

# Development Plan for the Institute of Genomics 2022-2027

## *Introduction*

The mission of the Institute of Genomics (IG) is to conduct research (including transdisciplinary research) on genomics and omics data and method development; raise new outstanding scientists; contribute to the application of research through genomics-related innovation in both the public and the private sector; and promote a worldview based on science and facts. In accordance with the Human Genes Research Act, the Institute of Genomics maintains the Estonian Biobank (EstBB). The Institute of Genomics takes great responsibility in the safe-keeping of the genetic data of biobank participants at the Estonian Biobank.

This development plan is structured along the functions (science, teaching, implementation of personalised medicine, development of Estonian Biobank) of the Institute rather than across its structural units. The development plan is subject to yearly reassessment by the council of IG.

## *Science*

### *Challenge*

Genomics is a highly competitive and rapidly developing area of research that requires world-class scientists and a great working environment. Being at the forefront also requires innovative ideas and initiative to develop new areas of research. The increasing need for health care and medicine in the ageing society requires input from the field of human genomics in order to understand the underlying biological mechanisms leading to disease and to use genomic and omics information for disease prediction, prevention and better treatment.

### *Objectives & Actions*

1. **RESEARCH.** Conduct cutting-edge scientific research and publish the results in high-impact, international peer-reviewed journals. Explore innovative and impactful research topics building on existing infrastructure, scientific excellence, interdisciplinarity and international networks.
  - a. Conduct research into the molecular aetiology of diseases using genomic (human and metagenomic) and other omics datasets. Conduct research into the implementation of personalised medicine:
    - i. Establish and advance phenome-wide methods for personalised risk estimation and treatment, including for admixed individuals and populations, based on polygenic risk score models that consider

- gene-gene, phenotype, epidemiological and environmental influences and sex specific aspects.
- ii. Adopt and develop state-of-the-art machine learning and artificial intelligence applications and methods to deepen and broaden the scientific scope of genomic studies, accompanied with data mining from other data sources such as electronic health records and databases (including free-text fields and imaging data).
  - iii. Test and evaluate several models for returning results to biobank participants and monitoring their health behaviour.
  - iv. Conduct applied research into the development and implementation of personalised medicine services in Estonia.
  - v. Collaborate with clinicians to help focus on clinically relevant research problems and better understand the aetiology of both complex and genetic diseases and to broaden the use of (gen)omic data in clinical practice.
- b. Study the genesis of the genetic diversity of humankind through reconstructions of genetic drift and natural selection:
- i. Develop theory and methods to analyse large modern and ancient genomic and biomedical datasets and to understand the demographic history (migrations, admixture patterns, kinship, pathogen load) of humans, concentrating on the sex-specific aspects thereof and the role of natural selection and cultural and social background on population structure and health.
  - ii. Promote the integration of genetic, environmental (including paleo-climate) and cultural (including material culture and oral/written traditions) data to achieve an integrated understanding of human past.
  - iii. Develop analytical tools and generate data from populations of interest to clarify complex mixtures of ancestry, natural selection and other evolutionary processes.
  - iv. Expand the interdisciplinary research of ancient biomolecules by a) adopting new source materials for ancient DNA-based demographic inferences and b) widening the scope of the research towards ancient metagenomics and immunomics.
  - v. Quantify past genetic selection and study genetic, environmental and gene-environment interaction effects in complex traits.
- c. Functional studies of genetic variants that are computationally identified to contribute to phenotype or to have been under natural selection:
- i. Functional studies of key proteins of interest.
  - ii. Establishment of iPSC cell cultures to measure the functional consequences of genetic variation linking genotype and phenotype.
  - iii. Map causal genetic variation underpinning population differences in immune regulatory and metabolic networks.

- iv. Conduct multi-omic studies across multiple modalities, such as microbiome, metabolome, transcriptome, proteome and others.

## **2. Build capacity**

- a. Attract outstanding scientists who can lead internationally recognised research projects and attract financial support from international funding bodies.
- b. Promote the development of early career researchers with internationally recognised research and leadership skills through the provision of supportive infrastructure, high-quality training, dedicated peer support and career-building mentorship.
- c. Create an environment that encourages and supports the proposal of novel and out-of-the-box ideas and that promotes and increases interaction across the three main sections of IG and between research groups and senior and junior scientists.

