2018 was a hallmark year for Swiss Bioinformatics. In 20 years, the Institute has grown into a fully fledged organization, recognized nationally and internationally, with 70 research and service groups scattered across the country in its most distinguished schools of higher education. Once a case of creating a Swiss Bioinformatics Institute to secure funding and build a community, two decades on, SIB has definitively shed its adolescence to dive into adulthood.

And the occasion was not overlooked: several celebratory events and projects honoured those who have shaped SIB over all these years. The most memorable event was the 20th anniversary evening in June, placed under the patronage of State Secretary Mauro Dell’Ambrogio. The celebration took place on the lakeside in Biel during SIB’s two-day conference and was attended by hundreds of SIB Members as well as eminent guests. Several outreach projects were also launched, in particular a short offbeat institutional video, an animated history of SIB, a photography book and an educational mobile game. All these initiatives, fully financed by our sponsors, are still nurturing visibility in the press and have helped to consolidate our ties with our partners.

On another note, a few major changes occurred on the Institute’s structural front— notably the co-directorship of its executive management in June. The Board of Directors also changed; Manuel Peitsch left to be succeeded by Jérôme Wojcik (undersigned), and Torsten Schwede stepped down—with Christophe Dessimoz succeeding him—to devote himself to his new role as Vice President for Research at the University of Basel. As for SIB’s pioneer core facility and competence centre Vital-IT, Mark Ibberson, one of its Senior Scientists, has been appointed as new director.

In 2019, SIB will pursue its mission by providing the life science community with state-of-the-art bioinformatics resources, expertise, services and training. Dealing with major challenges raised by the development of personalized health, the Institute will continue to lay the foundations for a data exchange infrastructure within the context of the Swiss Personalized Health Network, while lending biocuration expertise to health-related projects.

We are grateful to our partner institutions who have helped SIB thrive, as we are to the State Secretariat for Education, Research and Innovation, the Federal Assembly, the Swiss National Science Foundation and those in funding roles. SIB, however, would not have reached its 20th anniversary without the expertise, work and dedication of its employees and members, to whom we express our sincere gratitude.

And now, let us dive headfirst into the next two decades!
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CHAPTER 1

Why bioinformatics?

Computer-based approaches are indispensable to science, by allowing researchers to advance their understanding of complex systems. Find out what bioinformatics is about, and how we bring this discipline and its applications to society.
Life scientists and clinicians have always tried to assemble data and evidence to find the right answers to fundamental questions. In 2019, 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 – 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 collected in a clinical context, using computer science and dedicated data experts. This multidisciplinary field brings together scientists from a variety of backgrounds: biologists, computer scientists, mathematicians, statisticians and physicists.

Bioinformatics encompasses:

- DATABASES for storing, retrieving and organizing information to maximize the value of biological data;
- SOFTWARE TOOLS for modelling, visualizing, analysing, interpreting and comparing biological data;
- COMPUTING AND STORAGE INFRASTRUCTURE to process and safeguard large amounts of data;
- ANALYSIS of complex biological datasets or systems in the context of particular research projects;
- RESEARCH in a wide variety of biological fields using computer- and data science and leading to applications in diverse areas, from agriculture to precision medicine. (See p. 46)
Converting biological questions...

...into answers with various applications

Dedicated multidisciplinary experts

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

Life sciences and health actors

SIB
Swiss Institute of Bioinformatics

Data management planning
Secure computing and storage infrastructure
Databases and software tools
Cutting-edge analyses and research

Tailoring treatment to cancer patients
Understanding how chameleons change colour
Predicting the spread of bird flu

Hospitals and clinics
Research institutes
Private sector
International institutions

Basic research
Medicine
Environmental sciences
Agriculture

Why Bioinformatics - In our Institutions - Activities
Bringing bioinformatics to society

Bioinformatics is increasingly tied to health, ethics, and societal issues. Through its outreach activities, SIB informs the public about the potential applications of the discipline.

“In now see how absurdly large the human genome truly is!”

“Using the scientific tool, I was able to unleash my creativity and design a molecule with a potential use against cancer”

Feedback from high school students who attended the Drug Design workshop

In 2019, meet the team at:

MYSTÈRES DE L’UNIL
University of Lausanne,
33-36 May
The University’s open days will once again feature bioinformatics activities!
wsp.unil.ch/mysteres

UNCT SCIENTIFICA
Zurich Science Days,
ETH Zurich,
31 August – 1 September
What is the usefulness of bioinformatics in the identification of variants in oncology?
scientifica.ch

EXPANDING YOUR HORIZONS
University of Geneva,
16 November
Biennial event to encourage girls aged 11-14 to study science, technology, engineering and mathematics.
elargisteshorizons.ch/en

In 2018, the SIB Outreach Team met you at:

PLANÈTE SANTÉ LIVE
…the largest health fair in Switzerland: hundreds of visitors learned about the latest advances in clinical bioinformatics in the field of personalized health.
planetesante.ch/valon

NUIT DE LA SCIENCE
…two days and nights to discover the role of bioinformatics in drug design, personalized health and metagenomics.
ville-ge.ch/lanuitdelascience

SIB BIOHACKATHON
…at the second edition of the Open Geneva Innovation Festival, children aged 9-14 used the SCRATCH programming language to tackle biological problems.
opengeneva.org

About

2,400 participants in our activities each year including
1,600 students (12-18 years old)
Take a look back on the 20th anniversary celebrations of the SIB Swiss Institute of Bioinformatics and discover its key themes for the future. But first, take a backstage tour of the Institute’s organization, partners, people and finances.
**SIB in brief**

**OUR MISSION AND OBJECTIVES**

The mission of our Institute is to lead and coordinate the field of bioinformatics in Switzerland, in order to advance biological and medical research and to enhance health.

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 worldwide a large portfolio of databases and software tools.

**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.

---

**Infrastructure**

70 research and service groups

810 members, including

190 employees

20 institutional partners across Switzerland

13 core facilities and competence centres

Over 150 databases and software tools developed by our members and accessible via the ExPASy web portal

Over 2,450 peer-reviewed articles published since SIB’s creation in 1998

**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, etc.

Bottom

Collaboration is at the heart of science. And it all starts with a strong community.
Organization and governance

Federating and leading such a pervasive domain as bioinformatics, even across a modestly sized country such as Switzerland, requires a unique structure with multiple bridges across institutions.

As a non-profit foundation unifying bioinformatics across Switzerland and with 20 partner institutions (see opposite), SIB has a robust governance structure 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 entirely composed of SIB Employees, include: Clinical Bioinformatics (see p. 38), Core-IT (see p. 34), Digital Humanities+ (see p. 45), Personalized Health Informatics (see p. 45), Swiss-Prot (see p. 56), Training (see p. 42) and Vital-IT (see p. 76).

Finally, SIB Affiliated Groups include academic groups from partner institutions, including those maintaining and developing an SIB-supported infrastructure, such as an SIB Resource or an SIB Core facility – they can thus include SIB Employees as well.

A dual management at the head of SIB

As of 22 June 2018, Christine Durinx, previously Associate Director, and Ron Appel have been leading SIB together as Joint Executive Directors. Christine is also a member of the Executive Board of the BioMedIT project, and co-chair of the ELIXIR Data Platform.

Becoming an SIB Group Leader

The main criterion for groups to join SIB is that they are producing new bioinformatics methods or software, or act as a core facility providing computing infrastructure or bioinformatics support. In addition, their Group Leader must have a professorial appointment in one of SIB’s 20 partner institutions, or be a senior scientist leading her/his own group.

“The SIB Swiss Institute of Bioinformatics has considerable influence in Europe and successfully operates a national bioinformatics community with over 20 partner institutes.”

Evaluation of SIB (Swiss Node of ELIXIR) by ELIXIR’s Scientific Advisory Board, March 2019
LIST OF GOVERNING BODIES

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 SIB Executive Directors. Members of the BoD are appointed for a renewable five-year period.

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.

The Council of Group Leaders
The Council consists of the Group Leaders and the SIB Executive Directors.

Foundation Council
Highest authority in the Institute, with supervisory powers. Its responsibilities include changes to SIB’s statutes, nomination of Group Leaders, and approval of the annual budget and financial report.

Board of Directors
Takes the decisions necessary to achieve the aims of the Institute, such as defining the strategic and internal procedures, and allocating federal funds to service and infrastructure activities.

Scientific Advisory Board
Acts as a consultative body, providing recommendations to the Board of Directors and the Council of Group Leaders. Its main tasks consist in monitoring service and infrastructure activities, such as SIB Resources.

Management and support teams
External members from the political and industry sector

Council of Group Leaders
Defines and implements the Institute’s strategic and scientific goals, and ensures the organization’s representation at the national and international level. Support teams include financial, legal, human resources and communication specialists dedicated to institutional and activity matters.

