

# The ICPerMed vision for 2030

How can personalised approaches pave the way to  
Next-Generation Medicine?



# Imprint



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This Vision Paper has been developed on behalf of ICPerMed and its members: <https://www.icpermed.eu/en/icpermed-members.php>

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# 1. Executive Summary

This document presents the vision of the International Consortium for Personalised Medicine (ICPerMed) on personalised medicine (PM) research and implementation by 2030.

ICPerMed connects more than 40 European and international institutions that aim to:

- Establish ICPerMed members as global leaders in personalised medicine research.
- Support the personalised medicine science base through a coordinated approach to research.
- Support research to investigate the potential benefits of personalised medicine approaches to citizens and healthcare systems.
- Pave the way for personalised medicine approaches for citizens.

The notion that individuals can experience unique clinical manifestations for the same disease, as well as variable responses to treatment, has long been recognized by the medical community. There are many long-standing paradigmatic examples of the use of such knowledge in medicine, for example the testing of blood type before blood transfusions or the neonatal screening programs. However, the prolific technological advancements in biomarker detection over the last few decades, including not only genomics but also other “omics” and body imaging methods, have spurred the development of novel approaches to health and disease management that are specifically optimised for each individual. The term PM, and its subtle variations, such as precision medicine or stratified medicine, today generally describe an approach to medicine that integrates an individual's characteristics for early disease diagnosis, prognosis, optimal choice of treatment, accurate disease risk estimation, and targeted prevention<sup>1</sup>.

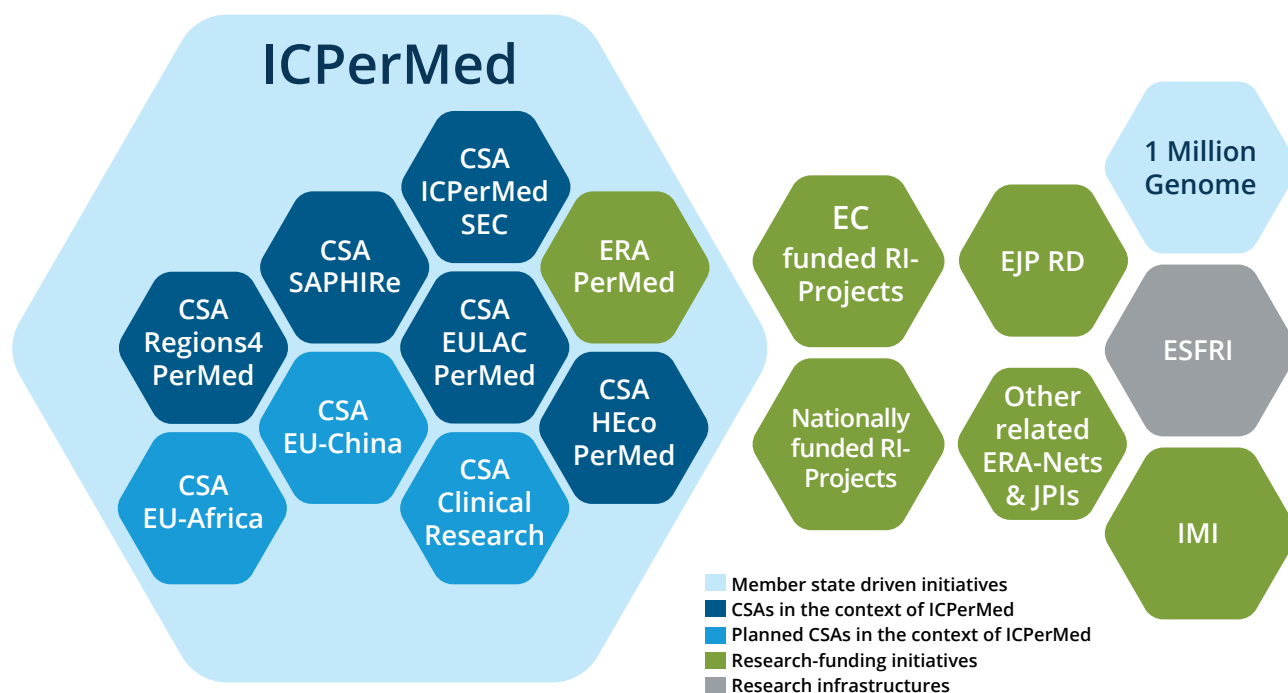
PM shows great potential to improve and optimise health promotion, disease prevention, and disease management. The upcoming challenges for this field are translating research results into clinical practice, facilitating their adoption by healthcare systems, and developing cost calculations and reimbursement models.

Formally established in 2016, ICPerMed builds on previous initiatives that enabled the identification of major challenges in PM<sup>2</sup>. Operating on the definition of Horizon 2020 and the European Council Conclusions on PM for patients<sup>1</sup> and covering the entire healthcare value chain, ICPerMed developed an Action Plan of actionable research and support activities that was released in March 2017<sup>3</sup>. The ICPerMed Action Plan is expected to guide strategic discussions of research and health funders around Europe and globally, as well as joint efforts of its members and the EC towards the implementation of PM.

With the support of the European Commission (EC), ICPerMed has grown into a family of consortia, programs, and actions. The “ICPerMed family” (Fig. 1), together with associated and related initiatives, plays a very important role in supporting the research and implementation of PM in Europe and beyond. In the future, ICPerMed will continue to foster such initiatives and activities in close collaboration with the EC and carry a leadership role to fulfil the vision for PM in 2030 presented herein. The overall aim is to pave the way for the next generation of medicine.

