ICPerMed
INTERNATIONAL CONSORTIUM

State of the Art Report 2022

Imprint

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Contents

Executive Summary

The International Consortium for Personalised Medicine publishes a state-of-the-art report on an annual basis and seeks to facilitate the follow-up of the development of personalised medicine research and its implementation throughout Europe and beyond.

ICPerMed is continuously mapping activities in the field of personalised medicine to promote achievements and successes in research and implementation as well as to honour activities of ICPerMed member organisations. A set of initiatives, activities and infrastructures supporting PM development and implementation are presented in this 2022 report.

Furthermore, in this 2022 report, ICPerMed outlines shortly its activities of the past year 2022 (conference, workshop, and education and training activities, presentation of Best Practices) as well as the ongoing preparations for the European Partnership for Personalised Medicine (EP PerMed) proposal and the supporting Strategic Research and Innovation Agenda for Personalised Medicine (SRIA for PM).

State of the Art Report 2022

1. International Consortium for Personalised Medicine

The International Consortium for Personalised Medicine (ICPerMed) serves since 2016 as a strategic platform, bringing together high-level policy makers such as research and innovation ministries, health ministries, and regional authorities as well as European regional and national funding organisations, to foster a common understanding of personalised medicine and the defragmentation of the personalised medicine environment by aligning strategies in research funding and at the policy level. ICPerMed initiates and drives strategic reflections and the development of strategic documents, i.e. the ICPerMed Action Plan in 2017 and the Vision Paper in 2019.

The ICPerMed consortium warmly welcomes two new members:

- The South African Medical Research Council (SAMRC)[1], and
- Genome Canada[2].

Therewith, ICPerMed currently brings together 49 public and private 'not-for-profit' health research funding and policy organisations from 30 countries, seven European regions and five continents. The European Commission (EC) participates in ICPerMed as observer.

ICPerMed provides a platform to initiate and support communication and exchange on personalised medicine research, funding and implementation. To foster these exchanges, the following internal meetings, public events and activities took place between November 2021 and October 2022:

- **ICPerMed Executive Committee meetings:** The ICPerMed member and observer organisations met on November 29-30, 2021 (online meeting), on May 10-11, 2022 (online meeting) and on October 7, 2022 (Paris, hybrid meeting) to discuss and plan strategy and future activities.
- **The 1st ICPerMed Family meeting:** The "ICPerMed Family Meeting – Joining Forces" took place as online event on November 9-10, 2021. The aim of the meeting was to foster collaboration of the involved initiatives and to collect input on future research and implementation approaches. The meeting report is available on the ICPerMed website.[3]
- **Biannual ICPerMed events:**
  - ICPerMed Workshop "Personalised Medicine: How to Ensure Value-based Implementation", on invitation only (hybrid event), in Brussels on June 21-22, 2022 - With this workshop, ICPerMed exchanged and find agreements on the mechanisms and key aspects of value-based cross cutting effects to guide an appropriate implementation of personalised medicine for the benefits of patients and citizens. The workshop report[4] is available on the ICPerMed webpage.
  - ICPerMed Conference "Prelude to the Future of Medicine" (hybrid event) on October 5-6, 2022, in Paris – Four keynote lectures, offered a focus on promising examples of targeted treatments or a personalised approach for diagnosis. Five sessions addressed crucial aspects for the successful implementation of personalised medicine. The video recordings[5] and the report[6] of the ICPerMed conference are available on the ICPerMed website.

ICPerMed representatives also participated in numerous external meetings to present the work and activities of ICPerMed, the so called "ICPerMed Family", but also preparations regarding the upcoming EP PerMed proposal.

Training courses and summer schools fostering exchanges with and the active involvement of the personalised medicine community:

- 1st ICPerMed Training Event "Personalised Communication in Personalised Medicine" took place on June 17, 2022 (online event) - New and comprehensive strategies around PM communications: Personalised communication could be defined as "the right message for the right citizen at the

1 https://www.samrc.ac.za/
2 https://genomecanada.ca/
3 https://www.icpermed.eu/en/905.php
4 https://www.icpermed.eu/en/ICPerMed-Workshop-2022-946.php
5 https://www.icpermed.eu/en/ICPerMed_Conference_2022.html
6 https://www.icpermed.eu/media/content/ICPerMed_Conference_Short_Report.pdf

right time"[7]. The training offered healthcare professionals and researchers the opportunity to learn new skills to communicate about PM approaches available in a more personalised way, especially in the crowded informational environment marked by incomplete scientific reporting, fake news, conspiracies, infodemic[8] and disinformation.

- 2nd ICPerMed Training Event "Personalised Medicine Research: Ethical and Legal aspects of Biobanking and Data Management" took place on October 10, 2022 (online event) – The course provided training to health professionals and personnel dedicated to biomedical, technological and scientific research to update their knowledge regarding data management and the establishment and maintenance of biobanks.

