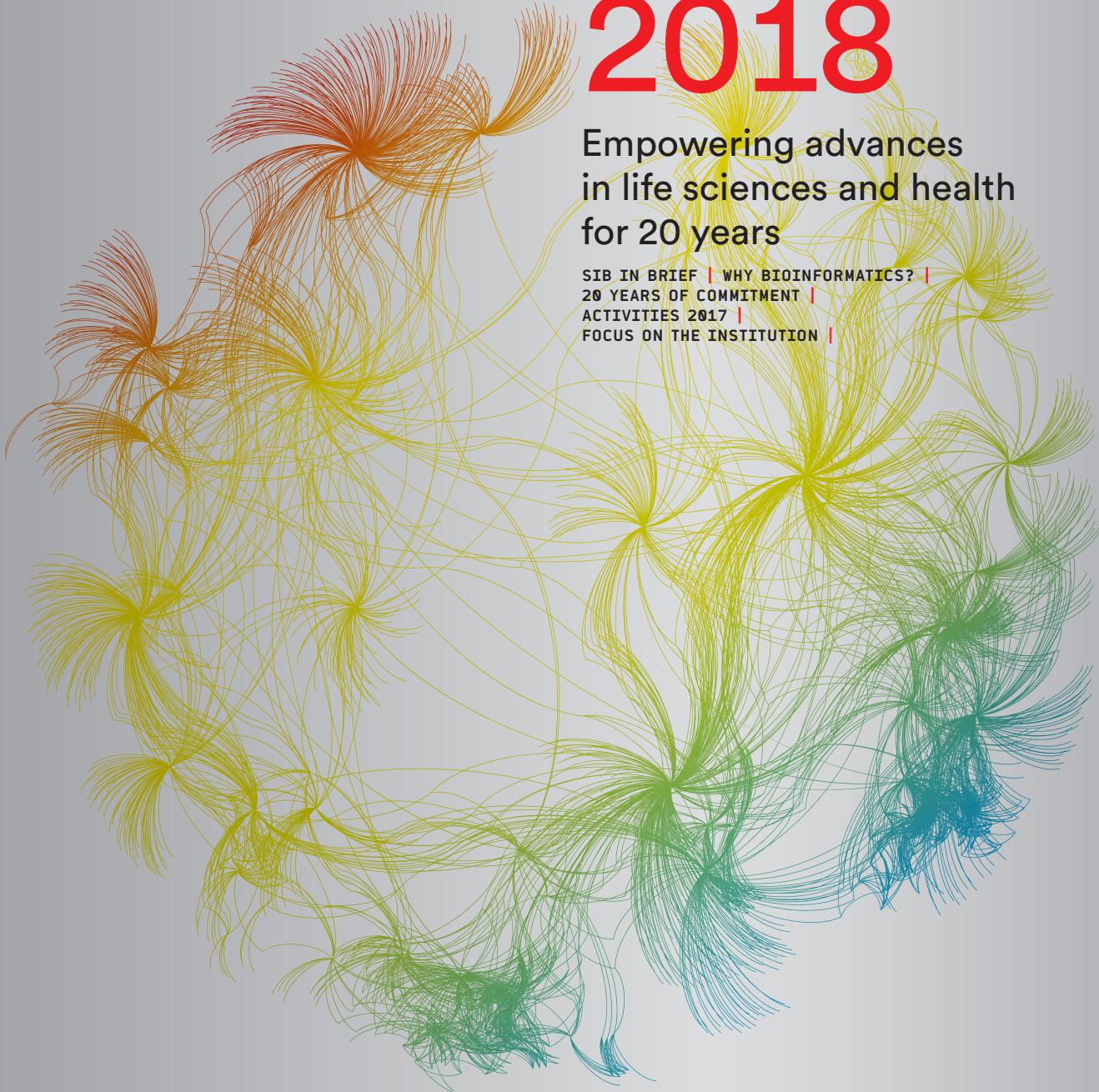


SIB Profile

2018

Empowering advances
in life sciences and health
for 20 years

[SIB IN BRIEF](#) | [WHY BIOINFORMATICS?](#) |
[20 YEARS OF COMMITMENT](#) |
[ACTIVITIES 2017](#) |
[FOCUS ON THE INSTITUTION](#) |



Foreword



Felix Gutzwiller
President of the
Foundation Council



Manuel Peitsch
Chairman of the
Board of Directors



Ron Appel
Executive Director

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 (SPHN). 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.

**“Defending
the long-term
sustainability
of data resources
is rooted in our
Institute’s DNA”**

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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.

67

research and service groups

Some **800**

members, including

200

employees

20

institutional partners across Switzerland

12

core facilities and competence centres

Over **150**

databases and software tools accessible via the ExPASy web portal

Over **2,200**

peer-reviewed articles published since 1998

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

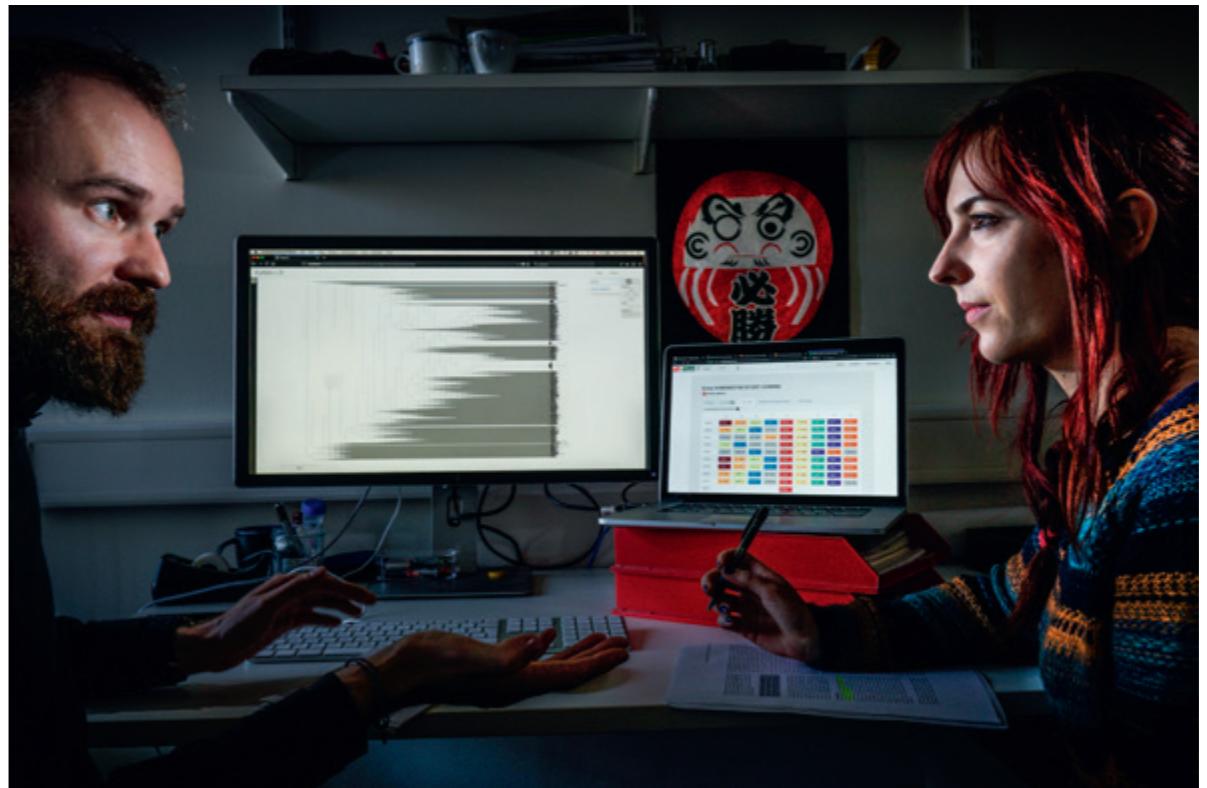
SIB brings together world-class researchers based in Switzerland and delivers training in bioinformatics.

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.

CHAPTER 1

Why bioinformatics?



Answering biological questions using computer- and data science

Bioinformatics: a definition

**FIGHTING DISEASES.
DISCOVERING SPECIES.
UNDERSTANDING LIFE.**

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

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)

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.

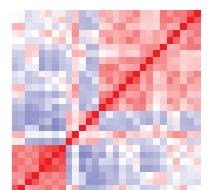
Bioinformatics is the application of computer technology to the understanding and effective use of biological and clinical data.

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.

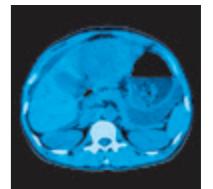
Computer-based approaches have become indispensable to science, by allowing researchers to advance their understanding of complex systems

**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

R3 and the α -D-glucosidase gene (α -GDP) are transcribed from the same promoter and therefore share common regulatory elements. The α -GDP gene encodes homologues of the transport protein genes (BtuA, BtuB, and BtuC) that specify the $E. coli$ glucose transporter, the glucose-6-phosphate transporter, and the glucose-6-phosphate permease, respectively. The α -GDP gene is also under catabolite control and grows best in the β -D-glucosidase medium and is suppressed by transport inhibitors of the β -D-glucosidase pathway. This result was found to be similar to the regulation of the α -GDP gene by glucose-6-phosphate and glucose-6-phosphate permease. Regulation studies with a mutant of α -GDP AMP-activated protein kinase show that it can bind AMP-activated protein kinase α -chain and it is weakly inhibited by the α -GDP gene product. These results suggest that α -GDP gene is co-expressed with its own synthase, transcript and coductase chain, and is subject to transcriptional control. The α -GDP gene indicated a single strongly predicted binding site.

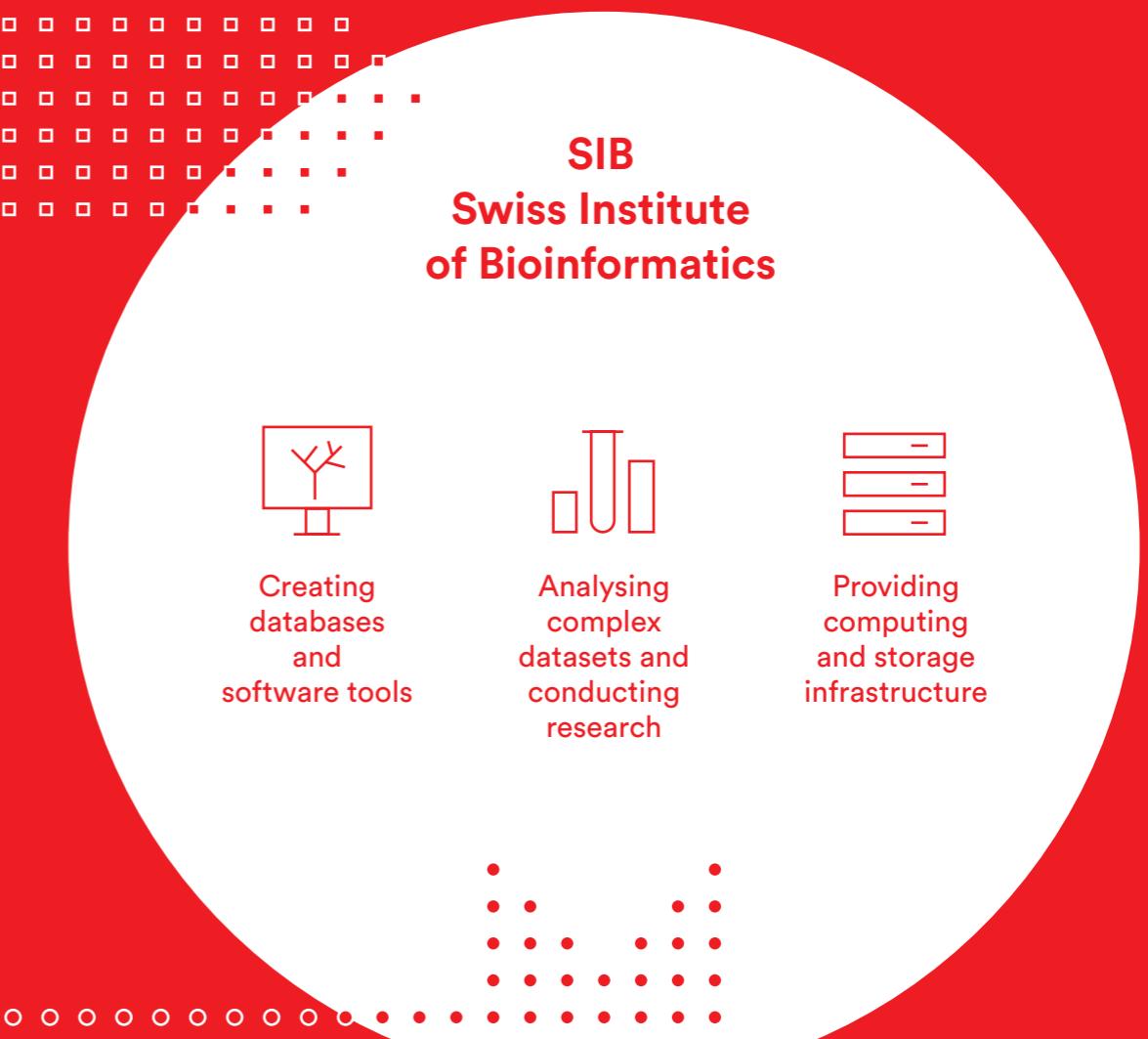
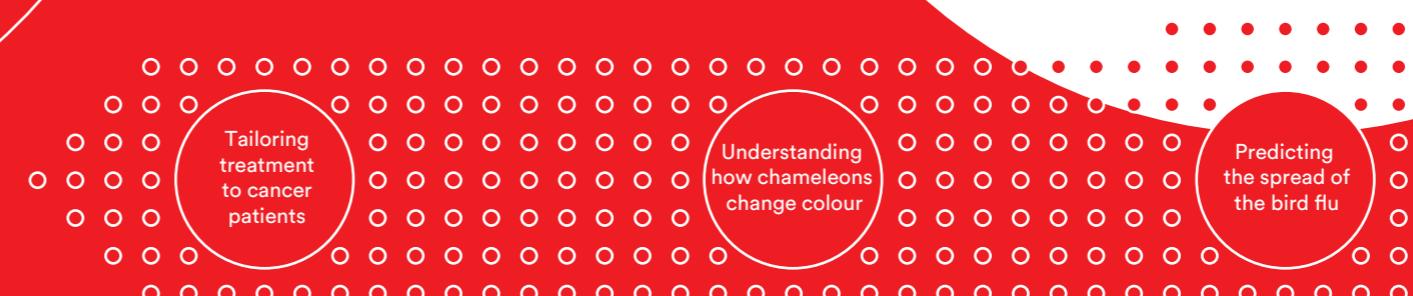
Text data
And more...

Converting biological questions ...



with various applications

- Basic research
- Medicine
- Ecology
- Agriculture



SIB
**Swiss Institute
of Bioinformatics**

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.



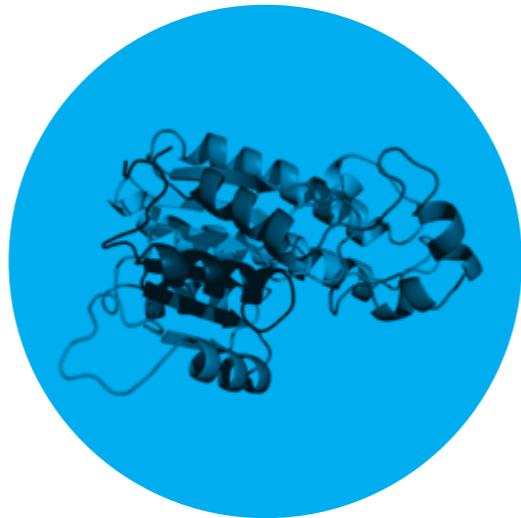
Detecting susceptibility to a disease

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](#)



Optimizing 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, **PHYLOGENETIC 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](#)

“Bioinformatics is everywhere but no one can see it.”

“Bioinformatics helps you find a needle in a haystack.”

“Bioinformatics is another way of creating better drugs.”

Bioinformatics defined in visitors' own words at SIB's *Planète Santé 2016* booth



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.

NUIT DE LA SCIENCE

Geneva, Museum of Science History & Campus Biotech, 7-8 July

What is the role of bio-informatics in drug design, personalized health and metagenomics?



Follow us on Facebook
Our dedicated outreach channel in French and English.



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

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...*

BIOINFORMATICS IN THE CLASSROOM

SIB has designed several websites and workshops to introduce bioinformatics to schoolchildren, teachers and students:

CHROMOSOMEWALK

An award-winning website to explore human chromosomes and the world of DNA, genes and proteins.
www.chromosomewalk.ch

DRUG DESIGN

A web-based workshop to acquire a simple yet realistic picture of how drugs are designed with the help of computers.
www.drug-design-workshop.ch

THE METAGENOMICS PIZZA

A workshop to grasp the concept of metagenomics, and identify all the species present in a pizza, based on their DNA.



2,100
participants
including 1,600 students
(12-18 years old)

CHAPTER 2

20 years of commitment



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.

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 is Switzerland positioned internationally in terms of its bioinformatics infrastructure?

MDA Thanks to SIB, 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.

“SIB is a good example of how we can reconcile competition and cooperation in research”

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.

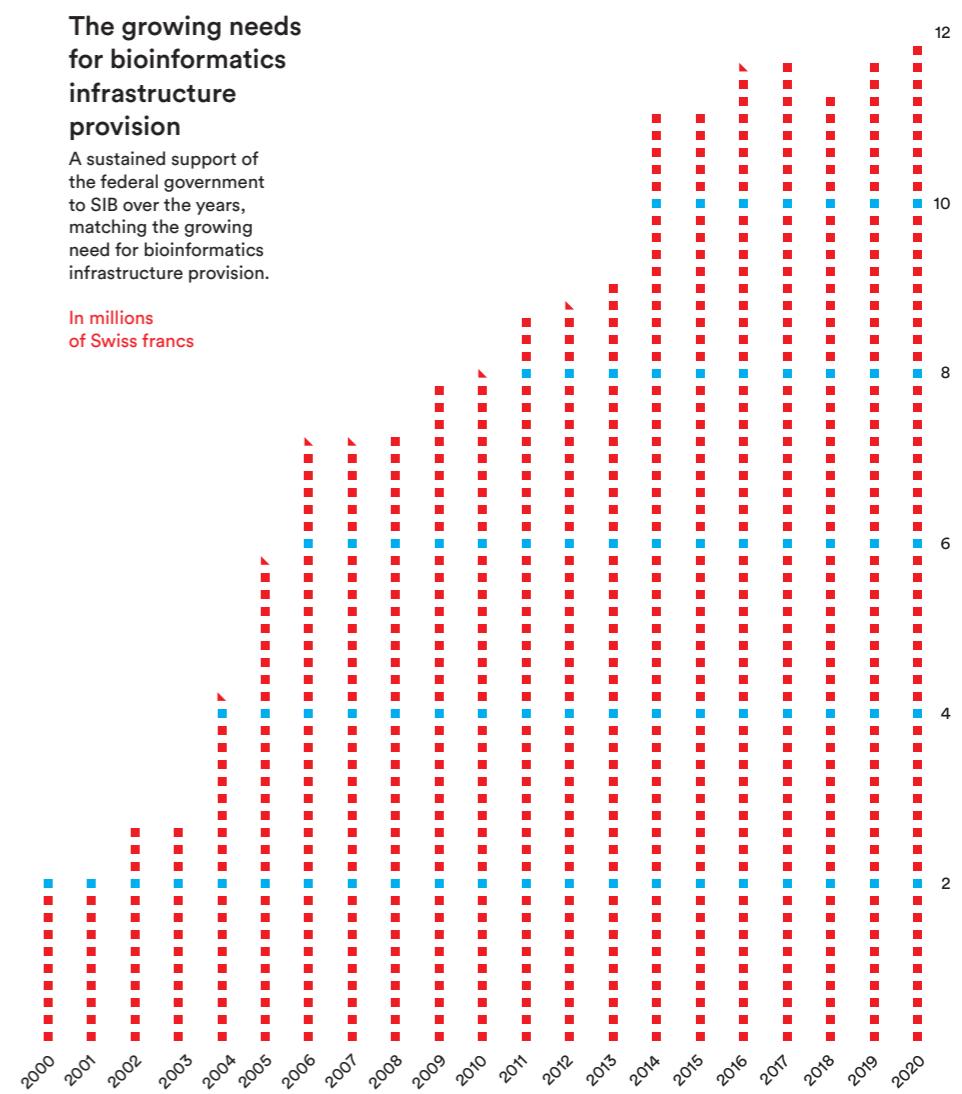
I would therefore like to thank all those at SIB who have helped to strengthen Switzerland’s standing as a location for top research for the past 20 years. Their work benefits society and the economy – today and tomorrow.

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

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.

In millions of Swiss francs



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

Representing bioinformatics

IN SWITZERLAND...

All roads lead to bioinformatics

SIB Members come from a wide range of educational backgrounds

Biology

■ 48%

Other (engineering, bioinformatics, administrative...)