ICPerMed considers our increased understanding of biological, lifestyle, and environmental factors that regulate disease onset and progression to be the driving force for the implementation of PM, and thus aims to align and encourage joint efforts in research and implementation. In this context, ***ICPerMed has developed a vision of how the use of PM approaches will promote “next-generation” medicine in 2030***, more firmly centred on the individual's personal characteristics, leading to increased effectiveness,

## ICPerMed “Family” and Related Initiatives



**Figure 1. Projects and initiatives which are either directly part of the ICPeMed platform (big hexagon) or indirectly related to ICPeMed.** Ongoing coordination and support actions (CSAs) in the context of ICPeMed consist of the CSA for the secretariat (ICPeMed SEC), the two regional CSAs on adoption of PM (Regions4PerMed and SAPHIRe), the Europe/Latin America and the Caribbean CSA (EULAC PerMed) and the CSA HEcoPerMed performing research on health economics and payment aspects related to PM. The ERA-Net ERA PerMed funds projects on several topics of PM. Further CSA calls, which have been published but not yet evaluated and funded, will be integrated into the ICPeMed platform.

In addition, there are research and innovation projects related to the aims of ICPeMed, either funded by the European Commission (EC) or national and regional entities and funding agencies. The European Joint Programme on Rare Diseases (EJP RD) and other European Research Area networks (ERA-Nets) and Joint Programming Initiatives (JPIs), jointly supported by the EC and participating national and regional partners, are listed.

Other programs aligned with the aims and goals of ICPeMed include the “1 Million Genome” and associated initiatives, the European Strategy Forum on Research Infrastructures activities (ESFRI), and projects funded via the Innovative Medicines Initiative (IMI).

Note: This is just a selection of initiatives and activities and the list is not exhaustive.

economic value, and equitable access for all citizens to the best possible healthcare.

ICPerMed has defined this vision by consulting with European and international experts, covering the entire range of relevant sectors and professional backgrounds. The consulted experts and stakeholders were presented with a framework of five core perspectives of PM: citizen literacy and empowerment; education, capacity building, and engagement of healthcare providers; optimised utilisation and equal access to the health system; safe data management and usage; and understanding of economic and societal value and novel business models. The experts' comments concerning opportunities and challenges helped to refine the overall vision, which was further shaped by the outcome and conclusions of the ICPerMed conference Personalised Medicine in Action<sup>4</sup> in November 2018.

The experts generally agreed with the five proposed ICPerMed core perspectives, while highlighting certain specific concerns and challenges, as well as proposing solutions. This helped identify transversal aspects that define crucial pillars of the vision for PM in 2030: data and technology, intersectoral synergies, health systems reforms, education, and literacy. The Personalised Medicine in Action conference in Berlin in November 2018 provided a number of best practice examples, strategies, and other ongoing activities in Europe and beyond, illustrating important and promising developments for the way forward for PM. ICPerMed will build on this experience in future activities.





## 2. ICPeMed Perspectives for Personalised Medicine in 2030

The perspectives that ICPeMed will address going forward are:

### **Perspective 1: Informed, empowered, engaged, and responsible citizens**

- a) All health-related data is controlled by the citizen.
- b) Health data input and access is controlled, supported, and monitored by the citizen.
- c) Easily accessible, reliable, and understandable sources of medical information are available, e.g. tested and approved Apps or internet tools.

### **Perspective 2: Informed, empowered, engaged, and responsible health providers**

- a) The safe (i.e., privacy-protective), responsible, and optimal use of health-related information and research results to identify the best health promotion, disease prevention, diagnosis, and treatment options for each patient, supported by suitable information and communication technology (ICT) solutions, has become routine in the clinic.
- b) Personalised treatment of multi-morbid patients, non-responders, the elderly, and other vulnerable groups, with the minimisation of adverse effects, is routine, requiring clinical decisions by multidisciplinary teams and the development of novel health-related professions to support personalised healthcare.
- c) The education of healthcare professionals has adopted the interdisciplinary aspects of PM, such as the use of ICT and policy/regulatory questions, including equity and ethics, to access and adequately utilise all available information.
- d) Clinicians and researchers work closely to support the rapid development and implementation of personalised clinical solutions.

### **Perspective 3: Healthcare systems that enable personally tailored and optimised health promotion, prevention, diagnosis, and treatment for the benefit of citizens and patients**

- a) Equitable access to personalised healthcare for all citizens, independently of socioeconomic status, age, gender, ethnicity, and insurance coverage, is a reality.
- b) Personalised healthcare services are optimised in terms of effectiveness and equity.
- c) Fair and reasonable allocation of resources within healthcare systems is consistent with societal values.
- d) Secure health and related data flow from citizens and healthcare systems to regulatory authorities and researchers, among others, is in place.

### **Perspective 4: Availability and optimal use of health-related information for optimised treatment, care, prevention, and research**

- a) A combination of imaging and diagnostic, genomic and other molecular data, and information on lifestyle, environment, and socioeconomic status, displayed in electronic health records (EHRs), are used by healthcare providers and researchers for more efficient healthcare.
- b) Harmonised solutions to ensure data privacy, safety, and security are used in a transparent way throughout the health-data management process.
- c) Optimised treatment and prevention benefit citizens, while minimising costs and risks.

**Perspective 5: Economic value by establishing the next generation of medicine**

- a) A reasonable balance between investment, profit, and shared-benefit for the citizen is a reality in PM.
- b) Innovative and appropriate business concepts and models are in place for PM.
- c) Telemedicine solutions and mobile Apps promote PM and are of economic value.
- d) New jobs in healthcare systems are created, including data-related professions, such as “Information/Data Technicians”.