New examples of best practices[9] in personalised medicine are presented and promoted on the ICPerMed website and through the **ICPerMed Recognition**[10]**:**

- Best Practice Examples:
  - *"Implementation of Personalised Medicine in Estonia"* – A 5-year programme for the Implementation of Personalised Medicine in Estonia was launched in 2019. Its main goal is to create the fundament to employ genetic data more extensively in clinical practice, e.g. for personalised genetic risk-based prevention.
  - *"The Ubiquitous Pharmacogenomics (U-PGx) Education Programme"* – A "serious game" was developed in the context of the European project U-PGx. It offers a more intuitive and practical training of medical students in recognising adverse drug effects that may be caused by pharmacogenomics.
  - *"Centers for Personalized Medicine (ZPM)"* – Four university hospitals in the German region of Baden-Wuerttemberg joined forces to offer access to state-of-the art personalised medicine approaches to all patients in the region, starting in the oncology field and expanding to further disease indications.
  - *"Swiss Personalized Health Network (SPHN)"* – Coordinated infrastructures to make health data interoperable and shareable for research in Switzerland and to establish a data-driven health ecosystem. Such an ecosystem is key for research in personalised medicine, and, eventually, for personalised clinical applications, such as more precise diagnostic, therapeutic, and prevention strategies.
- ICPerMed Recognition 2021 winners – congratulations![11]
  - *"Alterations in serum glutamate levels are presented as a new risk factor in Metabolic associated fatty liver Disease"*, Jorge Simon, Spain
  - *"PHARMANAGEN"*, Jose Beloqui, Spain
  - *"A higher platelet-to-lymphocyte ratio is prevalent in the presence of circulating tumor microemboli and is a potential prognostic factor for non-metastatic colon cancer"*, Ludmilla Thomé Domingos Chinen, Brazil

The latest ICPerMed Recognition 2022 was launched in April 2022 and the winners will be presented in the next ICPerMed state-of-the-art report 2023.

NEW ICPerMed Vice-Chair elected

ICPerMed is glad to announce that Gianni D'Errico (representing the Tuscany Region, Italy) was elected Vice-Chair of the ICPerMed Executive Committee.[12] He will succeed Astrid Vicente, whom ICPerMed thanks her for their valuable and enthusiastic support.

Elected ICPerMed Chair: Ejner Moltzen (Innovation Fund Denmark)

Elected ICPerMed Vice-Chair: Hemma Bauer (Federal Ministry of Science, Research and Economy, Austria)

7 https://www.icpermed.eu/en/icpermed-training-june-17-2022-personalised-communication-medicine-944.php

8 "An infodemic is too much information including false or misleading information in digital and physical environments during a disease outbreak. It causes confusion and risk-taking behaviours that can harm health. It also leads to mistrust in health authorities and undermines the public health response". World Health Organisation

9 https://www.icpermed.eu/en/best_practice_examples.php

10 https://www.icpermed.eu/en/icpermed-academy.php

11 https://www.icpermed.eu/en/887.php

12 https://www.icpermed.eu/en/icpermed-governance.php

2. European Partnership for Personalised Medicine – EP PerMed

Within the European Union's 9th Framework Programme for Research and Innovation, Horizon Europe, the set-up of a co-funded European Partnership for Personalised Medicine (EP PerMed) is proposed. This partnership will promote European regional and national priority setting and support the alignment and funding for research, innovation and implementation projects in all areas of personalised medicine between the EU Member States, European regions and Associated Countries to Horizon Europe as well as international partner countries.

The EP PerMed development stages:

1. First preparations started in 2020, with a preparatory group of representatives from ICPerMed and ERA PerMed members that developed a concept paper, guidance documents and an information day (see also "The ICPerMed – State of the Art Report (2022)"). Finally, personalised medicine was supported by the Member States and validated as topic for a co-funded European Partnership under Horizon Europe.

2. In 2021, a Drafting Group was formed on Member States level that developed two important documents: (1) A Draft Proposal that outlined a first framing of the EP PerMed and that was published by the EC in February 2022[13], and (2) the Strategic Research and Innovation Agenda for Personalised Medicine to be published in spring 2023. The so called SRIA for PM will provide guidance to a wide range of stakeholders and experts to further develop programmes, activities, and research towards personalised medicine and care, as well as prevention. Its main intention is of course to draw a roadmap supporting the planned activities of the European Partnership for Personalised Medicine.

3. In 2022, a Writing Group was established in anticipation of the respective EC call for applications expected to be published in December 2022. The Writing Group already represented the EP PerMed consortium core group and started the EP PerMed proposal development, in parallel of the SRIA development coordinated by the Drafting Group. The proposal will be submitted to the EC in April 2023 and will be complemented by the SRIA for PM published also on the ICPerMed webpage latest by the end of April 2023.