■ 18%

Informatics

■ 16%

Physics

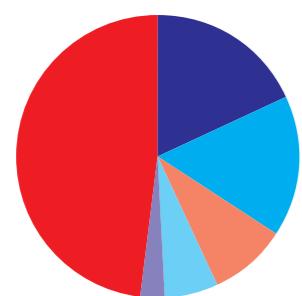
■ 9%

Chemistry

■ 6%

Mathematics

■ 3%



Bioinformatics as a career

SIB Members' current positions are diverse too

Bioinformatician, Computational Biologist

■ 45%

Others (Biochemist, Structural Biologist, Mathematician...)

■ 16%

N/A

■ 15%

Biocurator

■ 8%

Software Developer

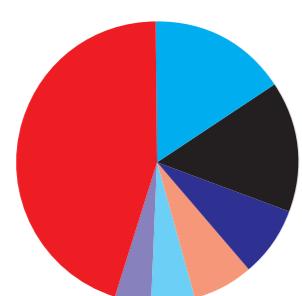
■ 7%

Administrative Staff

■ 5%

Statistician

■ 4%

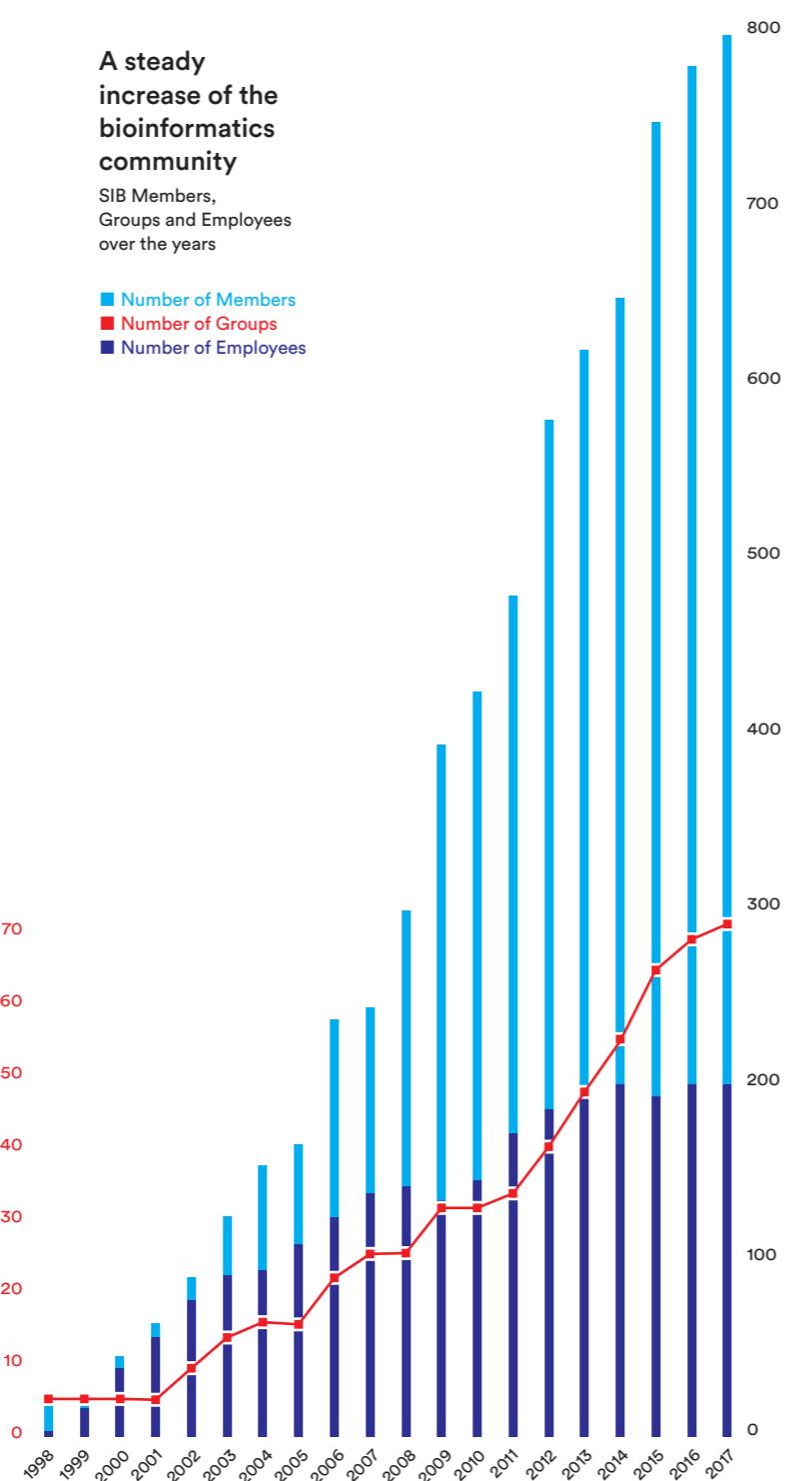


* Data collected from 110 SIB Members as part of an internal survey (2017)

A steady increase of the bioinformatics community

SIB Members, Groups and Employees over the years

■ Number of Members
■ Number of Groups
■ Number of Employees



...AND BEYOND

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.

I believe biocuration is the key for a better understanding of human genomic variation, which is fundamental for the future of precision medicine. To me, SIB's growth over the past 20 years shows the essential role played by bioinformatics in this new era of big data."

"I am an SIB embedded bioinformatician in the Clinical Pathology Division at the Geneva University Hospitals. My background is in computer science with a solid grounding in biology, and thus allows me to understand and address biological questions by developing novel computing approaches, such as clinical diagnostics tools and pipelines. Bioinformatics

Faces of Swiss bioinformatics



Cristina Casals Casas, PhD

UniProtKB/Swiss-Prot biocurator
Swiss-Prot Group

"I am a biocurator in SIB's Swiss-Prot Group, specializing in the manual annotation of proteins of human origin, as well as those from other vertebrates. My job is to ensure that knowledge is extracted from the best-quality biological data generated from the literature, and that such knowledge becomes accessible to the community.

I believe biocuration is the key for a better understanding of human genomic variation, which is fundamental for the future of precision medicine. To me, SIB's growth over the past 20 years shows the essential role played by bioinformatics in this new era of big data."



Gerardo Tauriello, PhD

Development team Leader
Computational Structural Biology Group

"I am an SIB software developer in the Computational Structural Biology Group of Torsten Schwede. I apply my programming skills to the development and optimization of bioinformatics resources such as SWISS-MODEL – an SIB core resource – for the life science community.

I believe bioinformatics has the power to provide the necessary tools and methods to improve our understanding of the complex machinery of life and enhance health. In that context, I am always excited to see how our users employ SWISS-MODEL-generated 3D models of proteins to help explain their function in pathogens, viruses, cancers and many more.

To me, SIB's growth over the past 20 years shows the importance of maintaining and improving reliable and top-quality services and resources to support life scientists in their research."

has the power to bring the best of computer science to medicine and biology. For instance, many advances in cancer biology were only made possible through the development of bioinformatics tools and algorithms.

To me, SIB's growth over the past 20 years shows that bioinformatics has become a driving force in biological research. Furthermore, the recent addition of a "clinical bioinformatics" unit at SIB highlights a change of scope: bioinformatics is no longer limited to research but is now being used to treat patients on a regular basis."



Yann Christinat, PhD

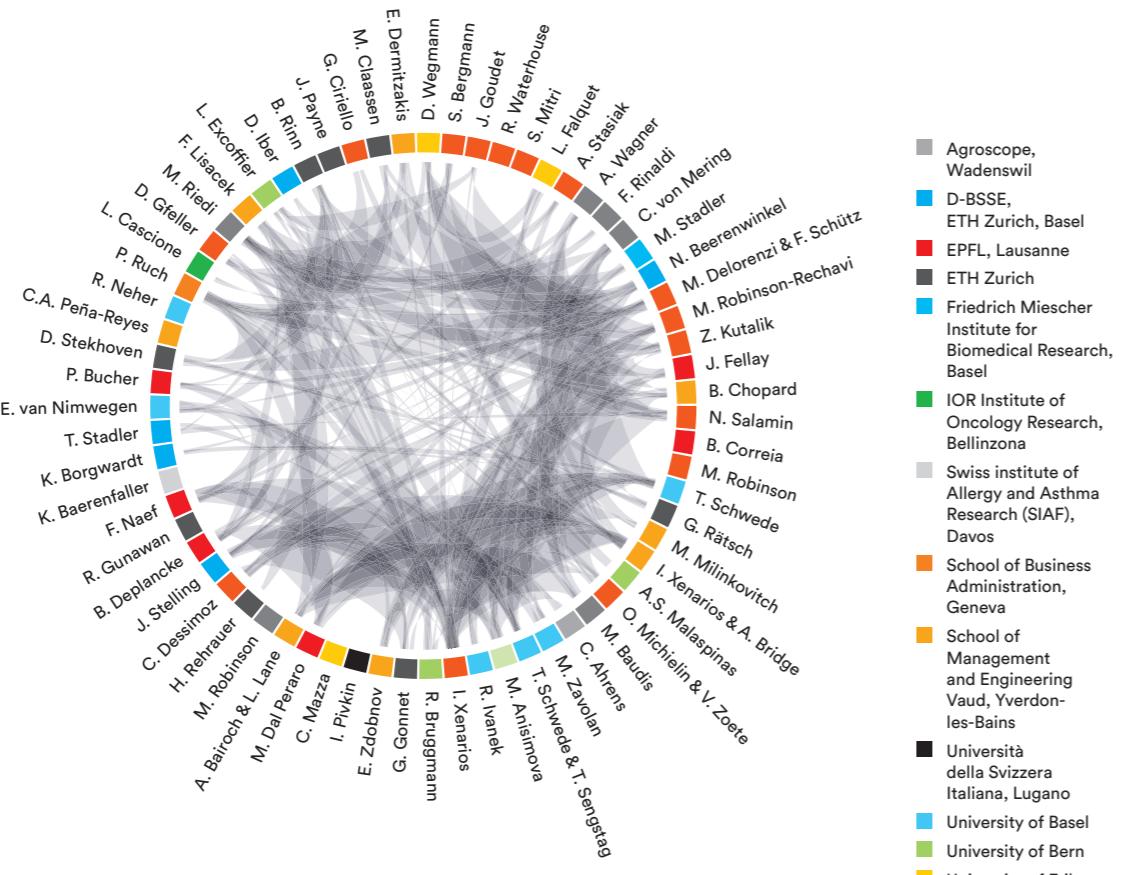
Clinical bioinformatician
Clinical Pathology Division, HUG

"I am an SIB embedded bioinformatician in the Clinical Pathology Division at the Geneva University Hospitals. My background is in computer science with a solid grounding in biology, and thus allows me to understand and address biological questions by developing novel computing approaches, such as clinical diagnostics tools and pipelines. Bioinformatics

Fostering scientific collaboration...

The SIB collaboration network

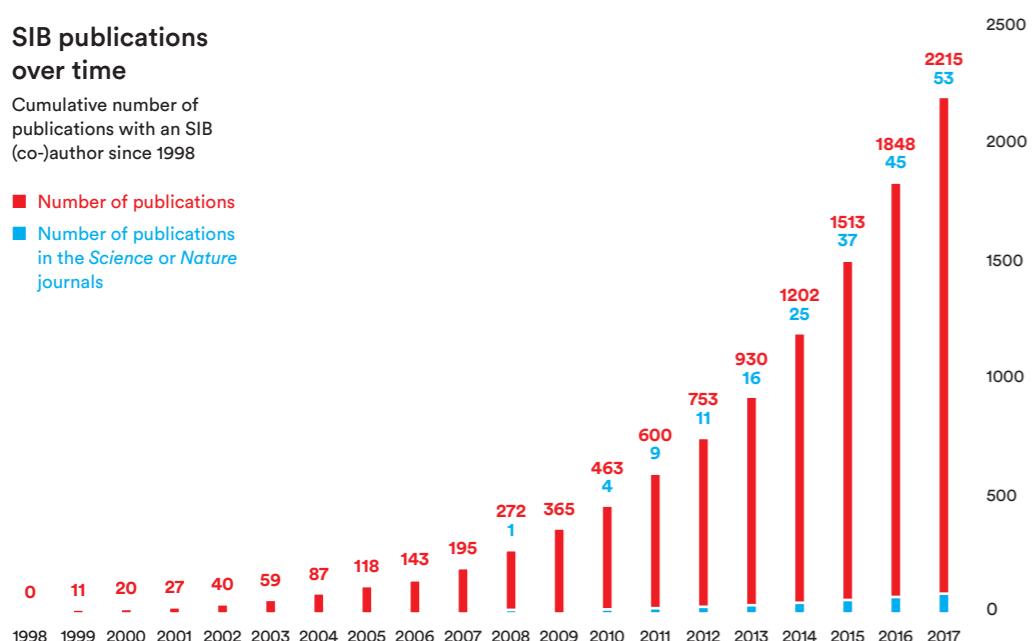
Past or ongoing collaborations between SIB Groups: the more collaborations, the broader the connection.



SIB publications over time

Cumulative number of publications with an SIB (co-)author since 1998

- Number of publications
- Number of publications in the Science or Nature journals



THIS COLLABORATION NETWORK WAS GENERATED WITH A PROGRAMME DEVELOPED BY SIB GROUP LEADER MICHAEL BAUDIS: PROGENETIX.ORG/COLLABPLOTS

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 “somatic mutation calling” and “microbe 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 OncoBench™, 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 EMBO 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.

<http://sourcedata.embo.org/>

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 v10: Protein-protein interaction networks, integrated over the tree of life.
By Szklarczyk D et al.
NUCLEIC ACIDS RES. 2015

STRING v9.1: Protein-protein interaction networks, with increased coverage and integration.
By Franceschini A et al.
NUCLEIC ACIDS RES. 2013

The STRING database in 2011: functional interaction networks of proteins, globally integrated and scored.
By Szklarczyk D 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
MOL ECOL RESOUR. 2010

Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index.
By Spelioles E et al.
NAT GENET. 2010

Transcriptome-wide identification of RNA-binding protein and microRNA target sites by PAR-CLIP.
By Hafner M et al.
CELL 2010

STRING 8 – a global view on proteins and their functional interactions in 630 organisms.
By Jensen L et al.
NUCLEIC ACIDS RES. 2009

A mammalian microRNA expression atlas based on small RNA library sequencing.
By Landgraf P et al.
CELL 2007

SWISS-MODEL: An automated protein homology-modelling server.
By Schwede T et al.
NUCLEIC ACIDS RES. 2003

The SWISS-PROT protein knowledgebase and its supplement TrEMBL in 2003.
By Boeckmann B et al.
NUCLEIC ACIDS RES. 2003

ExPASy: The proteomics server for in-depth protein knowledge and analysis.
By Gasteiger E et al.
NUCLEIC ACIDS RES. 2003

* SOURCE: EUROPEPMC EXTRACTION FROM 30.03.1998 TO 17.01.2018. CITATIONS ARE BASED ON OPEN CITATIONS DATA.

... 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.

PHD TRAINING NETWORK

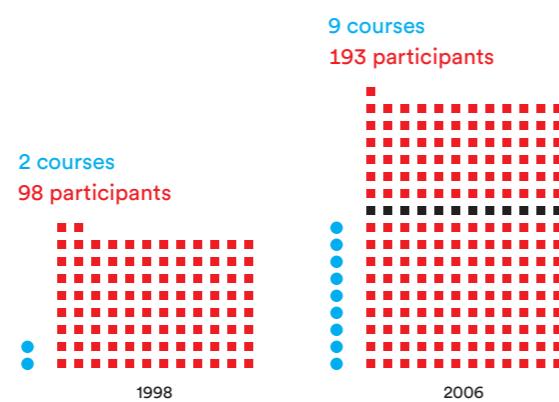
The PhD Training Network was created as a community support for doctoral students carrying out their research in bioinformatics or computational biology in Switzerland. The Network aims to foster interactions and exchanges among SIB PhD students and train them in the most up-to-date methods necessary 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.

Development of training activities over time

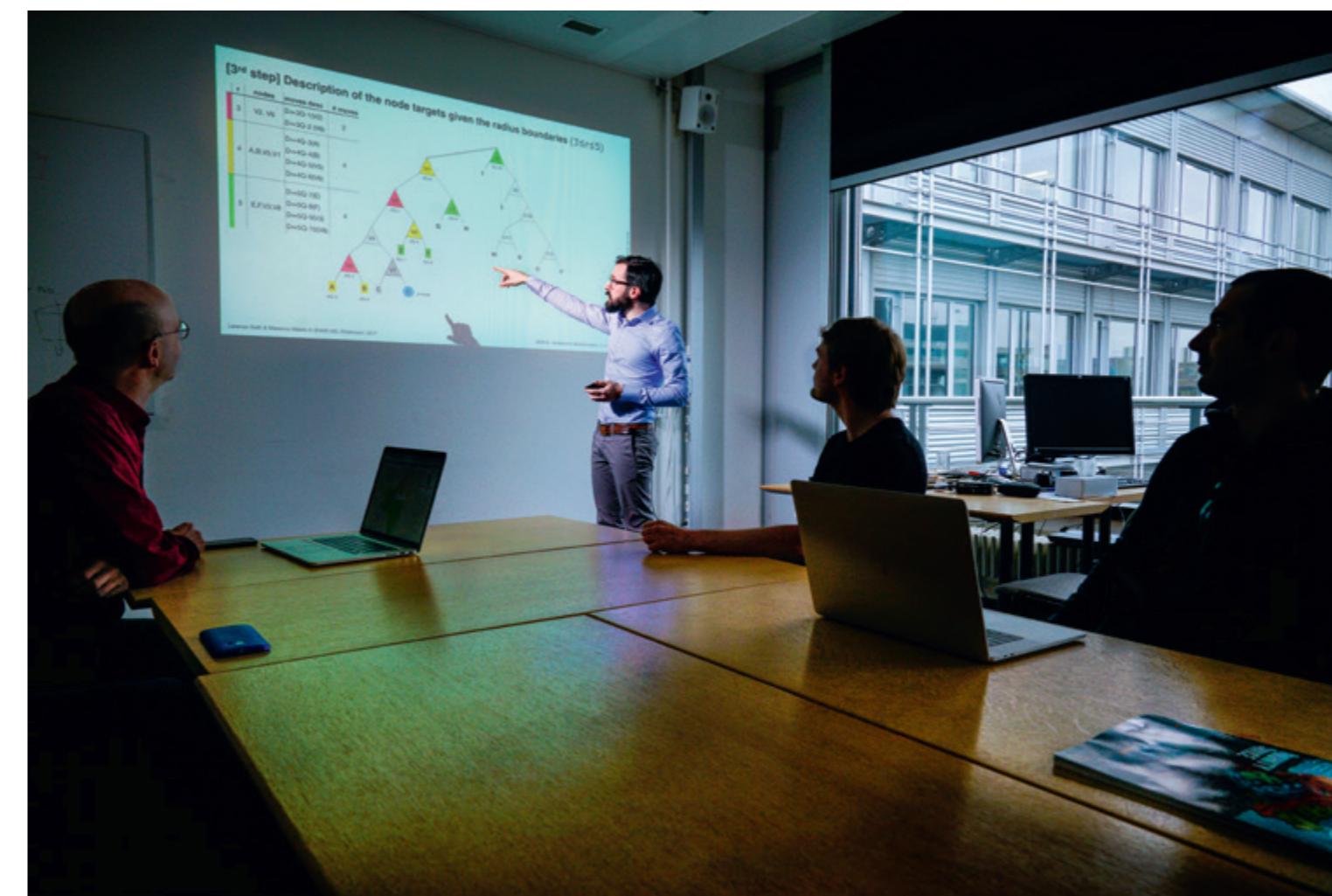
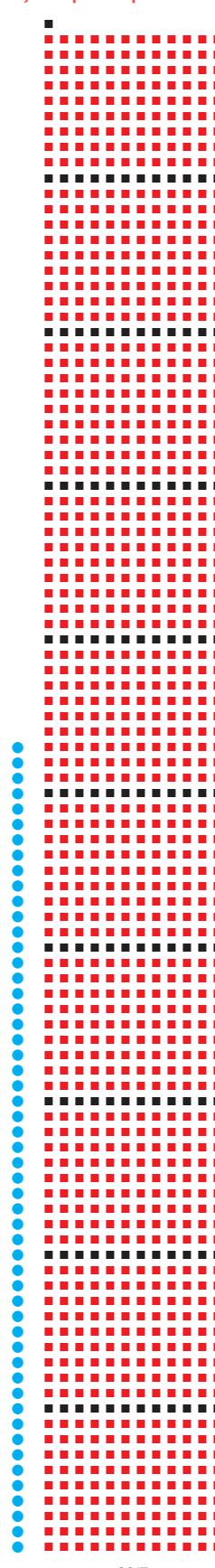
In the early years, the courses organized by SIB consisted mostly of introductions to sequence analysis, protein structure, statistics, phylogeny or evolution. Over time, more advanced and technical topics joined the training portfolio, such as high-performance computing and programming (machine learning, 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).

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.



53 courses
1,191 participants



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

FIGURES FROM 2017

Some of the most taught skills

Best coding practices HPC Machine learning

ChIP-seq R software Metabolomics Protein databases

Statistics Long read sequencing

Experimental design Variant annotation Regular expressions

Next-generation sequencing RNA-Seq

UNIX Python Proteogenomics and immunopeptidomics

Computational biology

Computer-aided drug design Snakemake

Sensitive data management Perl 6

Making open data sustainable

Open access has been a buzzword in the scientific community for some years now. SIB is committed to making this concept a reality for the information and data contained in its resources, thereby enabling science to make the most of bioinformatics discoveries. But the Institute is also concerned about the sustainability of open data in the long run.



