Health Genomics within the Swiss Personalized Health Network (SPHN) initiative

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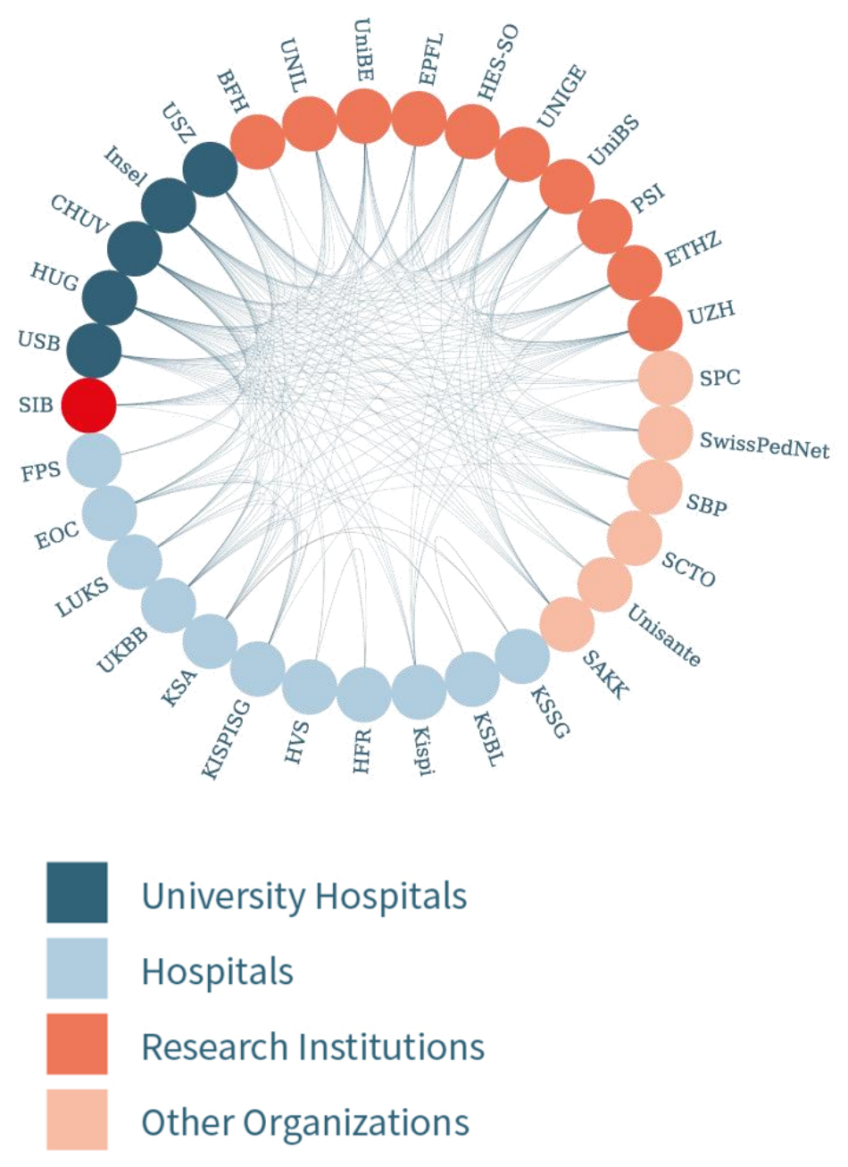
SPHN: A national initiative for F.A.I.R health-related data

Enable use and exchange of interoperable health data for research is the overall objective of the Swiss Personalized Health Network (SPHN) Initiative. SPHN is a national research infrastructure initiative mandated to the Swiss Academy of Medical Sciences (SAMS) and the Swiss Institute of Bioinformatics (SIB) and funded by the Swiss government for the period of 2017-2024. Within SPHN, all decision makers from key clinical, research and research support institutions are rallied together to promote the development, implementation and validation of coordinated data infrastructures in order to make health-relevant data shareable for research in Switzerland



Streams for data-driven biomedical research and Personalized Health

The first phase of SPHN focused on FAIRification of clinical routine data for personalized medicine, notably thanks to its infrastructure development and multi-domain driver projects. Starting at the end of phase one and to be continued in phase two, SPHN started to include other data streams such as clinical research data or omics.

