

# Genomic-based health care

Professor Mark A Rubin describes how the University of Bern and the Bern University Hospital are poised to lead the way to this next frontier of clinical care, dramatically improving medical outcomes both for patients and society

**T**he overarching goal of precision medicine is to identify the right therapy at the right time for each patient. Since 2003, when the results of the first-ever complete sequence of the human genome were revealed from the Human Genome Project, our understanding of the genome – and the therapeutic promise that it holds – has exploded. We now have a much greater understanding of how genes can cause or influence the progression of some diseases and are using this knowledge to develop targeted therapies.

Using genetics to find the right treatment has created an entirely new way to fight disease and though tremendous progress has been made to date, genomic-based health care is still in the early stages. Together, the University of Bern and the University Hospital (Inselspital) are poised to lead the way to this next frontier of clinical care, dramatically improving medical outcomes both for patients and society.

Following an initiative of the canton, the Bern Center for Precision Medicine (BCPM) was founded in January 2019 as a platform for the researchers of active in the field of Precision Medicine from both the University of Bern and the Inselspital. The Center is led by Professor Mark A Rubin, an expert in prostate cancer genomics and a pioneer in precision oncology. Rubin founded the Englander Institute for Precision Medicine at Weill Cornell and he will bring this New York experience to Bern.

## A robust precision medicine approach

By incorporating basic and translational research, education of the next generation of physicians, and exceptional care for the large and diverse patient population in Bern, the BCPM will create the collaborative organisational structure required to discover the genetic causes of diseases, develop novel, customised treatments, and deliver those therapies effectively. The BCPM will

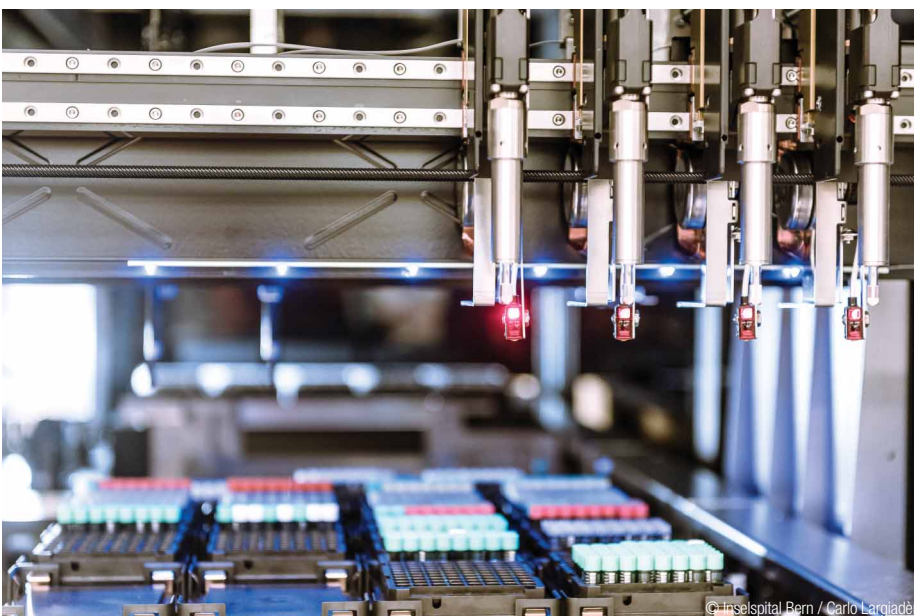
establish the regulatory, technical, clinical, ethical, and economic framework to create a robust precision medicine approach to treating patients in health and disease. With a new medical school curriculum, the University of Bern will also be training the first generation of doctors in the Precision Medicine era.

Existing organisational units from the University and Inselspital will be leveraged within precision medicine projects and new research and infrastructure projects in the area of precision medicine will be supported. Initially, the Center will focus on oncology, cardiology, and neurology. These are areas of great clinical strength for the University of Bern and Inselspital, and also offer great promise for the development of genetically-focused pharmaceutical therapies.

Over time it will expand to other disease areas, benefitting all patients within reach of medical knowledge. Training and further education about precision medicine will be offered, in co-operation with the existing graduate schools. Establishing tight interactions with existing research and clinical projects and programmes in Switzerland and elsewhere will allow the Center to create new standards in how patients are treated.

At the BCPM, a multidisciplinary approach will be adopted, combining the efforts of doctors, geneticists, scientists, and data analytics, thus leveraging everyone's expertise to empower clinicians with the best diagnosis and treatment options.

Specifically, the BCPM will incorporate: **Genetic testing and sequencing**  
The BCPM will incorporate new methods and technologies for genetic testing and genome sequencing to apply this powerful diagnostic tool to day-to-day clinical care. The results of these tests will help physicians identify which patients are at higher risk for developing disease, make



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quicker and more accurate diagnoses, and more precisely customise care plans and treatments.

The introduction of genetic testing into common medical practice will not only produce better outcomes, but will also increase the satisfaction of patients whose therapies are delivered more quickly, with greater specificity, and with increased safety. The information gathered through genetic testing and sequencing will also become part of larger genomic research efforts of Swiss Personalized Health Network (SPHN).

