource REPURPOSEDB - REFERENCE DATABASE OF DRUG REPOSITIONING INVESTIGATIONS, represented by Khader Shameer. This resource includes computational drug discovery tools such as drug similarity searches, and was developed at the Dudley Lab, led by Joel Dudley, at the Institute for Genomics and Multiscale Biology, Icahn School of Medicine at Mount Sinai (USA).

[BC]2 - Basel Computational Biology Conference
Co-organized by SIB and the Biozentrum University of Basel.
Key computational biology event of the year in Switzerland.
This 13th conference attracted over 500 participants from all over the globe. Among them were world-leading researchers alongside promising junior scientists, who returned home with prestigious awards.

LS² Annual meeting, Zurich
International scientists from all backgrounds gather to explore the diverse range of life science topics.
A symposium on personalized medicine in the context of the Swiss Personalized Health Network was chaired by SIB Group Leader Torsten Schwede.

Biology17, Bern
The Swiss conference on organismic biology and joint congress of the Swiss Zoological Society, the Swiss Botanical Society, and the Swiss Systematics Society.
First incursion of computational biology at the conference, with an SIB workshop during which Patricia Palagi, Joshua Payne, Alexandria Gavryushkina, and Christophe Dessioz respectively presented the institution and the following resources: Genonets, BEAST2 and OMA.

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A toolbox to improve genome annotation

With thousands of prokaryotic genomes newly sequenced each year, solutions for accurately annotating these genomes and thereby providing the basis for uncovering the functions of genes and their encoded proteins are urgently needed. SIB scientists have developed an open proteogenomics toolbox to allow researchers to obtain more accurate and complete genome annotations.

Harmful mutations and range expansion: computers got it right

Organisms that are expanding their spatial range suffer from a drastic decrease of fitness over time due to the accumulation of harmful mutations. A study led by a team of scientists from SIB and the University of Bern now provides the first experimental evidence of this theory.

Published in Genomics Research

SIB Groups involved

Bioinformatics and Proteogenomics Group

Population Genetics Group

Interfaculty Bioinformatics Unit (IBU)

A gene increases the severity of common colds

Colds that are not linked to influenza are generally benign. Nonetheless, 2% of each generation of children have to go to hospital following a virulent infection. An international research collaboration led by scientists from SIB and EPFL has discovered the reason for some of these infections: they are caused by mutations of a gene that plays a part in recognizing certain cold-inducing viruses.

Supercolling pushes molecular handcuffs along chromatin fibres

As it squeezes down the chromatin fibre, the cohesion protein complex extrudes a growing loop of DNA—a bit like the quick-lacing system of trail-running shoes. But what is powering the movement of the protein? A team of SIB scientists has found that the driving force could be the supercolling of upstream DNA. Their research is thereby adding a key piece to the puzzle of gene expression regulation.

Published in Nuclear Acids Research

SIB Group involved

DNA and Chromosome Modelling Group

A thorough 'catalogue' of an aggressive type of childhood cancer

Improving cancer precision therapy requires a greater ability to identify and describe groups of patients who share the same molecular and clinical particularities. An SIB group recently played a pivotal role in building the most thorough 'catalogue' of an aggressive type of childhood cancer—thus providing a basis for novel therapeutic treatments.

Published in Cancer Cell

SIB Group involved

Computational and Molecular Biology Group

Population Genetics Group

Supercoiling pushes molecular handcuffs along chromatin fibres

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Published in Nucleic Acids Research

SIB Group involved

DNA and Chromosome Modelling Group

A study led by researchers at SIB, the University of Lausanne and EPFL has revealed that the genome of an emblematic tree of the University campus, the Napoleon oak, has changed little during its first 234 years of existence. This surprising result suggests that the tree is capable of protecting itself from an accumulation of harmful genetic mutations.

Published in Genome Research

SIB Groups involved

Computational and Molecular Biology Group

Population Genetics Group

Interfaculty Bioinformatics Unit (IBU)

The genome of a 234-year-old oak tree, young as ever

A study carried out by researchers at the University of Lausanne and SIB has revealed that the genome of an emblematic tree of the University campus, the Napoleon oak, has changed little during its first 234 years of existence. This surprising result suggests that the tree is capable of protecting itself from an accumulation of harmful genetic mutations.

Published in Genome Research

SIB Groups involved

Computational and Molecular Biology Group

Population Genetics Group

Interfaculty Bioinformatics Unit (IBU)

From cancer evolution to personalized therapies

A study led by researchers at SIB, the University of Lausanne and EPFL provides a promising framework to anticipate drug resistance in cancer, by predicting the co-occurrence of tumour alterations and their response to common cancer drugs. The research showed that while some co-occurrences between genomic alterations confer a resistance on tumour cells to particular drugs, they also make them sensitive to others.

Published in Cancer Cell

SIB Group involved

Computational Systems Oncology

The quick-change artist strategy of a fungus that causes pneumonia

A study led by a team of researchers from the Institute of Microbiology of CHUV-UNIL and SIB reveals the mechanisms of antigenic variation used by the pathogen Pneumocystis jirovecii to escape the human immune system.