### *Performance indicators*

1. The quality and quantity of scientific output measured bibliometrically is constant or increases.
2. The share of papers with IG researchers as leading authors increases.
3. Hire five outstanding senior researcher level scientists who have completed successful postdoctoral research internationally and attract independent funding.
4. Hire ten outstanding postdoc-level researchers who have defended a PhD internationally.
5. PhD students whose defence takes place in IG are successful in obtaining good postdoctoral opportunities or industry jobs.
6. Funding from international and domestic research grants is constant or increasing.

### *Teaching*

#### *Challenge*

The excellence in genomics in IG is currently not fully engaged in teaching activities in the University of Tartu. This situation is suboptimal considering a) students' access to state-of-the-art genomics education, b) the sustainability of genomics research in UT and c) the lack of visibility of IG research within the student community.

The role of human genomics and other omics in healthcare is rapidly increasing and societal need for human genomic-related knowledge and professionals is expanding. Specifically, there is a lack of genetic counsellors with experience in genetic risk scores as well as a lack in the interdisciplinary training of specialists in the fields of medicine, informatics and genomics in Estonia.

## *Objectives & Actions*

### **1. DEVELOPMENT OF BSC AND MSC MODULES IN GENOMICS**

- a. Contribute to the development and management of the faculty's BSc and MSc curricula, where IG researchers are involved in teaching.
  - b. Development of transdisciplinary studies within the framework of the Collegium of Transdisciplinary Studies of Archaeology, Genetics and Linguistics and the Centre for Personalised Medicine and in collaboration with various institutes at the University of Tartu. We wish to support the development and implementation of new curricula and modules within current curricula (e.g. genetic counselling, medical bioinformatics, digital life sciences, entrepreneurship and innovation transfer).
  - c. Encourage researchers/PhDs to actively recruit BSc/MSc students.
- 2. TEACHING AND SUPERVISING CAPABILITIES.** Develop the teaching and supervision skills of our research staff.
- a. Teaching is valued as an important pillar of IG's development.
  - b. Encourage and fund participation in training courses on problem-based learning and other innovative methods.
  - c. Encourage the participation of researchers in colleagues' lectures in order to achieve a more comprehensive feedback system.
- 3. COURSES IN PERSONALISED MEDICINE FOR CLINICIANS**
- a. Offer clinicians access to cutting-edge research and the possible implementation of personalised medicine for improved disease prevention and treatment.

## *Performance Indicators*

1. One to two modules in BSc and MSc curricula at the University of Tartu are managed by the Institute of Genomics.
2. 25% of Institute of Genomics research staff has taken training courses to improve teaching and supervision skills.
3. 500 medical doctors have taken additional training in genomics and personalised medicine co-organised by the Institute of Genomics.

## *Contribution to the Implementation of Personalised Medicine*

### *Challenge*

The Estonian biobank is one of the largest population biobanks in the world. Estonia, due to its size and e-governance, offers the perfect setting for the development of new and innovative personalised medicine solutions. Despite this, the implementation of personalised medicine has been slow.

## *Objectives & Actions*

1. **PERSONALISED MEDICINE IN ESTONIA.** Contribute to the synergistic development and implementation of personalised medicine in Estonia to ensure the large-scale use of genomic data in health care (in collaboration with relevant institutes of the University of Tartu and leading healthcare and public health institutions in Estonia).
  - a. Develop and test the implementation of algorithms for decision support software, including pharmacogenetic recommendations and disease risk models that incorporate polygenic risk scores, total genetic risk assessment and complex risk models.
  - b. Certify and validate existing Estonian Biobank genomic profile data for healthcare applications.
  - c. Contribute to the development of the joint Centre for Personalised Medicine at the University of Tartu & Tartu University Hospital.
  - d. Contribute to the development of treatment guidelines for incidental findings (ACMG list of genes) identified among biobank participants.
  - e. Advance the genetic literacy of the general public.
  
2. **PERSONALISED MEDICINE GLOBALLY.** Contribute to the development and best practices for the implementation of personalised medicine globally.
  - a. Play an active role in international organisations and consortia in the research and implementation of personalised medicine, including GA4GH (Global Alliance for Genomics and Health), ICDA (International Common Disease Alliance), CPIC (The Clinical Pharmacogenetics Implementation Consortium), 1+MG (1+ Million Genomes Initiative), ELIXIR (European Life sciences Infrastructure for biological Information), BBMRI-ERIC (Biobanking and BioMolecular resources Research Infrastructure – European Research Infrastructure Consortium), and MMHP – (Million Microbiome of Humans Project) and join other rising relevant initiatives.