Clinical research, cohort and registry data

High-quality patient-oriented health data

Problems: Although this data is (mostly) captured for research purposes, it is merely vertically standardized and interoperability between studies is not given. Also, there is currently only minimal cross-use of routine data for this data stream. There are existing obstacles regarding the re-use of existing clinical study, registry and cohort data, but also need for action with a view to a more efficient re-use of data that will be collected in the future (fulfil FAIR criteria by design).

SPHN approaches: Connect important Swiss cohorts to an (inter)national Meta-Data Catalogue to increase findability; streamline compliance to governance processes; promote the use of routine data for clinical studies, cohort and registries,

Data from healthcare institutions

Routine data and specific routine data (e.g. diagnosis, medication, demographics, imaging data)

Problems: Data is not captured for research (but for care) in primary systems; data is widely unstructured, standards are lacking; there is limited explanatory information to routine health data.

SPHN approaches: Semantic Interoperability Framework, SNOMED CT coding of concepts and attributes, introduction of standards at the source (e.g. LOINC), nationally harmonized RDF specification, experimental NLP projects running

Molecular or *omics data

Genomics, transcriptomics, proteomics, metabolomics, DNA methylation, etc.

Problems: There are no agreed standards for omics data (and meta-data) in Switzerland; researchers using biobanking materials do not know if omics data of the particular samples would already be available; there is no central repository or source for the re-use of omics data.

SPHN approaches: Establish a local EGA for data from inside and outside SPHN projects; agree on data and meta-data standards in the Swiss omics landscape; SPHN expert taskforces received the mandate to develop concepts for a “Swiss Federated Omics Network”.

Healthy citizen data

Citizen/consumer health data, lifestyle data, social media data, wearable devices, etc.