ICPerMed highly supports the upcoming EP PerMed by fostering the reflections towards and providing content-driven input for the partnership and it is likely that the majority of members will also join the partnership proposal and consortium. In 2022, the ICPerMed Secretariat facilitated the work of the Drafting Group as well as Writing Group, e.g. by providing technical support, developing visuals related to the EP PerMed or reflections concerning impact pathways. Currently, the ICPerMed Secretariat is organising the next ICPerMed Workshop "Preparing the Future for Personalised Medicine: EP PerMed", on invitation only and taking place in Pamplona on January 17-18, 2023, that will be an important element in the finalisation of the SRIA for PM.

The next important steps in the preparation of the partnership are:

- November/December 2022: Launch of an open consultation in support of the SRIA for PM. Opportunity for all stakeholders to participate and contribute to future activities and strategies in personalised medicine.
- January 17-18, 2023: The ICPerMed Workshop "Preparing the Future for Personalised Medicine: EP PerMed", on invitation only, in Pamplona.
- Spring 2023: Publication of the Strategic Research and Innovation Agenda for Personalised Medicine.
- April 2023: Submission of the EP PerMed proposal to the EC.

If positively evaluated, the EP PerMed consortium will support personalised medicine research, innovation and implementation as well as overarching activities starting from the end of 2023. A first joint transnational call to support research in

13 https://www.icpermed.eu/en/ep-permed.php

the field of personalised medicine is expected to be launched already in 2024.

Contact details to reaching out to the EP PerMed Drafting and Writing Group for any question around the EP PerMed: eppermed@dlr.de.

ICPerMed is looking forward to the upcoming European Partnership and contributes to the development at every stage to support the process and establish a continuous collaboration with the Drafting and Writing Groups as well as with the future Partnership.

3. Snapshot: Initiatives in the field of personalised medicine

ICPerMed is continuously mapping activities and advancements in the field of personalised medicine in order to promote achievements and successes in the field as well as to honour activities of ICPerMed member organisations. A set of significant personalised medicine related or supporting initiatives is presented in this section:

I. Genomic Initiatives (pages 9-12)

a. Danish National Genome Center (Denmark)
b. Norwegian initiatives for precision cancer medicine (Norway)
c. The Pan-Canadian Genomics Strategy (Canada)
d. The Slovenian Genome Project (Slovenia)
e. MedeA (Extremadura, Spain)

II. Driving innovations (pages 12-13)

a. The Danish Life Science Cluster (Denmark)
b. MEDVIA (Flanders, Belgium)
c. The Innosasun Programme and the Medtech Initiative (Basque Country, Spain)

III. Infrastructures (pages 13-15)

a. The Eric Kandel Institute - Centre for Precision Medicine (Austria)
b. MIDGAM (Israel)
c. Basque Biobank (Basque Country, Spain)

IV. Disease-specific initiatives (pages 15)

a. PreciDIAB – The National Center for Precision Diabetic Medicine (France)
b. IMPERA – Precision Medicine in Respiratory and Allergic Diseases Initiative (Spain)

V. International Collaborations initiatives (pages 16-17)

a. Human Cell Atlas
b. The European Genome-phenome Archive
c. The International Cancer Genome Consortium

Genomic Initiatives

Danish National Genome Centre (Denmark)

The Danish National Genome Centre (NGC), a government agency and authority within the Danish healthcare system, was established in 2019 to implement the Danish Government's National Personalised Medicine Strategy. The purpose of the Danish NGC is to support the development of personalised medicine to the benefit of patients in collaboration with the regions and the entire Danish healthcare system, research institutions, patient associations etc.

The Danish government and Danish regions have updated their strategy for personalised medicine in the healthcare system for the period of 2021-2022. The focus of the strategy will be on patient and clinical needs. During the first phase of the strategy, the Danish NGC and its collaborators will recruit and sequence whole genomes of 60,000 patients diagnosed with cancer, autoimmune disorders and rare diseases by 2024.

In the short term, the centre will develop and operate a joint, national infrastructure to ensure that doctors throughout the country have access to whole genome sequencing and storage of information in a National Genome Database. The Danish NGC will also develop during a second phase a national research infrastructure for personalised medicine consisting a National Whole Genome Sequencing Centre and a National High-Performance Computing Centre (supercomputer system) enabling research projects and clinical activities. Lastly, the Centre will develop further personalised medicine by including additional data sources.

Source: https://eng.ngc.dk/

Norwegian initiatives for precision cancer medicine (Norway)

Three initiatives in Norway are working to transform the way healthcare is delivered, by providing more effective and personalised approaches to disease management: InPreD, IMPRESS, and INSIGHT.

InPreD (Infrastructure for Precision Diagnostics) is a national infrastructure for advanced molecular diagnostics that aims to facilitate clinical cancer trials on a national level by providing

equal access for patients to advanced diagnostics, state-of-the-art competence, and technology. By doing so, InPreD aims to give patients access to medicines they otherwise would not receive, and to increase the precision medicine experience of clinicians and researchers nationwide.