Published in Microbiology

SIB Group involved

Vital-IT

Evolutionary Bioinformatics Group

Bioinformatics Core Facility (BCF)

The 16 genetic markers that can cut a life story short

The answer to how long each of us will live is partly encoded in our genome. Researchers have identified the largest set of markers of lifespan uncovered to date. About 10% of the population carries some configurations of these markers that shorten their life by over a year compared with the population average. Spearheaded by scientists from SIB, the Lausanne University Hospital (CHUV), the University of Lausanne and EPFL, the study provides a powerful computational framework to uncover the genetics of our time of death, and ultimately of any disease.

Published in Nature Communications

SIB Groups involved

Statistical Genetics Group

Laboratory of Systems Biology and Genetics

Evolutionary Bioinformatics Group

Research at SIB in 2017

Full references of the papers mentioned are available at www.sib.swiss/about-us/media-homepage/news-2017
Fields of activity of SIB Groups

Bioinformatics is an interdisciplinary field, where the encounter of genetics, physiology, chemistry or physics leads to many fields of activities and applications.

Genes and genomes
Life’s instruction manual

A genome is the sum of genetic material of an organism, including all of its genes. It is composed of DNA and contains all the information needed to create and maintain an organism, as well as the instructions on how this information should be expressed.

Bioinformatics develops tools able to read genomes, and store, analyse and interpret the resulting data.

SEE P. 52

Proteins and proteomes
More than meets the eye

A proteome is the sum of proteins expressed by a cell, a tissue or an organism, at a given time. Proteins are the products of genes, and are involved in nearly every task carried out within an organism – from carrying oxygen to fighting off pathogens.

Bioinformatics develops tools to understand the role of proteins.

SEE P. 58

Evolution and phylogeny
Splitting ends

Changes that occur in genomes tell life scientists how an organism has evolved over time. Comparisons made between genomes of different species or populations tell us how species are related to one another – this is the field of phylogenetics.

Bioinformatics develops tools able to compare the genomes of organisms, as well as computing methods to reconstruct their past and build their ‘family’ trees.

SEE P. 60

Structural biology
The third dimension

Macromolecules such as DNA and proteins have specific 3D structures that are dictated by their sequence. A protein’s function is defined by its 3D structure, or architecture, which in turn defines the way it reacts with other molecules.

Bioinformatics develops software to create 3D models of proteins to study their interactions with other molecules, such as drugs.

SEE P. 63

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Bioinformatics develops tools able to compare the genomes of organisms, as well as computing methods to reconstruct their past and build their ‘family’ trees.

SEE P. 60

Core facilities and competence centres
The means to an end

The quantity of data generated by the life sciences has grown exponentially over the years, and needs to be stored and processed. Researchers also need help in interpreting their data. Core facilities centralize research resources, and provide tools, technologies, services and expert consultation to this end.

SIB coordinates 12 core facilities and competence centres located in the major Swiss academic institutions.

SEE P. 68

Systems biology
Never alone

Life occurs and is sustained by a mesh of interactions within and between cells, tissues, organisms, and their environment. Understanding how these complex systems function allows scientists to predict what happens if one of the components changes or the conditions are altered.

Bioinformatics develops models to delineate metabolic pathways.

SEE P. 64

Text mining and machine learning
Rise of the machines

Text mining extracts high-quality information from texts. Algorithms are designed to recognize patterns and trends within text so that computers can read and extract the information required. Computers can also be given the ability to learn without being explicitly programmed: this is the promising field of machine learning.

Bioinformatics develops text-mining tools that can be of immense value to complement expert biocuration. It can also rely on machine learning techniques to predict the outcome of complex interactions such as in antibiotic resistance.

SEE P. 71

Many areas of applications, including...

Agriculture
from predicting the spread of bird flu outbreaks and understanding the lifecycle of agricultural pests, to improving crop productivity.

Basic research
from unravelling the evolutionary processes that have shaped today’s biodiversity, to solving the equation behind a lizard’s scale colour pattern.

Medicine and health
from designing optimized proteins in cancer immunotherapy, to creating biomedical decision-support tools using text mining.

Ecology
from understanding how insects adapt to specific ecological niches, to documenting the biodiversity of fungi in the soil.
The Computational Oncogenomics Group works on the computational analysis of structural genome variations in cancer. Our work centers around our array-Map and Progenetix resources of curated molecular cytogenetic and sequencing data. Specific projects explore computational methods, genomics of selected tumor entities as well as computational features with patterns across malignancies. As a member of the Global Alliance for Genomics and Health, the group is developing standards in biocuration and data sharing for genome variants and phenotypic data. Other projects are related to genome data epistemology, e.g. geographic and diagnostic sampling biases.

Michael BAUDIS

In the Computational Oncogenomics Group, we are interested in gene regulation in both healthy and diseased cells. Breakthroughs in genomics technologies have led to the production of large volumes of data that could potentially tell us something about how gene regulatory instructions are encoded in our DNA. Our group develops new algorithms, computer programs, web services and databases that will help us and others to extract knowledge and understand from such data. We also collaborate with experimental biologists on interdisciplinary projects and organize postgraduate courses in genomic data analysis.

Philipp BUCHER

Mauro DELORENZI

Frédéric SCHÜTZ

In the Bioinformatics Core Facility (BCF), we promote the exchange of knowledge and computational approaches 1) to decipher the regulatory code in cell function, adipose biology and diseased cells. Breakthroughs in genomics technologies have led to the production of large volumes of data that could potentially tell us something about how gene regulatory instructions are encoded in our DNA. Our group develops new algorithms, computer programs, web services and databases that will help us and others to extract knowledge and understand from such data. We also collaborate with experimental biologists on interdisciplinary projects and organize postgraduate courses in genomic data analysis.