# 3. Critical pillars sustaining Personalised Medicine

The ICPeMed vision for 2030 is aligned with the 3rd Sustainable Development Goal of the United Nations 2030 Agenda for Sustainable Development, which sets out a vision for good health and well-being, promoting healthy lifestyles, preventive measures, and modern, efficient healthcare for everyone. ***To support these goals, the ICPeMed vision relies on four critical pillars: data and technology, intersectoral synergies, healthcare system reforms, and education and literacy, each of which is essential for PM implementation in 2030.***

## Data and technology

By 2030, digital technology will be a critical enabler of all aspects of society, including the health and well-being of citizens. Data generation will be continuous and rapidly evolving, requiring novel approaches to ICT in areas such as data storage capacity, management, access, protection and safety, harmonization, quality control, and sharing. Personal health data available through EHRs will be much more complete and extensive in 2030. An exponential increase in the use of wearable devices and apps will allow continuous tracking of health parameters and related behaviours, generating 'Big Data' fundamental for informing disease prevention, precise diagnostics, and therapeutic strategies. Concomitantly, attitudes towards digital technology and the sharing of personal data will change, as a new generation comes of age for whom digital technology is embedded in daily life.

EHR information will be supplemented by major developments in biomarker generation technology. Global efforts to generate knowledge of genomic variants in millions of individuals will allow a much deeper understanding of population-wide genomic variation, leading to the definition of individual genomic risk profiles associated with common multi-genic disorders and a greater emphasis on prevention. Other levels of biological information, including epi-genomics, proteomics,

and metabolomics, will complement genomic-risk estimates and provide monitoring tools for individuals at risk for disease. Modelling and systems medicine approaches will be further developed to interpret this data, especially as the data types will be heterogeneous.

The challenge by 2030 will be to develop and deploy innovative and flexible ICT solutions to address the needs of PM models. Significant investments in artificial intelligence methods will be needed for integration and interpretation of multilevel data coming from multiple sources. This will require that interoperability and harmonisation concepts are embedded in healthcare and research systems through more homogeneous data collection tools. Furthermore, creative and trustworthy ICT solutions will be available to support clinical decisions by healthcare providers at the point of care.

## Inter-sectoral synergies

In 2030, strong synergies between healthcare and research will be essential for the application of PM approaches. Large volumes of routine healthcare data will provide a rich source of material for research, allowing patient stratification and the definition of profiles, and supporting clinical trials. Conversely, a close alignment between healthcare providers and research will facilitate much faster turnaround of research results for clinical implementation.

Lifestyle and behaviours, socio-economic status, employment, and environmental exposure are all relevant to health outcomes. Integrating these related parameters with personal health and biomarker data will clarify the impact of policies in other sectors and enable the establishment of valuable cooperation. Inter-sectoral synergies will be particularly important for health promotion and disease prevention.

Synergies with the private sector will be driven by the need for rapid technological progress aligned with benefits to citizens, creating important business opportunities. PM will drive innovation, particularly in areas such as digital technology, predictive medicine, biomarker detection, and the development of molecular-targeted drugs suitable for smaller groups of patients with particular characteristics. By 2030, novel business models will be established, linking healthcare and industry, to accommodate the rapid generation of personal data and strategies for risk definition and personalised prevention and intervention. These developments will be led by digital technology and the pharmaceutical, biotech, and other industries.

## Healthcare systems reforms

Important reforms of healthcare systems will have taken place by 2030 to accommodate PM models. Policy makers will have confidence in PM models, in terms of optimisation of patient treatment, early diagnosis, improved prevention and health promotion for citizens, minimisation of health risks, economic sustainability, and societal benefits.

By 2030, the focus will shift from treatment to risk definition, patient stratification, and personalised health promotion and disease prevention strategies. This paradigm shift will be reflected in the organisation of healthcare systems and is particularly important in an ageing society. An increasing ageing population will need healthcare systems to incorporate both strands, i.e., health promotion and prevention, as well as treatment.

Technological reform and innovation will prompt significant investments in centralised data infrastructure and digital platforms to support data management, interoperability and access, and data sharing between citizens, health professionals, and researchers. Additionally, technological advances will create the need for new

skills. Health professionals trained in technologies related to digital platforms, biomarker examination and data analysis will be needed as part of multidisciplinary teams that make shared clinical decisions. Importantly, healthcare systems will have developed more flexible working models to accommodate individual needs for prevention or intervention and will also be able to accompany the rapid turnover of technological and scientific innovations streaming from PM approaches and needs.

In this novel context, new economic paradigms and business models will have been developed, incorporating a broader societal perspective, underpinned by shared ethical values and guaranteeing equity of access.

## Education and literacy

By 2030, there will be a paradigm shift in education and health literacy for citizens, in particular for patients, healthcare providers, healthcare managers, and policy makers.

Major changes in medical and other healthcare provider curricula (e.g. pharmacists, nurses, and therapists) will result in a new generation of informed, empowered, engaged, and responsible healthcare providers. There will be a strong focus on digital literacy and the skills needed to interpret biological and molecular information. The value of multidisciplinary in clinical and healthcare decisions will be well understood and embedded in practice. Given the fast turnover of technologies and their potential impact on healthcare, lifelong education and training will be essential for healthcare providers. On the other hand, professionals with a digital background will have a better understanding of healthcare and clinical issues, facilitating interactions among clinical teams encompassing professionals with different backgrounds. Finally, training in PM approaches

will be extended to healthcare managers and policy makers so that potential benefits can be rapidly and fully understood.