IMPRESS (Improving public cancer care by implementing precision medicine in Norway) is a prospective, non-randomised clinical trial evaluating the efficacy of commercially available anti-cancer drugs prescribed for patients with advanced cancer and diagnosed with potentially actionable alterations revealed by standardised molecular diagnostics. IMPRESS-Norway is a nationwide study, and all hospitals with an oncology or haematology department are invited to participate. The study aims to generate data and insights important for analysing the outcomes and adopting health technology assessments and reimbursement schemes for these novel, personalised treatment concepts.

INSIGHT (Regulatory framework for implementing precision medicine into the Norwegian healthcare system) aims to develop an analytic framework for using synthetic control data to evaluate the effects of small-scale, one-armed clinical trials, such as those conducted in IMPRESS-Norway. It will use the developed control arms and data from IMPRESS-Norway and InPreD to evaluate the cost-effectiveness of the precision medicine model and suggest a new reimbursement scheme that reflects the uncertainty in precision medicine.

Overall, these initiatives in Norway are working towards the common goals of increasing access to precision medicine for patients, improving the precision medicine experience of clinicians and researchers, and generating data and insights for evaluating and implementing precision medicine in the healthcare system.

Sources:

https://oslocancercluster.no/tag/inpred/

https://oslocancercluster.no/connect/

https://impress-norway.no/en/impress-norway-front-page/

The Pan-Canadian Genomics Strategy (Canada)

Over the past 20 years, Canada has established itself as a leader in genomics research, thanks in part to its strong research ecosystem, well-regarded public healthcare system, diverse ecosystems, and abundant natural resources and agri-food sectors. The Canadian government has also supported genomics research and development (R&D) at all stages of the innovation process, from basic research to commercialisation, through a variety of initiatives.

However, to bridge the gap between research and commercialisation in the genomics field, Canada must take advantage of the global interest in genomics R&D and leverage its own research strengths to drive the implementation of genomics technologies and innovations.

To that end, the Canadian government has announced the $400-million Pan-Canadian Genomics Strategy (PCGS) in the 2021 budget. The PCGS is intended to advance the commercialisation and adoption of genomics and related technologies, strengthen Canada's global leadership, and position Canada for long-term success in the global bio economy, while also promoting coherence and coordination among key players.

To refine the goals of the PCGS, the Canadian government launched in 2022 a consultation from stakeholders on the proposed themes of the strategy, as well as potential actions and interventions to address the opportunities and challenges facing the Canadian genomics ecosystem. The five proposed themes of the PCGS are:

- Canada's genomics landscape: opportunities and challenges;
- Attracting, developing, and retaining talent;
- Management, standardisation, and use of genomics data;
- Commercialisation and use of Canadian genomics technology and innovations;
- Adoption of genomics in key sectors: healthcare, environment, clean tech, food, and natural resources.

To unite these efforts, and provide a single-entry point for all Canadian genomes (clinical and research), a pan-Canadian

Genomic Library is being launched. The development of this national asset will enable all human sequencing efforts to be united for the benefit of all the people living in Canada. The objectives of the pan-Canadian Genomic Library are to:

- Develop a federated pan-Canadian framework that will enable access to and analysis of genomes and associated health/environmental information, governed in a transparent, accountable and consistent way;
- Support equity, diversity and inclusion by capturing the Canadian genomic "variome" to truly represent the richness of the genomic diversity of people living in Canada, and in all its diversity by promoting the inclusion of detailed information on key identity characteristics, such as sex, gender, and other intersecting identity factors, that shape the health;
- Position the framework as a key mechanism to improve effectiveness, both from an economic and health perspective, of healthcare delivery in a learning health system context;
- Provide means and opportunities to facilitate clinical trials based on genomic medicine;
- Position Canada as a key player in international genomic research endeavours.

Sources:

https://ised-isde.canada.ca/site/genomics/en/consultation-paper-developing-pan-canadian-genomics-strategy

https://cihr-irsc.gc.ca/e/53218.html

The Slovenian Genome Project (Slovenia)

The Slovenian Genome Project (SGP) is a ground-breaking initiative in Slovenia that aims to sequence the entire genome of its citizens improving diagnosis of rare diseases, as well as making it easier to distinguish between the changes that cause the disease and the changes that are part of the natural genetic diversity. It will also facilitate the discovery of new genes and mechanisms of genetic diseases and the development of predictive models for common polygenic diseases such as cardiovascular disease, diabetes, predisposition to cancer and other diseases. By collaborating with top research and medical institutions, the SGP will develop a bioinformatics platform containing the normal and pathological genetic variability of the Slovenian population. This platform will be used to improve the diagnosis and prevention of rare diseases through the implementation of genome sequencing, as well as to establish a personalised genetic approach to predicting, preventing, and treating common multifactorial diseases and cancers.

The use of new genomic technologies, such as whole-genome sequencing, allows for the affordable and fast analysis of the entire human genome, which can be applied in the diagnosis of diseases as well as in the study of normal genetic variability, or the biological inheritance of a population. While genomic projects have previously focused on populations in the USA, Western and Northern Europe, and some populations in Africa and Asia, there have not yet been any systematic genomic studies using whole-genome sequencing for the Slovenian population.