Christine Durinx, PhD
SIB Associate Director



Chiara Gabella, PhD
SIB Project Manager

Open access does not mean free of cost, especially for databases that need significant human resources. With the current rate of 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?

Christine Durinx Imagine a centralized encyclopaedia of the function of hundreds of thousands of proteins, made accessible to life scientists throughout the world. It allows you to discover, for example, within a few minutes, a new form of an enzyme for the industrial production of biofuel. With 900,000 user requests per month, the UniProtKB/Swiss-Prot knowledgebase 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.

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."

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

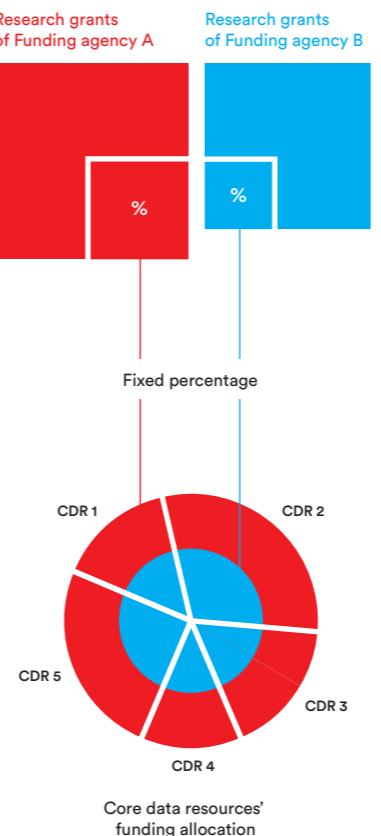
We should bear in mind that scientists have entrusted their research results to these databases. So the fact that their existence is so unstable could result in the loss of an immense wealth of biological information, and the associated investments.

What is the study about and what is the outcome?

Chiara Gabella In this study, we analysed 12 funding models for databases, and applied them to the UniProt knowledgebase.

The objective was to find the ideal funding model, by comparing

The Infrastructure model - a sustainable funding scheme for core data resources (CDR)



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. ●

A blog to go further

Read the discussion between C. Gabella and reviewer Helen Berman, Rutgers, The State University of New Jersey <https://blog.f1000.com/2018/02/07/how-best-to-fund-knowledgebases/>

Reference

Funding knowledgebases: Towards a sustainable funding model for the UniProt use case
By Gabella C et al.
F1000Research 2018, 6(ELIXIR):2051
[version 2; referees: 3 approved]

Expert curation is 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 biological knowledge, to support researchers in identifying the most relevant and substantiated evidence.

On expert curation and scalability: UniProtKB/Swiss-Prot as a case study
By Poux S et al.
Bioinformatics, 2017



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

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 150 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.

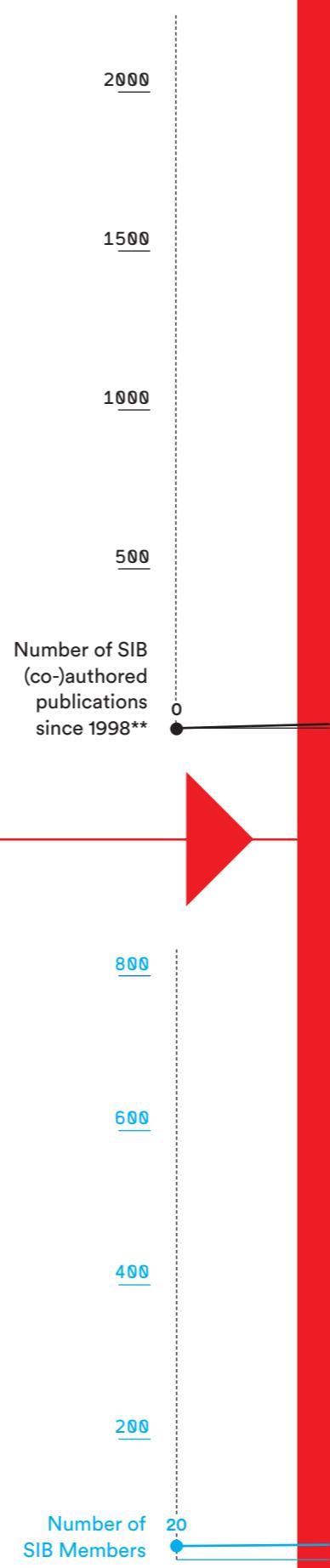
CREATION OF THE SIB SWISS INSTITUTE OF BIOINFORMATICS

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.*

5
groups

20
members

6
institutional partners



CREATION OF GENEBIO

Created to facilitate fund matching between industries and the Swiss government, GeneBio, SIB's commercial entity, was first focused on protein identification and characterization as well as proteome imaging.



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 (SIAF), DAVOS

67
groups

800
members

20
institutional partners

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

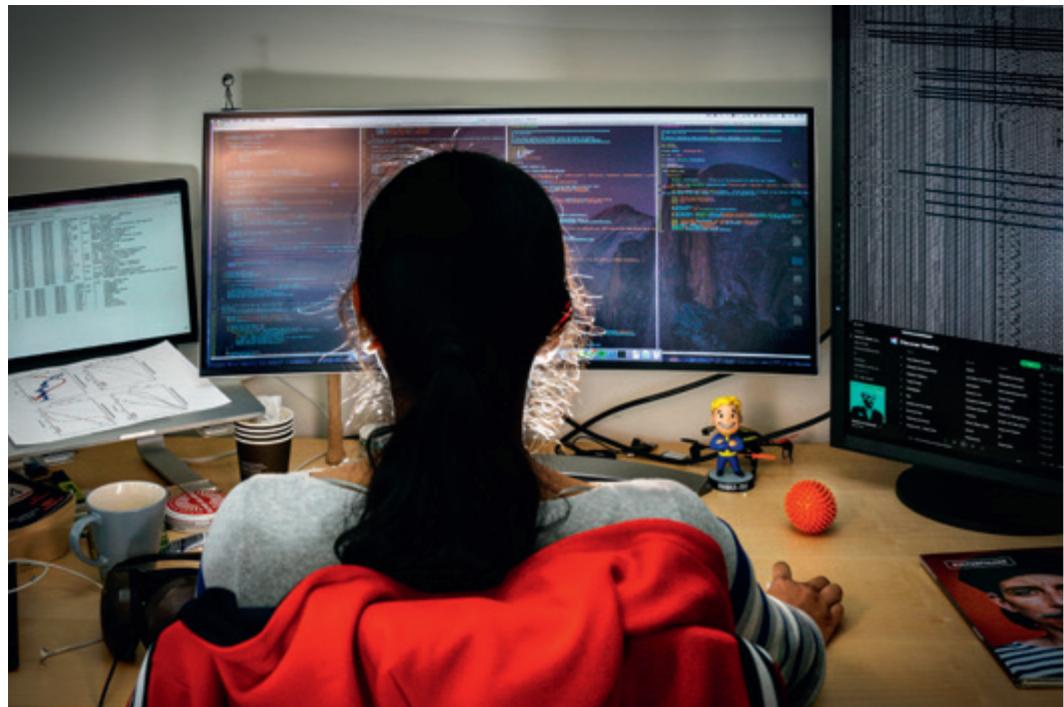
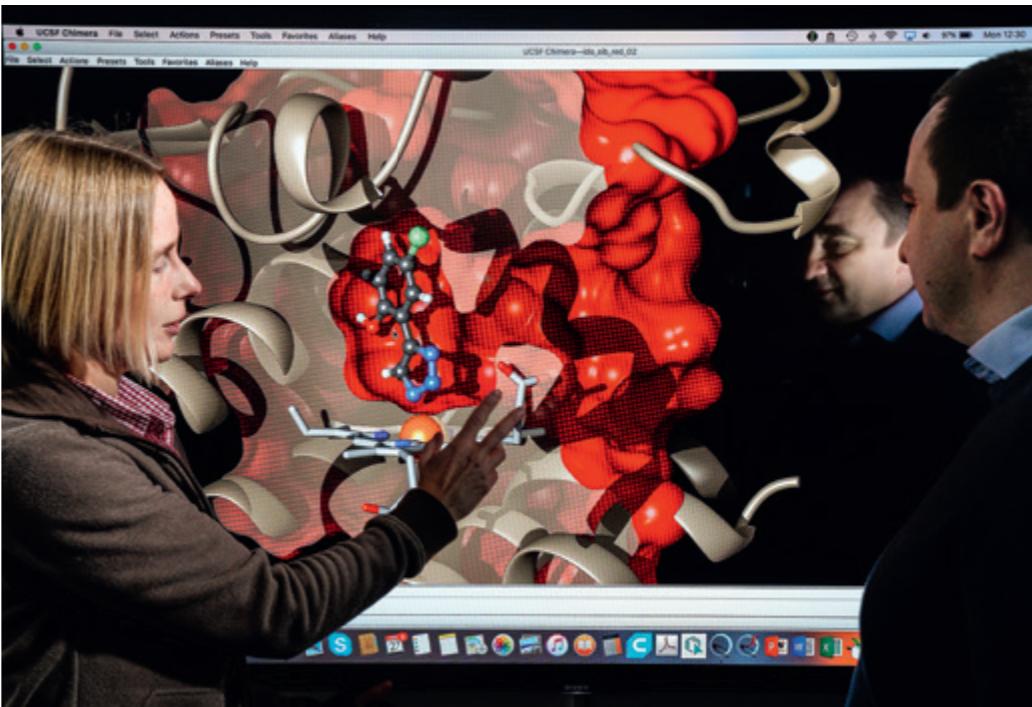
* The HUG remained an institutional partner of SIB until 2006, before joining again in 2016; ISREC remained until 2008 when it was integrated into EPFL, and Glaxo Wellcome Experimental Research until 2002.

** SOURCE: EUROPEPMC EXTRACTION FROM 30.03.1998 TO 17.01.2018



CHAPTER 3

Activities 2017



SIB's activities revolve around five pillars:

Infrastructure

**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:

| DOMAIN | SUB-DOMAIN | DATABASE | SOFTWARE TOOL |
|---|---------------------------------------|--|---|
|  Genes and genomes | Sequence alignment | LALIGN, Newick Utilities, T-Coffee | |
| | Similarity search | BLAST, BUSCO, LALIGN, Phylogibbs | |
| | Characterization/annotation | EPD, miROrtho, OMA, OpenFlu, OrthoDB, SwissRegulon | ChIP-Seq, ISA, PACMAN, QuasR, V-pipe |
| | Transcriptomics | Bgee, CleanEx, SwissRegulon | ISMARA, PPA, SNP2TFBS, TopAnat |
|  Proteins and proteomes | Protein sequences and identification | neXtProt, UniProtKB/Swiss-Prot, ViralZone | HAMAP, PeptideMass, Terminus, Translate |
| | Mass spectrometry and 2-DE data | SWISS-2D PAGE, WORLD-2D PAGE Repository | FindPept, GlycoMod, MSight |
| | Protein characterization and function | ENZYME, neXtProt, iPtgxDbs, Rhea, Swiss-Lipids, UniProtKB/Swiss-Prot | AACompSim, Biochemical Pathways, ProtScale |
| | Families, patterns and profiles | MyHits, PROSITE | MyDomains, pftools, PRATT |
| | Post-translational modification | SugarBind, UniCarb-DB, UniProtKB/Swiss-Prot | FindMod, GlycanMass, ISMARA |
| | Protein-protein interaction | STRING, UniProtKB/Swiss-Prot | |
| | Similarity search/alignment | UniProtKB/Swiss-Prot | BLAST, ClustalW, MyHits |
| | Imaging | | ImageMaster / Melanie, MSight |
|  Evolution and phylogeny | | Bgee, ImmunoDB, miROrtho, Swiss Orthology (OMA, OrthoDB), Selectome | |
|  Structural biology | | SWISS-MODEL Repository, SwissSideChain, SwissBiosostere | CAMEO, Click2Drug, SWISS-MODEL Workspace, Swiss-PdbViewer, SwissADME, SwissDock, SwissParam, SwissSimilarity, SwissTargetPrediction |
|  Systems biology | | Cellosaurus, Progenetix, SwissRegulon | arrayMap, CRUNCH, MetaNetX, The Systems Biology Research Tools |

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



Proteins & proteomes

UniProtKB/Swiss-Prot

Protein knowledgebase

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](#)

STRING

Protein-protein interactions

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, (conserved) co-expression and the literature.

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

Human protein knowledgebase

TYPE Knowledgebase with manual expert curation

DESCRIPTION Information on human protein biology such as function, involvement in diseases, mRNA/protein expression, protein/protein interactions, post-translational modifications, protein variations and their phenotypic effects.

HIGHLIGHT Advanced search options combining data at genomic, transcriptomic, proteomic and phenotypic levels.

[SEE P.58](#)

Glycomics@ExPASy

Glyco(proteo)mics resource portal

TYPE Web tools and knowledgebases with manual expert curation

DESCRIPTION Extensive set of resources on sugar structures (UniCarb-DB), their relationships with the proteins that carry them (GlyConnect) and the proteins that bind them, with a special focus on interactions between pathogen proteins and mammalian glycans (SugarBind).

HIGHLIGHT Unique portal for glyco(proteo)mics data analysis and interpretation.

[SEE P.59](#)



Structural biology

SwissDrugDesign

Drug design

TYPE Software tools

DESCRIPTION Web-based computer-aided drug design tools, from molecular docking (SwissDock) to pharmacokinetics and druglikeness (SwissADME), through virtual screening (SwissSimilarity) and target prediction of small molecules (SwissTargetPrediction).

HIGHLIGHT Comprehensive and integrated web-based drug design environment.

SEE P.63



Lipids

SwissLipids NEW

A knowledge resource for lipids

TYPE Knowledgebase

DESCRIPTION Information about known lipids, including knowledge of lipid structures, metabolism, interactions, and subcellular and tissular 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



Genes and genomes

Bgee

Gene expression evolution

TYPE Knowledgebase with manual expert curation

DESCRIPTION Information on gene expression evolution (incl. all types of transcriptomes), allowing retrieval and comparison of expression patterns between animals including human, model organisms and diverse species of evolutionary or agronomical relevance.

HIGHLIGHT Only resource to provide homologous gene expression between species.

SEE P.61

V-Pipe NEW

Viral genomics pipeline

TYPE Software tool

DESCRIPTION Pipeline integrating existing and in-house pieces of software, designed for the clinical application of next-generation sequencing to viral pathogens.

HIGHLIGHT Enabling reliable and comparable viral genomics and epidemiological studies and facilitating clinical diagnostics of viruses.

SEE P.52

SwissRegulon Portal

Tools and data for regulatory genomics

TYPE Software tools and knowledgebases

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

EPD

Eukaryotic Promoter Database

TYPE Knowledgebase with manual expert curation and software tools

DESCRIPTION Quality-controlled information on experimentally defined promoters of higher organisms, as well as web-based tools for promoter analysis.

SEE P.53

OMA

Orthology MAtrix browser

TYPE Automated phylogenomic database and software tool

DESCRIPTION High-quality orthology predictions among complete genomes.

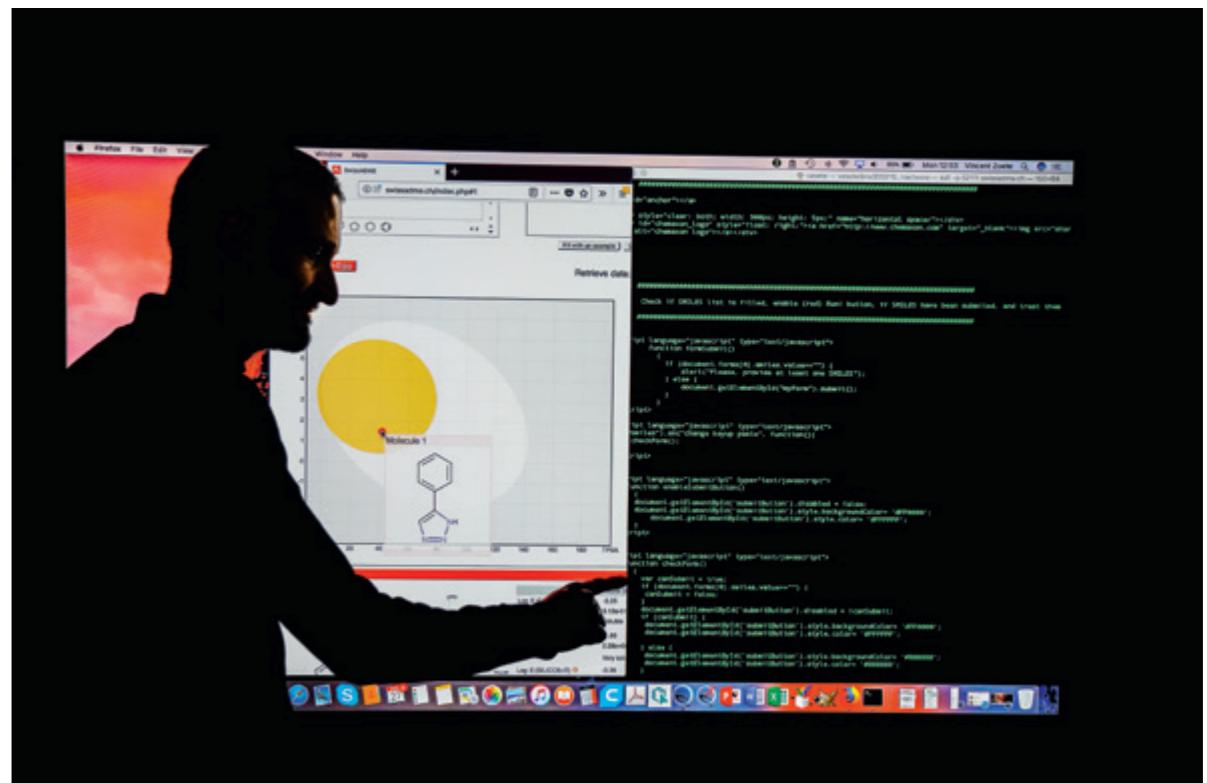
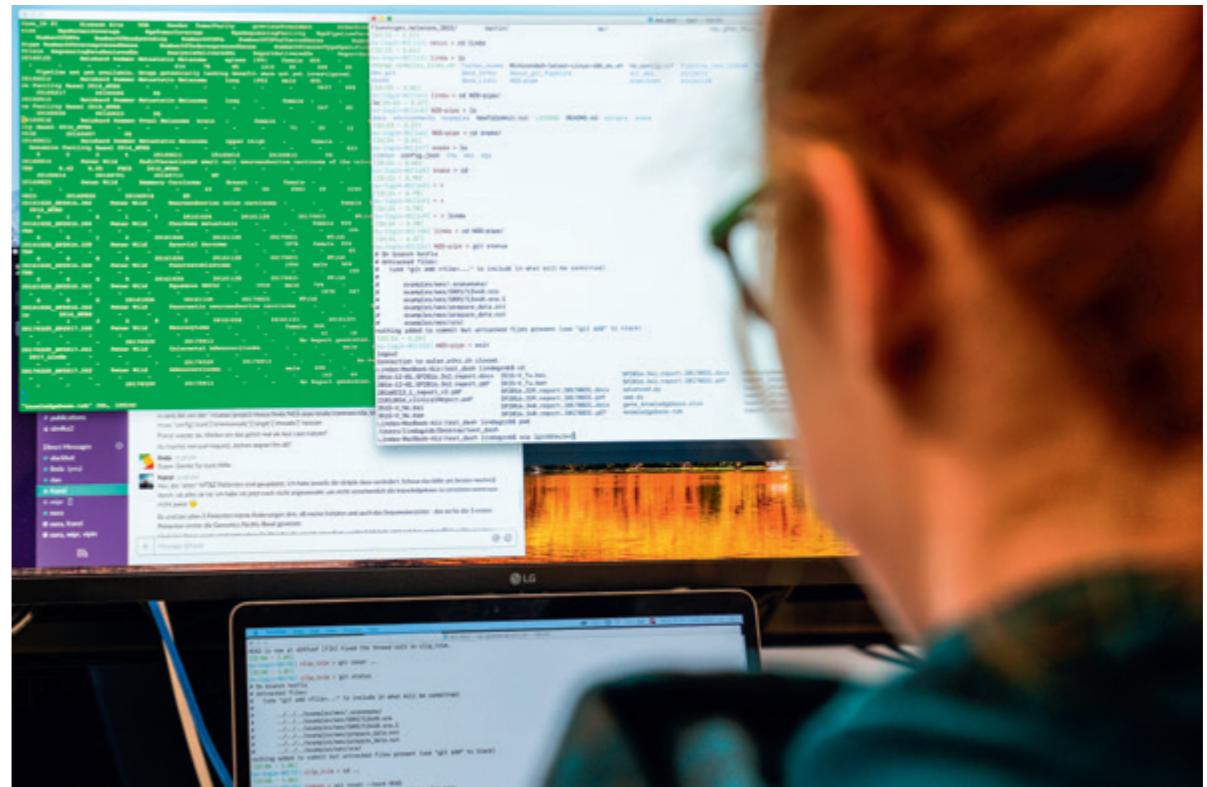
HIGHLIGHT Broad scope and size,

feature-rich web interface, availability in

a wide range of formats and interfaces,

frequent update schedule.