SIB Internal Groups
Staffed by SIB employees, they focus on SIB’s core databases or infrastructure such as the Swiss-Prot, Clinical Bioinformatics, Vital-IT, Care-IT or Personalized Health Informatics Group.

SIB Affiliated Groups
Spread across Switzerland’s leading institutions, these groups are headed by SIB Members. Some have SIB employees as a support to their infrastructure or service activities.
SIB funds remained stable in 2018, thanks to the continued support of its funders.

**Key figures**

**Allocation to SIB’s core missions**

- Infrastructure: CHF 7.6 million
  - Swiss Government: CHF 6.6 million
  - Sales of services / private contributions: CHF 4.2 million
- Databases & software tools: CHF 1.7 million
  - National Institutes of Health (NIH): CHF 0.2 million
  - Swiss Universities: CHF 0.5 million
- Core facilities: CHF 7.4 million
  - Swiss Universities: CHF 1.7 million
  - Swiss Government: CHF 0.6 million
- Personalized health: CHF 2.6 million
  - Swiss Government: CHF 0.2 million
  - Sales of services / private contributions: CHF 0.5 million
  - European funds: CHF 0.8 million

**Detail of funding sources**

- 50% Swiss Government – SERI: CHF 16.2 million
- 27% Sales of services / private contributions: CHF 9.0 million
- 7% National Institutes of Health (NIH): CHF 2.2 million
- 6% Swiss Universities: CHF 2.1 million
- 5% European Funds: CHF 1.6 million
- 4% Swiss National Fund (SNF) / Innosuisse: CHF 1.3 million
- 1% Swiss Hospitals: CHF 0.4 million

**Total: CHF 32.7 million**

**Allocation by activities**

- 78% Infrastructure
- 11% Community
- 11% Management & support teams

**71%** of financial resources are allocated to the payment of salaries, reflecting the expertise-driven activities of the Institute.

**SIB Members**

- SIB has 810 Members including 190 Employees from 23 different nationalities

**They are based in:**

- Basel: 182
- Bellinzona: 10
- Bern: 31
- Davos: 2
- Fribourg: 11
- Geneva: 145
- Lausanne: 286
- Lugano: 4
- Wädenswil: 19
- Yverdon: 10
- Zurich: 110
Perspectives
Where will SIB be adding value in the years to come? Here are some of the Institute’s major development themes for the near future, resulting from the 2018 strategic roadmap.

Securing Health Data Infrastructure
In the context of the Swiss Personalized Health Network (SPHN), SIB plays a central role as Data Coordination Centre and was mandated by the Swiss Confederation to implement a secure network, enabling data to be shared for research in personalized health. The first steps in this secure data architecture are being implemented through the BioMedIT project, thanks to the joint work of SIB Employees and affiliated core facilities known as BioMedIT nodes. The nodes provide access to a remote data analysis environment, thereby enabling scientists to analyse data securely.

Safeguarding Data Resources
For SIB Resources with sensitive data, penetration testing is conducted on a regular basis and guidelines on how to resolve security vulnerabilities have been established. Further work is underway to ensure security policies are adapted to the sensitivity of databases and extended to SIB Resources outside the health field. This is all part of SIB’s mission to add value to bioinformatics resources dedicated to the scientific community.

A Trusted Partner for Health Actors
SIB has become a partner of choice for health professionals, both for its strategic position between research and clinical needs, and for its expertise related to data security.

Biocuration, a Key Building Block of Personalized Health
In a tumour sample, up to several hundred variants can be detected. But how can one filter out those that are particularly pathogenic, or code for an accrued sensitivity — or resistance — to a specific treatment?

With the advent of molecular medicine, clinicians from around the world are turning to carefully annotated knowledgebases to assess the pathogenicity of a specific variant, guide their diagnosis, or pinpoint associated drugs.

In this context, the role of biocurators — these expert scientists who collect, annotate and validate publicly available information from the growing biomedical literature and enter it into knowledgebases — will take on increasing importance.

Biocurators will form an essential building block of pluridisciplinary personalized medicine programs in Switzerland and internationally.

Leveraging biocuration expertise for health projects
Be it at the genomic level or increasingly in other omics areas, SIB is leveraging its biocuration expertise in the context of personalized health.

A Curated Platform to Harmonize Variant Interpretation
A concrete example is the SVIP project that has recently started under the co-leadership of the SIB Clinical Bioinformatics Group. Its purpose is to harmonize the clinical interpretation of variants across Swiss hospitals.

Initially targeting variants associated with cancer, the platform will combine clinical data from partner hospitals with publicly available information from international projects (e.g. ClinVar, CIViC) and published literature, sourced by expert biocurators from SIB Groups such as SwissProt (See p. 54), neXtProt (See p. 40), CALIPHO (See p. 51), and BASE (See p. 56).

FROM GENOMIC VARIANTS TO PROTEINS — AND BEYOND
The human protein knowledgebase neXtProt has also been working on leveraging its information towards the clinical world: initially designed to help researchers find out what human proteins do in our bodies (see above), the Group developing the resource, CALIPHO (See p. 51), has been generating a corpus of annotations on the impact of protein sequence variations in cancers and genetic diseases.

These so-called ‘phenotypic’ effects encompass changes in the activity of the protein, its stability and its interactions with other proteins or drugs.

While most information currently used by clinicians is at the genomic level, the rise of integration of other ‘omics’ such as proteomics, lipidomics or transcriptomics means that expert-curator resources developed by SIB Groups will be crucial to advance our understanding of diseases, improve biomarker discovery or support the development of personalized medicine programmes.

Why Bioinformatics - Institution - Activities
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Increasing the impact of our bioinformatics resources

What does it imply to increase the impact of a bioinformatics resource when its scientific relevance is already established? Since February 2018, a dedicated SIB Team has been working hand-in-hand with resource developers to create a best-practice toolkit that will help them to grow their user base and share their know-how with the community.

To conduct their daily work, life scientists and clinicians have to wade through an ever-thickening swamp of tools, software and databases. At the same time, the next generation of researchers – both tech-savvy and ardent app-consumers – is raising the bar of expectations in terms of resource usability. “Scientific excellence is no longer the sole criterion for a resource to be competitive in the long term,” says Chiara Gabella, SIB Scientific Coordinator and User eXperience (UX) Architect.

To keep pace with this highly competitive context, SIB – whose mission includes providing the life science community with state-of-the-art resources, as well as ensuring their long-term sustainability – launched an initiative to further increase the impact of its resources. Indeed, beyond an established reputation for scientific excellence, much can still be done to ensure that the user community is aware of these tools, and that they are used to their full potential while generating a positive experience.

Led by Chiara Gabella and Séverine Duvaud, SIB Software Developer and UX Designer, the initiative aims to develop a best-practice toolkit available to SIB Resources’ teams, as well as to the wider community of SIB Developers.

In addition to improving resource design by taking into account the experience of the end user, this toolkit will also include instruments for the in-depth interpretation of analytics used to monitor and improve a resource. Among these: analysing the user base, finding out how much time is necessary for a user to find relevant information, or what is the most useful type of information on the platform. Cross-referencing between resources involved in the same analytical workflow will also be part of this strategic review. “Ultimately, such a toolkit will help target development efforts and increase the return rate of users,” says Duvaud.

To achieve its goal, the team is relying on a wide set of interactive approaches that have been successful in similar contexts, such as workshops and live user testing at conferences. It also released several community-based initiatives, including an internal competency network and a specialized blog dedicated to resource developers.

What is User eXperience (UX) design in a life science context?

UX encompasses the end user’s interaction with a product, tool, or system. It involves deeply understanding users through research, organizing information, visual design and more, all with the goal of meeting user needs. This is especially important in the life sciences, where users are scientists in a complex environment and where more usable, efficient tools can accelerate innovation.

Source: Pistoia Alliance
SIB’s 20th anniversary in images

Movies, events, a mobile game: this special year for bioinformatics was well celebrated! And these initiatives still continue to generate visibility and public interest for the field and its experts.

A BRIEF HISTORY OF SIB
An animated movie to retrace the steps of the Institute and its evolution over time.
gee.gl/PMAlLU

SIB DAYS SPECIAL EVENING
Hundreds of SIB Members and eminent guests gathered on the shores of Lake Biel, Switzerland, for our biennial conference. The special evening was placed under the patronage of the SERI (State Secretariat for Education, Research and Innovation) State Secretary.

SIB, THE MOVIE – SWISS BIOINFORMATICS IN ACTION
This short offbeat clip has enthused a broad audience, from scientists to the general public: if you haven’t already, meet Sophie the bioinformatician, Jürgen the software developer and Fred the biocurator.
gee.gl/RaekA2

SEQUENCING SIB’S BIRTHDAY CAKE
Hundreds of visitors at the Open Days of the University of Lausanne and at the Nuit de la Science used metagenomic approaches to analyse the ingredients of the SIB birthday cake, designed for the occasion.