### Pharmacogenomics

Each patient's unique genetic makeup can affect how that patient will respond to various pharmaceutical options. Pharmacogenomics uses genetic testing to predict the efficacy of a given therapy in each patient's case. For example, pharmacogenomics can help guide the choice of chemotherapies for a cancer patient. Similarly, pharmacogenomics can also help identify patients who can have potentially life-threatening reactions to certain medications. This burgeoning field promises to usher in a new era of truly personalised, precise medicine, and it has already begun to deliver results.

### Insel Data Science Center

Inselspital have led the effort to bring medical record keeping to the digital age through the development and implementation of the Insel Data Science Center. Now, for the precision medicine effort, our system must be adapted so that it can store a patient's genomic information. By upgrading the Electronic Health Record (EHR) to enable the linkage of genomic information with clinical information, we will be able to provide clinicians with even more powerful tools to diagnose and treat disease.

A fully equipped institute will provide the technologies, talent, and funding required to upgrade existing systems to incorporate this data and make actionable recommendations for clinical care based on the information in the EHR.

### Patient communication and education

The genomic medicine revolution will have a great impact on the patient experience, and healthcare institutions must translate this new information into a language that is easy for patients to understand. This will involve training physicians and other clinical staff to communicate clearly with patients. Additionally, the centre will expand the capabilities of existing IT platforms to enable patients to view information from their medical record, the results of genetic testing, and relevant educational content.

### Physician education, training and support

To enable the BCPM to effectively deliver cutting-edge patient care, the centre will educate the next generation of physicians as well as our current doctors about the role of genomics in the clinical

care that they provide, and it will provide the resources to help them make the most effective healthcare decisions. The centre will also feature a leadership program to identify promising junior faculty and train these scientists and clinicians to be the next generation of leaders in the field.

The centre will be supported by a robust research infrastructure, which will enable it to deliver leading-edge clinical care. This includes basic research to identify the genetic drivers of disease, translational research to develop new targeted therapies, and population-based research to better understand the impact of genetic variation.

### Core elements

The core elements of the research program of the BCPM will include:

#### Scientific talent

The centre already has a world-renowned group of scientists working to make precision medicine a reality. This talented group will be expanded through the targeted recruitment of additional scientists who are conducting studies on the cutting edge of the genomic revolution. These will include scientists discovering new genetic risk factors, researchers creating new algorithms and computational tools to handle the 'big data' of precision medicine, and investigators developing new gene-specific treatments and therapies. With their support, even more information, from the overwhelming amount of data from genomic testing that is generated, can be retrieved and used.

#### Biobanks

Understanding the role that genomics plays in human health requires analysing a large amount of genetic information. The centre will collect biological samples from more than 25,000 patients over five years. To unlock the vast amount of genetic information they contain, these samples will be sequenced for studying individual mechanisms of tumour formation and evolution and used to test drug responses.

The BCPM will take advantage of the biobanks that already exist in the University of Bern and Inselspital (see <https://www.biobankbern.ch/>). These biobanks have the appropriate infrastructure, processes, technology, and staff to store fluids, solid tissue, DNA, RNA, and protein molecules for clinical and research use. The dissemination of established samples to the greater research community will allow dissecting the molecular aspects and therapeutic options in the fastest way possible.

#### Clinical data repository

Effective genomic research requires pairing the genetic information in each sample with information about that individual's physical characteristics, their disease, and their family history. The clinical data repository will use both traditional patient surveys and cutting-edge natural language processing technology to collect

this information and store it in a structured format for use in genomic research. This repository will be integrated in the Swiss co-ordinated data infrastructure developed by SPHN to enable effective exchange and access of patient data (e.g. disease phenotypes).

#### Bioinformatics

One of the many advantages of building a robust research infrastructure is that the BCPM will be uniquely poised to make groundbreaking discoveries that will deepen our understanding of the function of the human genome, the interplay between genes and disease, and the therapies that can target specific gene-based disease processes. This will be accomplished by building a powerful bioinformatics data analytics capability that will allow researchers to identify and study individuals with similar diseases, identify genomic commonalities that cause disease, and determine precisely the efficacy of an intervention for a particular disease.

#### SOCIBP

A pilot project funded by SPHN to build such bioinformatics framework has already been launched. The Swiss Molecular Pathology and Tumor Immunology Breakthrough Platform (SOCIBP) project will create a breakthrough genomics platform to manage and share data across Switzerland to help our clinicians and scientists untangle the complexity of cancer therapy resistance. The Swiss oncology community will have a uniform genomics language to easily share and visualise data that is compatible and potentially extendable with international data.

Our goal is to make every patient's plan of care as unique as their genes. With the confluence of relatively inexpensive and dramatically faster genome sequencing, enhanced computing power, sophisticated analytical tools, and broader adoption of electronic health records, our knowledge and clinical abilities will continue to increase at an exponential pace. The BCPM will improve patient care and it will create value for the university, Inselspital, and also the patients.

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