Niko BEERENWINKEL

The Computational Biology Group is involved in research and teaching in the areas of computational biology, bioinformatics, and systems biology. Our activities include the development of mathematical and statistical methods, their implementation in computer programs, and their application to biomedical problems. We are conducting active research projects on HIV drug resistance, the evolutionary selection of cancer, haplotype inference from ultra-long sequencing data, reconstruction of signalling pathways from perturbation screens, and computational analysis of single-cell sequencing data.

Sevn BERGMANN

We develop concepts and tools for the analysis of large-scale biological and clinical data. We participate in many genome-wide association studies (GWAS) for human traits and have a particular interest in the integration of genotypic and complex phenotypic datasets (such as gene expression or metabolomics). A key approach is the reduction of complexity through modular and network analysis. A complementary direction of our research pertains to relatively small genetic networks, whose components are well known.

Bart DEPLANCHE

In the Laboratory of Systems Biology and Genomics (LSBG), we use high-throughput sequencing, single cell genomics, microfluidics, and computational approaches 1) to decipher the regulatory code in Druschmlus and mammals with a specific focus on monochromatic stem cell function, adipocyte biology and gut immunity, and 2) to examine how variations in this code affect molecular and organismal diversity. In addition to our research interests, we are actively pursuing the development of new research tools and computational pipelines that enable a better characterization of gene regulatory networks.

Brice DeLattier

Thomas Elsg
d

Davide Righi, Sibylle Karin et al.

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Davide Righi, Sibylle Karin et al.
Our group has a strong interest in population genetics and genomics of complex traits. We use various methodologies to understand the role of genetic variation in phenotypic variation and what factors affect genetic variation harboured within and between functional elements of the human genome. Our main focus is on genome-wide association studies and gene expression and cellular phenotypes and their association with functional variation. We attempt to detect functional genetic variation in regulatory elements and use the functional variation and precociously measured gene expression variation in animal models, to bridge the genotype with disease phenotypes in association studies.

Emmanouil DERMITZAKIS  
GENOMICS OF COMPLEX TRAITS  
BIOLOGY GROUP  
UNIVERSITY OF GENEVA

In the Host-Pathogen Genomics Group, we explore the genetic roots of inter-individual differences in response to infections, with a particular focus on the genomic interactions between pathogens and human host. At the crossroads between basic sciences and the clinical world, we are committed to transdisciplinary genomics research, aimed at identifying, validating and bringing to clinical use genomics markers of susceptibility to infectious diseases – e.g. host genomics of HIV infection, joint analyses of interactions between human and viral genomes, and immune responses in patients with extreme infectious disease phenotypes.

Jacques FELLAY  
HOST-PATH GENOMICS GROUP  
EPFL  
LAUSANNE

In the Computational Cancer Biology Group, our aim is to study the interactions between cancer and immune cells. To this end, we develop machine learning algorithms to analyze large-scale genetic and genomic data.

David GELLER  
COMPUTATIONAL CANCER BIOLOGY  
UNIVERSITY OF LAUSANNE

In the Statistical Genetics Group, we are interested in the development of statistical methodologies in order to decipher the genetic architecture of complex human traits related to obesity. To do this, we efficiently combine large-scale genome-wide association studies (GWAS) with various -omics data. Our methods improve genetic fine-mapping, reveal gene environment interactions, dissect genetic subpopulations of obesity, enhance causal effect estimation and detect various statistical interactions. Furthermore, we are involved in large consortia researching the genetic basis of autoimmune diseases (GIANT) and longevity (LIFEs).

Zoltán KUTALIK  
STATISTICAL GENETICS GROUP  
CHUV  
UNIVERSITY OF LAUSANNE

Our goal is to understand how evolution works at the molecular level and how organisms adapt to changing conditions through random mutations and recombination. Pathogen provide excellent model systems for studying these processes. We apply modern sequencing techniques to decipher the genome of thousands of HIV particles and develop new algorithms to elucidate the interactions between HIV and the immune system. We have developed a method that can predict the evolution of influenza viruses from their phylogenetic trees (treeorify). Such predictions can help to ensure that the seasonal influenza vaccine matches the viruses in circulation.

Richard NEHER  
MICROBE DIVERSITY  
UNIVERSITY OF BASEL
In the Statistical Bioinformatics Group, we develop robust data analysis solutions, including new or improved methods, for the analysis of genome-scale data. We develop statistical methods for interpreting high throughput sequencing and other technologies in the context of genome sequencing, gene expression and regulation, and analysis of epigenomes. We are largely data- and problem-driven, and ultimately the methods we develop are geared to the characteristics of the technology platform generating the data. We develop publicly available open-source software tools, generally through the Bioconductor project.

We develop and apply statistical methodologies to large human phenotypy-genotype datasets, to find long-standing questions in population and quantitative genetics. These have improved the prediction accuracy of disease risk in personalized medicine, quantified the contribution of genotype-environment interaction to effects on obesity risk, and provided frameworks to examine the genetic basis of phenotypic differences among people. Our current work focuses on improved testing for sex-, age-, or environment-specific genetic effects and investigating the role of interactions between microbial and host genotypes in shaping human phenotypes.