For the citizen and patient, information is equally empowering. By 2030, health data education, and literacy on ethical issues, regulations, and data control raised by PM will be provided through schools and specific literacy programs. Such improved literacy will be complemented by interfaces capable of providing required information on demand while preserving the patient-clinician interaction.

Politicians and policy makers will require access to evidence that PM can provide a benefit to citizens and patients and economic benefits to healthcare systems. This in turn will enable the establishment of political frameworks to tackle effectiveness, efficiency, equity, and ethical issues underlying the implementation of PM approaches.



## 4. Benefits and Challenges of the ICPeMed Perspectives

Below we discuss the benefits and challenges related to each of the five perspectives based on the experts' responses.

### Perspective 1: Informed, empowered, engaged, and responsible citizens

Perspective 1 concerns citizens and their relationship with the wide availability of health and medical information, as well as their own health and medical data. As such, the following can be envisioned for 2030:

- a) All health-related data is controlled by the citizen.
- b) Health data input and access is controlled, supported, and monitored by the citizen.
- c) Easily accessible, reliable and understandable sources of medical information are available, e.g. tested and approved apps or internet tools.

A citizen can be defined as: 1) an inhabitant of a city or town, especially one entitled to the rights and privileges of a freeman; 2.a) a member of a state; 2.b) a native or naturalized person who owes allegiance to a government and is entitled to protection by it. There are some concerns about the definition of citizen, as the term can be more inclusive and thus changed to "society member", "civilian", or "healthcare user". In the spirit of this document, "citizen" is meant to be used as "individual" to reflect the inclusive nature of this approach, rather than to limit its application to a restricted group of the population.

In this context, all health-related data is legally owned by the citizen, at least in Europe. Therefore, citizens should be able to access and control their own personal data. Deeper reflection is needed to address the question of monitoring and controlling

the data, as it can have a negative impact on data sharing and accessibility, with consequences for research and the healthcare system and maximization of the value of data for the common good.

Citizens must have confidence that data are securely stored and processed. Thus, there must be clear regulations to store, manage, and control access to health data. These regulations should protect the personal rights of the citizen, while at the same time enabling data sharing with healthcare providers and researchers and supporting the digitalization, standardization, harmonization, and interoperability of health data. There are many national and regional platforms in which personal health data is provided and accessed by citizens. However, these platforms should be made interoperable through common harmonisation, building on examples of best practices<sup>5,6,7,8</sup>. Efforts should allow the sharing of relevant information when needed. There are already successful examples of data sharing, e.g. the MatchMaker Exchange<sup>9</sup>, which was developed under the leadership of the Global Alliance for Genomics and Health and allows data sharing in a delimited and secure manner.

Education is crucial if citizens are to be informed, empowered, engaged, and responsible. Citizens, as well as healthcare professionals and government officials, should be informed about the benefits and challenges of PM. There should be campaigns to raise awareness and forums to discuss PM involving various stakeholders. For example, various media could be used to increase awareness of general topics of health/research in partnership with research institutes, universities, and industry<sup>10,11</sup>. This could be specifically applied to PM. Resources could include booklets and specific information for the public, researchers, and research staff.

## Perspective 2: Informed, empowered, engaged, and responsible health providers

Implementation of PM will require the engagement and commitment of healthcare providers. Their contribution, as well as their needs and expectations, to PM are contemplated in the four main areas that form Perspective 2:

- a) The safe (i.e., privacy-protective), responsible, and optimal use of health-related information and research results to identify the best health promotion, disease prevention, diagnosis, and treatment options for each patient, supported by suitable ICT solutions, has become routine in the clinic.
- b) Personalised treatment of multi-morbid patients, non-responders, the elderly, and other vulnerable groups, with the minimisation of adverse effects, is routine, requiring clinical decisions by multidisciplinary teams and the development of novel health-related professions to support personalised healthcare.
- c) The education of healthcare professionals has adopted the interdisciplinary aspects of PM, such as the use of ICT and policy/regulatory questions, including equity and ethics, to access and adequately utilise all available information.
- d) Clinicians and researchers work closely to support the rapid development and implementation of personalised clinical solutions.

One main concern to achieving Perspective 2 is the investment needed for the literacy, education, and capacity building of healthcare professionals. This comprises the inclusion of PM-related issues in the *curricula* of basic healthcare degrees, as well as lifelong training for medical doctors and other healthcare professionals. Research and innovation

need to be an integrated component and commitment of the healthcare sector and must address a variety of disciplines, from clinical aspects to digital and ICT topics. Equally vital is the fact that many future clinical decisions will involve multidisciplinary teams composed of members with various medical specialities, including general practitioners and non-medical professionals, and that there is utility and value in shared medical decision-making processes. Such intersectoral and multidisciplinary cooperation will most likely lead to the establishment of novel professional profiles in clinical teams and among healthcare professionals.

The leveraging of health-related information and research results for prevention, diagnosis, or treatment requires an improvement of digital literacy among healthcare providers and the effective use of ICT infrastructure and ICT solutions in clinical contexts. For example, networking and data sharing platforms that are less fragmented and more compatible and interoperable across healthcare facilities and research institutions will allow better data accessibility for clinical and research purposes. More and better tools and solutions for medical decision support, adequate EHRs for PM delivery, health data systematisation platforms, and the control of data quality are further highlighted as major steps towards the implementation of Perspective 2.