SEE P.60



Top

Programming is at the heart of many of our bioinformaticians' activity. It enables them to analyse data more swiftly or to develop softwares...

Bottom

...such as computer-aided drug design tools, which in turn enable life scientists to create the drugs of tomorrow.

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.



IOANNIS XENARIOS
SIB's Swiss-Prot and Vital-IT Group Leader
Professor at the Centre for Integrative Genomics, University of Lausanne and at the University of Geneva

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?

IX Depending on the needs of the groups, we provide three types

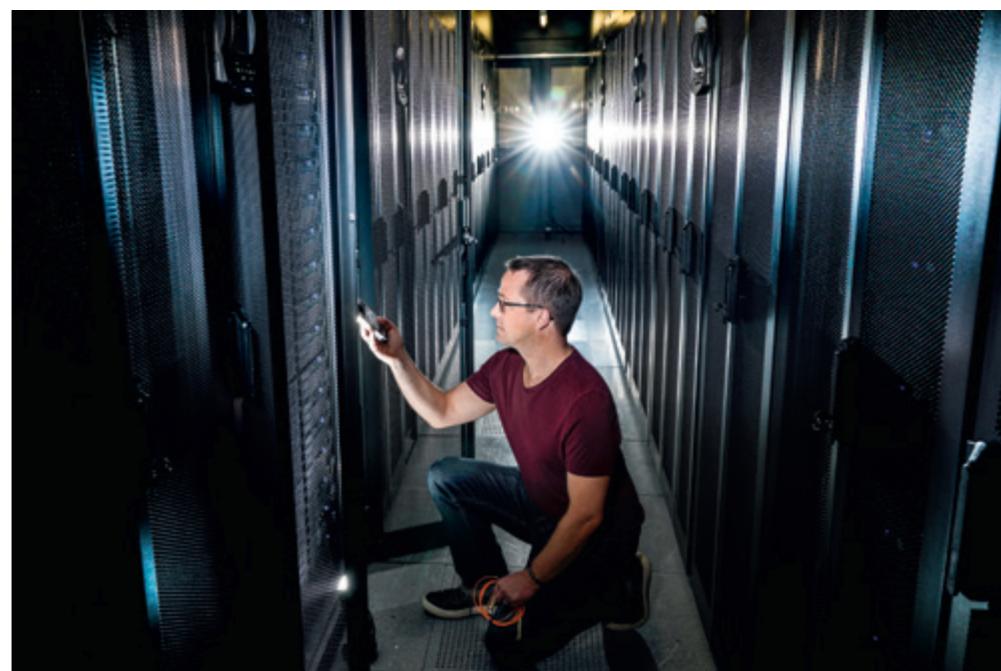
“Competence centres thus offer an economy of scale and a coordinated environment, where expertise is shared amongst their members.”

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



**How does it work at...
the DBM Bioinformatics Core Facility?**

“We provide a special type of scientific services, which focus on the complete analysis of next-generation sequencing data – or indeed any other type of high-throughput data, such as flow or mass cytometry. Each of these projects starts with a discussion with the users to define the appropriate experimental design (number of replicates, selection of technology or protocol). Once the data are generated, we perform an initial analysis and meet the users again to discuss the results. Most often there are new questions or additional analyses. Such projects typically last for months – sometimes even years.”

ROBERT IVANEK
SIB Group Leader,
DBM Bioinformatics Core Facility



Top
From high-performance computing resources...

Bottom
...to multidisciplinary expertise, gathered under a single roof: Swiss bioinformatics core facilities and competence centres offer a match to every need from life scientists.

researchers to include the costs of infrastructure usage for their projects in their grant application. What does this move represent, in your view?

IX 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 on 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

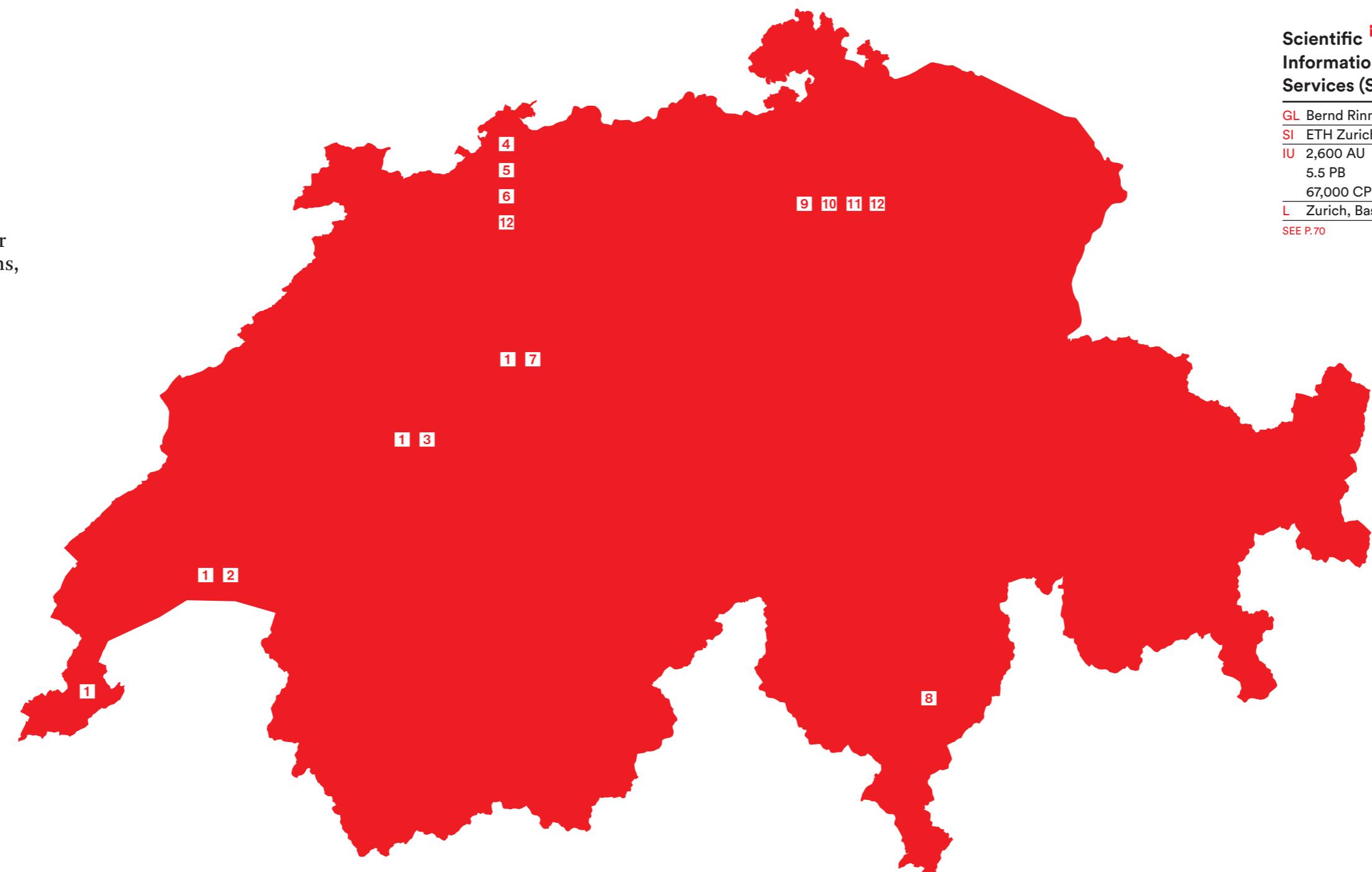
Twelve core facilities and competence centres are distributed in all the major Swiss academic institutions, and coordinated by SIB.

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

Vital-IT ¹

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 Core Facility (BCF) ²

GL Mauro Delorenzi and Frédéric Schütz
 SI SIB, University of Lausanne
 L Lausanne
 SEE P.53

Bioinformatics Unravelling Group (BUGFri) ³

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

FMI Computational Biology Group (FMICBG) ⁴

GL Michael Stadler
 SI Friedrich Miescher Institute
 IU 170 AU
 1 PB
 144 CP
 L Basel
 SEE P.56

DBM Computational Bioinformatics Core Facility ⁵

GL Robert Ivanek
 SI University of Basel, University Hospital Basel
 IU 170 AU
 1 PB
 144 CP
 L Basel
 SEE P.69

Centre for Scientific Computing (sciCORE) ⁶

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

Interfaculty Bioinformatics Unit (IBU) ⁷

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

Bioinformatics Core Unit (BCU) ⁸

GL Luciano Cascione
 SI Institute of Oncology Research
 IU 20-40 AU
 0.05 PB
 80 CP
 L Bellinzona
 SEE P.68

Service and Support for Science IT (S3IT) facility ¹¹

| |
|-----------------|
| GL Bernd Rinn |
| SI ETH Zurich |
| IU 2,600 AU |
| 5.5 PB |
| 67,000 CP |
| L Zurich, Basel |
| SEE P.70 |

NEXUS ¹⁰

Personalized Health Technologies

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

FGCZ Genome Informatics (FGCZ-GI) ⁹

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



Infrastructure, expertise and collaboration between bioinformatics and health experts, for the patients' benefit.

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.

Clinical bioinformatics

NAME SIB CLINICAL BIOINFORMATICS GROUP

TAGS personalized health; hospital collaborations; pipeline optimization; clinical training; practice harmonization; interoperability; genomics; oncology; infectious diseases; medical genetics.

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)

2017 highlights

| | |
|-------------|--|
| NAME | SIB PERSONALIZED HEALTH INFORMATICS GROUP (PHI) |
| TAGS | interoperability; personalized health; data management; standardization; precision therapy; information security |

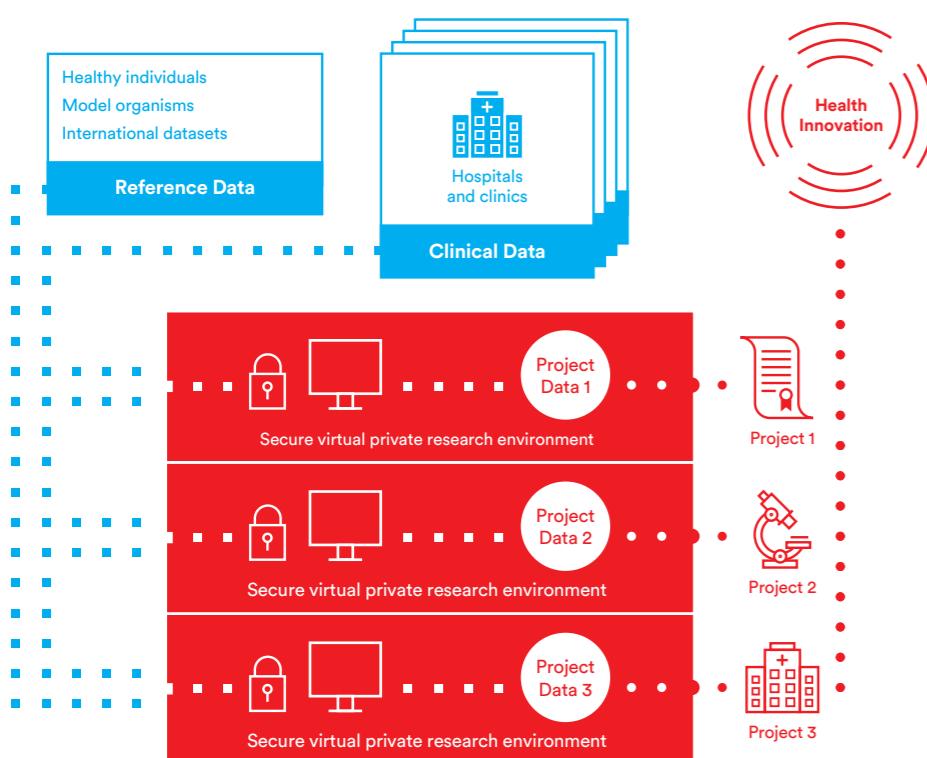
The group contributes to the SPHN initiative in a number of ways. As the group in charge of the Data Coordination Centre (DCC), it establishes standards for the interoperability of health-related data. These standards will allow for efficient nationwide collaborative research across hospitals and academic 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).

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. Swiss 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.



BioMedIT
Network of IT competence centres led by SIB

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 15 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 PIs 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').

A "driver" project for oncology treatment and research

SIB PI OLIVIER MICHELIN
(IN ASSOCIATION WITH SIB'S JACQUES FELLAY, PATRICH 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.

* PHRT: Personalized Health and Related Technology www.sfa-phrt.ch

Creating a strong bioinformatics community

IV. TRAINING IN BIOINFORMATICS

23
SIB groups engaged in teaching activities

1,191
trainees

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: Rémy 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.

SIB PhD Training Network

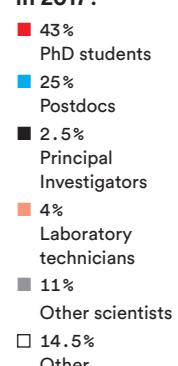
In 2017, the network celebrated its 10th anniversary with a special conference for its 240 members in Biel/Bienne. Other events in 2017 included the autumn school in "Machine Learning Applied to Systems Biology", jointly organized with SystemsX.ch, and the course on "Best practices in programming", which 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?



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

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.

[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, Alexandra Gavryushkina, and Christophe Dessimoz respectively presented the institution and the following resources: Genonets, BEAST2 and OMA.

14.12.2017

2,000th follower on Twitter

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

SIB International Young Bioinformatician Award – CHF 10,000

SEBASTIAN WASZAK, 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 REPPOSEDB - 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).

SIB Best Swiss Bioinformatics Graduate Paper Award – CHF 5,000

JOSÉ AGUILAR-RODRIGUEZ, University of Zurich, for his paper "A thousand empirical adaptive landscapes and their navigability", published in January 2017 in *Nature Ecology & Evolution*.

This paper, which explores the topography of adaptive landscapes in transcription regulation using high-throughput empirical data, was published as a part of his PhD thesis in Andreas Wagner's group.

[BC]² Best Poster Award, 1st prize

ANNA MARCIONETTI, SIB & University of Lausanne, with "Investigating the genetic basis of clownfish adaptive radiation using comparative genomics"

[BC]² Best 'Shared Poster' Award, 1st prize

LUKAS FOLKMAN, SIB & ETH Zurich, with "Kernelized Rank Learning for Personalized Drug Recommendation"

Research at SIB in 2017

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 function(s) 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.

Published in Genome Research

SIB Groups involved
Bioinformatics and Proteogenomics Group P. 58
Statistical Bioinformatics Group P. 56

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.

Published in Proceedings of the National Academy of Sciences

SIB Group involved
Host-Pathogen Genomics Group P. 54

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 Genetics

SIB Groups involved
Computational and Molecular Population Genetics Group P. 60
Interfaculty Bioinformatics Unit (IBU) P. 68

Supercoiling pushes molecular handcuffs along chromatin fibres

As it squeezes down the chromatin fibre, the cohesin 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 supercoiling of upstream DNA. Their research is thereby adding a key piece to the puzzle of gene expression regulation.

Published in Nucleic Acids Research

SIB Group involved
DNA and Chromosome Modelling Group P. 57

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

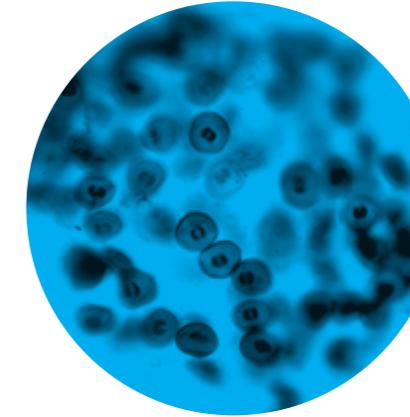
SIB Group involved
Computational Oncogenomics Group P. 52

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 P. 64



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 Nature Plants

SIB Groups involved
Vital-IT P. 71
Evolutionary Bioinformatics Group P. 61
Bioinformatics Core Facility (BCF) P. 53

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 mBio

SIB Group involved
Vital-IT P. 71

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 P. 55
Laboratory of Systems Biology and Genetics P. 53
Evolutionary Bioinformatics Group P. 61

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.



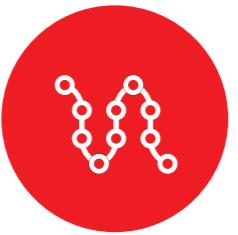
Genes and genomes

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](#)



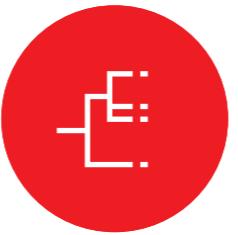
Proteins and proteomes

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 from different species or populations tell them 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](#)



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](#)

...ECOLOGY
from understanding how insects adapt to specific ecological niches, to documenting the biodiversity of fungi in the soil.



...MEDICINE AND HEALTH
from designing optimized proteins in cancer immunotherapy, to creating biomedical decision-support tools using text-mining.



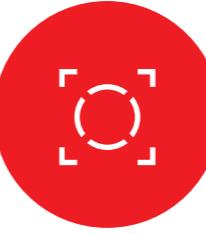
BASIC RESEARCH
from unravelling the evolutionary processes that have shaped today's biodiversity, to solving the equation behind a lizard's scale colour pattern.



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



Many areas of applications, including...



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](#)



Text mining and machine learning NEW

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](#)



Michael BAUDIS

GROUP NAME COMPUTATIONAL ONCOGENOMICS GROUP

INSTITUTION UNIVERSITY OF ZURICH CITY ZURICH

The Computational Oncogenomics Group works on the computational analysis of structural genome variations in cancer. Our work centres around our array-Map and Progenetix resources of curated molecular cytogenetic and sequencing data. Specific projects explore computational methods, genomics of selected tumour entities as well as genomic variant 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 genomic variants and phenotypic data. Other projects are related to genome data epistemology, e.g. geographic and diagnostic sampling biases.

KEY PAPER IN 2017
Mackay *et al.*, Integrated molecular meta-analysis of 1,000 pediatric high-grade and diffuse intrinsic pontine glioma. *Cancer Cell*

Niko BEERENWINKEL

GROUP NAME COMPUTATIONAL BIOLOGY

INSTITUTION ETH ZURICH, D-BSSE CITY ZURICH & BASEL

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 models, their implementation in computer programs, and their application to biomedical problems. We are conducting active research projects on HIV drug resistance, the somatic evolution of cancer, haplotype inference from ultra-deep sequencing data, reconstruction of signalling pathways from perturbation screens, and computational analysis of single-cell sequencing data.

KEY PAPER IN 2017
Mohammadi *et al.*, Automated design of synthetic cell classifier circuits using a two-step optimization strategy. *Cell Systems*

Sven BERGMANN

GROUP NAME COMPUTATIONAL BIOLOGY

INSTITUTION UNIVERSITY OF LAUSANNE CITY LAUSANNE

We develop concepts and algorithmic 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.

KEY PAPER IN 2017
Lamparter *et al.*, Genome-wide association between transcription factor expression and chromatin accessibility reveals regulators (...). *PLoS Comput Biol*.

MAIN DOMAIN OF ACTIVITY

Genes and genomes

SECONDARY DOMAINS OF ACTIVITY

Biocuration; Biomarkers; Knowledge-base; Microarrays; Next-generation sequencing; Oncology; Precision therapy; Software development; Text mining; APIs

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**Emmanouil DERMITZAKIS**

| GROUP NAME | GENOMICS OF COMPLEX TRAITS GROUP | MAIN DOMAIN OF ACTIVITY | Genes and genomes |
|-------------|----------------------------------|-------------------------------|---|
| INSTITUTION | UNIVERSITY OF GENEVA | SECONDARY DOMAINS OF ACTIVITY | Comparative genomics; Population genetics; Epigenetics; Functional genomics; Genome structure; GWAS; Next-generation sequencing; Oncology; Systems biology; Transcriptomics |
| CITY | GENEVA | DOMAIN OF APPLICATION | QTLtools |

Our group has a strong interest in population genetics and genetics of complex traits. We use various methodologies to understand the role of genetic variation in phenotypic variation and what fraction of genetic variation is harboured within functional elements of the human genome. Our main focus is on genome-wide analysis of gene expression and cellular phenotypes and their association with nucleotide variation. We attempt to detect functional genetic variation in regulatory elements and then use functional variation and accurately measured gene expression variation to bridge the genotype with disease phenotypes in association studies.