Our main research interest in the Genome Systems Biology (GBS) Group is the study of genome-wide regulatory systems, in order to reconstruct them from high-throughput molecular data, understand and model how they have evolved, and search for design principles in their construction. In particular, we are developing and applying new algorithmic tools for the automated reconstruction of genome-wide regulatory networks from comparative genomic, deep sequencing, and other high-throughput data. In addition, methods are being developed for studying genome evolution, and the evolution of regulatory networks, in particular.

In the Evolutionary Systems Biology Group, we study the evolution and modularity of biological systems at all levels of biological organization, from genes and genomes to biological networks and whole organisms. We develop bioinformatics tools to integrate data from a variety of sources, including comparative whole-genome sequencing, high-throughput RNA sequencing, and systems biology approaches to reconstruct regulatory networks and assess their navigability.

Using the mathematical concept of knotoids, we were able to characterize the topology of protein structures without the necessity of imposing a closure on analyzed protein chains. New simulation approaches permitted us to emulate the effect of torsional tension introduced during transcriptions into chromatin fibres, as well as the effects of torsional stress relaxation by DNA topoisomerases. Our modeling indicated that TAD-like self-interacting chromatin domains in chromosome of S. pombe can simply arise as a result of torsional stress accumulated during dividing transcriptional units.

Our work uses comparative analysis of genome data, labor on evolutionary experiments, and mathematical modeling.
MAIN DOMAIN OF ACTIVITY

Evgeny ZDOBNOV

Computational and Systems Genomics Group
University of Geneva
Geneva

Synergy of Genomic and Proteomic Approaches to Understanding the Function of the Human Prostate and Related Diseases

Our research focuses on the development of bioinformatics and computational systems biology methods to study the functions and relevant clinical applications of the human prostate. We aim to understand the regulatory mechanisms controlling gene expression and protein synthesis in the human prostate, with a particular emphasis on prostate cancer and related diseases.

We employ bioinformatics and computational approaches to analyze large-scale biological datasets generated by high-throughput technologies, such as RNA sequencing, proteomics, and metabolomics. Our research is supported by computational and experimental data from our own laboratories, as well as collaboration with clinical and research partners worldwide.

Secondary Domains of Activity

Bioinformatics; Computational genomics; Systems biology; Cancer biology

INSTITUTION

University of Geneva, Geneva

CITY

Geneva

GENEVA

UNIVERSITY OF GENEVA

INSTITUTION

University of Geneva, Geneva

CITY

Geneva

MAIN DOMAIN OF ACTIVITY

Christian AHRENS

Protein Informatics and Systems Biology Group
Institute for Medical Informatics and Biometrics
University of Zurich
Zurich

Computational Proteomics and Systems Biology

Our research focuses on the development of computational methods to understand the function of proteins and their interactions in biological systems. We aim to develop integrative models that can predict protein function and interactions across different biological contexts.

We employ computational and experimental approaches to analyze large-scale biological datasets generated by high-throughput technologies, such as mass spectrometry, proteomics, and transcriptomics. Our research is supported by computational and experimental data from our own laboratories, as well as collaboration with clinical and research partners worldwide.

Secondary Domains of Activity

Bioinformatics; Computational genomics; Systems biology; Cancer biology

UNIVERSITY OF ZURICH

Institute for Medical Informatics and Biometrics

CITY

Zurich

GENEVA

University of Zurich, Zurich

CITY

Zurich

MAIN DOMAIN OF ACTIVITY

Amos BAIROCH

Computational and Laboratory Proteomics Group
University of Geneva
Geneva

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Bioinformatics; Computational genomics; Systems biology; Cancer biology

UNIVERSITY OF GENEVA

Computational and Laboratory Proteomics Group

CITY

Geneva

GENEVA

University of Geneva, Geneva

CITY

Geneva

MAIN DOMAIN OF ACTIVITY

Frédérique LISACEK

PROTEOME INFORMATICS GROUP
University of Geneva
Geneva

Proteome Informatics

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Geneva

GENEVA

University of Geneva, Geneva

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Iannios XENARIOS

Alan BRIDGE

Swiss Institute for Bioinformatics

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SWISS INSTITUTE FOR BIOINFORMATICS

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Grenchen

GENEVA

Swiss Institute for Bioinformatics, Grenchen

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Institute for Medical Informatics and Biometrics

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Zurich

GENEVA

University of Zurich, Zurich

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Computational and Laboratory Proteomics Group

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University of Geneva, Geneva

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Proteome Informatics Group

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University of Geneva, Geneva

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Secondary Domains of Activity

Bioinformatics; Computational genomics; Systems biology; Cancer biology

SWISS INSTITUTE FOR BIOINFORMATICS

CITY

Grenchen

GENEVA

Swiss Institute for Bioinformatics, Grenchen

CITY

Grenchen
The Applied Computational Genomics Team focuses on theoretical and computational aspects of modeling the process of genome evolution and adaptive change. With the growing size and complexity of molecular data, we strive to keep pace by providing accurate, scalable, and practical computational solutions that enable a wide range of scientists to analyze patterns of evolution and natural selection in large genomic and omics data. We develop bioinformatic methods for real applications, ranging from biotechnology to biomedical research, ecology and agricultural fields.