In many areas, robust evidence of the benefit and cost effectiveness of PM approaches is still failing to gain the full confidence of healthcare providers in their value. Although diagnosis and disease classification, as well as therapeutic protocols involving genomic information, are currently used for many cancers and rare diseases, evidence of the potential benefits of personalised preventive approaches is still insufficient. Faster evaluation of preventive approaches will require the bidirectional exchange of health data between clinicians and researchers to be an explicit component of Perspective 2. The transfer of clinical and biomedical research to routine healthcare needs to be facilitated and organ-



ised. For example, clinical data should be available and used for clinical-trial design and research. Conversely, research results need to be available for use by healthcare providers in clinical settings. Transparent and secure data access procedures must be established to attain this goal.

The involvement of healthcare professionals as main players in this synergy is vital for the implementation of PM. They are fundamental partners, as research plays an important role in healthcare, for example in longitudinal predictive studies aiming to gather evidence to support the health benefit for patients, analyse cost effectiveness, and tackle the challenges of transforming healthcare systems. Continuous education of healthcare providers concerning the methodological issues, activities, and outcomes of research will improve mutual understanding and allow a better and more complete flow of information.

A need to understand the impact of educational systems on the use of PM approaches by health professionals is crucial for the development and adequate definition of education and literacy programmes. Educational materials need to be easily accessible and certified medical education on PM technologies needs to be standardised and compulsory.

Furthermore, crucial policy frameworks, including national guidelines, plans and strategies, and funding and resources will be needed to fully implement Perspective 2. It is clear that the “best” prevention, diagnosis, and treatment options need to refer to not only effectiveness but also efficiency, equity, and ethical considerations. A commitment to health promotion and prevention, as opposed to disease care, also needs to be stressed to fulfil this vision. Finally, Perspective 2 includes a component explicitly focusing on population groups that present specific challenges to the implementation of PM, such as multi-morbid patients, non-responders, the elderly, minorities, and children. In this sense,

expertise and awareness of policy and regulatory questions, including equity and ethical issues and the health versus disease focus, require great attention and education for healthcare professionals should be extended to healthcare managers, stakeholders, and policy makers.

The improvement of the literacy and education of healthcare professionals concerning PM, as well as a supportive infrastructure, will be fundamental to enable the use of available information for medical and healthcare decisions. The terms “engaged” and “empowered” ensure the involvement and participation of healthcare professionals at several levels.

### Perspective 3: Healthcare systems that enable personally tailored and optimised health promotion, prevention, diagnosis, and treatment for the benefit of citizens and patients

Perspective 3 concerns the implementation of PM in healthcare systems, focusing on four main components:

- a) Equitable access to personalised healthcare for all citizens, independently of socioeconomic status, age, gender, ethnicity, and insurance coverage, is a reality.
- b) Personalised healthcare services are optimised in terms of effectiveness and equity.
- c) Fair and reasonable allocation of resources within healthcare systems is consistent with societal values.
- d) Secure health and related data flow from citizens and healthcare systems to regulatory authorities and researchers, among others, is in place.

Healthcare systems and other actors within the life sciences sector will undergo fundamental changes to implement stronger PM approaches and multiple aspects of such reforms will have an impact on the attainment of the perspectives above.

The first concern regarding the implementation of PM in healthcare systems is one of equitable access to healthcare. Access to adequate healthcare should be independent of age, sex, ethnicity, wealth, or insurance coverage. Healthcare systems need to retain access to high-quality healthcare that is affordable at the point of care for all and

ensure facilitated access to marginalised sectors and under-served populations. As PM depends on state-of-the-art technology, the costs of PM need to incorporate the long-term value of innovative approaches with justifiable reimbursement models to ensure equitable access.

Healthcare services will have to be optimised for the wide inclusion of personal data in individual healthcare strategies. This can be achieved, for example, through the establishment of centres of excellence in primary care for PM, specialised diagnostic testing services, and data centres. The optimisation of healthcare services will depend on the availability of resources, including infrastructure and specialised professionals to build multidisciplinary teams, and may require a less fragmented organization. Sustainability of healthcare systems will demand the prioritisation of resources and investment, with a global perspective according to value-based approaches based on an economic analysis of PM strategies. This will allow validation of novel business models, including information-based strategies for health promotion, disease prevention, and treatment selection.

PM is based on personal health data, which includes not only biomarkers, but also environmental factors, social determinants, and lifestyle. In this framework, it is important to consider the impact of policies from other sectors on health, as well as adopt a health in all policies approach, to maximize the value of the implementation of PM strategies.

Healthcare systems will make wide use of personal data to strategically and systematically integrate primary prevention in healthcare, with more preventive and predictive services for citizens. This will require improvements in the availability of advanced infrastructures and novel tools for data sharing and analysis, including harmonised databases at the national and international levels and digital frameworks to connect data generated by individuals, health providers, researchers, and

healthcare system managers. Decision support systems and continuous monitoring of outcomes also need to be integrated into healthcare systems. This will contribute to ensuring that appropriate preventive measures are put in place or that early diagnoses are made or optimal therapies selected, with continuous monitoring of outcomes.