GENOME JUMPER, A MOBILE GAME TO EXPLORE HUMAN DIVERSITY
Why is it some people don’t have fingerprints? Or some are more at risk of gluten intolerance? And what does bioinformatics have to do with it? Over 2,500 downloads already for this educational game:
genome-jumper.sib.swiss

SCIENCE FICTION BOOK
Combining the portraits of our Group Leaders and an overview of significant advances made possible by bioinformatics, this book is an aesthetic and fantasy-like journey to the land of biological data science.

deep into the world of data

coding the secrets of life

discovering the future of medicine
CHAPTER 3
Activities 2018

Discover our latest developments and achievements on the bioinformatics infrastructure and community fronts.

Infrastructure
I. DATABASES AND SOFTWARE TOOLS
...a large range of bioinformatics resources
II. CORE FACILITIES AND COMPETENCE CENTRES
...easily accessible, in-depth expertise and support in bioinformatics
III. PERSONALIZED HEALTH
...dedicated know-how, resources and secure data infrastructure for researchers and clinicians

Community
IV. TRAINING IN BIOINFORMATICS
...a diverse portfolio of courses and workshops for life scientists and clinicians
V. SCIENTIFIC COLLABORATION
...a unique ecosystem for bioinformatics groups from Swiss universities and research institutes

Discover our latest developments and achievements on the bioinformatics infrastructure and community fronts.
A complete bioinformatics infrastructure

From research tools and computing power to data science expertise and dedicated infrastructure for personalized health, SIB’s portfolio addresses the bioinformatics needs of life scientists and clinicians, in academia or the private sector.

I. DATABASES AND SOFTWARE TOOLS

Among the 150 databases and software tools developed and maintained by SIB Groups, the so-called SIB Resources are deemed of particular importance to the life-science community, and thus benefit from the Institute’s specific support.

SWISS-MODEL celebrates its 25th anniversary

Once the first automated homology modelling service on the internet, SWISS-MODEL today processes over one million model requests and generates about 2,000 literature citations each year. It is one of the most widely used structure modelling servers worldwide. Although it is 25 years old, it has lost nothing of its original philosophy: to make protein modelling accessible to non-experts, thanks to an intuitive and interactive interface. It celebrated its anniversary with an international symposium in Basel, held in October 2018, which explored the state of the art of computational structural biology and its latest exciting developments, with new players such as Google DeepMind entering the field.

Sustainability of knowledgesbases

In 2018, SIB pursued its efforts to find solutions to the long-term sustainability of knowledgebases with an F1000 Research blog on the study led by SIB and supported by ELIXIR on that topic. SIB’s Chiara Gabella, first author on the study, and reviewer Helen Berman, Director Emerita and Associate Director of the RCSB Protein Data Bank (Rutgers, The State University of New Jersey) discussed funding models, open access data and the future of biocuration.

Life scientists often rely on dozens of databases or software tools to reconstruct DNA sequences, interpret the function of genes, or predict the 3D structure of proteins. It is therefore crucial that key resources are maintained in the long term while taking into account the fast-changing needs of researchers. It is SIB’s mission to support such goals, and it has set up a process to identify and support such key resources.

Every four years, the Institute’s external Scientific Advisory Board (SAB) is in charge of providing recommendations on the portfolio of SIB-supported Resources. By taking into account competitive criteria – such as the impact and scientific return on investment of candidate resources – together with SIB’s global portfolio management and strategic orientation, the SAB identifies best-in-class resources, and provides guidelines for their continuous development.

Based on these recommendations, the Board of Directors decides the allocation of funding, and SIB Management then assists the groups developing the resources in implementing their action plan and exchanging best practices. For instance, since 2018 a dedicated team has assisted developers in increasing the visibility and impact of their resource (SEE P. 24).

Swiss-Prot: a world flagship of biocuration

Thirty-three years after its creation and with some 50 employees, SIB’s Swiss-Prot Group continues to be internationally recognized for its expertise in biocuration, ranging from annotating protein sequences, their functions, and their sequence variants (e.g. UniProtKB/Swiss-Prot, ViralZone, HAMAP, PROSITE), to metabolites and metabolic reactions – also featuring prominently in many human diseases – with SwissLipids.

ExPASy, SIB’s bioinformatics resource portal, turned 25 years old in 2018: to mark the occasion, SIB initiated its in-depth redesign to better meet the community’s recent needs and add value to the scientific resources it annotates and connects. The launch of ExPASy 3.0 is expected in 2019.

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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**: Expert-curated part of UniProt, the most widely used protein information resource in the world, with over six million page views per month. Recognized as an ELIXIR Core Data Resource.

**NEW IN 2018**: Includes the resource Rhea, a comprehensive expert-curated knowledgebase of biochemical reactions, to annotate and represent enzyme-catalyzed reactions.

---

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**: Over 25,900,000 protein interactions at the highest confidence level. Recognized as an ELIXIR Core Data Resource.

**NEW IN 2018**: Deemed overall best-performing network resource according to a Californian study (Huang J-K et al. Cell Systems 2018)

**COMING UP**: Next major release (version 11.0), doubling the organism coverage. New features for improved detection of functional enrichments in user-provided inputs.

---

Glycomics@ExPaSy

**Glycoproteomics 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 glycoproteomics data analysis and interpretation.

**NEW IN 2018**: Release of UniLectin, a collaborative platform integrating information on non-enzymatic carbohydrate-binding proteins, aka lectins.

---

SwissLipids

**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 lipodermic data with biological knowledge and models.

**HIGHLIGHT**: Unique portal for glycoproteomics data analysis and interpretation.

**NEW IN 2018**: Release of UniLectin, a collaborative platform integrating information on non-enzymatic carbohydrate-binding proteins, aka lectins.

---

SwissLipids

**SwissLipids**

**TYPE**: Knowledgebase with manual expert curation

**DESCRIPTION**: Human protein knowledgebase

**HIGHLIGHT**: 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. Tools specifically designed for the proteomics community.

**NEW IN 2018**: The SwissProt Cancer Variants portal contains over 6,300 annotated observations at the molecular and/or cellular level for BRCA1 variants.

---

SwissDrugDesign

**Gene expression expertise**

**TYPE**: Knowledgebase with manual expert curation and software tool

**DESCRIPTION**: Gene expression data (including all types of transcriptomes), allowing retrieval and comparison of expression patterns between animals, human, model organisms and diverse species of evolutionary or agronomical relevance.

**HIGHLIGHT**: Only resource to provide homologous gene expression between species.

**NEW IN 2018**: Publication of the R package BgeeDB for data access and TopAnal analyses.

---

V-Pipe

**SwissRegulon Portal**

**Tools and data for regulatory genomics**

**TYPE**: Software tools and knowledgebases

**DESCRIPTION**: Web resource for regulatory genomics, including genome-wide annotations of regulatory sites and motifs, and the webserver ISMARA for automated inference of regulatory networks and CRUNCH for automated analysis of ChIP-seq data.

**HIGHLIGHT**: User can upload raw microscopy, RNA-seq or ChIP-seq data to automatically infer the core regulatory networks acting in the system of interest.
Switzerland, offering high-performance computing, SIB Internal Groups (Core-IT and Vital-IT) and two SIB Affiliated Groups (BCF and sciCORE), which are respectively either entirely or partly composed of SIB Employees.

Sciences to medicine. Among them, SIB is strongly involved in four: two SIB Affiliated Groups (BCF and sciCORE), which are respectively either entirely or partly composed of SIB Employees.

15 years down the line, over 40 scientists and experts in data analysis, software development, database management, information technology or statistics, backed up by SIB’s powerful computational power and storage, are involved in a wide diversity of research projects on the national and international scene. Recognized for its ability to deliver on a wide variety of projects, the team’s activities have long revolved around three main areas: HPC and bioinformatics services for SIB’s partner institutions; developments for SIB’s activities in the domain of biomedical data; and research collaborations. Finally, the Vital-IT Group also manages over 1,000 software packages, which are highly used by researchers in its partner institutions.

Mark Ibberson, a new director from within

Mark, one of the group’s senior scientists, was appointed as Vital-IT’s director in November. Mark’s expertise in the private sector as well as his recognized experience as co-PI for several international research projects – including the pan-European IMI projects mentioned above – will be invaluable assets for the group. After a PhD in medical genetics at the University of London, Mark has spent 10 years as bioinformatician in a major pharmaceutical company before joining SIB in 2010.
A national network to support the data science needs of life scientists

Thirteen core facilities and competence centres are distributed in all the major Swiss academic institutions, and managed by SIB Group Leaders.