Maria ANISIMOVA
APPLIED COMPUTATIONAL GENOMICS TEAM
INSTITUTE OF BIOLOGY AND GENOMICS GROUP
UNIVERSITY OF LAUSANNE
CITY

Christophe DESSIMOZ
COMPUTATIONAL AND MOLECULAR POPULATION GENETICS GROUP
UNIVERSITY OF LAUSANNE
CITY

Evolution; Gene ontology; Selection; Software development; Text mining; Semantic web

In the Computational and Molecular Population Genetics (CMPOP) Group, we develop new methodologies for the simulation and analysis of molecular polymorphism within species with a particular focus on humans. We also develop and maintain the Argoquin software, a popular package for the analysis of multi-locus genetic diversity with and between populations, as well as statistical methods to reconstruct and infer evolutionary processes from genomic data. The team focuses on the effect of range expansions on genomic and functional diversity, and the detection of signatures of adaptation and selection at the molecular level.

Laurent EXCOFFIER
COMPUTATIONAL AND MOLECULAR POPULATION GENETICS GROUP
UNIVERSITY OF LAUSANNE
CITY

In the Evolutionary Bioinformatics Research Group, we are interested in the modeling and analysis of biological problems at the molecular level. In particular, our expertise lies in searching algorithms, optimizing algorithms, mathematical modeling, and computational systems. Most of our research efforts are concentrated on the Orthologous Matrix (OMA) project. This particular project aims to produce, automatically, reliable orthologue groups of proteins that are derived from entire genomes.

Gaston GONNET
COMPUTATIONAL BIOINFORMATICS RESEARCH GROUP
ETH ZURICH
CITY

In the Computational Bioinformatics Group, we are mainly concerned with determining the role of evolutionary innovation and constraint in animals. For this, we develop methods and databases to extract reliable information from genome and transcriptome data. These include Bape, a database for gene expression evolution, and Selectome, a database of positive selection. While developing these resources, we also conduct research on ontologies, biocuration, and high-performance computing. Our biological focus is on Drosophila melanogaster with Drosophila. Notably, we study the role of gene duplication in the divergence between genes and between species.

Marc ROBINSON-RECHAVI
EVOLUTIONARY BIOINFORMATICS RESEARCH GROUP
UNIVERSITY OF LAUSANNE
CITY

Bonif oscillator theory, data mining, and comparative genomics.

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**CHAPTER 3 Activities 2017**

**MAIN DOMAIN OF ACTIVITY**
Daniel WEGMANN
Evolution
EVOLUTION
ETH ZURICH
D-BSSE
INSTITUTION

**SECONDARY DOMAINS OF ACTIVITY**
- Biostatistics; Population genetics
- Infectious diseases; Next-generation sequencing; GWAS; Machine learning; Next-generation sequencing; Genome-wide association studies (GWAS)

**DOMAINS OF APPLICATION**
- Phylogeny; Selection; Transcriptomics
- Human genetics

**RESOURCES**
- BEAST—A community for BEAST 2 resource
- SwissDock, SwissTargetPrediction

**KEY PAPERS IN 2017**

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**Bruno CORREIA**
LABORATORY OF PROTEIN MODELLING
EPFL
LAUSANNE
INSTITUTION

The Laboratory of Protein Design & Immunomodulation started in March 2015. We have established both the computational and experimental branches of our research. We currently perform our computational calculations at the High Performance Computing facility at the EPFL. The experimental arm of the laboratory is prepared to perform molecular biology, proteomics expression and purification, and protein biophysical characterization.

**Matteo DAL PERARIO**
LABORATORY FOR BIOMOLECULAR MODELING
EPFL
LAUSANNE
INSTITUTION

Our main goal at the Laboratory for Biomolecular Modeling is to understand the physical and chemical properties of complex biological systems, in particular their function with regard to structure and dynamics. To this end, we use and develop a broad spectrum of computational tools, fully integrated with experimental data. Multi-scale simulations and dynamic integrative modelling are used to investigate the function of molecular assemblies, mimicking the conditions of the native cellular environment.

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**Matthias Käser et al.**
Chemoproteomics: profiling and discovery of new targets of drug resistance in human cells
Nat Chem.

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**Olivier MICHELIN**
MOLECULAR MODELLING GROUP
UNIVERSITY OF LAUSANNE
LAUSANNE
INSTITUTION

The Molecular Modelling Group studies molecular interactions using modelling techniques such as homology-modelling, molecular dynamics, docking and free energy simulations. Our main activity consists in developing and applying methods for computer-aided protein design and drug design, as well as in computer-aided protein design and drug design. Notably, we develop novel inhibitors that target cancer therapy and design optimized proteins, such as T-cell receptors, for cancer immunotherapy. We develop and maintain web tools for drug design, such as SwissDock, SwissTargetPrediction and SwissADME. We also run the Protein Modelling Facility of the University of Lausanne.

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**Teresa Gandolfi et al.**
Diprogramm: an integrative description and docking of biomolecular complexes
Bioinformatics.

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**Chaba P. et al.**

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**Abraham et al.**

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**Daniel WEGMANN**
STATISTICAL AND COMPUTATIONAL EVOLUTIONARY BIOLOGY GROUP
UNIVERSITY OF FRIBOURG
FRIBOURG
INSTITUTION

When observing nature, it is easy to be impressed by the huge diversity seen on any biological scale. Our primary aim is to better understand the underlying evolutionary processes that have been shaping this diversity over the course of evolution on our planet. To achieve this, we design and evaluate new statistical and computational approaches to infer complex evolutionary histories. For this, we develop and apply machine-learning algorithms, with a particular focus on likelihood-free methods. We then apply these approaches to the wealth of data currently being generated.
**Torsten SCHWEDE**

Computational and Systems Biology Group
University of Basel

In the Computational Structural Biology (CSB) Group, we focus on the development of methods and algorithms to model, simulate, and analyse three-dimensional protein structures and their molecular properties in order to apply these techniques to the understanding of biological processes at a molecular level. Our main emphasis is on homology modelling – using evolutionary information to model protein tertiary and quaternary structures. Applications in biomedical research include the study of protein-ligand interactions, drug discovery, structure-guided protein engineering, and the interpretation of disease-causing mutations.