The constant incorporation of new knowledge and innovative solutions will require healthcare systems to be more adaptable to enable end user-driven biomedical and clinical research and to more easily assimilate research results. This can be achieved by better connecting routine healthcare with clinical and biomedical research, through close cooperation between researchers, clinicians, and patients. Conversely, a strong relationship with academia and industry should make the data from academic or pharma industry clinical trials accessible to the medical community, providing better opportunities for access to innovative medicines. Funding for research will be crucial and healthcare funders need to be encouraged to support research (basic and applied) to strengthen the evidence base of novel PM therapies, diagnostic methods, and preventive approaches and the effectiveness, efficiency, and value of PM. Health technology assessment (HTA) is an important component of innovation and translation to healthcare. The engagement of independent research and HTA institutions that can fully examine the benefits and harms of PM interventions for patients is thus warranted. The assessment of the true value of technologies in its context of application will incentivize PM. The research community also needs to further engage in health economics studies, as well as the impact of PM on health inequalities.

It is crucial that healthcare system managers and policy makers can accurately evaluate the long-term societal health and economic benefits of PM approaches. Measures to improve the education and literacy of all stakeholders, including patients and citizens, clinicians, managers, and policy mak-

ers, will be fundamental in generating the required political support. Education and literacy concerning PM across the entire value chain will also contribute to the definition of PM strategies and the creation of guidelines and regulations for their implementation in healthcare systems, with a strong focus on equity, sustainability, infrastructure, and value.

Transparency in governance is a key element for the successful implementation of PM. Such transparency needs to involve the public and be present at multiple levels: in the setting of priorities, health assessments, decision-making processes, treatment-expenditure criteria, involvement of patients on drug-price decisions, and use of evidence in decision-making. Such transparency will facilitate the implementation of PM by increasing trust in the evolution and changes made to healthcare systems.

## Perspective 4: Availability and optimal use of health-related information/data for optimised treatment, care, prevention, and research

One of the cornerstones of PM is the availability of high-quality data. This aspect, together with issues related to data privacy, is mainly covered by Perspective 4 and its components.

- a) A combination of imaging and diagnostic, genomic and other molecular data, and information on lifestyle, environment, and socioeconomic status, displayed in EHRs, are used by healthcare providers and researchers for more efficient healthcare.
- b) Harmonised solutions to ensure data privacy, safety, and security are used in a transparent way throughout the health-data management process.
- c) Optimised treatment and prevention benefit citizens, while minimising costs and risks.

PM relies on complete datasets, incorporating lifestyle and other personal data together with genomic and molecular (-omics) data, that are harmonised and of sufficient quality. Several initiatives, such as the National Institute of Health Electronic Medical Records and Genomics Network<sup>12</sup> and the Genomic Health Initiative at NorthShore<sup>13</sup>, which aim to integrate diagnostics results with genomic data, are already in place, whereas some others are just starting (e.g. the Estonian Genome Center<sup>14</sup>, FinnGen<sup>15</sup>), suggesting that such integrated databases could become common in the near future. Harmonisation and the exchange of complex health data across borders is one of the challenges of the near future and has been one of

the long-term goals of the European Union (EU). In 2017, the European Commission set up a Task Force to establish the European Ecosystem for Healthcare, which is expected to examine the incentives and obstacles to achieve the secure exchange of data across the EU<sup>16</sup>. This was succeeded by the Digital Health Society Declaration<sup>17</sup>, aiming to achieve the digital transformation of healthcare systems, and compliant with the European Science Cloud Initiative<sup>18</sup>, which foresees high-capacity cloud solutions with super-computing capacity for future common data-storage platforms. On Digital Day 2018, the 1M Genomes Initiative<sup>19</sup> was launched, aiming to delivering cross-border access to at least one million genomes in the EU by 2022.

The complexity of voluminous personal health, genetic, and lifestyle datasets presents health professionals with the challenge of interpreting data to produce results of medical relevance. Appropriate mathematical modeling and computer simulation for integration, analysis, and interpretation of such massive amounts of biological and clinical data are critical for the personalization of medical interventions and understanding disease, as they deliver knowledge about the underlying mechanisms and pathways affected in disease states and their treatment. As a result, computational models render collected data manageable in clinical practice, since they can be used to predict important endpoints, such as individual disease risk, disease evolution, or treatment response. Data modelling will allow accurate prioritisation of each individual's health needs at every point throughout the course of their lives, thus enabling evidence-based innovative solutions for precise diagnoses and personalised prediction, prevention, and treatment.

As analytical tools, models help to organize, explain, and interpret data, but will only work on data of adequate quality, quantity, density, and structure (e.g. of sufficiently deep phenotyping or curated datasets). Therefore, data management and data handling protocols that are compliant with inter-

national state-of-the-art standards, such as FAIR<sup>20</sup> (Findable, Accessible, Interoperable, Re-usable) and the GDPR<sup>21</sup> (General Data Protection Regulation) are crucial components of PM research, addressing data accessibility, format and storage, stewardship and curation, as well as the quality of metadata and data security.

The protection of health-related data to ensure their safe and responsible use and raising public awareness among citizens and healthcare providers on data protection issues has been an essential topic throughout the ICPeMed vision. Recent surveys on public attitudes towards the use of health data have indicated a high level of support. For example, a 2018 survey by the publicly-funded organization Understanding Patient Data in the UK showed that 96% of respondents agreed with sharing of their health data for individual patient care and 77% for medical research<sup>22</sup>. Another pan-European study<sup>23</sup> indicated that 50% of people were concerned about the use of their data by non-medical personnel and 60% by private companies. However, in spite of the questions regarding data sharing, it is clear that attitudes towards digital technology and the sharing of personal data will be different in the generation that will be entering adulthood in 2030, which comprises a large percentage of the overall global population. Digital technology is an essential part of daily life for this generation, which understands its value and tends to be much more open regarding the sharing of personal data through digital media.