The SGP will not only accelerate our understanding of the epidemiology of genetic diseases in Slovenia, but it will also standardise the transfer and gathering of data between Slovenian institutions (adhering to the FAIR principles: "Findable, Accessible, Interoperable, and Reusable") and facilitate collaboration with European genome initiatives, including the "1+ Million Genomes" initiative. Overall, the SGP has the potential to greatly improve the field of genomic medicine in Slovenia and enhance the health of its citizens.

Source: http://genom.si/en/index.html

MedeA (Extremadura, Spain)

The Clinical Implementation of Personalised Medicine in Health Services (MedeA) is a health innovation programme that aims to improve patient care by promoting individualised, patient-centred drug prescription. To achieve this goal, the project aims to integrate genetic analysis with other relevant data on a patient's response to drugs, such as their medical history, analytical data, and other relevant factors. This information will be used to develop information and communication technology (ICT) support tools that can assist healthcare professionals in making informed decisions about drug prescriptions.

One of the key components of the MedeA project is the development of a personalised prescription system that includes patient information and utilises a decision algorithm to provide the best possible treatment options. This system will also be applied to the clinical research process, allowing for the development of individualised selection strategies that minimise unnecessary risks and increase the effectiveness of studies.

At the conclusion of the project, a validated personalised prescription system will be in place within the Extremadura Health System, with connection to the electronic prescription system. To achieve this goal, the project involves the development of business innovation in four key areas, known as technological challenges.

Overall, MedeA is a project that seeks to optimise drug prescription for patients through the use of personalised medicine, combining genetic information with other relevant data to provide the best possible treatment options. By involving private companies in the development of this programme through the use of Innovative Public Procurement and an open innovation model, the project aims to promote research, development, and innovation in the healthcare industry.

Source: https://www.proyectomedea.es/en/home/

Driving innovations

The Danish Life Science Cluster (Denmark)

The Danish Life Science Cluster is a national organisation that aims to connect and drive innovation within the life science and welfare technology sectors in Denmark. The organisation works to convert Danish research and knowledge in these areas into new commercial solutions that benefit businesses, healthcare systems, municipalities, and citizens in Denmark.

Life science and welfare technology are significant areas of strength in Denmark, with skilled companies, a strong healthcare system, and internationally renowned research environments. The sector plays a significant role in the Danish economy and is supported by close partnerships between public and private organisations.

Through knowledge sharing and collaboration, the Danish Life Science Cluster helps to convert the most recent research and healthcare needs into new competitive solutions that generate value for companies, patients, municipalities, and the healthcare system. The organisation works to build bridges across sectors and in collaboration with companies, academic institutions, the healthcare system, and municipalities.

The Danish Life Science Cluster is designated as the national cluster for life science and welfare technology by the Danish Board of Business Development and receives funding from the Danish Ministry of Higher Education and Science, the Danish Business Authority, and other sources such as the Ministry of Industry, Business and Financial Affairs, regional cluster organisations, and membership fees. The organisation has a high ambition to compete with the best clusters in the world in the development of future healthcare solutions.

Source: https://www.danishlifesciencecluster.dk/en/

MEDVIA (Flanders, Belgium)

MEDVIA is a public-private partnership based in Flanders, Belgium that aims to stimulate innovation in the healthcare sector. It does this by facilitating and supporting collaborations between various stakeholders in the healthcare ecosystem, including companies, hospitals, universities, and other healthcare institutions. These collaborations focus on addressing unmet medical needs and creating solutions for better health at the crossroads of medical biotechnology, medical technology, and digital technology.

In addition to facilitating these collaborations, MEDVIA also supports the growth and development of digital health innovators and medtech innovators in the region. It does this by providing resources and support to help these innovators overcome challenges such as regulation, finance, and go-to-market issues. MEDVIA also works to bring all stakeholders together, including clinicians, technicians, entrepreneurs, and patients, to address these challenges and promote the development and adoption of new technologies.

One of the main goals of MEDVIA is to become the leading ecosystem in Europe for better health, bringing together

biotech, medtech, and digital technologies. It also aims to build a competitive network with strong economic potential for Flanders and create a pool of highly skilled employees. To achieve these goals, MEDVIA is working to develop a strategic healthcare roadmap for the region and provide tailored services to its members in a collaborative approach.

Overall, MEDVIA is committed to improving patient health and wellbeing through the development and adoption of innovative technologies in the healthcare sector. It does this by bringing together various stakeholders in the ecosystem and providing resources and support to help them overcome challenges and achieve success.

Source: https://medvia.be/about-medvia/

The Innosasun Programme and the Medtech Initiative (Basque Country, Spain)

The Basque Government's Ministry of Health created the Innosasun Programme in 2014 to support the development and validation of health technologies by companies and other parties. The main activities of the programme are counselling and guidance to companies and other agents on the specifications and requirements of their health technologies, as well as conducting pre-clinical and clinical studies for the development and validation of these technologies.