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**Manfred CLAASSEN**

Computational Single Cell Biology Group
ETH Zurich

Our research in the Computational Single Cell Biology Group aims at elucidating the composition of heterogeneous cell populations and how these implement functions in the context of cancer and immune biology. To accomplish this task, we build on concepts from statistics, machine learning and mathematical optimisation to develop probabilistic approaches to describe biological systems, learn these descriptions from data, and design experiments to validate hypotheses following computational analyses. Our research can be used to pin point therapeutic targets with a view to designing drugs.

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**Rudiwang o GUNAWAN**

Chemical and Biological Systems Engineering Laboratoy
ETH Zurich

In the Chemical and Biological Systems Engineering Laboratory, we develop tools for systems modelling and the analysis of chemical and biological networks. Our mission is to create enabling theories and computational methods for the generation of systems insights, as well as for understanding and acquiring knowledge in chemical, biological and medical applications. Our research spans multiple length and time-scales of cell biology, from gene networks and cellular networks in single cells to the ageing process in human and cell culture biomaterials in the pharmaceutical industry.

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**Bagas I. BFER**

Computational Biology Group
ETH ZURICH & Basel

The Computational Biology Group (CBG) develops computational models of developmental processes. We place a particular focus on mechanistic 4D image-based in silico models of organogenesis (mouse lung, kidney, pancreas, limb, brain, Drosophila wing and eye) and on the delineation of fundamental cell mechanisms, such as those that restrict the size of organs and those that maintain the proportions of structures in different-sized embryos. We use the established set of methods to progressively refine those models and develop spatially-organized tissue bioreactors in the pharmaceutical industry.
Mathematical modelling is becoming more and more instrumental in life sciences; the data complexity and the high number of interacting components from molecules to animals, render it more and more difficult. The idea of formulating mathematical equations to describe how certain biological units affect each other and how these interactions affect the whole system, is not straightforward. To achieve this, we combine Evolutionary and Developmental Biology as well as genomic approaches with the study of physical processes to understand the mechanisms generating life’s complexity and diversity. The self-organizational capabilities of small bacterial communities in the lab can be used to understand the interactions between physical (e.g., mechanics, cell signalling, proliferation, migration) parameters, which generate patterns and shapes during development. Our research team consists of biologists, computer scientists, engineers, and physicists.

Our research interests in the Scientific Computing Group lie in the area of multi scales models of cellular processes and their simulations of biological systems. We focus on the development of new computational models and corresponding numerical methods suitable for the next generation of supercomputers. We are working on stochastic multi-scale modelling of motion, the interaction, deformation and aggregation of cells under physiological flow conditions, biofilm growth, and coarse-grained molecular dynamics simulations, as well as the modelling of transport processes in healthy and tumour-induced microcirculation.
The individual cells of a body exhibit a stunning diversity of phenotypes, despite carrying a largely identical genetic makeup. This is due to the distinct ways in which the same genetic information is read, interpreted and translated into function. At the RNA Regulatory Networks (RRN) Group at the Biozentrum in Basel, we combine computational modelling with big data and experimental analysis to discover and understand the regulatory networks governing the interpretation of genetic information at the level of tissues and single cells.

In the Interfaculty Bioinformatics Unit of the University of Bern (IBU), we provide services and expertise to assist researchers in the three “Life-Science” Faculties (i.e. Sciences, Medicine, and VetSciences) in designing and project planning for large-scale experiments (e.g. next-generation sequencing, qPCR, mass spectrometry). Furthermore, we have our own research programme and collaborate on large and complex projects. We develop methods to analyse high-throughput data. We have a high-performance computing cluster and a data storage system that we use for our own research, collaborations and service projects.

Our group is located in the Department of Biomedicine (DBM) at the University of Basel. We collaborate with scientists from DBM on projects covering a broad spectrum of research topics, from cellular differentiation and evolution to the identification of a molecular basis for various human diseases. To do this, we focus on the analysis, interpretation and visualization of expression, epigenetic and genomic data, which are mainly derived from next-generation sequencing experiments, or any large-scale biological experiment requiring bioinformatics resources. We focus on genome assembly, annotation and comparison, as well as on mutant and structure variant identification by resequencing. We also perform metagenomics, RNAseq and ChIPseq data analysis, proteome clustering and functional clustering classification, as well as pathway and gene set enrichment analysis.
In the Service and Support for Science IT (S3IT) unit, we provide support for science in general, and for life sciences and medicine in particular. S3IT serves as a partner for both local and national projects to enable competitive research with the advanced use of computational tools and resources.

Our group in Scientific IT Services (SIS) is an interdisciplinary bioinformatics and scientific IT group, which develops computational tools and services. We propose high-performance computing infrastructure in support of high-performance computing and cloud computing. With our services, we support life science and biomedical research, as well as other basic and applied research.