Integrated data should be interpretable by health professionals and patients. For citizens, it is crucial to emphasize their role in controlling and providing health-related data and to empower patients to make decisions in partnership with health professionals. Citizens are expected to have the right to voluntarily provide data, assuming that they have a sufficient level of understanding of both the positive and negative implications, and ensuring this is the

responsibility of PM advocates. The voluntary provision of data required must inherently allow the patient to choose whether or not to engage with PM services.

The use of integrated health information by healthcare providers in daily practice and the routine availability and use of data as an integral part of healthcare requires the motivation of clinicians to use the data and share their findings with other healthcare providers. This can only occur when a clear benefit of using personalised data (including genomic information) is evident to clinicians and/or when the respective legislative framework is in place. The motivation and engagement of clinicians are closely linked with citizen empowerment: as citizens become more knowledgeable and are able to make informed decisions to request personalised services, clinicians will also be more motivated to use the data provided for PM approaches. Also, user-friendly interphases for viewing the data and ICT tools, such as decision-support tools that enable rapid and comprehensive analysis of complex personal data, will facilitate the actual use of the data in healthcare.

Revealing the potential of personalised health data in improving the efficacy and precision of clinical diagnosis, prevention, and practice is central for the implementation of PM. Clarifying the positive impact of using personalised data on health outcomes to healthcare providers will facilitate the implementation of data-intensive approaches.

## Perspective 5: Economic value by establishing the next generation of medicine

Perspective 5 concerns multiple aspects of the economic value for societies of implementing PM and thus includes several components:

- a) A reasonable balance between investment, profit, and shared-benefit for the citizen is a reality in PM.
- b) Innovative and appropriate business concepts and models are in place for PM.
- c) Telemedicine and mobile health solutions promote PM and are of economic value.
- d) New jobs in healthcare systems are created, including data-related professions, such as “Information/Data Technicians”.

The scope of Perspective 5 extends from the value of PM for healthcare systems and society to actual economic growth. There is a broad range of potential viewpoints, from strict development of the private sector to a broad societal perspective. The true value of PM may come from looking beyond short-term benefits and the direct benefit to healthcare costs. By integrating a societal perspective, the benefits and resource allocation beyond PM may be evaluated, allowing the optimisation of resources dedicated to healthcare. The efficiency of the system and the value of healthcare investments could thus be maximised. However, doing so will require a shift in the way many systems are currently organised.

In Europe and beyond, the existence of publicly funded healthcare systems could facilitate such optimisation. Public ownership of data probably constitutes the biggest value of such systems. Ensuring that structures are in place to extract

knowledge from these data and combine multiple sources, from genomic data to EHRs and social media, will allow maximization of the economic value of this common good. Moreover, international data sharing and harmonisation will allow further comparisons and the optimisation of healthcare systems.

Demonstrations of the potential value of PM approaches will need to be made to achieve their large-scale implementation and uptake. This can be accomplished through the development of exemplars, areas such as cancer and rare diseases that are more likely to be ready to integrate genomic medicine and PM approaches. Careful consideration will need to include appropriate healthcare and macroeconomic methodology to demonstrate the value of PM approaches for these exemplars in a publicly funded healthcare context. The approaches used must consider the “economics” at the systemic level, beyond the mere aspects of health economics, to truly address the societal value. These approaches could also integrate unemployment and social-care systems, new risk sharing methods, and take into consideration the entire life cycle and value chain of the innovative PM approaches, rather than be based on a single point-in-time evaluation.

A better understanding of the notion of value in various health and social care systems, optimisation of the resources within these publicly-funded systems, and development of a robust evidence base could drive the implementation of appropriate PM approaches to healthcare and deliver benefits for patients and society. Although new PM approaches have sometimes been described as a paradigm change, they can also be described as the simple evolution of medicine in a data-rich era. Such evolution comes with required adjustments in the way medicine is delivered. This requires new types of data specialists and quality experts, skill sets for healthcare professionals, and tools for delivery.

## 5. Developing the Vision

To reflect the views of multidisciplinary and inter-sectoral experts, ICPeMed surveyed European and international experts concerning their opinion about the progress of PM in Europe and their vision for PM in 2030. The survey focused on three main questions:

1. How applicable are the innovative tools required to implement medical care that is based on large amounts of personal data, including omics data?
2. How prepared are stakeholders and healthcare systems to adopt such approaches?
3. What challenges lie ahead and how might they be solved?

For this survey, ICPeMed proposed a framework of five perspectives, based on the perceptions gained from previous work and targeting core issues and challenges for healthcare implementation.

These five perspectives cover main aspects related to i) equitable and fair access to healthcare systems and the optimal use of resources, ii) public literacy and empowerment, iii) engagement and involvement of healthcare providers, iv) safe data management and usage, v) and economic value and novel business models. Experts from several professional sectors were invited to comment on these five core perspectives and whether they reflect their own vision for PM for the next decade, whether there were issues that were not fully contemplated, and whether they could identify additional challenges, solutions, and best practices, and share their opinions from a national and global perspective.

Along with the experts' views, the conclusions from the first ICPeMed conference, Personalised Medicine in Action<sup>4</sup>, which took place in November 2018, helped to develop the ICPeMed vision for PM in 2030. Figure 2 represents the vision-building process.