KEY PAPERS IN 2017

- Ongen H *et al.* Estimating the causal tissues for complex traits and diseases. *Nat Genet.*
Brown AA *et al.* Predicting causal variants affecting expression by using whole-genome sequencing and RNA-seq from multiple human tissues. *Nat Genet.*

| GROUP NAME | HOST-PATHOGEN GENOMICS GROUP | MAIN DOMAIN OF ACTIVITY | Genes and genomes |
|-------------|------------------------------|-------------------------------|--|
| INSTITUTION | EPFL LAUSANNE | SECONDARY DOMAINS OF ACTIVITY | GWAS; Immunology; Infectious diseases; Next-generation sequencing; Precision therapy |
| CITY | CITY | DOMAIN OF APPLICATION | Be |

| GROUP NAME | COMPUTATIONAL CANCER BIOLOGY GROUP | MAIN DOMAIN OF ACTIVITY | Genes and genomes |
|-------------|------------------------------------|-------------------------------|---|
| INSTITUTION | UNIVERSITY OF LAUSANNE | SECONDARY DOMAINS OF ACTIVITY | Immunology; Machine learning; Peptidomics; Single-cell biology; Transcriptomics |
| CITY | CITY | DOMAIN OF APPLICATION | Be ☀ |

| GROUP NAME | UNIVERSITY OF LAUSANNE | MAIN DOMAIN OF ACTIVITY | Genes and genomes |
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| INSTITUTION | CITY | SECONDARY DOMAINS OF ACTIVITY | GWAS; Immunology; Infectious diseases; Next-generation sequencing; Precision therapy |
| CITY | CITY | DOMAIN OF APPLICATION | Be ☀ |

| GROUP NAME | LAUSANNE | MAIN DOMAIN OF ACTIVITY | Genes and genomes |
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| INSTITUTION | CITY | SECONDARY DOMAINS OF ACTIVITY | Immunology; Machine learning; Peptidomics; Single-cell biology; Transcriptomics |
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| GROUP NAME | LAUSANNE | MAIN DOMAIN OF ACTIVITY | Genes and genomes |
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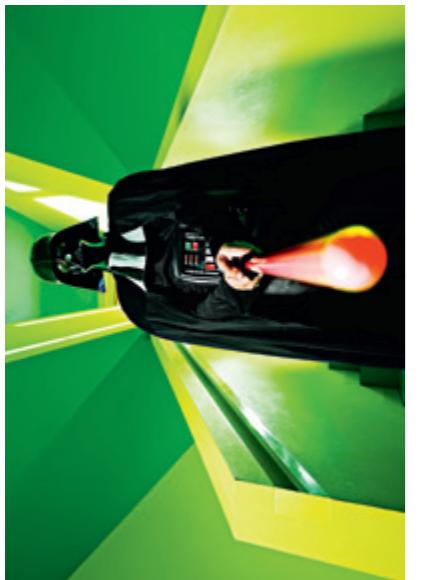
| GROUP NAME | LAUSANNE | MAIN DOMAIN OF ACTIVITY | Genes and genomes |
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| INSTITUTION | CITY | SECONDARY DOMAINS OF ACTIVITY | Immunology; Machine learning; Peptidomics; Single-cell biology; Transcriptomics |
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| GROUP NAME | LAUSANNE | MAIN DOMAIN OF ACTIVITY | Genes and genomes |
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| INSTITUTION | CITY | SECONDARY DOMAINS OF ACTIVITY | Immunology; Machine learning; Peptidomics; Single-cell biology; Transcriptomics |
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Michael STADLER

| MAIN DOMAIN OF ACTIVITY | | MAIN DOMAINS OF ACTIVITY | |
|-------------------------|---------------------------------|--|-------------------|
| GROUP NAME | FMI COMPUTATIONAL BIOLOGY GROUP | Genes and genomes | Genes and genomes |
| INSTITUTION | FRIEDRICH MIESCHER INSTITUTE | CITY | BASEL |
| | | Core facilities and competence centres | |

Activities 2017

We study gene regulation through the analysis and modeling of genome-wide datasets. We collaborate closely with experimental researchers on various biological topics, including cancer progression, chromatin biology and cellular differentiation, using data from various aspects of gene expression, such as DNA methylation and RNA transcription in single cells, measured by high-throughput sequencing. Our aim is to better understand how the different layers of epigenetic, transcriptional and post-transcriptional regulation interact and contribute to the control of gene expression.

KEY PAPERS IN 2017

Krebs AR *et al.*, Genome-wide single-molecule footprinting reveals high RNA polymerase II turnover at paused promoters. Mol Cell

SECONDARY DOMAINS OF ACTIVITY

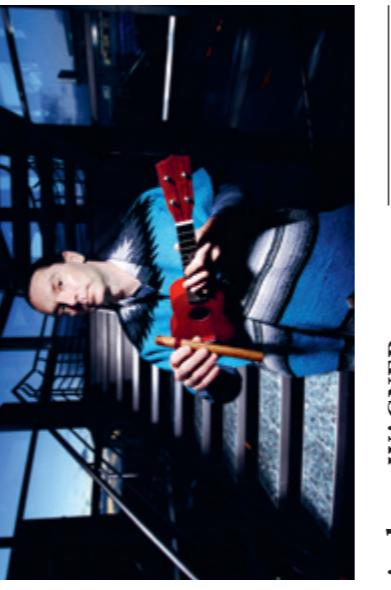
Epigenetics; Functional genomics; Genetic networks; Microarrays; Next-generation sequencing; Oncology; Single-cell biology; Software development; Systems biology; Transcriptomics

DOMAINS OF APPLICATION

DATA MINING; GWAS; Machine learning; Selection; Software development; Bayesian random regression models

RESOURCES

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Andreas WAGNER

| MAIN DOMAIN OF ACTIVITY | | MAIN DOMAIN OF ACTIVITY | |
|-------------------------|------------------------------------|--|-------------------|
| GROUP NAME | EVOLUTIONARY SYSTEMS BIOLOGY GROUP | Genes and genomes | Genes and genomes |
| INSTITUTION | UNIVERSITY OF ZURICH | CITY | ZURICH |
| | | Evolution; Experimental biology; Genetic networks; Molecular evolution | |

Activities 2017

In the Evolutionary Systems Biology Group, we study the evolution and evolvability 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 sequence data, microarray expression data, and high-throughput protein interaction data. Our work uses comparative analysis of genomic data, laboratory evolution experiments and mathematical modelling.

KEY PAPERS IN 2017

Fortuna MA *et al.*, The genotype-phenotype map of an evolving digital organism. PLoS Comput Biol.

SECONDARY DOMAINS OF ACTIVITY

Evolution; Experimental biology; Genetic networks; Molecular evolution

DOMAIN OF APPLICATION

DATA MINING; GWAS; Machine learning; Selection; Software development; Systems biology; Transcriptomics

RESOURCES

SwissRegulon ISMARA CRUNCH



Matthew ROBBINSON

| MAIN DOMAIN OF ACTIVITY | | MAIN DOMAIN OF ACTIVITY | |
|-------------------------|------------------------------|--|-------------------|
| GROUP NAME | COMPLEX TRAIT GENETICS GROUP | Genes and genomes | Genes and genomes |
| INSTITUTION | UNIVERSITY OF LAUSANNE | CITY | LAUSANNE |
| | | Bioinformatics; Epigenetics; Immunology; Microarrays; Next-generation sequencing; Oncology; Single-cell biology; Software development; Transcriptomics | |

Activities 2017

We develop and apply statistical methodologies to large human phenotype-genotype datasets, to address 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 effects to 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.

KEY PAPERS IN 2017

Minoux M *et al.*, Gene bivalency at Polycomb domains regulates cranial neural crest positional identity. Science

SECONDARY DOMAINS OF ACTIVITY

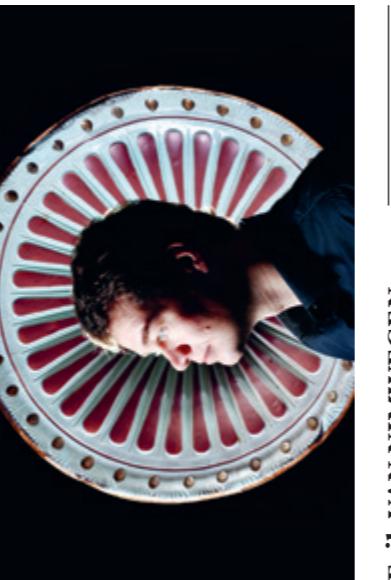
Data mining; Population genetics; GWAS; Machine learning; Selection; Software development

DOMAIN OF APPLICATION

DATA MINING; GWAS; Machine learning; Selection; Software development

RESOURCES

Cytometry dATA analYSIS Tools (CATALYST) CrisPRVariants conquer



Erik VAN NIMWEGEN

| MAIN DOMAIN OF ACTIVITY | | MAIN DOMAIN OF ACTIVITY | |
|-------------------------|------------------------------|---|-------------------|
| GROUP NAME | GENOME SYSTEMS BIOLOGY GROUP | Genes and genomes | Genes and genomes |
| INSTITUTION | UNIVERSITY OF BASEL | CITY | BASEL |
| | | Comparative genomics; Population genetics; Evolution; Genetic networks; Next-generation sequencing; Phylogeny; Systems biology; Transcriptomics; Gene regulation; Single-cell biology | |

Activities 2017

Our main research interest in the Genome Systems Biology (GSB) 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.

KEY PAPER IN 2017

Aguilar Rodriguez J *et al.*, 1000 empirical adaptive landscapes and their navigability. Nat Ecol Evol.

SECONDARY DOMAINS OF ACTIVITY

Bioinformatics; Evolution; Genetic networks; Molecular evolution

DOMAIN OF APPLICATION

DATA MINING; GWAS; Machine learning; Selection; Software development

RESOURCES

SwissRegulon ISMARA CRUNCH



Mark ROBINSON

| MAIN DOMAIN OF ACTIVITY | | MAIN DOMAIN OF ACTIVITY | |
|-------------------------|----------------------------------|--|-------------------|
| GROUP NAME | STATISTICAL BIOINFORMATICS GROUP | Genes and genomes | Genes and genomes |
| INSTITUTION | UNIVERSITY OF ZURICH | CITY | ZURICH |
| | | Bioinformatics; Epigenetics; Immunology; Microarrays; Next-generation sequencing; Oncology; Single-cell biology; Software development; Transcriptomics | |

Activities 2017

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 data from 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.

KEY PAPERS IN 2017

Robinson MR *et al.*, Genotype–covariate interaction effects and the heritability of adult body mass index. Nat Genet.

SECONDARY DOMAINS OF ACTIVITY

Population genetics; Evolution; Comparative genomics; Population genetics; Evolution; Genetic networks; Next-generation sequencing; Phylogeny; Systems biology; Transcriptomics; Gene regulation; Single-cell biology

DOMAIN OF APPLICATION

DATA MINING; GWAS; Machine learning; Selection; Software development

RESOURCES

Sonegon C and Robinson MD, Towards unified quality verification of synthetic count data with countsimQC. Bioinformatics



Andrzej STASIAK

| MAIN DOMAIN OF ACTIVITY | | MAIN DOMAIN OF ACTIVITY | |
|-------------------------|------------------------------------|--|-------------------|
| GROUP NAME | DNA AND CHROMOSOME MODELLING GROUP | Genes and genomes | Genes and genomes |
| INSTITUTION | UNIVERSITY OF LAUSANNE | CITY | LAUSANNE |
| | | Protein structure; Protein reconstruction; Protein structure and interaction | |

Activities 2017

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

KEY PAPER IN 2017

Omidí S *et al.*, Automated incorporation of pairwise dependency in transcription factor binding site prediction using dinucleotide weight tensors. PLoS Comput Biol.

SECONDARY DOMAINS OF ACTIVITY

Protein structure; Protein reconstruction; Protein structure and interaction

DOMAIN OF APPLICATION

DATA MINING; GWAS; Machine learning; Selection; Software development

RESOURCES

Benedetti F *et al.*, Transcription explains supercoiling formation of self-interacting chromatin domains in *S. pombe*. Nucleic Acids Res.



Evgeny ZDOBNOV

| GROUP NAME | COMPUTATIONAL EVOLUTIONARY GENOMICS GROUP |
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The group is active in the fields of comparative genomics and shotgun metagenomics. We study molecular evolution, develop approaches to genomics data analyses, and implement computational pipelines. We apply sequencing data, and revise these models using the novel data. We study functional genomic elements on the basis of sequence variability among different species and within populations. Our interests range from arthropod genomics, including invertebrate vectors of human pathogens, to the evolution of viruses and clinical microbiology.

| KEY PAPERS IN 2017 | Djordjevic M et al. |
|--------------------|--|
| | Scoring targets of transcription in bacteria rather than focusing on individual binding sites. |
| | Mol Biol Evol. |

Waterhouse RM et al. BUSCO applications from quality assessments to gene prediction and phylogenomics. Mol Biol Evol.

Ioannidis P et al. Genomic features of the damselfly *Calopteryx splendens* representing a sister clade to most insect orders. Genome Biol Evol.

Omasits U et al. An integrative strategy to identify the entire protein coding potential of prokaryotic genomes by proteogenomics. Genome Res.



Christian AHRENS

| GROUP NAME | BIOINFORMATICS AND PROTEOGENOMICS GROUP |
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Our research revolves around the bioinformatic integration and analysis of datasets from state-of-the-art omics technologies, which we obtain through close collaboration with experimental biologists. These datasets include genome sequences, gene and protein expression, as well as metabolomics data. One particular focus is to exploit the unique advantages of proteomics data, including strategies to identify all proteins encoded in a genome (proteogenomics). Recently, we started to study the role of microbiomes – e.g. for plant protection – by applying metagenomics, genomics and transcriptomics approaches.

| KEY PAPERS IN 2017 | Hiller-Bodmer M et al. |
|--------------------|--|
| | Competition assays and physiological experiments of soil and phyllosphere yeasts identify <i>Candida subashii</i> (...). |
| | BMC Microbiol. |

Montes Vidal D et al. Long-chain alkyl cyanides: unprecedented volatile compounds released by pseudomonads (...). Angew Chem Int Ed Engl.

Montes Vidal D et al. Long-chain alkyl cyanides: unprecedented volatile compounds released by pseudomonads (...). Angew Chem Int Ed Engl.



Amos BAIRROCH

| GROUP NAME | COMPUTER AND LABORATORY INVESTIGATION OF PROTEINS OF HUMAN ORIGIN (CALIPHO) |
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We aim to use a combination of bioinformatics and experimental methodologies to increase knowledge about the function of the 20,000 protein-coding genes in the human genome. Our main mission is the development of neXt-Prot, a human protein knowledge resource. We also annotate the effects of human protein variations in the context of cancers and genetic diseases. We are part of the HUPO Human Protein Project, which aims to validate the existence of all predicted human proteins in biological samples by mass spectrometry. We are active in the development of ontologies/standardization resources such as the Cellosaurus and ICEPO.

| KEY PAPERS IN 2017 | Hinard V et al. |
|--------------------|---|
| | Annotation of functional impact of voltage-gated (...). |
| | Hum Mutat. |

Ommen GS et al. Progress on the HUPO draft human proteome: 2017 Metrics (...). J Proteome Res.

Schaeffer M et al. The neXtProt peptide uniqueness checker: a tool for the (...) Bioinformatics



Frédérique LISACEK

| GROUP NAME | PROTEOME INFORMATICS GROUP |
|------------|----------------------------|
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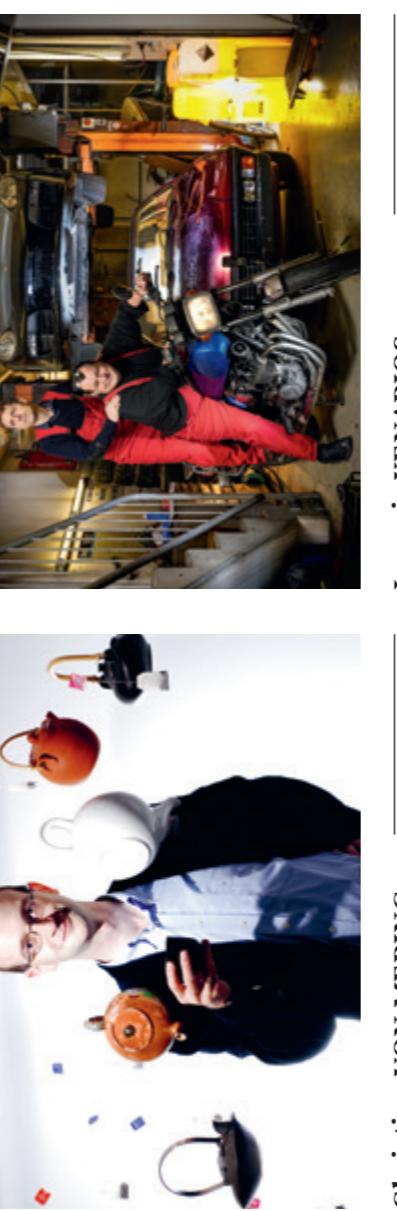
In the Proteome Informatics Group (PIG), we are involved in software and database development for the benefit of the proteinomes and glycomics communities. These resources are made available through the ExPASy server. Software tools support experimental mass spectrometry data analysis, mainly for the detection of post-translational modifications, with a strong focus on glycosylation. We also develop integrated tools and databases to query knowledge of carbohydrates attached to proteins, carbohydrate structures and protein-carbohydrate interactions.

| KEY PAPERS IN 2017 | Matias Rodrigues JF et al. |
|--------------------|--|
| | highly efficient N-terminal search with confidence estimates, for RNA (...). |
| | Bioinformatics |

Feigelman R et al. Sputum DNA sequencing in cystic fibrosis: non-invasive access to the lung microbiome (...).

Szklarczyk D et al. The STRING database in 2017: quality-controlled protein-protein association networks (...).

Mortag A et al. Updates in Rhea - an expert curated and sealable Uni-ProteKB/Swiss-Prot as a case study.



Ioannis XENARIOS

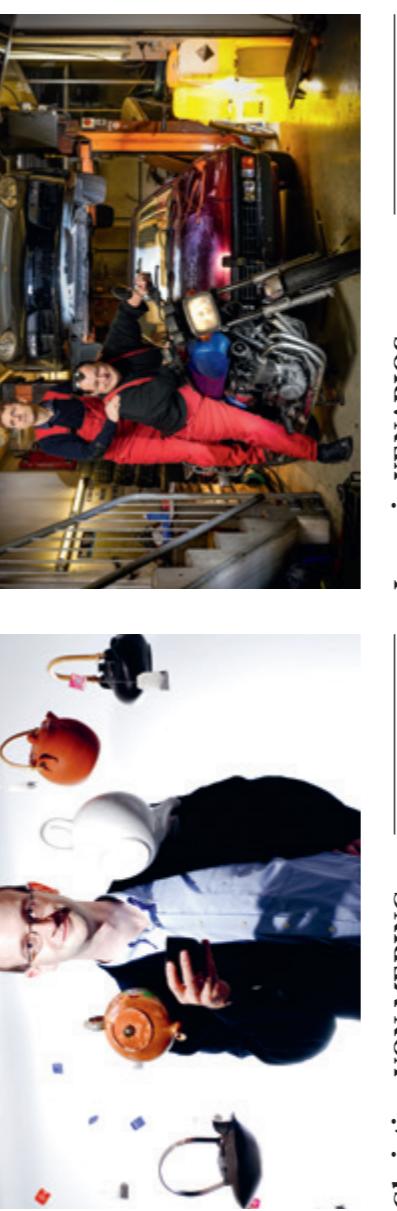
| GROUP NAME | SWISS-PROT |
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In the Swiss-Prot Group, we develop, annotate and maintain the UniProtKB/Swiss-Prot protein sequence database, the most widely used protein information resource in the world and an ELIXIR Core Data Resource, as well as a number of other internationally renowned expert-curated knowledge resources. These include the HAMAP and PROSITE databases of protein families and domains, the ENZYME database of enzyme nomenclature, the Rhea database of biochemical reactions, the SwissLipids database of lipid structures and biological knowledge, and the ViralZone portal. The group also participates in the development and maintenance of the ExPASy website.

| KEY PAPERS IN 2017 | Hulo C et al. |
|--------------------|--|
| | Bacterial virus ontology; coordinating across databases. |
| | Virus |

Poux S et al. On expert curation and sealability: Uni-ProteKB/Swiss-Prot as a case study.

Bioinformatics



Lydie LANE

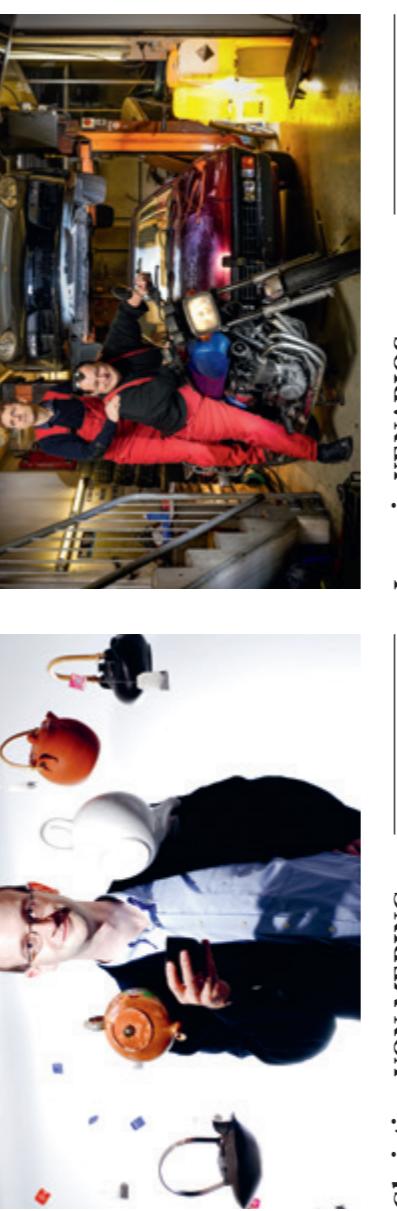
| GROUP NAME | COMPUTER AND LABORATORY INVESTIGATION OF PROTEINS OF HUMAN ORIGIN (CALIPHO) |
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| KEY PAPERS IN 2017 | Hinard V et al. |
|--------------------|---|
| | Annotation of functional impact of voltage-gated (...). |
| | Hum Mutat. |

Ommen GS et al. Progress on the HUPO draft human proteome: 2017 Metrics (...). J Proteome Res.