Service and Support for Science IT (S3IT) facility

- **GL:** Marcel Riedi
- **I:** University of Zurich
- **KS:** science IT infrastructure; consulting; training; project collaborations
- **L:** Zurich

**SEE P. 72**

**Scientific Information Services (SIS)**

- **GL:** Bernd Rinn
- **I:** ETH Zurich
- **KS:** research data management; scientific software engineering; data management plan (DMP); research data pipelines; data co-analysis
- **L:** Zurich, Basel

**SEE P. 72**

**NEXUS**

**Personalized Health Technologies**

- **GL:** Daniel Stekhoven
- **I:** ETH Zurich
- **KS:** bioinformatics/biostatistical data analysis; translational research support; clinical decision support; software development
- **L:** Zurich

**SEE P. 75**

**FGCZ Genome Informatics (FGCZ-GI)**

- **GL:** Hubert Rehrauer
- **I:** ETH Zurich
- **KS:** next-generation data analysis; analysis of long-reads; bioinformatics training; clinical genomics
- **L:** Zurich

**SEE P. 73**

**Centre for Scientific Computing (sciCORE)**

- **GL:** Torsten Schwede and Thierry Sengstag
- **I:** SIB, University of Basel
- **KS:** scientific services hosting; bioinformatics analysis and training; data management support
- **L:** Basel

**SEE P. 72**

**Interfaculty Bioinformatics Unit (IBU)**

- **GL:** Raimy Bruggmann
- **I:** University of Bern
- **KS:** next-generation data analyses; biostatistical analyses; training
- **L:** Bern

**SEE P. 70**

**Bioinformatics Core Unit (BCU)**

- **GL:** Luciano Cascione
- **I:** Institute of Oncology Research
- **KS:** biostatistical analyses; data mining; training
- **L:** Bellinzona

**SEE P. 70**

**Core-IT**

- **HEAD:** Heinz Stockinger
- **I:** SIB
- **KS:** secure HPC infrastructure; data protection (incl. training and documentation); quality assurance; IT services and support
- **L:** Lausanne, Geneva

**Vital-IT**

- **GL:** Mark Ibberson
- **I:** SIB
- **KS:** bioinformatics/biostatistical analyses; data integration and management; software and web development
- **L:** Lausanne

**Bioinformatics Core Facility (BCF)**

- **GL:** Mauro Delorenzi and Frédéric Schütz
- **I:** SIB, University of Lausanne
- **KS:** bioinformatics/biostatistical analyses; training and consulting; single-cell applications; profiling and sub-typing of tumours
- **L:** Lausanne

**Bioinformatics Unravelling Group (BUGFri)**

- **GL:** Laurent Falquet
- **I:** University of Fribourg
- **KS:** project planning and grant writing; software testing and development; data management plan (DMP) and deposition in OpenData repositories; data analysis; training
- **L:** Fribourg

**FMI Computational Biology Group (FMICBG)**

- **GL:** Michael Stadler
- **I:** Friedrich Miescher Institute
- **KS:** high-throughput data analysis, from experiment design to result interpretation; training; bioinformatics helpdesk; tool development
- **L:** Basel

**DBM Bioinformatics Core Facility**

- **GL:** Robert Ivanek
- **I:** University of Basel, University Hospital Basel
- **KS:** consultancy on experimental design of genomics experiments; data analysis and visualization with focus on large-scale genomics and proteomics; consulting in statistics and programming in R
- **L:** Basel

**Centre for Scientific Computing (sciCORE)**

- **GL:** Torsten Schwede and Thierry Sengstag
- **I:** SIB, University of Basel
- **KS:** scientific services hosting; bioinformatics analysis and training; data management support
- **L:** Basel

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**SEE P. 70**
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 for medical care

- Consultation
- Report
- Sample
- Interpretation
- Data generation
- Applications for the clinician include:
  - Diagnose
  - Anticipate risk
  - Adapt therapy
  - Monitor patient
  - Data integration
  - Data management
  - Software development
  - Consultation

Clinical bioinformatics

**SIB Profile 2019**

**CLINICAL BIOINFORMATICS GROUP**

**NAME**

Valérie Barbié

**CLINICAL BIOINFORMATICS GROUP**

**NAME**

Valérie Barbié

**CLINICAL BIOINFORMATICS GROUP**

**INTERVENTIONS**

- 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. Notably, it:
- Establishes consensus and good practices for diagnostic omics analyses across Swiss hospitals;
- Partners with Swiss hospitals to develop, implement and sustain state-of-the-art diagnostic tools;
- Provides clinical bioinformatics training across Switzerland;
- Facilitates interactions between SIB’s research groups and the medical realm for clinical research projects.

**THE SOMATIC MUTATION CALLING WORKING GROUP**

has benchmarked Swiss hospitals’ next-generation sequencing analysis and interpretation practices in cancer diagnosis;

**THE MICRONE TYPING AND CHARACTERIZATION WORKING GROUP**

has benchmarked clinical next-generation sequencing practices in the diagnosis of infectious diseases, both in virology and bacteriology.

The results of these two benchmarking studies will be published in 2019.

**2018 HIGHLIGHTS**

**WORKING GROUPS**

With each hospital using different bioinformatics methods to analyse their molecular data, such as DNA from tumour tissues, results are likely to vary significantly from one to another. Several benchmarking projects led by the Clinical Bioinformatics Group thus aim to identify the most appropriate methods and harmonize practices among hospitals, leading to faster diagnosis and better targeting of treatments.

**THE SOMATIC MUTATION CALLING WORKING GROUP**

has benchmarked Swiss hospitals’ next-generation sequencing analysis and interpretation practices in cancer diagnosis;

**THE MICRONE TYPING AND CHARACTERIZATION WORKING GROUP**

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**COLLABORATIVE CLINICAL PLATFORMS**

The Group is co-leading the SWISS VARIANT INTERPRETATION PLATFORM project (SVIP), funded by SfPH and BionmedIT, which aims to provide a central resource for clinicians and researchers to annotate and interpret clinically verified variants in cancer. The tool will be available in 2019. SVIP is a partner in the ELIXIR Annotation and Curation of Human Genomic Variations implementation study, which aims to understand the existing infrastructure, resources and protocols for human genome variation annotation and curation. svip.ch

The group is also actively involved in a multi-site National Research Programmes “Antimicrobial Resistance” (NRP72) project led by the University Hospital of Basel, on the development of a SWISS PATHOGEN SURVEILLANCE PLATFORM – a nationwide database that will make it possible to track intercontinental outbreaks of multiresistant pathogens in near real-time. Version 1 will be released in early 2019. svsp.ch

**PARTNER HOSPITALS**

At the Geneva University Hospitals, OncoBench+ – a tool for analysing and storing genomic data in the field of oncology – has been running since 2016, and version 3 will be released in early 2019. A tailored management and analysis platform for medical genetic data is currently under development for the Institute of Medical Genetics of the University of Zurich.

**TRAINING**

Organized jointly with the University Hospitals of Basel and Lausanne, the first Certificate of Advanced Studies in PERSONALIZED MOLECULAR ONCOLOGY has started in the fall of 2018 and continues over 2019 with 20 participants.

Other training activities included the second edition of a course specifically designed for hospital staff on next-generation sequencing quality control and annotation, and a contribution to a MOOC on Precision Medicine, both in collaboration with the Geneva University Hospitals.

On the international level, the Group also ran a training event in Bahrain, and is co-organizing a European technical workshop on microbial genomics with the Lausanne University Hospital.

In 2018, the Group continued to take part in outreach-related events such as the Planète Santé health fair, and a series of videos about personalized health, funded by the Leenaards Foundation.

What should family doctors know about personalized health?

Two articles in the Swiss Medical Newsletter (DE/FR) highlighted the relevance of the Group’s activities for general practitioners in 2018.
Focus on the BioMedIT project

To promote the development of personalized health research, the Swiss government launched the Swiss Personalized Health Network (SPHN) in 2017. In parallel, the BioMedIT project was funded to establish a coordinated network of core facilities at Swiss universities for the secure processing of biomedical data.

BIOMEDIT IN A NUTSHELL

The ability of life scientists to advance our knowledge of diseases and to identify novel and personalized therapies hinges on their access to large amounts of high-quality patient data. However, such data are associated with stringent levels of protection and information security, which impose strict requirements on the underlying information technology infrastructure, processes and expertise.

BioMedIT will establish a coordinated nationwide network of secure infrastructures (‘nodes’) to support biomedical research, by building on existing expertise and research infrastructure at the partner institutions. At this stage, three BioMedIT nodes are being set up, in Basel (operated by SIB and the University of Basel’s sciCORE Group), Lausanne (operated by ETH Zürich’s SIS Group) and Zurich (operated by ETH Zurich’s SIS Group). BioMedIT will thus provide the researchers with a secure high-performance computing environment – and the accompanying technical expertise – to leverage knowledge from patient data. Discover ‘The researcher’s new path to nationwide clinical data’ on the opposite page.