Through the Innosasun Programme, the healthcare system acts as a test bench or living lab in a real-world environment, where medical devices and technologies can be demonstrated, validated, and studied for cost effectiveness. This allows third parties, particularly small medium enterprises (SMEs), to overcome one of the main barriers to bringing their technologies to market, which is of high costs in clinical studies.

In 2018, the Ministry of Health of the Basque Government created the Medtech Initiative to economically support the collaboration of the Basque healthcare system with companies in the frame of Innosasun Programme for the development of health technologies that improve health outcomes and generate value. The Medtech Initiative is part of the RIS3 Euskadi strategy and the Health Research and Innovation Strategy 2020 (EIIS 2020) and aims to promote collaboration between the healthcare system and the Basque business and science and technology sectors.

Both, the Innosasun Programme and the Medtech Initiative, have been valuable to facilitate the collaboration between the healthcare system and companies and have helped accelerate the development and validation of technologies. Based on their success, these programmes could potentially be used as a model for other health systems looking to support the entrepreneurial health sector. The EIIS 2020 includes the development of open innovation programmes like Innosasun in its first Strategic Axis (Impact) and considers Innosasun one of the "Initiative Strategies" of the Biosciences-Health area of RIS3. The Innosasun Programme, coordinated by BIOEF and involving the entire healthcare system, offers personalised support to companies and science and technology agents in the development of products and services with potential applications in health. It aims to bring together the scientific, technological, and business capabilities of the Basque Country for the benefit of health, the improvement of the healthcare system itself, and the generation of wealth.

Sources:

https://www.interregeurope.eu/good-practices/the-basque-public-health-system-programmes-which-articulate-the-interaction-with-the-business-sector-innosasun-and-medtech-0

https://www.biocrucesbizkaia.org/en/web/guest/servicios/medtech

Infrastructures

The Eric Kandel Institute - Centre for Precision Medicine (Austria)

Funds from the European Resilience and Recovery Facility and donations are being used to create a centre for precision medicine at MedUni Campus AKH in Vienna. The new research infrastructure creates modern framework conditions for digital and personalised medicine. In future, diagnoses, therapies

and preventive measures adapted to individual factors will be developed here. The new research centre is named after Eric Kandel, a Nobel Prize winner from Vienna.

The Eric Kandel Institute - Centre for Precision Medicine is to become one of the leading centres for research and development of therapies in the field of customised medicine. A new building is being constructed as part of one of the largest investment projects at MedUni Campus AKH and as one of three centres shaping 21st century medicine: The Centre for Translational Medicine and Therapies, the Centre for Technology Transfer and the Eric Kandel Institute - Centre for Precision Medicine.

The focus of the new Eric Kandel Institute is particularly on biomedical research, clinical trials, genome technology, bioinformatics and Information Technology. The immediate vicinity to the AKH brings a significant advantage for patients: Clinicians and basic researchers work closely together and in close proximity to each other to develop and use the latest findings, enabling patients to be treated at the cutting edge of medicine.

Source: https://www.meduniwien.ac.at/web/en/forschung/eric-kandel-institut-fuer-praezisionsmedizin/

MIDGAM (Israel)

The Israeli National Biobank for Research, also known as MIDGAM, is a decentralised biobank that serves as a platform to support research aimed at improving patient care options and developing diagnostic tools and innovative drugs and therapies for personalised medicine. MIDGAM operates as a one-stop shop, offering researchers all the assistance they need and encouraging collaborations between medical centres and researchers from academia and industry in Israel and abroad.

MIDGAM has five collection centres located in leading medical centres in Israel, as well as a management office at the Weizmann Institute of Science. The collection centres are responsible for recruiting donors, collecting, processing, and storing human bio samples with annotated data. The management office maintains a central database and is responsible for relationships with researchers, establishing and updating standard operating procedures, maintaining quality control, regulatory issues, and finance.

Since 2014, MIDGAM has collected nearly 200,000 bio samples from around 20,000 donors, most of whom are oncology patients. The demand of bio samples for research is constantly increasing, and the bio samples and data collected by MIDGAM are currently being used in numerous studies in industry, academia, and medical centres in Israel and around the world.

Source: https://www.midgam.org.il/about-2/?lang=en

Basque Biobank (Basque Country, Spain)

The Basque Biobank is a hospital-based platform that aims to support basic and clinical research that may lead to the development of tools for the prevention, diagnosis, and treatment of various diseases. It is operated by the Basque Foundation for Health Innovation and Research (BIOEF) and has a network structure that involves all Osakidetza health centres, the Basque government's Department of Health, and Onkologikoa. This network organisation allows for standardisation of procedures and makes it easier to trace and follow-up with patients in the Basque health system.