Schaeffer M et al. The neXtProt peptide uniqueness checker: a tool for the (...) Bioinformatics



Amos BAIRROCH

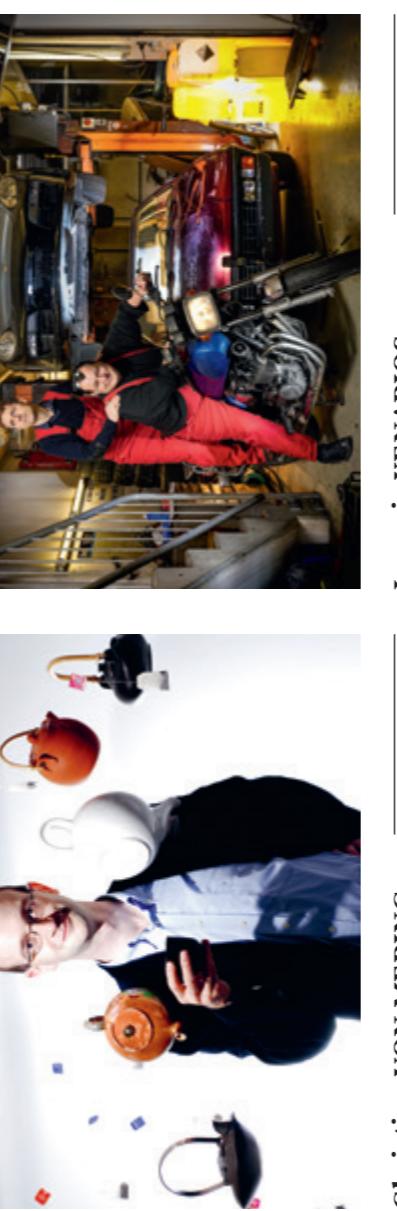
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|--------------------|---|
| | Annotation of functional impact of voltage-gated (...). |
| | Hum Mutat. |

Ommen GS et al. Progress on the HUPO draft human proteome: 2017 Metrics (...). J Proteome Res.

Schaeffer M et al. The neXtProt peptide uniqueness checker: a tool for the (...) Bioinformatics



Lydie LANE

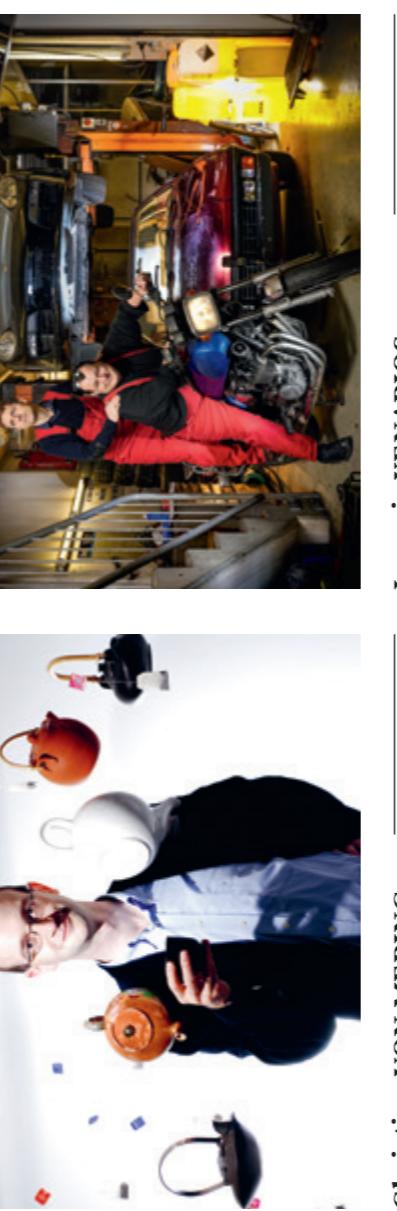
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| KEY PAPERS IN 2017 | Hinard V et al. |
|--------------------|---|
| | Annotation of functional impact of voltage-gated (...). |
| | Hum Mutat. |

Ommen GS et al. Progress on the HUPO draft human proteome: 2017 Metrics (...). J Proteome Res.

Schaeffer M et al. The neXtProt peptide uniqueness checker: a tool for the (...) Bioinformatics



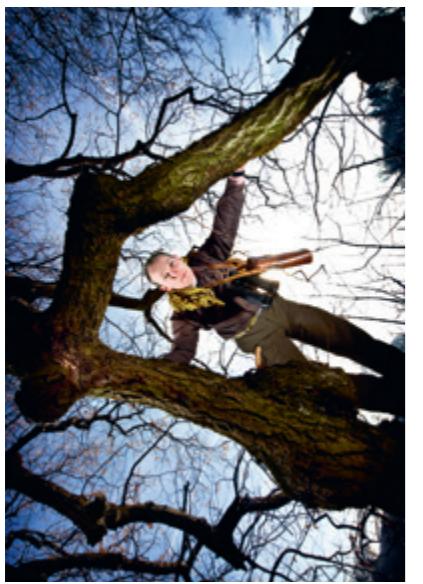
**Nicolas SALAMIN**

| GROUP NAME | COMPUTATIONAL PHYLOGENETICS GROUP | INSTITUTION | UNIVERSITY OF LAUSANNE |
|------------|-----------------------------------|-------------|------------------------|
| CITY | LAUSANNE | | |

We develop models and software to better understand the evolution of organisms and to test macroevolutionary hypotheses. We are looking at the ecological, genomic and morphological factors that constrain speciation and adaptation. We focus on phylogenetic methods, clownfish and plant selection, modelling the evolution of DNA sequences and phenotypes, the mode and tempo of species evolution and the spatially explicit evolution of diversity. Our aim is to develop better models to analyse sequence data and quantitative models to estimate macroevolutionary patterns and processes.

KEY PAPERS IN 2017

| | | | | | | |
|--|--|--|---|---|--|---|
| Meyer X et al. Accelerating Bayesian inference for evolutionary biology models. | Davydov I et al. State aggregation for fast likelihood computations in molecular evolution. | Meyer X et al. Scheduling finite difference approximations for DAG-modeled (...). | Bando-Sottani J et al. BEAST—A community teaching material resource for BEAST 2. | Müller NF et al. The structured coalescent and its approximations. | Rasmussen DA et al. Phylogenetics on local sexual contact networks. | Gros-Balthazard M et al. The discovery of wild date palms in Oman reveals a complex domestication history involving centers in the Middle East and Africa. |
|--|--|--|---|---|--|---|

**Tanja STADLER**

| GROUP NAME | COMPUTATIONAL EVOLUTION GROUP | INSTITUTION | ETH ZURICH, D-BSSE |
|------------|-------------------------------|-------------|--------------------|
| CITY | ZURICH, BASEL | | |

In the Computational Evolution Group, we develop phylogenetic tools in order to understand evolutionary processes. Using our phylogenetic methods, we aim to improve our understanding of past evolutionary and population dynamic processes on different scales. We address questions in a number of fields, focusing on epidemiology, public health and medicine, ecology and evolution, and language evolution. In our daily work, we define and analyse stochastic models, implement computational methods, analyse empirical data, and discuss our new insights with clinicians, and public-health policy makers, as well as ecologists and palaeontologists.

KEY PAPERS IN 2017

| | | | | | | |
|---|--------------------|---|----------------|--|-------------------|---|
| Bando-Sottani J et al. BEAST—A community teaching material resource for BEAST 2. | Systematic Biology | Müller NF et al. The structured coalescent and its approximations. | Mol Biol Evol. | Rasmussen DA et al. Phylogenetics on local sexual contact networks. | PLoS Comput Biol. | Gros-Balthazard M et al. The discovery of wild date palms in Oman reveals a complex domestication history involving centers in the Middle East and Africa. |
|---|--------------------|---|----------------|--|-------------------|---|

**Daniel WEGMANN**

| GROUP NAME | STATISTICAL AND COMPUTATIONAL EVOLUTIONARY BIOLOGY GROUP | INSTITUTION | UNIVERSITY OF FRIBOURG |
|------------|--|-------------|------------------------|
| CITY | FRIBOURG | | |

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 and ecological 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.

| MAIN DOMAIN OF ACTIVITY | Evo: Evolution and phylogeny | SECONDARY DOMAINS OF ACTIVITY | Biostatistics; Population genetics; Evolution; GWAS; Machine learning; Next-generation sequencing; Paleogenomics; Selection; Software development; Human genetics |
|-------------------------|------------------------------|-------------------------------|---|
| DOMAINS OF APPLICATION | BEAST add-ons | RESOURCES | ABCtoolbox ATLAS RASPerry |

KEY PAPERS IN 2017

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| Gros-Balthazard M et al. The discovery of wild date palms in Oman reveals a complex domestication history involving centers in the Middle East and Africa. |
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SIB groups

**Olivier MICHELIN**

| GROUP NAME | STRUCTURAL BIOLOGY | INSTITUTION | UNIVERSITY OF LAUSANNE |
|------------|--------------------|-------------|------------------------|
| CITY | LAUSANNE | | |

Inference of evolutionary jumps in large phylogenies using Levy processes. Genetics

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 engineering and drug design. Notably, we develop novel inhibitors of important targets for 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.

| MAIN DOMAIN OF ACTIVITY | Drug design: Immunology; Molecular dynamics; Oncology; Precision therapy; Protein structure and interaction; Software development; Systems biology | SECONDARY DOMAINS OF ACTIVITY | Drug design; Immunology; Molecular dynamics; Oncology; Precision therapy; Protein structure and interaction; Software development; Systems biology |
|-------------------------|--|-------------------------------|--|
| DOMAINS OF APPLICATION | SwissDock | RESOURCES | SwissTargetPrediction SwissADME |

KEY PAPERS IN 2017

| | | | |
|---|--|--|---|
| Rohrig UF et al. The binding mode of N-hydroxyamidines to indoleamine 2,3-dioxygenase 1 (IDO). Biochemistry Sci Rep. | Chaskar P et al. On-the-Fly QM/MM docking with attracting cavities. J Chem Inf Model. | Reidenbach AG et al. Conserved ciprofamide-molecule modulation of COQ8 reveals regulation of the ancient kinase-like UbB family. Cell Chem Biol. Sci Rep. | Daina A et al. SwissADME: a free web tool to evaluate pharmacokinetics, drug-likeness and medicinal chemistry friendliness of small (...). |
|---|--|--|---|

**Matteo DAL PERARO**

| GROUP NAME | LABORATORY FOR BIOMOLECULAR MODELING | INSTITUTION | EPFL |
|------------|--------------------------------------|-------------|------|
| CITY | LAUSANNE | | |

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. Multiscale simulations and dynamic integrative modeling are used to investigate the function of molecular assemblies, mimicking the conditions of the native cellular environment.

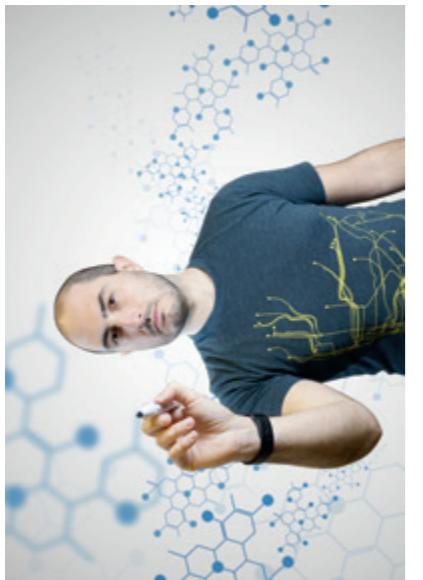
| MAIN DOMAIN OF ACTIVITY | Structural biology | SECONDARY DOMAINS OF ACTIVITY | Biochemistry; Drug resistance; Lipids; Protein dynamics; Protein structure and interaction; Software development |
|-------------------------|--------------------------------|-------------------------------|--|
| DOMAINS OF APPLICATION | pow ^{er} LipidBuilder | RESOURCES | LipidBuilder |

**Bruno CORREIA**

| GROUP NAME | LABORATORY OF PROTEIN DESIGN AND IMMUNOENGINEERING | INSTITUTION | EPFL |
|------------|--|-------------|------|
| CITY | LAUSANNE | | |

The Laboratory of Protein Design & Immunoengineering 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, protein expression and purification, and protein biophysical characterization.

| MAIN DOMAIN OF ACTIVITY | Abriata LA et al. Signal sensing and transduction by histidine kinases as unveiled through studies on a temperature sensor. Acc Chem Res. Sci Rep. | SECONDARY DOMAINS OF ACTIVITY | Reidenbach AG et al. Conserved ciprofamide-molecule modulation of COQ8 reveals regulation of the ancient kinase-like UbB family. Cell Chem Biol. Sci Rep. |
|-------------------------|--|-------------------------------|--|
| DOMAINS OF APPLICATION | Tano G et al. Disentangling constraints using viability evolution principles in integrative modeling of macromolecular assemblies. Cell Chem Biol. Sci Rep. | RESOURCES | Parker CG et al. Ligand and target discovery by fragmentation screening in human cells. Cell Chem. Sci. Rep. |

**Torsten SCHWEDE**

| GROUP NAME | COMPUTATIONAL STRUCTURAL BIOLOGY |
|---|----------------------------------|
| INSTITUTION | 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. | |

KEY PAPERS IN 2017
Bertoni M et al. Modelling protein quaternary structure of homo- and hetero-oligomers beyond homology. *Sci Rep.*
Biemert S et al. The SWISS-MODEL Repository - new features and functionality. *Nucleic Acids Res.*

Haas J et al. Continuous automated model evaluation (CAMEO) complementing the critical assessment of structure prediction in CASP12. *Proteins*

**Giovanni CIRIELLO**

| MAIN DOMAIN OF ACTIVITY | Systems biology |
|--|--|
| SECONDARY DOMAINS OF ACTIVITY | Data mining; Drug design; Knowledge base; Machine learning; Protein structure and interaction; Software development; Homology modelling; Benchmarking; Model quality; Protein engineering; Biotechnology |
| GROUP NAME | COMPUTATIONAL SYSTEMS ONCOLOGY |
| INSTITUTION | UNIVERSITY OF LAUSANNE |
| In the Scientific and Parallel Computing (SPC) Group, we develop new algorithms and methods to better understand and/or predict various phenomena in biology. We focus on multiscale modelling and computing, high-performance computing, cellular automata, lattice Boltzmann methods, multi-agent systems, and optimizing techniques and machine learning. A core activity of our group is the modelling and simulation of complex systems. In bioinformatics, the objective is to develop advanced numerical methodologies to model biological processes. | The Computational Systems Oncology lab integrates algorithmic design, numerical modelling, and molecular biology approaches to address relevant questions in cancer biology and therapeutics. We explore single and combinations of genetic and epigenetic alterations that emerge and are selected during cancer evolution, to understand cancer driving mechanisms and inform precision medicine approaches. |

KEY PAPERS IN 2017

Oiticchia E et al. Genetic and epigenetic inactivation of SESTRIN1 controls mTORC1 and response to EZH2 inhibition in follicular lymphoma. *Science Transl Med.*

Mina M et al. Conditional Selection of Genomic Alterations Dictates Cancer Evolution and Oncogenic Dependencies. *Cancer Cell*

**Bastien CHOPARD**

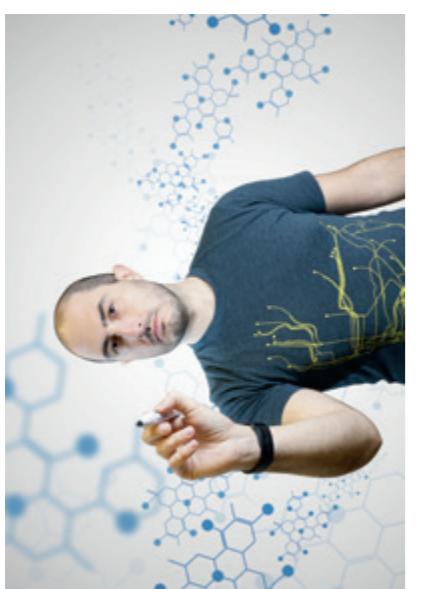
| MAIN DOMAIN OF ACTIVITY | Systems biology |
|--|--|
| SECONDARY DOMAINS OF ACTIVITY | Data mining; Drug design; Knowledge base; Machine learning; Protein structure and interaction; Software development; Homology modelling; Benchmarking; Model quality; Protein engineering; Biotechnology |
| GROUP NAME | SCIENTIFIC AND PARALLEL COMPUTING GROUP |
| INSTITUTION | UNIVERSITY OF GENEVA |
| In the Scientific and Parallel Computing (SPC) Group, we develop new algorithms and methods to better understand and/or predict various phenomena in biology. We focus on multiscale modelling and computing, high-performance computing, cellular automata, lattice Boltzmann methods, multi-agent systems, and optimizing techniques and machine learning. A core activity of our group is the modelling and simulation of complex systems. In bioinformatics, the objective is to develop advanced numerical methodologies to model biological processes. | In the Scientific and Parallel Computing (SPC) Group, we develop new algorithms and methods to better understand and/or predict various phenomena in biology. We focus on multiscale modelling and computing, high-performance computing, cellular automata, lattice Boltzmann methods, multi-agent systems, and optimizing techniques and machine learning. A core activity of our group is the modelling and simulation of complex systems. In bioinformatics, the objective is to develop advanced numerical methodologies to model biological processes. |

KEY PAPERS IN 2017

Dutta R et al. Parameter estimation of platelets deposition: Approximate Bayesian computation with high performance computing. *arXiv*

Li S et al. Model for pressure drop and flow deflection in the numerical simulation of stents in aneurysm. *Int J Numer Method Biomed Eng.*

Merzouki A et al. Influence of cell mechanics and proliferation on the buckling of simulated tissues using a vertex model. *NACO*

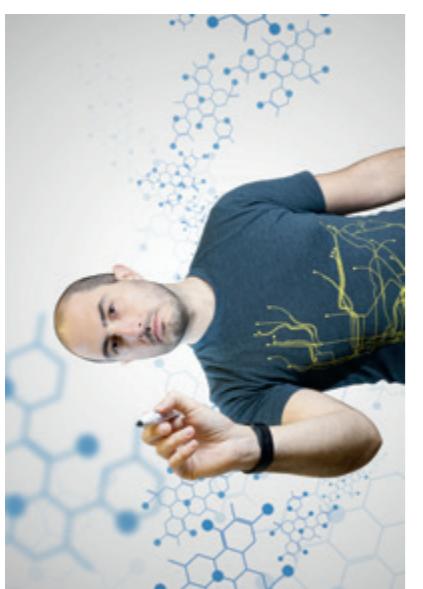
**Giovanni CIRIELLO**

| MAIN DOMAIN OF ACTIVITY | Systems biology |
|--|--|
| SECONDARY DOMAINS OF ACTIVITY | Data mining; Drug design; Knowledge base; Machine learning; Protein structure and interaction; Software development; Homology modelling; Benchmarking; Model quality; Protein engineering; Biotechnology |
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| INSTITUTION | UNIVERSITY OF LAUSANNE |
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KEY PAPERS IN 2017

Oiticchia E et al. Genetic and epigenetic inactivation of SESTRIN1 controls mTORC1 and response to EZH2 inhibition in follicular lymphoma. *Science Transl Med.*

Mina M et al. Conditional Selection of Genomic Alterations Dictates Cancer Evolution and Oncogenic Dependencies. *Cancer Cell*

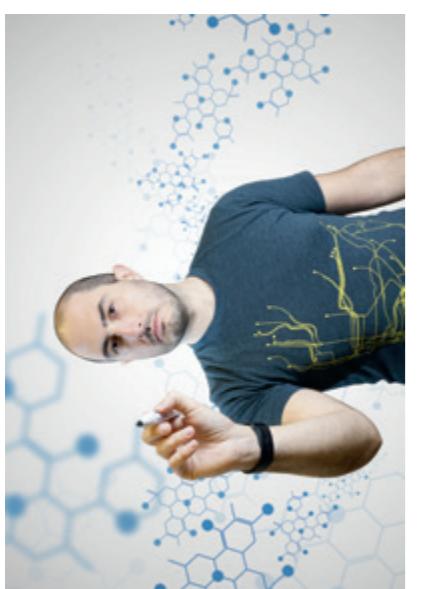
**Giovanni CIRIELLO**

| MAIN DOMAIN OF ACTIVITY | Systems biology |
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| SECONDARY DOMAINS OF ACTIVITY | Data mining; Drug design; Knowledge base; Machine learning; Protein structure and interaction; Software development; Homology modelling; Benchmarking; Model quality; Protein engineering; Biotechnology |
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| INSTITUTION | UNIVERSITY OF LAUSANNE |
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KEY PAPERS IN 2017

Oiticchia E et al. Genetic and epigenetic inactivation of SESTRIN1 controls mTORC1 and response to EZH2 inhibition in follicular lymphoma. *Science Transl Med.*

Mina M et al. Conditional Selection of Genomic Alterations Dictates Cancer Evolution and Oncogenic Dependencies. *Cancer Cell*