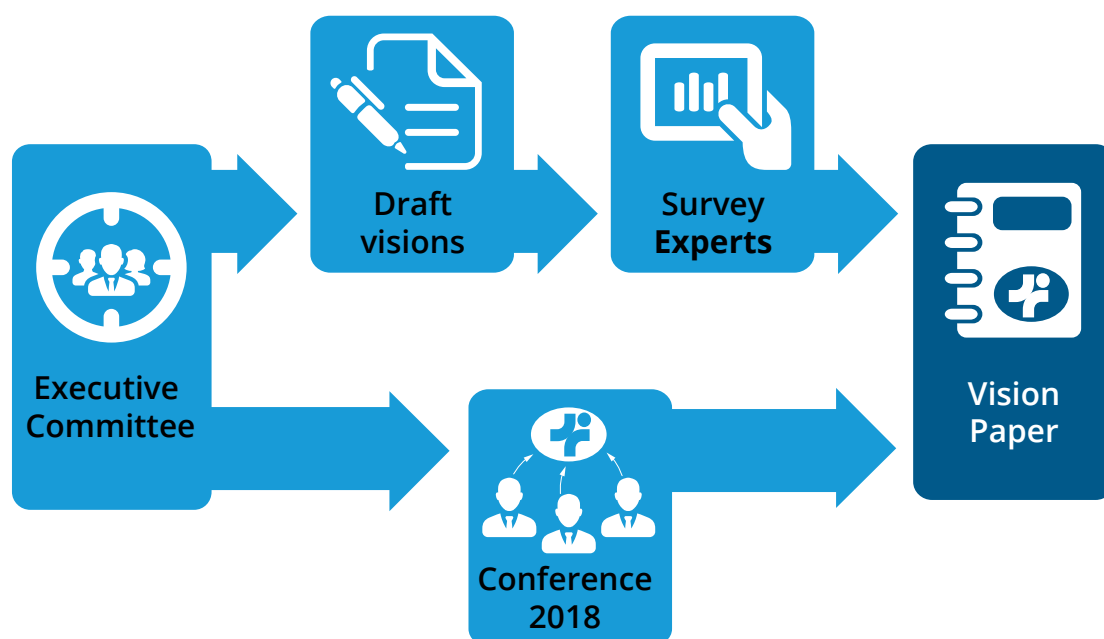


Figure 2. Development of the ICPeMed Vision Paper

## The ICPeMed survey – methodology and general outcomes

European and international experts were invited to participate in the survey and the 1st ICPeMed Conference, Personalised Medicine in Action, upon the suggestion of ICPeMed members. In total, 97 experts from various countries and professional sectors were invited to participate in the survey, of whom 70 fully completed the survey (72% response rate). The professional background of most of the experts was research (53%), whereas 13% were healthcare professionals, 6% members of patient organizations, 4% healthcare providers, 4% regulators, and 13% “other professions” related to healthcare policy, intergovernmental organizations, or information technology. Most of the experts (84%) were employed by public institutions and a few respondents (11%) worked in industry.

The survey asked questions specific to each of the five perspectives, including whether they shared the same vision, what might be changed, how far was the realization of the perspective in their country and when it might be fully achieved, and what would be needed for full achievement. Finally, the experts were asked to cite best practices for each perspective.

## Personalised Medicine in Action – The Berlin Conference

The ICPeMed conference in Berlin focused on the theme Personalised Medicine in Action<sup>4</sup>. The conference was an opportunity to consider how PM approaches are being implemented in different countries through case studies and best-practice examples. The Conference message supported the experts’ perceptions in relation to the current implementation of PM and the vision for 2030.

The conference included three panels, organised around translational research best practices, entitled PM Research & Implementation: From Basic Research to the Patient. Successful examples were presented of research-based initiatives that have been implemented in healthcare systems with a clear patient benefit and that are already meeting many of the above-described aspects envisioned for 2030.

A number of common themes were evident across all examples. Successful implementation of PM depended on collaboration and teamwork by all healthcare stakeholders. Traditional work practices had to be adapted to support new approaches and required new skills. It was clear that patient involvement positively influenced the development and implementation of PM approaches.

The examples demonstrated that implementing the outcomes of PM research in practice is not a linear process: although research drove changes in healthcare, healthcare also influenced research. At the political level, it was clear that politicians and policy makers are starting to develop a shared understanding of PM and the benefits it can provide.



## 6. Conclusions

ICPerMed and its members have already successfully established PM-specific research and health system-related programs and actions in their countries and regions. A selection of successful efforts was presented at the ICPerMed conference in November 2018 in Berlin and will continuously be collected in the ICPerMed database, which is accessible on the webpage.

Furthermore, there are plenty of initiatives or projects supported and funded by the European Commission (EC) that are consistent with the presented vision and represent the “ICPerMed Family”. Some of these activities are member-state driven, such as ICPerMed and the 1 Million Genomes Initiative, whereas several are strongly supported or fully financed by the EC. In addition, there are plenty of other initiatives and projects active in topics closely related to PM and interconnected aspects, such as those on rare diseases, cancer, genomic research and data management and analysis, and medical informatics and EHRs.

ICPerMed is confident that its members, as well as the EC, will take the perspectives presented herein into consideration when planning upcoming and future programs and activities. ICPerMed will continue to act as a communication platform for existing and future initiatives and organisations related to PM and the perspectives presented here.

After the ICPerMed Berlin conference, the consortium started to develop a concept for the upcoming conference, scheduled for autumn 2020 in Paris, France. This event will be inspired by the five perspectives presented herein and will be a good opportunity to discuss the advances in PM towards the ICPerMed 2030 vision.

In parallel, ICPerMed will continue to foster research and other activities to pave the way towards this vision in a realistic way. This will include multiple regional, national, and European strategies and approaches.

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