## *Performance Indicators*

1. Centre for Personalised Medicine at the University of Tartu has been launched and funding secured.
2. High stakeholder satisfaction
3. Our contributions to global efforts and best practices are documented and recognised.

## *Development of the Estonian Biobank*

### *Challenge*

Biobanks have become an essential part of healthcare innovation. The Estonian Biobank was one of the first national biobanks in the world. However, our competitive advantage is challenged in the areas of policy, research, innovation and commercialisation. Running an internationally competitive biobank, conducting cutting-edge research and contributing to

innovation in health care is costly and requires more resources than existing funding enables. In order to continue internationally as a top-grade biobank, the database size, depth, quality, access and interoperability must be increased.

### *Objectives & Actions*

1. **VALUE OF BIOBANK.** Increase the value of the biobank to maintain the position among leading biobanks globally.
  - a. Organise, update and enrich the growing EstBB database with information from national health databases and registries.
  - b. Enrich the EstBB database with DNA sequence data (ultimately whole genomes for the whole biobank) and other omics data (biomarkers, proteomics, epigenetic data, metagenomics, etc.).
  - c. Expand the EstBB database, both in terms of the number of participants and the biological samples to be collected. Develop the longitudinal aspect of the database.
  - d. Increase the applicability of the EstBB database by implementing data quality, harmonisation, cross-validation, clinical systematisation process standards and automated pipelines.
  - e. Establish broader collaborations with medical service providers in Estonia to explore possibilities for increased inter-usability of population and clinical biobanks.
  - f. Evaluate the present state of and future need for biobank equipment and infrastructure and plan acquisitions accordingly.
  
2. **EASE OF ACCESS.** Increase the accessibility and interoperability of EstBB repository across stakeholder verticals, including academic, public and private organisations, to maintain the position among leading biobanks globally.
  - a. Establish and run a secure and privacy-preserving cloud computation platform based on EU data protection standards and data processing best practices according to market studies across EU countries.
  
3. **ENGAGEMENT OF BIOBANK PARTICIPANTS.** Engage biobank participants more actively with biobank activities ensure their continuous support towards and willingness to contribute in future research projects.
  - a. Design, launch and continue to innovate and operate the participant portal to return both general and personally relevant research results to participants.
  - b. Establish processes for the selection of topics included in the return of results to biobank participants and to aid with the planning of resources and best practices.
  - c. Develop a strategy for participant engagement.

4. **COLLABORATION WITH PRIVATE COMPANIES.** Become a valuable, professional and resourceful R&D partner and academic Clinical Research Organisation for the private sector in the fields of MedTech, HealthTech and other deep tech and data-driven businesses.
  - a. Trust and knowledge building (both internal and external advisory team) – build a professional team of experts to facilitate and support industry-standard R&D collaborations.
  - b. Develop a standard portfolio of products and services that will be attractive to private companies covering contract research work and the commercialisation of intellectual property.
  - c. Create an environment and competences to support product development in different stages of development, from start-ups to big pharma. Actively promote licensing deals for prototype genetic and complex risk models developed by researchers.
  - d. Build trust towards commercial collaboration among all stakeholders (biobank participants, state officials, politicians, healthcare workers).
  
5. **DATA ACCELERATOR.** Become the leading Data Accelerator globally and facilitate knowledge transfer, innovation acceleration and the development of products/services for international entrepreneurs.
  - a. Build an international network of investors, mentors, funds, entrepreneurs, accelerators, incubators and corporate ventures to increase EstBB database global visibility.
  - b. Generate a deal flow for R&D contracts and Data Accelerator and UniTartu Ventures.
  
6. **ORGANISATIONAL EVOLUTION.**
  - a. Conduct an analysis to assess the roles and expectations of the structural units of the Institute of Genomics – the Estonian Biobank and Estonian Biobank Innovation Centre.
  - b. Restructure the structural units according to the results of the analysis.