After obtaining informed consent, the Biobank collects a variety of samples from patients, including peripheral blood, DNA, RNA, plasma, serum, cells, cerebrospinal fluid, saliva, tears, tumour tissues, and tissues obtained from autopsies. These samples are collected from a broad range of diseases, including cardiovascular disease, diabetes, metabolic disorders, respiratory diseases, cancer, neuromuscular diseases, mental illnesses, and rare diseases.

The research goals of the Basque Biobank include 1) creating a collection of samples from healthy individuals and treated patients with longitudinal follow-up, 2) ensuring the association of samples with accurate and relevant diagnoses to promote quality research, 3) providing methodological, legal, or technological support to hospital departments conducting

research projects, 4) promoting biotechnological research through collaboration forums, 5) maximising resources by sharing infrastructures, 6) complying with current legislation on sample transfer, 7) ensuring sample quality, quantity, and representativeness for common diseases, and 8) facilitating knowledge transfer.

Overall, the Basque Biobank is a valuable resource for researchers working on a variety of diseases, as it provides access to a wide range of samples and supports the development of innovative tools for the prevention, diagnosis, and treatment of these conditions.

Source: https://www.biobancovasco.org/en/

Disease-specific initiatives

PreciDIAB – The National Center for Precision Diabetic Medicine (France)

PreciDIAB, the National Center for Precision Diabetic Medicine, is an organisation that aims to find new solutions to prevent and treat all types of diabetes and their complications. The organisation combines translational and clinical approaches to research and develop concrete, scientifically proven solutions. The knowledge gained from this research is shared with future generations of researchers and healthcare professionals, and may also lead to the development of innovations through public-private partnerships.

PreciDIAB brings together multidisciplinary specialists, researchers, and clinicians who share a common goal: to develop new methods for preventing, treating, and managing diabetes and its consequences, and to offer diabetic patients a quality of life and life expectancy that is equal to that of non-diabetics.

One of the programmes at PreciDIAB focuses on preventing the incidence of diabetes and obesity in children, as well as their co-morbidities, by targeting those at highest risk in the general population and those who are most vulnerable among diabetic patients. The overall objective of this programme is to develop personalised medicine that is based on stratifying patients according to advanced markers such as biomarkers and lifestyle markers. By targeting prevention and treatment efforts at those most at risk, PreciDIAB hopes to make a significant impact on the lives of diabetic patients.

Source: https://www.precidiab.org/en/

IMPERA – Precision Medicine in Respiratory and Allergic Diseases Initiative (Spain)

IMPERA is a community of health professionals dedicated to improving the lives of individuals with respiratory and allergic disorders through research and the dissemination of scientific medical knowledge. The organisation recognises that the traditional "one-size-fits-all" approach to treating respiratory and allergic diseases is inadequate, particularly in the case of complex, heterogeneous diseases that can differ from person to person in terms of triggers, mediators, inflammation, and treatment response.

To address these challenges, IMPERA advocates for a personalised medicine approach that considers the unique genetic, pharmacological, physiological, and immunological factors that can impact treatment response. This approach aims to produce knowledge, encourage continuous training, and promote the democratic circulation of knowledge in the field of respiratory and allergic disease treatment.

In addition to its focus on research and education, IMPERA is committed to ethical principles and the right of health for individuals suffering from respiratory and allergic diseases. The organisation works to train reflective, critical, and qualified professionals who are open to dialogue and service, and who are willing to establish partnerships with pharmaceutical companies, official institutions, and community organisations at the local, national, and regional level.

Source: http://www.iniciativa-impera.org/index.php

International Collaborations

Human Cell Atlas

The Human Cell Atlas (HCA) is an international collaborative consortium that aims to create a comprehensive map of all the cells in the human body. This is being done by collecting data from a wide range of tissues and organs using a variety of technologies, including single-cell genomics, computational techniques and microscopy.

The data collected from these technologies is being used to create a detailed map of the human body at the cellular level. This includes identifying the different types of cells that exist in the body, how they are organised and arranged within tissues and organs, and how they interact with each other.

The goal of the HCA project is to provide a resource for researchers to better understand the function and behaviour of individual cell types, and how they work together to maintain health and disease. It is expected to have a number of applications in the fields of biology and medicine, including helping to identify the cellular changes that occur during the development of diseases such as cancer, and helping to develop new treatments and therapies.

One way the HCA could contribute to personalised medicine is by providing insights into how cells respond to different stimuli and how they communicate with one another. This information could be used to tailor treatment strategies to individual patients, based on the specific characteristics of their cells.

For example, if a person has a particular type of cancer, the HCA data could be used to identify the specific types of cells that are involved in the disease and how they are behaving. This could help researchers develop targeted therapies that are more likely to be effective for that patient, rather than using a one-size-fits-all approach.

Additionally, the HCA data could be used to identify markers or "biomarkers" that are associated with specific diseases or health conditions. These biomarkers could be used to develop diagnostic tests or to predict an individual's risk of developing a particular condition.