WHERE ARE WE TODAY?

The first successful steps towards a coordinated encryption/decryption workflow were made in September 2018. They included a computational analysis workflow in the form of a container to demonstrate the feasibility of robust, distributed data analysis across the BioMedIT network. An “SPHN information security policy” clarifying the roles and responsibilities of the various parties was also rolled out in September, and a Security Awareness training workshop was held in Zurich at the end of last year.

Today, the high-performance computing infrastructure (including both storage and computing) dedicated to BioMedIT is already operational in Zurich. It is currently being deployed in Basel, and will follow in Lausanne in the coming months.

Martin Fox, BioMedIT Project Manager at PHI: “Not all BioMedIT nodes evolve at the same pace, each location having its own technical constraints. While it is one thing to have the individual nodes ready to operate and welcoming users, it is another to have the full network interoperable and securely connected to the hospital data warehouses. I am confident that by the end of 2019 we will have three BioMedIT nodes on line, and the frameworks in place to efficiently process data for personalized health research.”

WHY IS SIB LEADING THIS PROJECT?

SIB has been mandated by the Swiss Government to lead the Data Coordination Centre of the SPHN initiative as well as the BioMedIT project because of its unique position as a non-profit, nationwide foundation with no interest other than the advancement of life science and health. Thanks to 20 years of experience in research infrastructure, its involvement as Data Coordination Centre for several public-private Europe-wide health projects and its established partnerships with university hospitals for clinical bioinformatics activities, SIB is indeed well positioned to bridge the gap between research and the clinical world.

2018 highlights

* The city refers to the coordination headquarter of the node
A strong bioinformatics community

IV. TRAINING IN BIOINFORMATICS

One of SIB’s missions is to train the next generation of bioinformaticians and to ensure that scientists make the most out of bioinformatics resources. The SIB Training Group promotes and coordinates training in bioinformatics, both in Switzerland and internationally.

FAIR training

Embracing FAIR principles will directly benefit SIB courses, trainers and course participants, by making training events and materials “Findable, Accessible, Interoperable and Reusable”. In order to tackle the challenges and developments required in this context, SIB Training co-led an ELIXIR workshop in 2018. The practical aspects of FAIR training implementation are currently being set up and should be integrated into SIB courses in the next years.

Reaching out to Europe and the world

While the vast majority of attendees at SIB courses originate from Switzerland, in 2018 participants also came from Belgium, Brazil, Canada, Czech Republic, France, Germany, Italy, the Netherlands, Poland, Qatar, Spain and Tunisia.

Metrics for training quality and impact

Assessing the quality and the long-term impact of training on the work of participants is of great importance to SIB, which has been systematically evaluating its courses since 2013. In 2018, SIB’s metrics were integrated into a pan-European analysis led by ELIXIR. The final outcome of this study will be released in 2019, and the preliminary results already show the benefits European and SIB’s bioinformatics training have in the researchers’ work, such as an improvement in their ability to handle and analyse scientific data.

Bioinformatics capacity building in South Africa

The development of bioinformatics expertise among future African scientists is key for finding solutions to local health problems but often relies on powerful and complex infrastructure. The Swiss Tropical and Public Health Institute (Swiss TPH) joined forces with SIB’s Training and sciCORE Groups to create a novel virtual educational infrastructure to support a bioinformatics course in South Africa. It is now being adapted to enable its use in additional courses.
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.

Key scientific events in 2018

At SIB, fostering scientific collaboration means creating opportunities for our scientists to exchange know-how, and to promote their work. Some of the actions implemented during the year are listed below, followed by a digest of some of the key research results on the next page.

SIB Days – special 20th anniversary edition, Biel

SIB’s two-day internal conference, allowing the Swiss Bioinformatics community to get together;

Keynote speakers included Nick Goldman (EMBL-EBI) on connected DNA and data storage and Fabian Theis (Institute of Computational Biology at the Helmholtz Zentrum) on single-cell RNA sequencing;

Lakeside celebrations on the first evening provided the hundreds of SIB Members and guests with a relaxed and informal moment to celebrate SIB’s anniversary.

Congratulations to the winners of the SIB Days Awards 2018

Best Lightning Talk Award

EMMA RICART, PhD student from the Proteome Informatics Group, Geneva (Group Leader: Frédérique Lisacek)

ADITHI VARADARAJAN, PhD student from the Bioinformatics and Proteogenomics Group, Wädenswil (Group Leader: Christian Ahrens)

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Key research outcomes in 2018

DNA loops and smoke rings have a lot in common

Why do DNA loops forming different knot types sediment at different speeds? Researchers from SIB, the University of Lausanne and EPFL, together with Polish colleagues, observed that this property resembled another, fascinating physics phenomenon, namely the way smoke rings behave in the air. Their results could improve our understanding of the shape and function of biomolecules at large.

Published in Physical Review Letters
SIB Group involved
DNA and Chromosome Modelling  P. 54

How the African elephant cracked its skin to cool off

An intricate network of minuscule crevices adorns the skin surface of the African bush elephant. By retaining water and mud, these micrometer-wide channels greatly help elephants in regulating their body temperature and protecting their skin against parasites and intense solar radiation. SIB Researchers at the University of Geneva found that African elephant skin channels are true fractures of the animal’s skin outermost layer.

Published in Nature Communications
SIB Group involved
Artificial & Natural Evolutionary Development of Complexity  P. 66

The genetic burden of pioneers

Using a unique combination of genomic and historical data, researchers from SIB at the University of Bern, in collaboration with Canadian colleagues, showed that having a pioneer ancestor could explain the excess of harmful mutations observed in recently colonized areas of Quebec. The findings have broad implications for disease incidence in expanding human populations.

Published in Genetics
SIB Group involved
Computational and Molecular Population Genetics  P. 59

Fat, a personal affair

Thirteen percent of the world’s population suffers from obesity, a particularly complex multi-factor disease. Understanding the different factors that bring it about, and defining adiposity subgroups, would offer the possibility of more personalized interventions. Together with colleagues from the University of Regensburg, SIB Scientists at the Lausanne University Hospital describe a first important step towards this goal.

Published in Nature Communications
SIB Group involved
Statistical Genetics  P. 52

Decoding the genome of the wheat stem sawfly, a major agricultural pest

What makes the wheat stem sawfly such a major pest in the grasslands of North America? An international study co-led by SIB Researchers at the University of Lausanne unraveled its genome and found clues on possible mechanisms it employs to select its host plants.

Published in Genome Biology and Evolution
SIB Group involved
Evolutionary-Functional Genomics  P. 51

Zooming into our metabolism

Every minute, each one of our cells carries out countless chemical reactions to convert food into energy, build proteins or eliminate toxic waste. Taken as a whole, these reactions make up our metabolism. Researchers from SIB and the Lausanne University Hospital found a new method to understand how metabolism is regulated at a far higher resolution.

Published in Bioinformatics
SIB Group involved
Vital-IT  P. 71

Cells are able to detect more shades of information than previously thought

Hacking a cell’s communication system is a common feat of information diseases, such as cancer. Researchers at SIB and the University of Lausanne provided the first direct experimental evidence that one of the most common cell receptors – until then thought to function only as on/off switches – can also reliably convey intermediate levels of signal concentrations, thus acting rather like a ‘dimmer’.

Published in Nature Communications
SIB Group involved
Computational Biology  P. 50

Cracking the ‘dark matter’ in bacterial genomes

Bacterial genomes are a treasure trove of information, be it for the development of novel antibiotics or the protection of crops against pathogens. SIB Scientists at the Agroscope in Wädenswil provided hints as to how to resolve the highly complex regions these genomes contain and revealed that they may contain functionally important features.

Published in Nucleic Acids Research
SIB Group involved
Bioinformatics and Proteogenomics  P. 55
Fields of activity of SIB Groups

Bioinformatics is an interdisciplinary field, where the combination of genetics, physiology, chemistry and 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. 50

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. 55

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

SEE P. 58

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. 62

Systems biology
Never alone

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

Bioinformatics develops models to delineate metabolic pathways.

SEE P. 64

Text mining and machine learning
Rise of the machines

Text-mining algorithms are designed to recognize patterns within text so that computers can extract the information of interest, such as biomedical terms. Using machine learning, computers can also acquire the ability to learn without explicit instructions.

Bioinformatics develops text-mining tools that complement expert bio-curation, as well as machine-learning techniques that can improve the prediction of protein structures.

SEE P. 68

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.

Bioinformatics coordinates 13 core facilities and competence centres located in the major Swiss academic institutions.