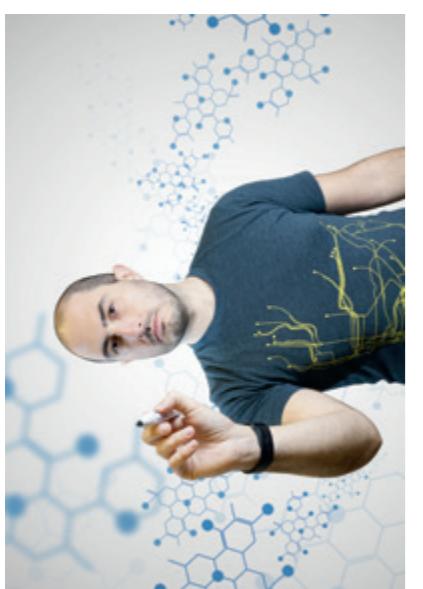
**Giovanni CIRIELLO**

| MAIN DOMAIN OF ACTIVITY | Systems biology |
|--|--|
| SECONDARY DOMAINS OF ACTIVITY | Data mining; Drug design; Knowledge base; Machine learning; Protein structure and interaction; Software development; Homology modelling; Benchmarking; Model quality; Protein engineering; Biotechnology |
| GROUP NAME | COMPUTATIONAL SYSTEMS ONCOLOGY |
| INSTITUTION | UNIVERSITY OF LAUSANNE |
| In the Scientific and Parallel Computing (SPC) Group, we develop new algorithms and methods to better understand and/or predict various phenomena in biology. We focus on multiscale modelling and computing, high-performance computing, cellular automata, lattice Boltzmann methods, multi-agent systems, and optimizing techniques and machine learning. A core activity of our group is the modelling and simulation of complex systems. In bioinformatics, the objective is to develop advanced numerical methodologies to model biological processes. | The Computational Systems Oncology lab integrates algorithmic design, numerical modelling, and molecular biology approaches to address relevant questions in cancer biology and therapeutics. We explore single and combinations of genetic and epigenetic alterations that emerge and are selected during cancer evolution, to understand cancer driving mechanisms and inform precision medicine approaches. |

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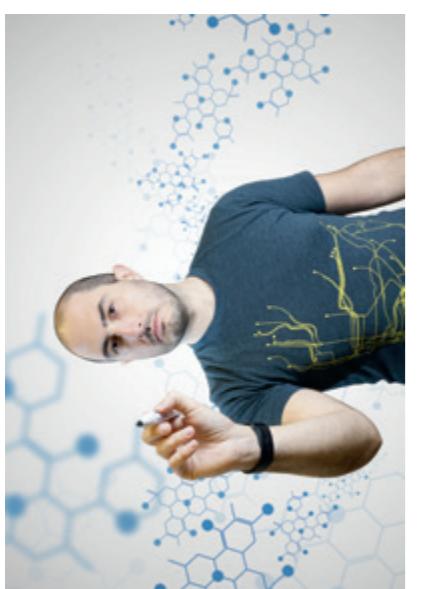
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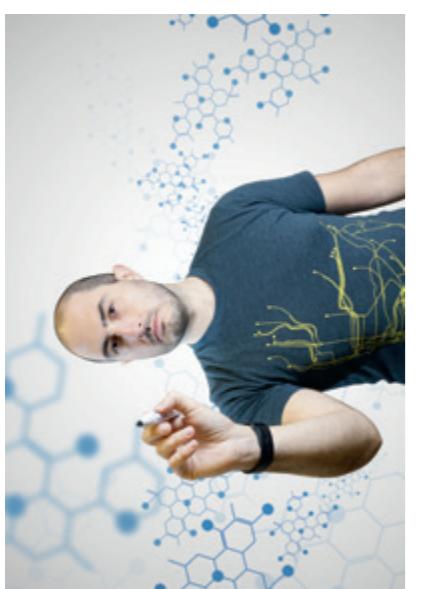
**Giovanni CIRIELLO**

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Mina M et al. Conditional Selection of Genomic Alterations Dictates Cancer Evolution and Oncogenic Dependencies. *Cancer Cell*

**Giovanni CIRIELLO**

| MAIN DOMAIN OF ACTIVITY | Systems biology |
|-------------------------|-----------------|
|-------------------------|-----------------|



Christian MAZZA

| GROUP NAME | BIO-MATHEMATICS AND COMPUTATIONAL BIOLOGY GROUP | INSTITUTION | UNIVERSITY OF FRIBOURG | CITY |
|---|---|-------------|------------------------|------|
| <hr/> | | | | |
| 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 intuitive reasoning very difficult. The idea consists in formulating mathematical units which are then implemented using computer software. We study phylotaxis by trying, for example, to explain the formation of geometrically regular patterns in plants, such as spirals in sunflowers. | | | | |
| <hr/> | | | | |



Sara MITRI

| MAIN DOMAIN OF ACTIVITY | Systems biology | SECONDARY DOMAINS OF ACTIVITY | Bioinformatics; Evolution; Genetic networks; Signaling pathways; Synthetic biology; Systems biology | DOMAINS OF APPLICATION |
|--|---------------------------------|-------------------------------|---|------------------------|
| GROUP NAME | EVOLUTIONARY MICROBIOLOGY GROUP | INSTITUTION | UNIVERSITY OF LAUSANNE | CITY |
| <hr/> | | | | |
| Our group focuses on studying social interactions in microbial communities and how they evolve. We follow an interdisciplinary approach, where we quantify and characterize interactions within small bacterial communities in the lab, and then use computational and mathematical models to explain and predict how these interactions will change over ecological and evolutionary time-scales. | | | | |
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|--------------------|--|---|--|---|
| KEY PAPERS IN 2017 | Dougoud M et al. Ultrasensitivity and sharp threshold theorems for multisite systems. <i>Journal of Physics A: Mathematical and Theoretical</i> | Rogg C. et al. Quantitative chemical biosensing by bacterial chemotaxis in microfluidic chips. <i>Environ Microbiol.</i> | Manukyan L et al. A living mesoscopic cellular automaton made of skin scales. <i>Nature</i> | Daijit Singh J et al. Bifurcation analysis of reaction diffusion systems on arbitrary surfaces. <i>Bull Math Biol.</i> |
| <hr/> | | | | |



Félix NAEFF

| MAIN DOMAIN OF ACTIVITY | Systems biology | SECONDARY DOMAINS OF ACTIVITY | Functional genomics; Single-cell biology; Transcriptomics | DOMAIN OF APPLICATION |
|---|-------------------------------------|-------------------------------|---|-----------------------|
| GROUP NAME | COMPUTATIONAL SYSTEMS BIOLOGY GROUP | INSTITUTION | EPFL | CITY |
| <hr/> | | | | |
| Systems biology is aimed at a quantitative and dynamic understanding of cellular networks by combining experimental data with theoretical and computational methodologies. Our interest lies in the regulatory and cellular networks involved in oncogenic signalling, cell-cycle regulation, and molecular oscillators. Data obtained from technologies such as microarrays, chromatin-immunoprecipitation (ChIP) and genome sequencing are brought together to discover regulatory dependencies between genes and regulatory proteins involved in cell proliferation. One thematic focus is the study of biomolecular oscillators, in particular the circadian clock. | | | | |
| <hr/> | | | | |



Igor PIVKIN

| MAIN DOMAIN OF ACTIVITY | Systems biology | SECONDARY DOMAINS OF ACTIVITY | Cell mechanobiology; Computational modelling; Particle-based methods | DOMAIN OF APPLICATION |
|---|----------------------------|-------------------------------|--|-----------------------|
| GROUP NAME | SCIENTIFIC COMPUTING GROUP | INSTITUTION | UNIVERSITÀ DELLA SVIZZERA | CITY |
| <hr/> | | | | |
| Our research interests in the Scientific Computing Group lie in the area of multiscale/multiphysics modelling and parallel large-scale simulations of biological systems. We focus on the development of new computational models and corresponding numerical methods suitable for the next generation of super computers. We are working on stochastic multiscale 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. | | | | |
| <hr/> | | | | |



Jörg STELLING

| MAIN DOMAIN OF ACTIVITY | Systems biology | SECONDARY DOMAINS OF ACTIVITY | Synthetic biology; Systems modelling | DOMAIN(S) OF APPLICATION |
|---|-------------------------------------|-------------------------------|--------------------------------------|--------------------------|
| GROUP NAME | COMPUTATIONAL SYSTEMS BIOLOGY GROUP | INSTITUTION | ETH ZURICH, D-BSSE | CITY |
| <hr/> | | | | |
| We develop and apply computational and – most recently – experimental methods to analyse and design complex cellular networks, with a focus on large-scale mechanistic approaches. The group comprises biologists, computer scientists, engineers, and mathematicians, who perform interdisciplinary research in systems and synthetic biology. We focus on developing and applying computational methods and mechanistic mathematical models to study complex cellular networks and their operating principles, and to enable their rational re-design. Applications rely on our experimental lab, which uses budding yeast as a model organism, and on collaboration. | | | | |
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|--------------------|---|---|---|---|
| KEY PAPERS IN 2017 | Young J et al. Transcription factor activity rhythms and tissue-specific chromatin interactions explain circadian gene (...). <i>Genome Res.</i> | Mauvoisin D et al. Circadian and feeding rhythms orchestrate the diurnal liver acetylome. <i>Cell Rep.</i> | Sobel JA et al. Regulatory logic of the diurnal cycle in the mouse liver. <i>PLoS Biol.</i> | Lykov K et al. Probing eukaryotic cell mechanics via mesoscopic simulations. <i>PLOS Comp Biol.</i> |
| KEY PAPERS IN 2017 | Otero-Muras I. Multi-objective design of synthetic biological circuits. <i>IFAC</i> | Lormeau C et al. Multi-objective design of synthetic biological circuits. <i>IFAC</i> | Hass H et al. Predicting ligand-dependent tumors from multi-dimensional signaling features. <i>NRJ Syst Biol Appl.</i> | Hass H et al. Predicting ligand-dependent tumors from multi-dimensional signaling features. <i>NRJ Syst Biol Appl.</i> |

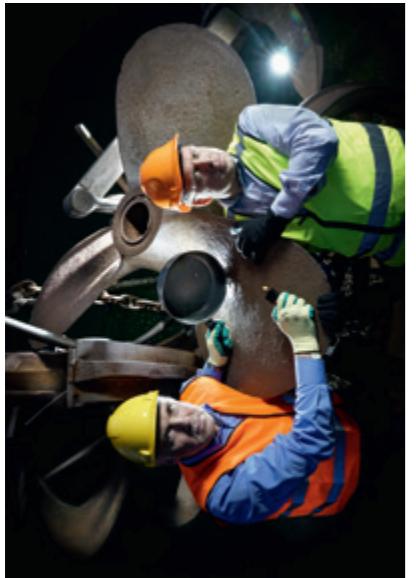


Michel MILINKOVITCH

| MAIN DOMAIN OF ACTIVITY | Systems biology | SECONDARY DOMAINS OF ACTIVITY | Comparative genomics; Evo-Devo; Evolution; Experimental biology; Machine learning; Next-generation sequencing; Phylogeny; Software development; Systems modelling; Transcriptomics; Biophysics | DOMAIN OF APPLICATION |
|---|---|-------------------------------|--|-----------------------|
| GROUP NAME | ARTIFICIAL & NATURAL EVOLUTIONARY DEVELOPMENT OF COMPLEXITY GROUP | INSTITUTION | UNIVERSITY OF GENEVA | CITY |
| <hr/> | | | | |
| 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 tissues are pertinent to EvoDevo at multiple spatial scales. We aim to understand the interactions between physical (e.g., mechanics, reaction diffusion) and biological (e.g., cell signalling, proliferation, migration) parameters, which generate patterns and shapes during development. Our multidisciplinary team includes Biologists, Computer Scientists, Engineers, and Physicists. | | | | |
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| KEY PAPERS IN 2017 | Niehuis R et al. The evolution of siderophore production as a competitive trait. <i>Evolution</i> |
| <hr/> | |

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| KEY PAPERS IN 2017 | Christel S et al. Multi-omics reveal the lifestyle of the acidophilic (...). <i>Applied Environ Microbiol.</i> |
| <hr/> | |



| MAIN DOMAIN OF ACTIVITY | | SECONDARY DOMAINS OF ACTIVITY | | DOMAINS OF APPLICATION | |
|---|-------------|-------------------------------|-------------|------------------------|-------------|
| GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION |
| Torsten SCHWEDE Thierry SENGSTAG | | | | | |
| scCORE | | | | | |

Core facilities and competence centres

Activities 2017

Bioinformatics; Data management plan; Data mining; Machine learning; Next-generation sequencing; Software development

scCORE is a centre of competence in scientific computing located at the University of Basel. Over 170 research groups in the fields of bioinformatics, computational chemistry, physics, biology, medicine, and economics use our services. Our expertise ranges from high-performance computing infrastructure, large-scale storage resources, scientific software and databases to scientific consulting and education. We help scientists with computational needs achieve their research goals. We operate the IT infrastructure behind several key SIB bioinformatics resources and, as a BioMedIT node, we actively support Swiss biomedical research.

KEY PAPERS IN 2017

Cabezón R *et al.*, SPHYNX: an accurate density-based SPH method for astrophysical applications. *Astronomy & Astrophysics*



| MAIN DOMAIN OF ACTIVITY | | SECONDARY DOMAINS OF ACTIVITY | | DOMAINS OF APPLICATION | |
|--|-------------|-------------------------------|-------------|------------------------|-------------|
| GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION |
| Karsten BORGWARDT | | | | | |
| Machine Learning and Computational Biology Lab | | | | | |

Text mining and machine learning

bioinformatics; Biomarkers; Data mining; GWAS; Machine learning; Software development; Systems biology

easyGWAS

R and Python packages for graph comparison

Arapheno

Sugiyama M *et al.*, graphkernels: R and Python packages for graph comparison.

Bioinformatics

Llinares-Lopez F *et al.*, Genomic-wide genetic heterogeneity discovery with categorical covariates.

Bioinformatics

Plant Cell

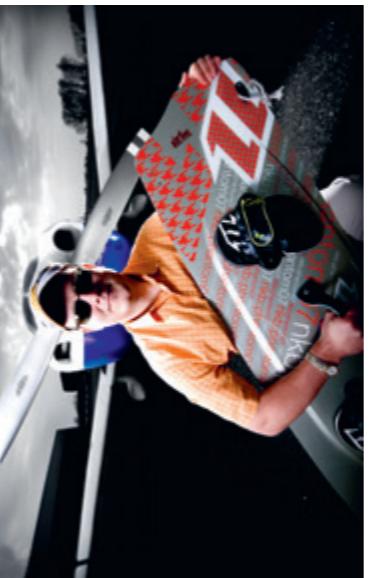


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|------------------------------|-------------|-------------------------------|-------------|------------------------|-------------|
| GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION |
| Bernd RINN | | | | | |
| Scientific IT Services (SIS) | | | | | |

Our group in Scientific IT Services (SIS) is an interdisciplinary bioinformatics and scientific IT group, which develops computational tools and services. We provide high performance computing infrastructure and services, improve and port scientific software, develop data management solutions and provide associated services. We also integrate and operate data analysis pipelines, and provide training and consulting in databases, scientific software development, high-performance and cloud computing. With our services, we support life science and biomedical research, as well as other basic and applied research.

KEY PAPERS IN 2017

Graf A *et al.*, Exome sequencing of healthy phenotypic extremes links TROY/E2 to emotional memory and PTSD. *Nat Human Behav.*



| MAIN DOMAIN OF ACTIVITY | | SECONDARY DOMAINS OF ACTIVITY | | DOMAINS OF APPLICATION | |
|-------------------------|-------------|-------------------------------|-------------|------------------------|-------------|
| GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION |
| Ioannis XENARIOS | | | | | |
| VITAL-IT | | | | | |

Genome structure; Data management plan; Data mining; Genome reconstruction; Machine learning; Mass spectrometry; Next-generation sequencing; Software development; Systems modeling; Advanced software optimization

MetalNetX

OpenFlu EMPIRES-i

SourceData platform

Grimm D *et al.*, easyGWAS: a semantic sourceData platform for curating and searching figures.

Nat methods

a novel transcriptomic signature of islets (...).

Diabetologia



| MAIN DOMAIN OF ACTIVITY | | SECONDARY DOMAINS OF ACTIVITY | | DOMAINS OF APPLICATION | |
|---|-------------|-------------------------------|-------------|------------------------|-------------|
| GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION |
| Marcel RIEDI | | | | | |
| Service and Support for Science IT (S3IT) | | | | | |

In the Service and Support for Science IT (S3IT) unit, we provide support for science in general, and for life science 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 methods and resources. Our team advises groups and projects about data management and data analysis, and cooperates to optimize their specific workflow. S3IT also takes part in national projects and cooperates with similar technology-oriented groups to ensure that its expertise is always up-to-date.

KEY PAPERS IN 2017

Graf U *et al.*, Pramel7 mediates ground-state pluripotency through proteasomal-epigenetic combined pathways. *Nat Cell Biol.*



| MAIN DOMAIN OF ACTIVITY | | SECONDARY DOMAINS OF ACTIVITY | | DOMAINS OF APPLICATION | |
|------------------------------|-------------|-------------------------------|-------------|------------------------|-------------|
| GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION | GROUP NAME | INSTITUTION |
| Daniel STEKHOWEN | | | | | |
| Clinical Bioinformatics Unit | | | | | |

Biostatistics; Drug resistance; Oncology; Precision therapy; Proteomics; Signaling pathways; Software development; Transcription; Molecular diagnostics

Metrich

Richnet MTB

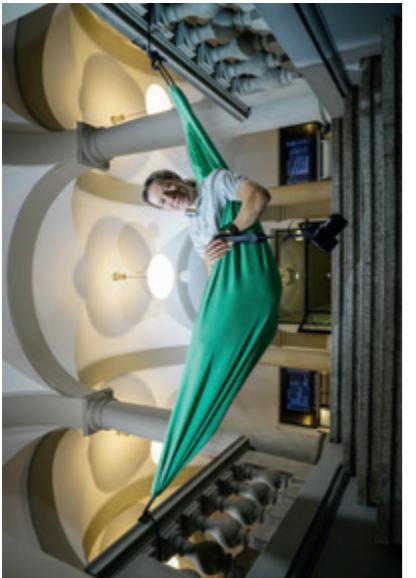
Schmid-Siegert E *et al.*, Low number of fixed somatic mutations in a long-lived oak tree. *Nat plants*

Solimena M *et al.*, Systems biology of the IM-DBA biobank from organ donors and pancreatic transplanted patients defines a novel transcriptomic signature of islets (...).