Overall, the HCA has the potential to provide a wealth of information that could be used to improve personalised medicine approaches, by helping researchers to understand the specific characteristics of an individual's cells and how they contribute to health and disease.

Source: https://www.humancellatlas.org/

The European Genome-phenome Archive

The European Genome-phenome Archive (EGA) is a service for the long-term secure archiving and sharing of all types of potentially identifiable genetic and phenotypic data from biomedical research projects. This data is subject to participant consent agreements, so it can only be shared with researchers for specific research purposes. The EGA was launched in 2008 and has grown rapidly, currently archiving over 4,500 studies from almost 1,000 institutions. Studies archived in the EGA represent a variety of research fields (e.g. cancer, rare diseases, infectious diseases, common/chronic diseases), data types (e.g. genetic/genomic, phenotypic, clinical) and technologies (e.g. whole genome/exome sequencing, bulk and single cell RNA sequencing, DNA methylation-sensitive sequencing) from researchers around the world.

The EGA's mission is to facilitate data reuse, enable reproducibility, and accelerate biomedical and translational research in line with the FAIR (Findable, Accessible, Interoperable, and Reusable) principles. The EGA operates a distributed data access model, in which requests are made to the data controller rather than the EGA. This means that the submitter maintains control over who has access to the data and under what conditions.

In addition to its role as a data archive, the EGA is also involved in the development and implementation of standards and methods necessary to deliver the full value chain of human health data. Notably, EGA is involved with international standards bodies as the Global Alliance for Genomics and Health (GA4GH), Biobanking and Biomolecular Resources Research Infrastructure (BBMRI) and ELIXIR.

To this end, it has been appointed as an ELIXIR Core Data Resource. The EGA is committed to providing the necessary

security measures to control access to data and maintain patient confidentiality, while also providing access to authorised researchers and clinicians. Access to data is granted by the appropriate data access-granting organisation (DAO), which is typically the same organisation that approved and monitored the initial study protocol or a designated representative of this organisation.

Sources:

https://ega-archive.org/

Mallory Ann Freeberg, et al., The European Genome-phenome Archive in 2021, Nucleic Acids Research, https://doi.org/10.1093/nar/gkab1059

The International Cancer Genome Consortium

The International Cancer Genome Consortium (ICGC) is a global initiative that aims to create a comprehensive catalogue of genetic abnormalities in various types of cancer. To achieve this goal, the ICGC has developed the Data Portal, a platform that allows users to easily visualise, analyse, and interpret large and diverse cancer datasets. The portal currently contains data from 84 cancer projects around the world, and includes information on over 77 million somatic mutations and molecular data from over 20,000 contributors.

The ICGC Data Portal uses scalable big data technologies to overcome the challenges of storing, annotating and exploring large and complex datasets efficiently. This enables researchers to perform powerful integrative analyses that could provide new insights into the biology of cancer. For example, the integration of a large number of tumour genomes in the portal could help identify rare molecular subtypes that have distinct clinical behaviours.

The ultimate goal of the ICGC is to use the data it collects to develop new and better diagnostic tools and more targeted therapies for cancer. To this end, the ICGC is launching the ARGO project, a new phase of the consortium, that will collect clinical and molecular data from over 100,000 cancer patients participating in therapeutic clinical trials. The ARGO project will work closely with the GA4GH to define and implement next-generation GA4GH protocols and technical standards.

Sources:

Zhang, J., Bajari, R., Andric, D. et al. The International Cancer Genome Consortium Data Portal. Nat Biotechnol 37, 367–369 (2019). https://doi.org/10.1038/s41587-019-0055-9

https://dcc.icgc.org/

https://www.icgc-argo.org/page/64/about-icgc-argo

Outlook

ICPerMed is looking forward to 2023 with:

- **biannual ICPerMed events and one ICPerMed Family meeting:**
  - ICPerMed Workshop "Preparing the Future for Personalised Medicine: EP PerMed", on invitation only and taking place in Pamplona on January 17-18, 2023
  - The 2nd ICPerMed Family Meeting "Staying connected to shape the future for Personalised Medicine" on invitation only, October 19, 2023
  - ICPerMed Workshop "Advancing Personalised Medicine through Technology Development", on invitation only and taking place in Siena on November 14-15, 2023
- **training courses** fostering exchanges with and the active involvement of the PM community;
- new **examples of best practices** in personalised medicine presented and promoted on the ICPerMed website and through the **ICPerMed Recognition**;
- the outcome of the preparations and evaluation of the **European Partnership for Personalised Medicine (EP PerMed) proposal.**

Stay tuned for ICPerMed's activities via our newsletters[14] (published quarterly), the ICPerMed website and through the ICPerMed YouTube channel, Twitter and via LinkedIn.

https://twitter.com/ICPerMed

New ICPerMed LinkedIn Account

@ICPerMed - The International Consortium for Personalised Medicine

14 https://www.icpermed.eu/en/services-newsletter.php