### Core facilities and competence centres

- **Data management plan:**
  - Machine learning
  - Software development
  - Systems biology

- **Other services:**
  - Machine learning
  - Software development
  - Cloud computing

- **Software development:**
  - Cloud computing
  - Machine learning

- **Software optimization:**
  - Cloud computing
  - Machine learning

### Key papers in 2017

1. **Astronomy & Astrophysics**
   - Pramet7 et al.
   - EasyGWAS: a cloud-based platform for genome-wide association studies.

2. **Bioinformatics**
   - Liechti R et al.
   - EasyGWAS: a cloud-based platform for genome-wide association studies.

3. **Bioinformatics**
   - Singer J et al.
   - EasyGWAS: a cloud-based platform for genome-wide association studies.

4. **Diabetologia**
   - Liechti R et al.
   - EasyGWAS: a cloud-based platform for genome-wide association studies.

5. **Nat methods**
   - Solimena M et al.
   - EasyGWAS: a cloud-based platform for genome-wide association studies.

### Remaining contact information

- **Email:** info@s3it.ethz.ch
- **Phone:** +41 44 633 11 11
- **Website:** www.s3it.ethz.ch

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**ETH ZURICH, D-BSSE**

**FOR SCIENCE IT (S3IT)**

**UNIVERSITY OF BASEL**

**BIOINFORMATICS UNIT**

**COMPUTATIONAL BIOLOGY LAB**

**VITAL-IT**

**SIB LAUSANNE**

**LAUSANNE**

**ZHURICH**

**ZURICH**

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**GROUP NAME:**

- **Core facilities and competence centres**
  - Data management plan
  - Data mining
  - Machine learning
  - Software development
  - Systems biology
  - Cloud computing

- **Data management plan:**
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  - Cloud computing
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- **Software optimization:**
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**INSTITUTION:**

- **ETH ZURICH**
- **D-BSSE**
- **FOR SCIENCE IT (S3IT)**
- **VITAL-IT**
- **SIB LAUSANNE**
- **LAUSANNE**
- **ZHURICH**
- **ZURICH**

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**CITY:**

- **ETH ZURICH**
- **D-BSSE**
- **VITAL-IT**
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- **ZHURICH**
- **ZURICH**

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- **SIB LAUSANNE**
- **ZHURICH**
- **ZURICH**
We are interested in modern machine-learning techniques suitable for the analysis of problems that arise in medicine and biology. In particular, we develop new learning techniques that are capable of dealing with large amounts of genomic data, allowing for very accurate predictions on the phenomenon at hand and are able to comprehensively provide reasons for their predictions, and thereby assisting in gaining new biomedlical insights.

In the Molecular Allergology group we are aiming to unravel the molecular basis of allergic diseases, antigen presentation, immune network, immune tolerance and tolerance breakage with a combination of functional genomics techniques, including proteomics, peptidomics and immunoproteomics. The acquired large-scale molecular data are analyzed with the aim to identify molecular markers with which risk factors, different disease phenotypes, their underlying pathogenic mechanisms, and different patient response groups can be distinguished.

We are interested in understanding the design constrains, robustness, and evolution of gene regulatory systems, particularly at the level of transcription, using both modeling and data-driven approaches.
Take a backstage tour of the SIB Swiss Institute of Bioinformatics: governance, partner institutions, finances, members and staff.
Organization

A ROBUST GOVERNANCE

As a non-profit foundation unifying bioinformatics across Switzerland and with 20 partner institutions, SIB has a robust governance mechanism ensuring both its scientific independence and optimal internal functioning.

A Foundation Council, a Board of Directors, an external Scientific Advisory Board and a Council of Group Leaders complement the management and support teams of the Institute.

SIB Internal Groups, which are fully staffed by SIB, include two of its research and service groups, Swiss-Prot and Vital-IT, as well as the Clinical Bioinformatics, Personalized Health Informatics, Training and Technology Groups. The latter is in charge of coordinating and optimizing technical activities across SIB.

SIB, a national bioinformatics organization model

SIB has become a reference throughout Europe with regard to its structure and organization. The ingredients to make an SIB were the focus of a 2017 publication by Vivienne Baillie Gerritsen (science writer at SIB), Patricia Palagi (team leader of SIB’s Training Group) and Christine Durinx (SIB’s associate director), in the journal Briefings in Bioinformatics.

Becoming an SIB Group Leader: some criteria

Aspiring Group Leaders in the field of bioinformatics must have a professorial appointment in one of SIB’s 20 partner institutions, or be a senior scientist leading her/his own group. Since SIB does not fund Group Leaders, they also must have a sufficient level of financial independence, such as that provided by research grants.
LIST OF GOVERNING BODIES

The Foundation Council

Each of SIB's partner institutions is represented in this Council.