#### *Performance indicators*

1. The results of the analysis of the roles of current structural units are implemented.
2. Steady annual increase of biobank participants while maintaining high public trust.
3. Maintain high partner satisfaction with access to the biobank repository.
4. Steady influx of contracts for generating sequencing and other molecular profiling data.
5. Increase private sector contract volume to 25% of GI's annual budget to support infrastructure modernisation and innovative research at the Institute.
6. Data accelerator portfolio of 25 innovative enterprises.

7. High level of participation in recall studies.

## *Core facility services*

### *Challenge*

1. Maintaining DNA/RNA sequencing technological excellence and integrating emerging innovation. Achieving industrial gold standards in managing, running and delivering services both in-house and commercially.
2. The commercial service strategy needs to be updated/targeted in order to increase competitiveness and service volume.
3. The laboratory information and quality management systems need to be developed/upgraded.

### *Objectives & Actions*

1. **INFRASTRUCTURE.** Significantly upgrade technological capability and excellence and develop a sustainability plan to support this.
  - a. Up-to-date/state-of-the-art high throughput sequencing technology
  - b. Up-to-date Sanger sequencing technology
  - c. Test and establish capabilities based on emerging sequencing technologies
2. **MARKETING.** Develop a marketing strategy for the core facility (advising clients, building a loyal customer base, etc.) and introduce new services.
  - a. Customer support in experimental design, analytical pipelines and access to data processing platform
  - b. Building and expanding a loyal customer base
  - c. Integration of ancient DNA, genetic ancestry feedback and metagenomic service into the Core facility
3. **QUALITY MANAGEMENT.** Update the laboratory information and quality management system.
  - a. Implement LIMS & data processing services
  - b. Implement ISO

### *Performance Indicators*

1. New DNA sequencing infrastructure is installed and operating in accordance with the sustainability plan.
2. The Core facility's departments have been fully integrated, including ancient DNA service.
3. Laboratory information and quality management systems have been implemented.
4. Annual increase in service volume, processing speed and product offering.
5. High customer satisfaction

## *People & working environment*

### *Challenge*

Our people are our main asset. Only content and satisfied people can sustain the challenges and demands of excellence in international science, teaching, maintaining and growing a client base, promoting substantial changes to the national healthcare system and supporting the younger generation of scientists. The Institute of Genomics strives to be an international, inclusive, environmentally conscious organisation supporting the development of personalities and a work-life balance and promoting a healthy lifestyle. The Institute of Genomics is an attractive workplace for researchers from Estonia and abroad with a safe and invigorating work culture and environment.

### *Objectives & Actions*

1. **PEOPLE.** The employees of IG feel valued, and the organisational culture is supportive of both personal and professional development.
  - a. Create a mentorship programme to support academic and entrepreneurial career models. Monitor personal development and workload through developmental interviews of staff.
  - b. Create a family-friendly organisation. We aim to support researchers, including during parental leave, to participate, in different forms, in seminars and scientific meetings with other researchers and supervisors. Part-time and flexible work solutions for parents with young children will be offered.
  - c. Organise regular team-building events, retreats and scientific writing courses both at the institute and working group level.
  - d. Support and promote mental and physical wellbeing of staff.
  - e. Support and value creativity by offering opportunities to engage with music, arts, etc. on Institute premises. We strive to acknowledge the efforts of our students and employees in these fields.
  - f. Promote moderate exercise in the workplace and reimburse sporting and rehabilitation activities for employees. Meeting while taking a walk should be encouraged.
  - g. Increase internal communication within IG to better inform people of the topics discussed and decisions made in executive meetings.
  
2. **WORK ENVIRONMENT.** IG strives to offer a modern working environment that suits the needs of employees.
  - a. Manage office space usage more creatively by encouraging working from home and using shared/scheduled workspaces at the Institute. Encourage digital meeting platforms and walking meetings.

- b. Contribute to climate goals by optimising the energy consumption involved in data storage and computation (through more efficient methods), optimising consumables (e.g. lab plastic), promoting waste sorting, etc.
- c. Eventual expansion of the premises of the Institute of Genomics.

*Performance Indicators*

- 1. High employee satisfaction monitored via annual questionnaire.
- 2. Mentorship programme implemented with high mentee satisfaction.
- 3. People use most of their annual leave; working during vacation and sickness is discouraged.