SEE P. 70
WHY BIOINFORMATICS

MAIN DOMAIN OF ACTIVITY

Genomes

Sven BERGMANN
Philipp BUCHER

UNIVERSITY OF ZURICH

LAUSANNE

COMPUTATIONAL BIOLOGY

GROUP NAME
ONCOGENOMICS GROUP

CITY
EPFL

INSTITUTION
LAUSANNE

SECONDARY DOMAINS OF ACTIVITY
Benchmarking; Comparative genomics; Database curation; Epigenetics; Genetic networks; Software development; Transcriptomics

We develop concepts and algorithmic tools for the analysis of selected tumour entities and genomic variation patterns across genomes. Our group develops new computational methods, genomics databases and network analysis. A complementary direction of our research pertains to relatively small genetic variants and phenotypic data. Other projects are related to genome data of single-cell genomics, microfluidics, and computational approaches. 1) to decipher the regulatory code in selected human systems infections with causative pathogens. 2) to understand the positioning and function of long non-coding RNA and small regulatory RNAs in mammalian fat depots.

KEY PAPERS IN 2018
Wang QA et al. Malaria infected red blood cells induce a stromal cell population promoting differentiation of mammary progenitor cells. Cell Metab. 2018


et al. Genetic separation of adipose tissue in animals. Microbiol. 2018

et al. Central nervous system infections causing small regulatory RNAs vesicles. Nature 2018
In the Host-Pathogen Genomics Group, we explore the genetic roots of inter-individual differences in response to infections, with a particular focus on the genetic interactions between pathogens and their human hosts. At the crossroads between basic science and the clinical world, we are committed to translational genomic research, aimed at identifying, validating and leveraging clinical use of genetic markers of susceptibility to infectious diseases — e.g., host genomics of HIV infection, joint analyses of interactions between human and viral genomes, and genome sequencing on patients with severe infectious disease phenotypes.

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

In the Statistical Bioinformatics Group, 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 gene-environment interactions to obesity risk, and provided frameworks to discover new height-susceptibility variants influencing human height.

We study gene regulation through the analysis and modelling of genome-wide datasets. We collaborate closely with experimental researchers on various biological topics, including cancer progression, chronic inflammation 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, transcriptomic and post-transcriptional regulation interact and contribute to the context of gene expression changes.
Andrzej STASIAK
DNA AND CHROMOSOME MODELLING GROUP
UNIVERSITY OF LAUSANNE
LAUSANNE

Using Brownian motion simulations, we model a chromosome structure in interphase nuclei of mammalian cells to test the possibility that transciptional-induced supercoiling is involved in the formation of TADs. In 2018, we proposed a new model of the chromatin loop extension, in which chromatin rings are passively pushed by formation of transcription product resulting from transcription-induced supercoiling. We also showed by numerical simulations that the chromatin loop extension is capable of directing DNA nicking for unknotting by DNA topoisomerase I.

Shinichi SUNAGAWA
MICROBIOME RESEARCH GROUP
ETH ZURICH

We are interested in studying ecological and evolutionary factors that determine the structure, function and diversity of microbial communities — with a focus on the gut, including the gastrointestinal tract of animals and humans. To this end, we develop and combine bioinformatic and experimental approaches to integrate quantitative "meta-omics" results with contextual information, with the goal of better understanding and predicting the role of environmental microorganisms and the underlying mechanisms of host-microbial homeostasis.

Erik VAN NIMWEGEN
GENOME SYSTEMS BIOLOGY GROUP
UNIVERSITY OF BASEL
BASEL

Our main research interest is the Genome Systems Biology (GSB) Group is the study of genome-wide regulatory systems in order to reconstruct them from high-throughput molecular data, genomes, and other high-throughput data. In addition, we are developing computational approaches and integration of the resulting datasets.

Robert WATERHOUSE
EVOLOUTORY-FUNCTIONAL GENOMICS GROUP
UNIVERSITY OF LAUSANNE
LAUSANNE

The group's research is focused on elucidating interactions between gene evolution and gene function through developing computational approaches to infer genic 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 and their adaptive responses to both ecological niches and extreme environments. Our interests range from arthropod genomics, including invertebrate vectors of human pathogens, to the evolution of viruses and microbial ecology.

Evgeny ZDOBNOW
COMPUTATIONAL EVOLUTORY GENOMICS GROUP
UNIVERSITY OF GENEVA

The group is active in the fields of comparative genomics and metagenomics. We study microbial evolution, develop approaches to genomics data analysis, and implement computational pipelines. We apply evolutionary models to digest sequencing data, and revise those 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.

Christian AHRENS
PROTEOMIC AND INFORMATICS GROUP
WADSWORTH WL

Our research revolves around the bioinformatic and integration of data sets from state-of-the-art omics technologies, which we obtain through collaborations with experimental biologists. These datasets include genome sequences, gene expression and proteomics. We focus in exploiting the unique advantage of proteomics data, including strategies to identify all proteins encoded in a genome (proteogenomics). Another focus is to study the role of secretomes — e.g. for plant protection — by applying metagenomic, comparative genomic and transcriptional approaches and integration of the resulting datasets.

Andreas WOESSNER
GENOMICS GROUP
AGROSCOPE
WÄDENSWIL

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Sarah Parry
INFORMATICS RESEARCH GROUP
CRUNCH

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Katja BAERENFALLER  
MOLECULAR PROTEIN BIOLOGY GROUP  
SWISS INSTITUTE OF ALLERGY AND IMMUNOLOGY RESEARCH (SIAR)  
ZURICH

In the Molecular Allergology group we are aiming to unravel the molecular basis of allergic diseases, antigen presentation, immunotherapies, immune tolerance and tolerance breaking with a combination of functional genomics techniques, including proteomics, peptidomics and immunochemistry. The acquired large-scale molecular data are analysed with the aim of identifying molecular markers which, in different disease phenotypes, underlie the pathogenetic mechanisms, and different patient response groups can be distinguished.

Amos BAIRROCH  
Lydie LANE  
COMPUTER AND LABORATORY INVESTIGATION OF PROTEINS OF HUMAN OR BEES (CLAPIH)  
UNIVERSITY OF GENEVA

We aim to use a combination of bioinformatics and experimental methodologies to gather knowledge about the function of the 20,000 protein-encoding genes in the human genome. Our main mission is the development of a database, a human protein knowledge resource. We also annotate the effects of human protein variation in the context of cancer and genetic diseases. We are part of the HUPRO 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 ontological standardization resources such as the Cellosaurus and ICEPPO.

David GELLER  
GROUP NAME  
COMPUTATIONAL CANCER BIOLOGY GROUP  
UNIVERSITY OF LAUSANNE

In the Computational Cancer Biology Group, our aim is to study interactions between cancer and immune cells. We are focusing on molecular and cellular aspects of cancer immune cell interactions. At the molecular level, we have developed tools to predict neo-antigen presentation from HLA peptide datasets and are currently working on TCR-peptide-HLA interaction predictions. At the cellular level, we are developing novel approaches to characterizing immune infiltrations and the different states of immune cells from single-cell gene expression data.

Frédérique LISACEK  
PROTEOME INFORMATICS GROUP  
UNIVERSITY OF GENEVA

In the Proteome Informatics Group (PIG), we are involved in software and database development for the benefit of the proteomics 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 posttranslational modifications, with a strong focus on glycosylation. We also develop integrated tools and databases to query knowledge of carbohydrate-attached proteins, carbohydrate structures and protein-carbohydrate interactions.

Alan BRIDGE  
SWISS PROTEIN DATABASES (SWISS-PROT)  
UNIVERSITY OF GENEVA

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. We also provide the ELIXIR Core Data Resources, as well as a number of other internationally renowned curated knowledge resources. These include the HAMAP and PROSITE databases of protein families and domains, the ENZYMBase database of enzymes, the Rhea database of biochemical reactions, the Swiss Lipids database of lipid structures and biochemical knowledge, and the ViriZone portal. The group also participates in the development and maintenance of the ExPasy website.

Christian VON MERING  
BIOMINFORMATICS / SYSTEMS BIOLOGY GROUP  
UNIVERSITY OF ZURICH

In the Bioinformatics / Systems Biology Group, we study the dynamics of entire biological systems, both at evolutionary time-scales and at shorter time-scales—down to a few minutes. We often work in close collaboration with laboratory scientists, focusing on the computational aspects of studying such systems, in fields ranging from genomics to genomics and proteomics. In addition, we produce and maintain several online resources for the life science community, including STRING (a protein network), EGGNOG (gene orthology relations), and IPA (gene abundance).