Nature Medicine



| | |
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| Fabio RINALDI <small>GROUP NAME BIOMEEXT: BIOMEDICAL INFORMATION EXTRACTION INSTITUTION UNIVERSITY OF ZURICH CITY ZURICH</small> | <p>The BioMeEXT group specializes in Information Extraction from the biomedical literature and other textual sources. Information extraction consists in automatically extracting structured information from textual documents.</p> <p>We specialize in the extraction of domain-specific entities (such as genes, drugs, diseases), and their semantic relationships (e.g. gene-disease associations). Our tools are often evaluated through participation in community-run evaluation challenges (e.g. BioCre-AT-TE-EN). We are involved in several international projects, such as BioCreative, BioNLP, and BioASAP.</p> |
|  Text mining and machine learning | <small>MAIN DOMAIN OF ACTIVITY</small> |
|  Knowledgebase; Machine learning; Text mining | <small>SECONDARY DOMAINS OF ACTIVITY</small> |
|  | <small>DOMAINS OF APPLICATION</small> |
| <small>RESOURCE</small> Bio Term Hub | <small>DOMAINS OF ACTIVITY</small> |



| Gunnar RÄTSCH | | MAIN DOMAIN OF ACTIVITY | SECONDARY DOMAINS OF ACTIVITY | DOMAIN OF APPLICATION | RESOURCES |
|------------------------|-------------|----------------------------------|---|-----------------------|---------------------------------------|
| GROUP NAME | INSTITUTION | | | | |
| BIOMEDICAL INFORMATICS | ETH ZURICH | | | | |
| ZURICH | ZURICH | | | | |
| | | Text mining and machine learning | Comparative genomics; Genome structure; Machine learning; Metagenomics; Next-generation sequencing; Oncology; Software development; Text mining; Transcriptomics; eHealth Records; Deep sequencing data | Be | RiboDiff SplAdder BRCA Exchange |



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| Carlos PEÑA | MAIN DOMAIN OF ACTIVITY | |
| | COMPUTATIONAL INTELLIGENCE FOR COMPUTATIONAL BIOLOGY (CICB) | Text mining and machine learning |
| GROUP NAME | SECONDARY DOMAINS OF ACTIVITY | |
| HEIG-VD | Biomarkers; Data mining; Machine learning; Metagenomics; Software development; Systems modelling | |
| INSTITUTION | DOMAINS OF APPLICATION | |
| YVERDON-LES-BAINS |  | |
| CITY |  | |
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| | RESOURCE | FUGE |
| | With the advent of high-throughput technologies and clinical information systems, the life sciences and clinical sciences now produce very large amounts of data (big data). Our goal is to uncover hidden patterns in these data, as well as build data-driven models as tools to discover biomarkers and assist clinicians in their decisions. Our projects encompass the fields of transcriptomics, systems biology, and clinical bioinformatics & analytics. | |

卷之三

| ROBERT WATKINSON | | Genes and genomes | |
|---|--|---|---|
| GROUP NAME | INSTITUTION | SECONDARY DOMAIN OF ACTIVITY | SECONDARY DOMAINS OF ACTIVITY |
| Proteins and proteomes | EVOLUTIONARY-FUNCTIONAL GENOMICS | Biomarkers; Biostatistics; Data mining; Functional genomics; Immunology; Peptidomics; Proteomics; Systems biology; Transcriptomics; Mass spectrometry | Biocuration; Comparative genomics; Evolution; Functional genomics; Genome structure; Phylogeny; Selection; Software development; Systems biology; Transcriptomics |
| SECONDARY DOMAIN OF ACTIVITY | INSTITUTION CITY | UNIVERSITY OF LAUSANNE LAUSANNE | DOMAIN OF APPLICATION |
|  |  |  |  |
| In the Molecular Allergology group we are aiming to unravel the molecular basis of allergic diseases, antigen presentation, immunotherapies, immunotolerance and tolerance breakage with a combination of functional genomics techniques, including proteomics, peptidomics and immunopeptidomics. The acquired large-scale molecular data are analysed 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. | The group's research is focused on elucidating interactions between gene evolution and gene function through developing computational approaches to interrogate evolutionary and functional genomics data. We are developing quantifications of gene evolutionary dynamics and functional properties to build models that link the underlying genetics to observable biological features. Our research projects focus on insects and other arthropods as their countless adaptations to exploit ecological niches mean that they are ideal for investigating how conservation or divergence of functional genomic elements give rise to the splendour of animal biology. | | |
| Natalya BALKEVITZALLER | | MOLECULAR ALLERGOLOGY | |
| GROUP NAME | INSTITUTION | SECONDARY DOMAIN OF ACTIVITY | SECONDARY DOMAINS OF ACTIVITY |
| SIAF | UNIVERSITY OF LAUSANNE LAUSANNE | | |
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| Joshua PAYNE | | COMPUTATIONAL BIOLOGY | |
| GROUP NAME | INSTITUTION | SECONDARY DOMAIN OF ACTIVITY | SECONDARY DOMAINS OF ACTIVITY |
| ETH ZURICH | ETH ZURICH | | |
|  |  | | |
| We are interested in understanding the design constraints, robustness, and evolution of gene regulatory systems, particularly at the level of transcription, using both modeling and data-driven approaches. | | | |
| Frédéric SCHÜTZ | | BIOINFORMATICS CORE FACILITY (BCF) | |
| GROUP NAME | INSTITUTION | SECONDARY DOMAIN OF ACTIVITY | SECONDARY DOMAINS OF ACTIVITY |
| UNIVERSITY OF LAUSANNE LAUSANNE | UNIVERSITY OF LAUSANNE LAUSANNE | | |
|  |  | | |
| Frédéric is joining Mauro Delorenzi as co-group leader. | | | |
| | | | SEE P.53 |



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|--|---|--|
| | | DOMAIN OF APPLICATION |
| Patrick RUCH GROUP NAME TEXT MINING INSTITUTION HES-SO - GENEVA SCHOOL OF BUSINESS ADMINISTRATION (HEG) GENEVA |  DOMAIN OF ACTIVITY Text mining and machine learning | DOMAIN OF APPLICATION Sequence exchange, proteomics, peptidomics, immunopeptidomics. The acquired large-scale molecular data are analysed 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. |
| Joshua PAYNE GROUP NAME COMPUTATIONAL BIOLOGY INSTITUTION ETH ZURICH CITY ZURICH |  DOMAIN OF ACTIVITY Data management; Data mining; Drug metabolism; Functional genomics; Gene ontology; Knowledgebase; Machine learning; Oncology; Precision therapy; Software development | MAIN DOMAIN OF ACTIVITY E: Evolution and phylogeny SECONDARY DOMAINS OF ACTIVITY Evolution; Functional genomics; Genetic networks; Microarrays; Next-generation sequencing; Systems biology; Systems modelling |
| Luciano CASCIONE GROUP NAME BIOINFORMATICS CORE UNIT INSTITUTION INSTITUTE OF ONCOLOGY RESEARCH BELINZONA CITY BELINZONA |  DOMAIN OF ACTIVITY Data mining; Drug metabolism; Functional genomics; Gene ontology; Knowledgebase; Machine learning; Oncology; Precision therapy; Software development | MAIN DOMAIN OF ACTIVITY E: Evolution and phylogeny SECONDARY DOMAINS OF ACTIVITY Evolution; Functional genomics; Genetic networks; Microarrays; Next-generation sequencing; Systems biology; Systems modelling |
| Frédéric SCHÜTZ GROUP NAME BIOINFORMATICS CORE FACILITY (BCF) INSTITUTION UNIVERSITY OF LAUSANNE CITY LAUSANNE |  DOMAIN OF APPLICATION Triage by ranking to support the curation of protein interactions. Database | MAIN DOMAIN OF ACTIVITY Text mining and machine learning |
| | | DOMAIN OF APPLICATION |
| Teodoro D'elalio GROUP NAME SCILITE INSTITUTION M.I.T. - MASSACHUSETTS INSTITUTE OF TECHNOLOGY CITY CAMBRIDGE |  DOMAIN OF APPLICATION Improving average search results for biomedical research datasets. Database | MAIN DOMAIN OF ACTIVITY Text mining and machine learning |
| Venkatesan A et al. GROUP NAME SCILITE INSTITUTION UNIVERSITY OF CALIFORNIA SAN DIEGO CITY SAN DIEGO |  DOMAIN OF APPLICATION Text mining and machine learning | MAIN DOMAIN OF ACTIVITY Text mining and machine learning |

CHAPTER 4

Focus on the Institution



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.

HEIG-VD
HAUTE ÉCOLE D'INGÉNIERIE ET DE GESTION DU CANTON DE VAUD
www.heig-vd.ch
School of Business and Engineering Vaud

UNIVERSITÉ DE GENÈVE
University of Geneva

HUG Hôpitaux Universitaires Genève

espeRare
h e g
Haute école de gestion de Genève
Geneva School of Business Administration

FMI

Friedrich Miescher Institute for Biomedical Research

University of Basel

Swiss TPH

Swiss Tropical and Public Health Institute

EPFL

ÉCOLE POLYTECHNIQUE FÉDÉRALE DE LAUSANNE
UNIVERSITÉ DE FRIBOURG
UNIVERSITÄT FREIBURG

Unil
UNIL | Université de Lausanne

LUDWIG CANCER RESEARCH

SIB'S PARTNER INSTITUTIONS

ETH

Eidgenössische Technische Hochschule Zürich
Swiss Federal Institute of Technology Zurich

University of Zurich

Agroscope

Zurich University of Applied Sciences

zhaw

Swiss Institute of Allergy and Asthma Research

SIAF

U^s
Università della Svizzera italiana

IOR
Institute of Oncology Research

LIST OF GOVERNING BODIES

The Foundation Council

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

| | |
|---|--|
| President Prof. Felix Gutzwiller Former Senator | Prof. Susan Gasser Director, Friedrich Miescher Institute for Biomedical Research |
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| Prof. Nicolas Fasel Vice-Dean for Research and Innovation, Faculty of Biology and Medicine, University of Lausanne | Dr Johannes R. Randegger Former National Councillor, Honorary President of the SIB Foundation Council |

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.

| | |
|--|--|
| Prof. Ron Appel SIB Executive Director, University of Geneva | Prof. Manuel Peitsch Chairman, Chief Scientific Officer Research at Philip Morris International |
| Ms Martine Brunschwig Graf Former National Councillor | Prof. Torsten Schwede Group Leader, SIB and University of Basel |

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.

| | |
|---|--|
| Prof. Alfonso Valencia Chairman Life Sciences Department Director, Barcelona Supercomputing Centre, Spain | Prof. Alexey I. Nesvizhskii Department of Pathology and Department of Computational Medicine & Bioinformatics, University of Michigan, Ann Arbor, USA |
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| Prof. Jean-Marc Piveteau President, Zurich University of Applied Sciences | Prof. Ron Shamir Computational Genomics Group at the Blavatnik School of Computer Science, Tel Aviv University, Israel |
| Prof. Alexandre Reymond Director, Centre for Integrative Genomics, Faculty of Biology and Medicine, University of Lausanne | Dr Laurent Duret CNRS Research Director, Laboratory of Biometry and Evolutionary Biology, Claude Bernard-Lyon 1 University, France |
| Prof. Eduardo Sanchez Dean, School of Management and Engineering Vaud (HEIG-VD), HES-SO | Prof. Melissa Haendel Director of the Ontology Development Group, Oregon Health & Science University, Portland, USA |

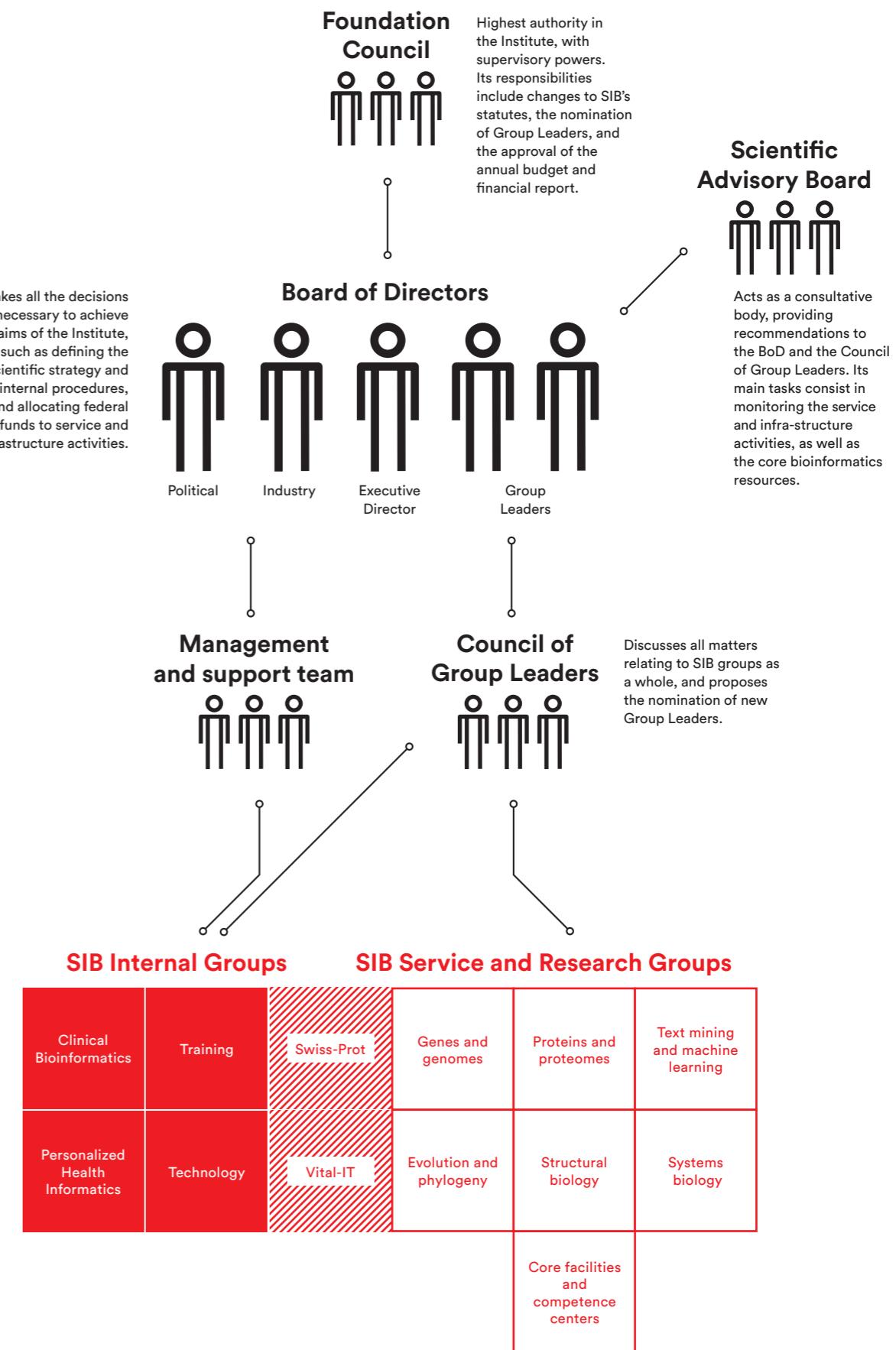
Council of Group Leaders

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

OsiriX, led by Prof. Osman Ratib, is an Affiliate Group.

Honorary Members

| | |
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| Prof. Ernest Feytmans Honorary Director | Dr Johannes R. Randegger Former National Councillor, Honorary President of the SIB Foundation Council |
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Key figures

FINANCES

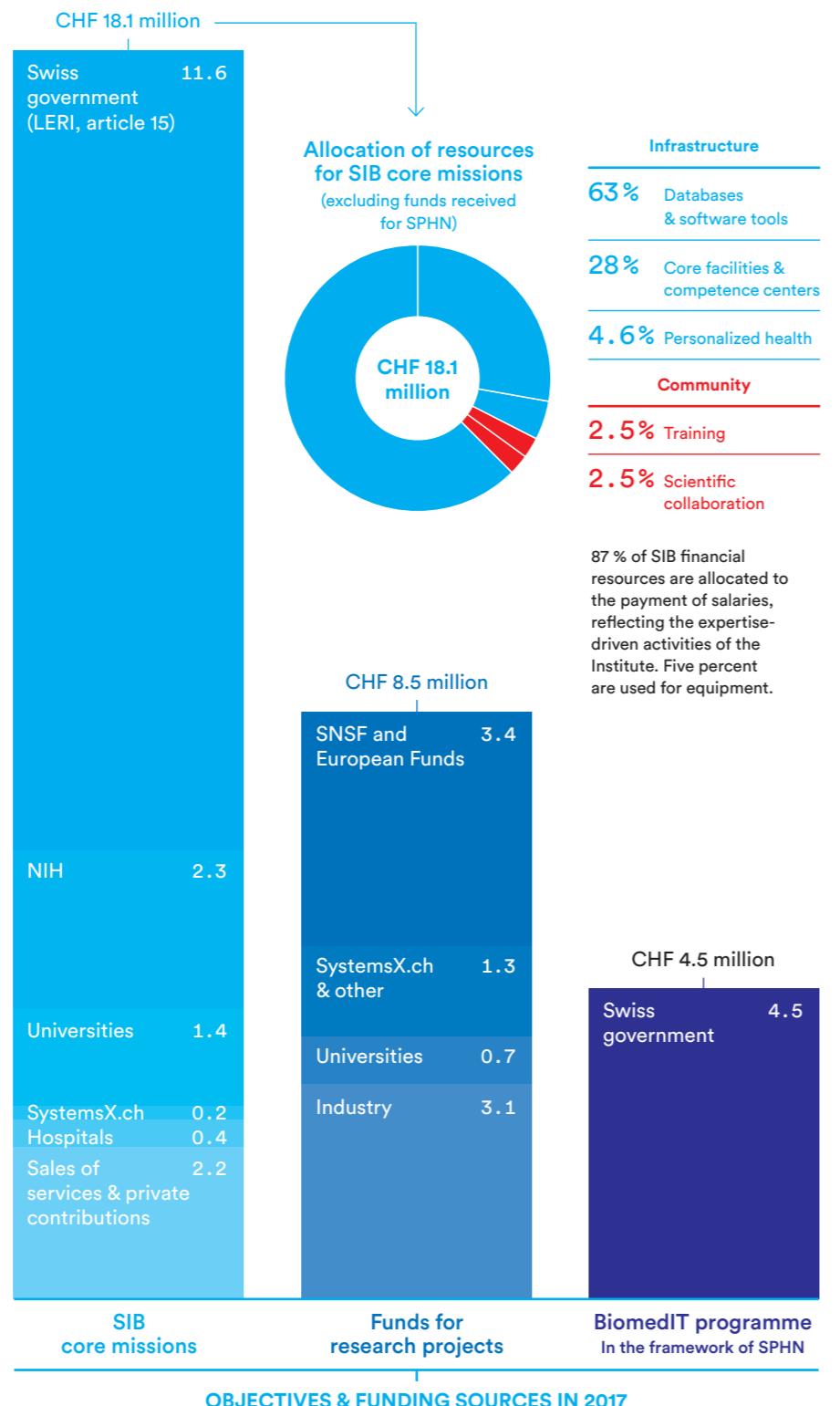
SIB funds remained stable in 2017, thanks to the continued support of its funders.

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):

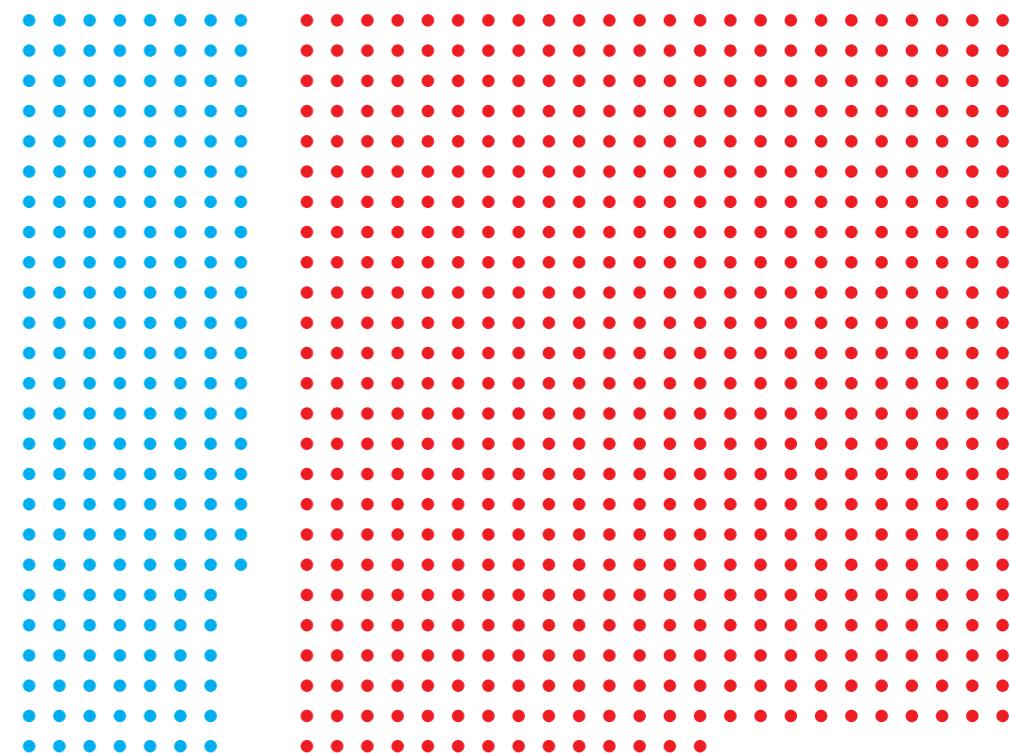
Being in charge of the BioMedIT project, CHF 18M have been allocated to SIB for the period 2017-2020. This amount goes to the core facilities of SIB and the Universities, for the work performed as data nodes in the project over these 4 years.

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.



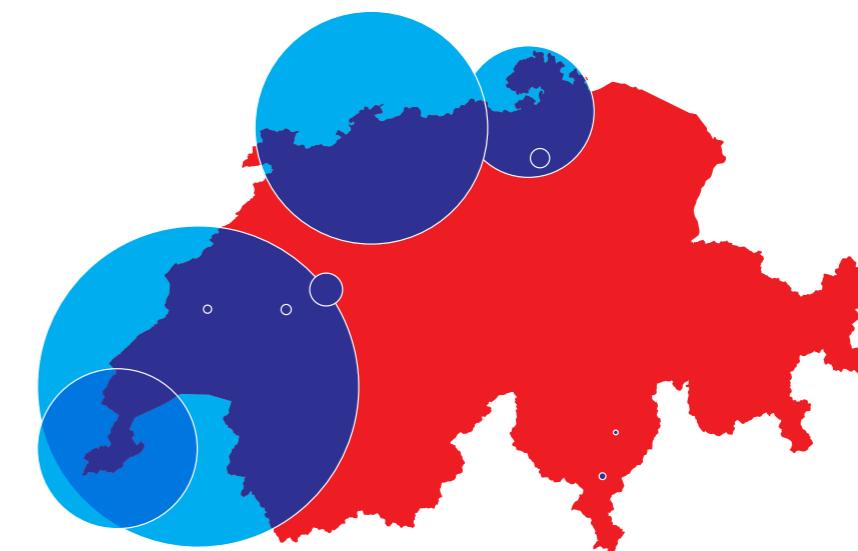
Key figures

SIB MEMBERS



Geographical distribution of SIB Members

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| Fribourg | 9 |
| Geneva | 141 |
| Lugano | 4 |
| Wädenswil | 17 |
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* As of 13 February 2018

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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

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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
Nicolas 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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SIB Swiss Institute of Bioinformatics
Quartier Sorge
Bâtiment Génopode
CH – 1015 Lausanne
T. +41 21 692 40 50

www.sib.swiss