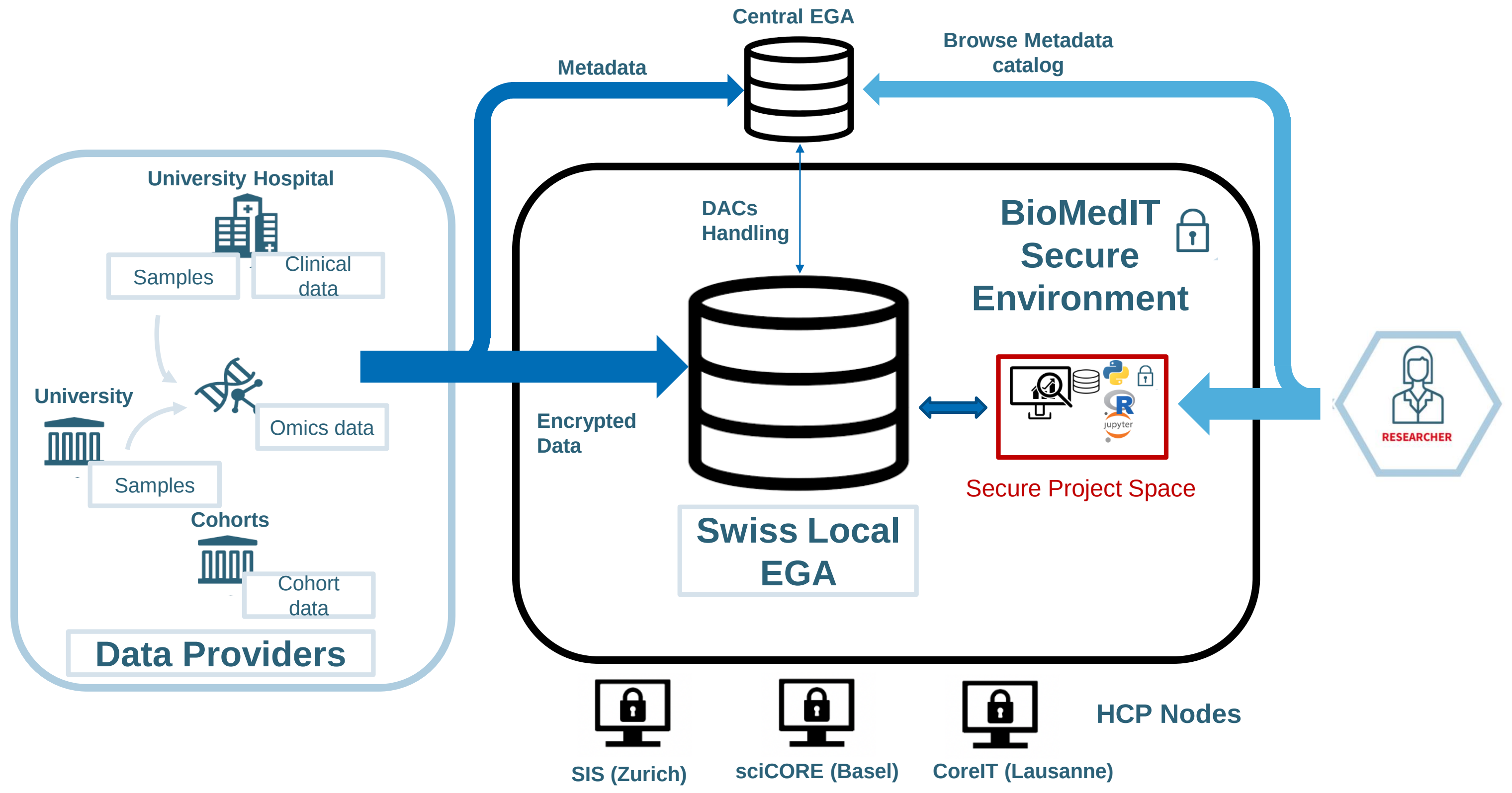
Problems: Data lack standardization as well as technical interoperability; data is often locked in apps.

SPHN approaches: Healthy citizen data is to be addressed in the second phase of SPHN. Development of a citizen-centred consent procedure is currently under development.

Swiss Local EGA - A local repository for genomic data

Switzerland is currently lacking a data repository fulfilling the privacy and security requirements needed to deal with sensitive genomic data generated within the biomedical domain. In order to fill this gap, SPHN explores the possible implementation of a local EGA hosted within a secure environment.

More information about the BioMedIT secure environment is available on the SPHN webpage (sphn.ch/network/projects/biomedit).