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WHY BIOINFORMATICS

MAIN DOMAIN OF ACTIVITY

Evolution

Evolution

Evolution

ETH ZURICH, D-BSSE

Niko BEERENWINKEL

Group Name

and phylogeny

APPLIED COMPUTATIONAL

COMPUTATIONAL

Group Name

ZHAW

INSTITUTION

Instituition

SECONDARY DOMAINS OF ACTIVITY

Comparative genomics;

sequencing data;

Infectious diseases;

Next-generation sequencing; Oncology;

Machine learning;

Ontology; Phylogenetic analysis natural selection;

Semantic web format

With the growing size and complexity of biological data, we seek to better understand the evolutionary history of species and the rational design of medical interventions. To achieve this, we use computational and statistical methods to analyse patterns of evolution and development. We aim to support the rational design of medical interventions and the development of new technologies, with a particular focus on humans. We also investigate the interplay of population structure, selection, and demographic changes on genomic and functional signatures of adaptation and diversity.

ORGANIZATION

The OMA orthology benchmarking service allows for the reliable orthologous groups of any pair of species, while the OMA orthology service provides predictions for orthologous groups. The team focuses on the effect of range expansions on genomic and functional diversity, as well as the interplay between population structure, selection, and demographic changes on genomic and functional signatures of adaptation and diversity.

KEY RESOURCES

PGDSpider (part of SwissOrthology) and between populations, as well as the interplay between population structure, selection, and demographic changes on genomic and functional signatures of adaptation and diversity.

PGDSpider (part of SwissOrthology)

Background selection

Relaxed selection

Complex demographic

Forward and backward simulations

Complex genetic

signature of adaptation

Machine learning

Ontology

Phylogenetic analysis

Semantic web format

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KEY RESOURCES

PGDSpider (part of SwissOrthology)
Sara MITRI  
MICROBIAL EVOLUTION GROUP  
UNIVERSITY OF LAUSANNE  

Our group focuses on studying social interactions in microbial communities and how they evolve. We follow an interdisciplinary approach, where we quantify and characterise interactions within and between microbial colonies, and then use computational and mathematical models to explain and predict how these interactions will change over ecological and evolutionary time scales.

Richard NEHER  
MICROBIAL EVOLUTION GROUP  
UNIVERSITY OF BASEL  

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

Joshua PAYNE  
EVOLUTIONARY BIOLOGY GROUP  
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.

Marc ROBINSON-RECHAVI  
EVOLUTIONARY BIOINFORMATICS GROUP  
UNIVERSITY OF LAUSANNE  

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

Nicolas SALAMIN  
COMPUTATIONAL EVOLUTION GROUP  
ETH ZURICH  

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, clade phylogenetics and plant genomics, the estimation of positive 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.

Tanja STADLER  
COMPUTATIONAL EVOLUTION GROUP  
ETH ZURICH  

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, as well as ecologists and palaeontologists.
In the Evolutionary Systems Biology Group, we study the evolution and evolvability of biological systems at all levels of biological organization, from genes and gene networks 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 modeling.

Matthew DAL PERA

Our main goal at the Laboratory for Biomolecular Modelling 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.

Olivier MICHIELIN

The Molecular Modelling Group studies molecular interactions using a range of modeling techniques such as homology modeling, 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 human neutral endopeptidase, such as T-cell receptors, for cancer immunotherapy. We develop and maintain web tools for drug design, such as SwissDock, SwisTargetPrediction, and SwissADME. We also run the Protein Modelling Facility of the University of Lausanne.

Matteo DAL PERA

Structural biology; Protein biophysical characterization.

Olivier MICHIELIN

Drug design; Drug development; Metabolic pathways; Structural biology; Toxicology; Protein engineering; Software development; Systems biology; Computer simulations; Drug design

The overall objective of our research is to understand the role that structure and dynamics play in the definition of the function of biomolecules. In order to perform their function, proteins and other biological molecules undergo a series of conformational changes that have been shaping 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-based methods. We then apply these approaches to the wealth of data currently being generated.

Daniel WEGMANN

Structural and statistical computational biology.

The Laboratory for Biomolecular Modelling at the EPFL is prepared to perform molecular biology, protein expression and purification, and protein biophysical characterization.

Bruno CORREIA

The Laboratory for 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.
NEW MAIN DOMAIN OF ACTIVITY

Torsten SCHWEDE
COMPUTATIONAL STRUCTURAL BIOLOGY GROUP UNIVERSITY OF BASEL, BASEL

In the Computational Structural Biology (CSB) Group, we focus on the development of methods and algorithms to model, simulate, and analyze 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.

Vincent ZOETE
COMPUTER-AIDED MOLECULAR BIOLOGY GROUP UNIVERSITY OF LAUSANNE, LAUSANNE

The laboratory specializes in the development of computer-aided algorithms, programs and databases for protein engineering and drug design, with applications in oncology, notably in immunotherapies of cancer. We also provide support in Molecular Modelling for the Molecular Tumour Board of the Lausanne University Hospital.

Basile CHOPARD
SCIENTIFIC AND PARALLEL COMPUTING GROUP UNIVERSITY OF GENEVA, 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-scale systems, and optimising 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.

Giovanni CIRELLO
COMPUTATIONAL SYSTEMS BIOLOGY GROUP UNIVERSITY OF LAUSANNE, LAUSANNE

The Computational Systems Oncology lab integrates algorithmic design, numerical modeling, and molecular biology approaches to address relevant questions in cancer biology and the development of treatments. 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.

Manfred CLAASSEN
COMPUTATIONAL SINGLE CELL BIOLOGY GROUP 6TH FLOOR ZURICH, ZURICH

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

Dagmar IBER
COMPUTATIONAL BIOLOGY GROUP ETH ZURICH, BASEL

The Computational Biology Group (CoBi) develops computational models of developmental processes. We place a particular focus on mechanistic 4D image-based models of organogenesis (mouse lung, kidney, pancreas, brain, Drosophila wing and eye) and on the delineation of fundamental mechanisms such as those that restrict the size of organs and those that maintain the proportions of structures in different-sized embryos. The group runs a wet lab to obtain image data, and collaborates with tissue engineers to develop spatially-organized tissue from stem cells, as well as with clinicians to apply the techniques to disease models.
Our group is located in the Department of Bioinformatics (DBM) at the University of Basel. We collaborate with scientists from DBM on projects covering a broad spectrum of research topics, from cellular differentiation and evolutionary biology to the identification of a molecular basis for various human diseases. To do this, we focus on the analysis, interpretation and visualization of expression, epigenetic and genomic data, which are mainly derived from next-generation sequencing experiments.


We combine Evolutionary and Developmental Biology as well as genetic approaches with the study of physical processes to understand the mechanisms generating life’s complexity and diversity. The self-organizational capacities of systems are pertinent to Evo-Devo 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 Evolutionary and Developmental Biologists, Computer Scientists, Engineers, and Physicists.

Our research interests in the Scientific Computing Group lie in the areas of multiscale/multiphysics 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, deconstruction and aggregation of cells under physiological flow conditions, host infection, and coarse-grained molecular dynamics, as well as the modelling of transport processes in healthy and tumour-affected biological systems.


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 controlling 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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The individual cells of a body contain a stunning diversity of phenotypes, despite carrying a largely identical genetic makeup. This is due to the distinct biological pathways in which the same genetic information can be read, interpreted and translated into function. At the Research Network (BCN) at the Biocenter in Basel, we combine computational modelling with big data and experimental analysis to discover and understand the regulatory networks governing the interpretation of genetic information at the level of tissues and single cells.

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## Karsten BORGWARDT

**Main Domain of Activity:** Biostatistics; Data mining; Machine learning; Mathematical modeling; Single cell biology; Transcriptomics;Uhura flows.

**Group Name:** RNA REGULATORY MACHINE LEARNING AND BIOCOMPUTATIONAL INTELLIGENCE GROUP

**Institution:** ETH Zurich

**City:** Zurich

Our lab acts as the bridge between big data analysis and biomedical research. We develop novel data-mining algorithms to detect patterns and statistical dependencies in large datasets from the fields of biology and medicine. Our main goals are two-fold: 1) to enable the automatic generation of new knowledge from big data through machine learning, and 2) to gain an understanding of the relationships between biological systems and their molecular properties. Such an understanding is of fundamental importance for personalized medicine, which tailors medical treatment to the molecular properties of a person.

## Gunnar RÄTSCH

**Main Domain of Activity:** API; Database curation; Information management; Web format; Software engineering.

**Group Name:** BIOMEDICAL INFORMATICS GROUP

**Institution:** UNIVERSITY OF ZURICH

**City:** Zurich

We are interested in modern machine-learning techniques suitable for the analysis of problems that arise in medicine and biology. In particular, we develop new learning techniques that are capable of dealing with large amounts of genomic data and medical data. These techniques aim to provide accurate predictions in a robust manner. This allows us to comprehensively provide reasons for their performance, and thereby assist in gaining new biomedical insights.