- **President**: Prof. Felix Gutzwiller, Former Senator
- **Founding Members**: Prof. Ron Appel, SIB Executive Director, University of Geneva; Prof. Amos Bairoch, Group Leader, SIB and EPFL; Prof. Denis Hochstrasser, Vice-rector, University of Geneva; Dr. Claire Baribaud, Research Director, Ludwig Institute for Cancer Genetics, University of Illinois, USA; Prof. C. Victor Jongeneel, Carl R. Woese Institute for Genomic Biology, University of Illinois, USA; Prof. Denis Hochstrasser, Vice-rector, University of Geneva; Prof. Edwin Constable, Vice-Rector of Research and Talent Promotion, University of Basel; Prof. Francois Bussy, International Relations and Administration (HEG-Geneva), Director, School of Business Management and support team, Agroscope; Mr. Thomas Baenninger, Ex officio Members Mr. Thomas Baenninger, Chief Financial Officer, Ludwig Institute for Cancer Research
- **Ex officio Members**: Dr. Caroline Kant, Co-opted Member, Foundation Switzerland; Prof. Felix Gutzwiller, Former Senator
- **Honorary Members**: Dr. Johannes R. Randegger, Honorary President of the SIB Foundation Council; Prof. Christian von Muralt, Group Leader, SIB and University of Zurich

The Board of Directors (BoD)

The BoD consists of two Group Leaders elected jointly by the Council of Group Leaders and the BoD, two external members elected by the Foundation Council on the recommendation of the BoD, and the Executive Director. Members of the BoD are appointed for a renewable five-year period.

- **Group Leaders**: Prof. Ron Appel, SIB Executive Director, University of Geneva; Prof. Christian von Muralt, Group Leader, SIB and University of Zurich
- **External Members**: Prof. Manuel Potisch, Chairman, Chief Scientific Officer Research at Philip Morris International; Prof. Martin Rüegsegger, Group Leader, SIB and University of Basel
- **Honorary Members**: Prof. Francois Bussy, International Relations and Administration (HEG-Geneva), Director, School of Business Management and support team, Agroscope; Mr. Thomas Baenninger, Ex officio Members Mr. Thomas Baenninger, Chief Financial Officer, Ludwig Institute for Cancer Research

The Scientific Advisory Board (SAB)

The SAB is made up of at least five members, who must be internationally renowned scientists from the Institute's fields of activity.

- **Group Leaders**: Prof. Alfonso Valencia, Chair, Life Sciences Department, Barcelona; Prof. Sami Boroch, Founding President, SIB, University of Geneva
- **Ex officio Members**: Prof. Michael Schaepman, Dean, School of Management of Zurich; Prof. Antoine Gras, Head of the Health and Telemedicine, Geneva University Hospitals

The Council of Group Leaders

The Council consists of the Group Leaders, the Affiliate Group Leaders and the Executive Director.

- **Honorary Members**: Prof. Dietmar Hopp, Dr. Johannes R. Randegger, Honorary President of the SIB Foundation Council
- **Group Leaders**: Prof. Susan Gasser, Director, Friedrich Miescher Institute for Biomedical Research; Prof. Marc Gailly, Director, Genomes and Proteomes, University of Geneva; Prof. Alain Fischer, Director, Institute of Molecular Genetics, University of Geneva; Prof. Laurent Duret, CNRS Research Director, Co-opted Member, Foundation Switzerland

Management and support team

- **SIB internal Groups**: Clinical Bioinformatics, Training, Genes and genomes, Proteins and proteomes, Text mining and machine learning, Genomics and proteomics, Text mining and machine learning, Evolution and phylogeny, Structural biology, Systems biology, Core facilities and competence centers
- **SIB Service and Research Groups**: Personalized Health Informatics, Technology, Viro-IT
Key figures

FINANCES

SIB funds remained stable in 2017, thanks to the continued support of its funders.

In 2017, the total amount of funds managed by SIB has reached CHF 26.6 million. This amount remained stable compared to last year.

SIB has also received funds from the Swiss Government in the framework of the Swiss Personalized Health Network (SPHN):

- Being in charge of the BioMedIT project, CHF 18M have been allocated to SIB for the period 2017-2020. This amount goes to the core facilities of SIB and the Universities, for the work performed as data nodes in the project over these 4 years.
- The Government has granted SIB with CHF 20M for the SPHN programme over the same period. This amount is however directly transferred to the Swiss Academy of Medical Sciences to be allocated to the projects as per the decisions of the National Steering Board. It therefore does not appear in SIB’s accounts.

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The Swiss government and in particular:
- The State Secretariat for Education, Research and Innovation (SERI)
- The Swiss National Science Foundation (SNSF)
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- Our institutional partners
- The European Commission
- The Leenaards Foundation
- The Medic Foundation
- The National Institutes of Health (NIH)
- The Research for Life Foundation
- SystemsX.ch

The SIB Fellowship programme continues, thanks to the generous support of:
- The R. Geigy Foundation
- The University of Geneva
- The University of Lausanne

We also thank all industrial and academic partners who trust SIB’s expertise.

IMPRESSUM
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SIB GROUP LEADERS AND ACTIVITIES PICTURES BY
Nicolee Righetti, www.lundi13.ch
DESIGN AND LAYOUT BY
Bogsch & Bacco, www.bogsch-bacco.ch

COVER IMAGE
Fireworks... and some birthday cake to celebrate SIB’s 20th anniversary!
While most people would gladly enjoy a slice—or two—of birthday cake, some others have to restrain themselves. Why is it that an extra dose of sugar leads to diabetes in some people? And once there, how does the disease evolve over time?
At SIB, data scientists are working together with experimental biologists to better understand the progression of diabetes over time, using advanced computational methods and visualization tools.
This image represents a network of relationships (arcs) between genes, cellular pathways and traits associated with diabetes.

SOURCE: MARK IBBERSON, SENIOR SCIENTIST AT SIB’S VITAL-IT COMPETENCE CENTRE (CC BY-NC 4.0)

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