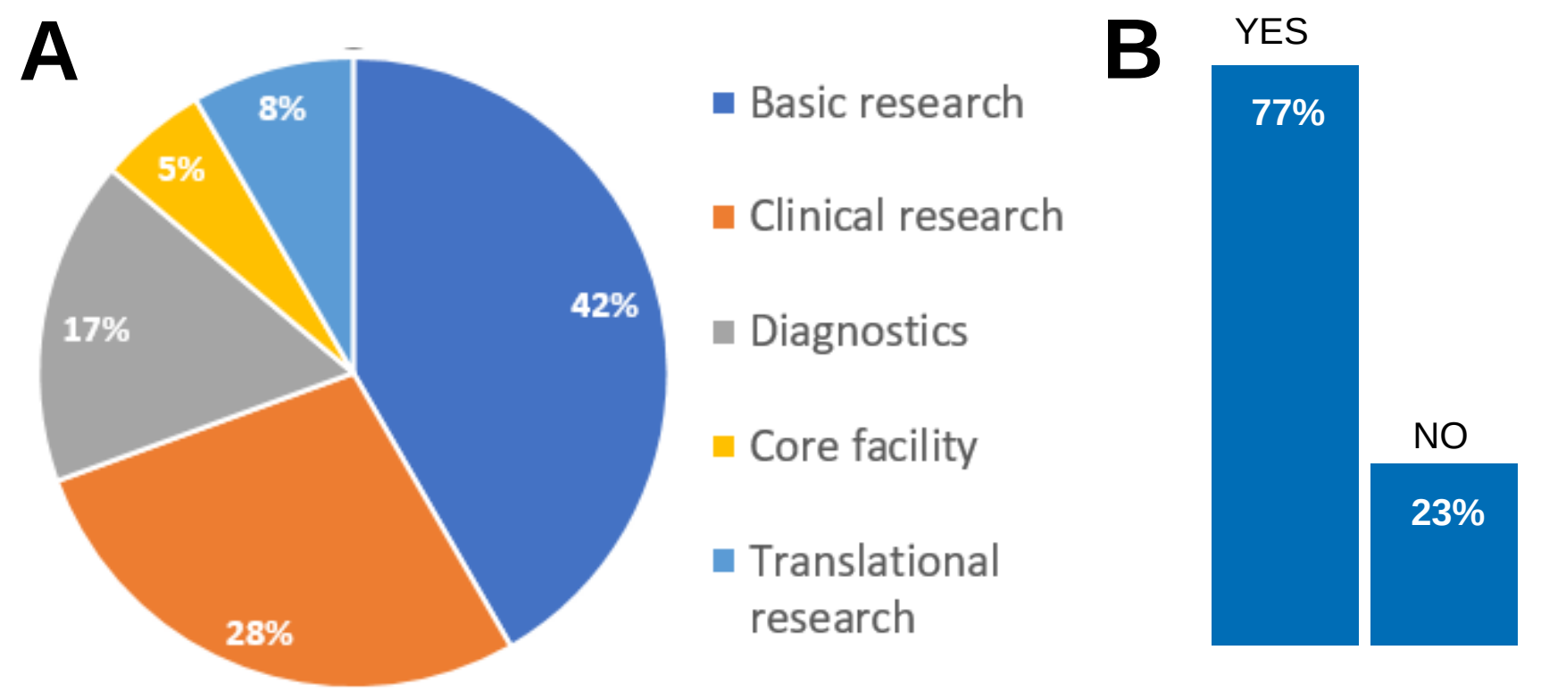
Improving data accessibility

Contribution to the GA4GH Beacon Protocol

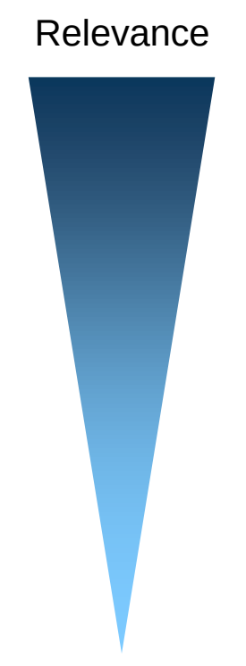
The beacon protocol delineate an open standard for genomics data discovery by providing a simple and federated solution to query against genomic data collections. The Beacon v1.n handover extension allows to overcome the direct data delivery limitations of the standard protocol by allowing the retrieval of the matched data. For more details see *Progenetix - A cancer genomics reference resource around GA4GH standards (M. Baudis)* poster.

Sharing Genomic data in Switzerland

SPHN, in close collaboration with the Health2030 Genome Center (https://www.health2030genome.ch), are actively mapping the current practices in genomic data usage and sharing in the Swiss genomics community. As part of the process, an in-person survey has been run in a set of representative experts in the field. Here we present a preliminary results from the survey. These data will help SPHN to adjust their future efforts in improving genomic shareability.



(A) Fields of expertise of the survey respondent. (B) Willingness to share data for secondary use. (n=18)



1. Ethical and Legal constraints.
2. Need to obtain a specific consent.
3. Time-consuming procedure.
4. Restrictions set by consortium agreement.
5. Lack of IT support.
6. Lack of incentives to share data.

Main perceived limitations for data sharing.