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## Fabio RINALDI

**Main Domain of Activity:** Artificial intelligence; Bioinformatics; Biological databases; Biological networks; Bioinformatics; Computational biology; Data mining; Database curation; Electronic health record; Machine learning; Oncomics; Drug discovery.

**Group Name:** BIOCOMPUTATIONAL INTELLIGENCE AND BIOMEDICAL INFORMATICS GROUP

**Institution:** UNIVERSITY OF ZURICH

**City:** Zurich

The BioMXIT group specializes in Information Extraction from biomedically relevant textural sources, such as the scientific literature, clinical records, or social media. We focus on particular topics, such as the extraction of domain-specific entities (such as genes, drugs and diseases), and their semantic relationships, e.g. gene-disease associations. Our tools are often evaluated through participation in community-run evaluation challenges (e.g. BioCreAtIvE). We also provide an environment for Assisted Curation (ODEN), which is used in the curation pipeline of the RegulonDB database, in an SHU-funded project.

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## Carlos PENA

**Main Domain of Activity:** Data mining; Machine learning; Metagenomics; Bioinformatics; Systems biology.

**Group Name:** DATA MINING AND MACHINE LEARNING GROUP

**Institution:** ETH ZURICH

**City:** Zurich

With the advent of high-throughput technologies and clinical data analysis 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.

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## Patrick RUCH

**Main Domain of Activity:** Data management; Database curation; Electronic health record; Computational biology; Computer science.

**Group Name:** TEXT MINING GROUP

**Institution:** UNIVERSITY OF ZURICH

**City:** Zurich

In the Text Mining Group, we carry out activities in semantic interoperability and text analysis applied to the health and life sciences. Previously hosted by the Radiology and Medical Informatics Department of the University of Zurich, our group moved to the University of Applied Sciences Geneva (HEG-VD). We develop text-mining solutions to support both the annotation of curated databases and the work of a wide range of biomedical professionals from drug discovery to clinicians. We thus develop specific biomedical decision-support systems, in particular in oncology and molecular pathology.

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## Mihaela ZOVANOL

**Main Domain of Activity:** Data mining; Deep learning; Inference in large scale multi-task learning problems; Interventional Conferences of Artificial Neural Networks (CUR). Systems biology.

**Institution:** UNIVERSITY OF ZURICH

**City:** Zurich

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**Main Domain of Activity:** Data management; Database curation; Electronic health record; Computational biology; Computer science.

**Group Name:** TEXT MINING GROUP

**Institution:** UNIVERSITY OF ZURICH

**City:** Zurich

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NEW WHY BIOINFORMATICS — INSTITUTION — MAIN DOMAIN OF ACTIVITY

Core facilities
BIOINFORMATICS CORE UNIT
GROUP NAME
MEDICAL DATA SCIENCE GROUP
BIOINFORMATICS UNIT (IBU)

INSTITUTION
UNIVERSITY OF BASEL
Berne

SECONDARY DOMAINS OF ACTIVITY
Machine learning
Biostatistics; Data management; Next-generation sequencing; Transcriptomics
Personalized medicine; Single-cell biology; Systems biology; Training; Translational research

DOMAINS OF APPLICATION
Workflows and project planning for large-scale cancer analyses. The field of action comprises many areas such as prediction of response to treatments in personalized medicine, (quasi-) biomarker discovery, tumour classification or the understanding of interactions between genes or groups of genes. We take a data- and problem-driven approach to these challenges. In close collaboration with physicians we identify gaps where current technologies fail and develop tailored solutions.

NEW WHY BIOINFORMATICS — INSTITUTION — MAIN DOMAIN OF ACTIVITY

Core facilities
BIOINFORMATICS CORE UNIT
GROUP NAME
VITAL-IT
LAUSANNE

INSTITUTION
UNIVERSITY OF LAUSANNE
Lausanne

SECONDARY DOMAINS OF ACTIVITY
Deep sequencing data; Mathematical modelling; Machine learning; Mass spectrometry; Next-generation sequencing; Software engineering; Systems biology; Transcriptomics

DOMAINS OF APPLICATION
We are a competence centre in bioinformatics with a multidisciplinary team of scientists and technical staff. Our mission is to provide bioinformatics support to life science projects in Switzerland and abroad through scientific collaborations with academic and industry partners. The services and expertise we offer include algorithm development and optimization, software engineering, data management including database expertise (Oracle certified), data analysis, genomics, transcriptomics, proteomics, metabolomics, lipidomics, development of web-based data mining and visualization tools and in silico modelling.

NEW WHY BIOINFORMATICS — INSTITUTION — MAIN DOMAIN OF ACTIVITY

Core facilities
BIOINFORMATICS CORE UNIT
GROUP NAME
DEPARTMENT OF ONCOLOGY
BELLASONIA

INSTITUTION
LOMBARDIA TELAMONIA
Legnano

SECONDARY DOMAINS OF ACTIVITY
Biostatistics; Drug resistance; Machine learning; Transcriptomics

DOMAINS OF APPLICATION
Our main task at the Bioinformatics Core Unit (BCU) is to support the research groups at the Institute of Oncology Research (IOR) with computational and statistical services. Our research is dedicated to the genomics and biology of cancer with a major emphasis on lymphomas and epithelial cancers, such as prostate, breast and ovarian cancer. Importantly, more than just a supporting role, we proactively identify and develop novel bioinformatics projects that can complement and in many cases drive our biologic research.

NEW WHY BIOINFORMATICS — INSTITUTION — MAIN DOMAIN OF ACTIVITY

Core facilities
BIOINFORMATICS CORE UNIT
GROUP NAME
FUNZIONE NAZIONALE GENOMICA CENTRE ZURICH (CNCG)
ZURICH

INSTITUTION
UNIVERSITY OF ZURICH
Zurich

SECONDARY DOMAINS OF ACTIVITY
Data management; Data mining; Genomic analysis; Machine learning; Mass spectrometry; Next-generation sequencing; Software engineering; Systems biology; Transcriptomics

DOMAINS OF APPLICATION
We are dedicated to the processing, analysis and interpretation of next-generation sequencing data. We interact closely with research groups, and provide tailored comprehensive bioinformatics solutions. Additionally, we provide standard analysis pipelines for the most frequent research questions. We train researchers and bioinformaticians on various aspects of data analysis, and provide access to our computing infrastructure for running analyses.

NEW WHY BIOINFORMATICS — INSTITUTION — MAIN DOMAIN OF ACTIVITY

Core facilities
BIOINFORMATICS CORE UNIT
GROUP NAME
HUBERT REHAUER
ZURICH

INSTITUTION
UNIVERSITY OF ZURICH
Zurich

SECONDARY DOMAINS OF ACTIVITY
Data management; Data mining; Deep sequencing data; Gene regulatory network analysis; Next-generation sequencing; Single-cell biology; Software engineering; Transcriptomics

DOMAINS OF APPLICATION
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Core facilities
BIOINFORMATICS CORE UNIT
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SECONDARY DOMAINS OF ACTIVITY
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In the Service and support for Science IT (S3IT) unit, we provide support for science in general, and for life sciences and medicine in particular. S3IT serves as a partner for both local and national projects to enable competitive research with the advanced use of computational methods and resources. Our team advises groups and projects about data management and data analysis, and cooperates to optimize their specific workflows. S3IT also takes part in national projects and cooperates with similar technology-oriented groups to ensure that its expertise is always up to date.

**Daniel STEKHOVEN**

We are the Clinical Bioinformatics Unit at the ETH Zurich 
core facility NEXUS Personalized Health Technologies. Our unit offers highly customizable bioinformatics and biostatistics services for projects in the field of biomedical research and development. We maintain close collaborations with hospitals in Switzerland, enabling and accelerating the use of top-notch data analysis, software development, and clinical decision support. Based on a fee-for-service or joint collaboration model, we aim for tailor-made full services for our clients, translating computational and analytical tools to meet their needs.

**Marcel RIEDI**

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**Bernd RINN**

S3IT is an interdisciplinary bioinformatics and scientific IT support group which builds upon the skills and experience of our team. We offer services to local and national projects, including bioinformatics and life sciences. Our work involves data analysis, data management, and computational tools – from lab databases to reusable framework components for data analysis and data management – for life science research and beyond. We improve and port scientific software, integrate and operate data analysis pipelines, and provide training and consulting in databases, scientific software development, high-performance computing, and software engineering.

**Tosten SCHWEDE**

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ACKNOWLEDGEMENTS

We gratefully acknowledge the following funders, sponsors and partners for their financial support and encouragement in helping us fulfill our mission in 2018.

The Swiss government and in particular:
The State Secretariat for Education, Research and Innovation (SERI)
The Swiss National Science Foundation (SNSF)

Innosuisse

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

We are very thankful to our 20th anniversary’s sponsors and partners (see p. 27) whose contribution fully financed the projects and events launched to mark the occasion.

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

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