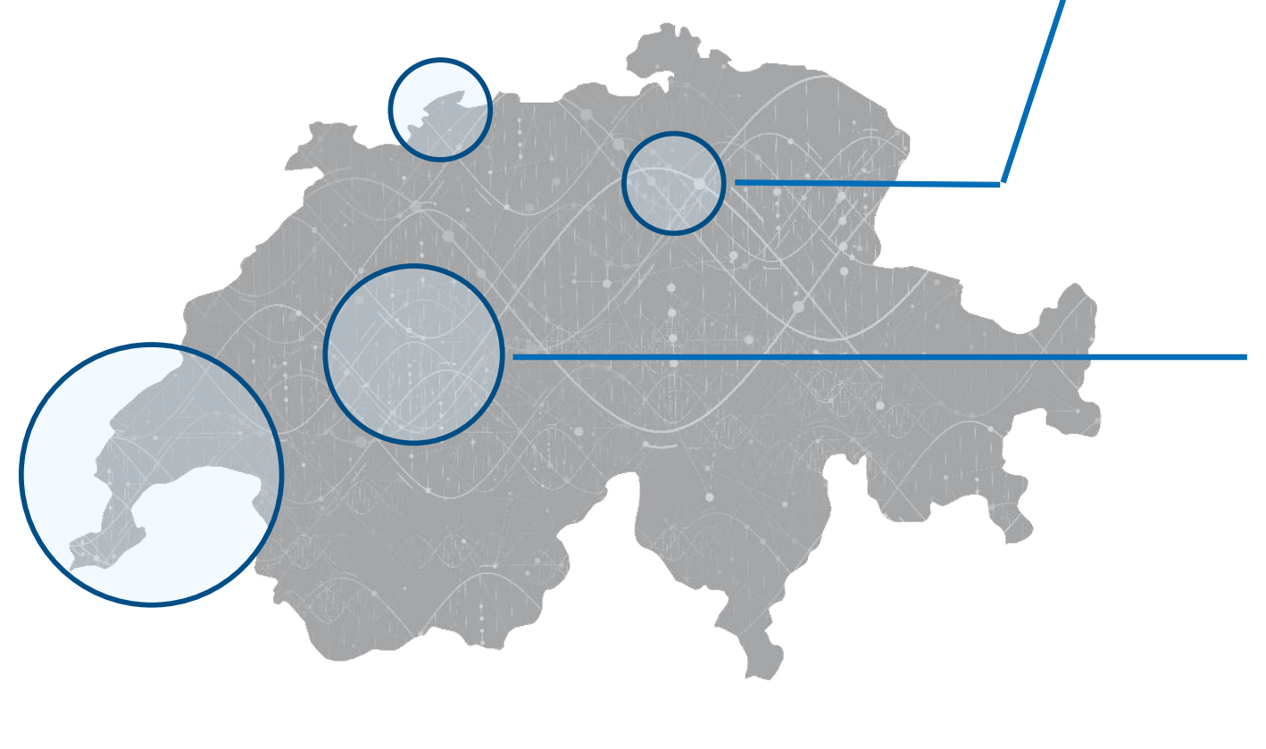
The Swiss Genomics Landscape

Genomics in Switzerland is mainly represented by genomics and metagenomics laboratories within the five university hospitals and dedicated core facilities relied to the main universities and polytechnics. With the foundation of the Health 2030 Genome Center in 2017, a multi-institutional organization for genomic medicine has been put in place.

Universities Core Facilities
University of Geneva
University of Lausanne
Swiss Federal Institute of Technology Lausanne

University hospitals
Geneva University Hospitals (HUG)
Lausanne University Hospital (CHUV)

Centers and platforms
Health 2030 Genome Center
Swiss Biobanking Platform



Universities Core Facilities
University of Basel
D-BSSE

University hospitals
Basel University Hospital

Universities Core Facilities
University of Zürich
Swiss Federal Institute of Technology Zurich

University hospitals
Zürich University Hospital (USZ)

Universities Core Facilities
University of Fribourg
University of Bern

University hospitals
Bern University Hospital (Insel)