e-ISSN 2544-1620

Medical Science Pulse 2020 (14) 2

Reviews

DOI: 10.5604/01.3001.0014.2475

PERSONALISED MEDICINE – BEST PRACTICES EXCHANGE AND PERSONAL HEALTH IMPLEMENTATION IN EUROPEAN REGIONS – A QUALITATIVE STUDY CONCEPT UNDER THE REGIONS4PERMED (H2020) PROJECT

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A – study design, **B –** data collection, **C –** statistical analysis, **D –** interpretation of data, **E –** manuscript preparation, **F –** literature review, **G –** sourcing of funding

ABSTRACT

Personalised medicine (PM) is the adaptation of medical treatment to an individual patient. More importantly, PM offers the potential to detect disease earlier when it is easier to treat effectively. PM is beginning to overcome the limitations of traditional medicine. In PM there are many potential benefits and facilitators but also many barriers. The goals of the Regions4PerMed project are to set up the first interregional cooperation on PM, align strategies and financial instruments, and most importantly, identify primary barriers in personal medicine adoption in the health care system and systematic actions to remove as many of them as possible to create a future where PM is fully integrated into real life settings. Each key action activity will be followed by a focus group or semi-structured qualitative interview. The questions asked during the research will concern barriers and facilitators of PM implementation in the country of a subject and will concern: medical big data and electronic medical records; health technology in connected and integrated care; the health industry; facilitate the innovation flow in health care; socio-economic aspects. The qualitative study outcomes are supposed to bring more qualitative data to the discussion. They could be implemented to the daily practice of the health care system's stakeholders through the best practices transferred to all five key strategic areas of the Regions4PerMed project.

KEYWORDS: personalised medicine, personalised health, prevention, regional policies, interregional cooperation

Background

In the early 20th century the first connection between genetic inheritance and susceptibility to disease was seen [1]. In 2003, the first sequencing of the human genome was completed and cost over £2 billion for a single sequence with the work of tens of thousands of scientists in the UK, the US, and around the world [2]. Today, because of new sequencing technology, there has been a dramatic drop in the cost, which coupled with the availability of the high-speed computing needed for analysis, means it is possible to consider this technology as part of routine healthcare [3].

In the face of this potential huge leap forward, the fact that personalised health lacks the cooperation and coordination needed to organise the still very fragmented field is a severe drawback to its development and to the placement of investments in an effective manner. For this reason, it is crucial to direct major efforts towards coordinating and aligning relevant stakeholders in personalised health action across Europe and beyond; create a participatory approach; build trust; enable a multi-stakeholder process; channel investments towards Personalised Health [4].

Personalised medicine (PM) is a move away from a "one size fits all" approach to the treatment and care of patients with a particular condition, to one which uses new approaches to better manage patients' health and target therapies to achieve the best outcomes in the management of a patient's disease or predisposition

to disease. It uses diagnostic tests, functional genomic technologies, molecular pathways, etc. [3].

PM addresses the challenges of common medicines not being effective in treating large numbers of patients. This adds to rising health care costs due to more prevalent chronic diseases and an aging population. However, PM is tailor-made prevention and treatment strategies for individuals or groups, so patients receive specific therapies that will work best for them and no money is wasted on trial and error treatments [5].

Concept of the project

The Interregional Coordination For A Fast And Deep Uptake Of Personalised Health (Regions4PerMed [6,7]) main goal is to increase the involvement of relevant stakeholders (regional authorities, researchers, policy makers, and cluster organisations) for the implementation of personalised health. Another predominant goal is to set up the first interregional cooperation on PM, align strategies and financial instruments, identify key investment areas, and release a European regional agenda in order to foster the delivery of PM services to patients and citizens. The project aims to: support the coordination of regional policies and innovation programmes in PM in order to accelerate the employment of PM for citizens and patients; strengthen cooperation between Horizon 2020 and ESIF on PM aspects; ensure complementarity between RIS3 diagnostics priority and RIS3 personalised medicine priority mappings; establish a permanent dialogue between European regions regarding a fast and full implementation of PM; strengthen industrial specialisation areas in Europe and allow PM to flourish as an emerging industry; enable interregional joint investment on PM including a stable link with the Vanguard Initiative and with the European Innovation Council; provide guidance to the EC for the next Multiannual Financial Framework (MFF) as well as Research Framework Programme; provide guidance to EC, Member States, and regional authorities on the next European Structural and Investment Funds (ESIF) Operational Programme.

The other specific objectives of the project are: organise technical dialogue among regions around five Key Strategic Areas (KA) and through five thematic workshops; provide a final action plan of strategic areas of investments; establish a HUB of European initiatives and partnerships on PM (PerMed HUB); contribute to the realisation of the IC PerMed action plan; provide guidelines in the form of a report to regional authorities on how PM can boost local economies and keep the EU competitive; provide guidelines on how to address PM within the Smart Specialisation Strategies (RIS3); build and maintain a database of PH research and innovation and monitor programmes and projects that can be easily replicated elsewhere.

At the core of the project are five regional authorities and organisations representing European regions strongly committed to PM and the Wroclaw Medical University as an academic partner of the consortium. These authorities act as the Executive Board for the interregional coordination and are mainly responsible for the implementation of the project activities concentrated around the five key strategic thematic areas [6].

Five strategic areas of the project vs. Barriers and facilitators of PM implementation in European regions

Medical big data and electronic medical records

With the broad adoption of electronic medical record (EMR) systems, researchers can mine vast amounts of patient data, searching for the best predictors of health outcomes. Many of these predictors may lie in the genome, the encoded representation of each person's DNA. As gene sequencing continues to evolve from a complex, expensive research tool to a routine, affordable screening test, most of us are likely to have our DNA fully digitised, vastly expanding the already large store of electronic health data preserved in or linked to our EMRs [8].

Technological innovation has triggered an explosion in data production that will soon reach exabyte proportions. There is great potential for "big data" to improve health, but at the same time, "big data" also prompts new challenges [6]. The main barriers in this strategic area are that the technologies to store and analyse big data and the ability to model them are not fully developed yet. Creating a system that makes big data robust will be the biggest challenge.

To study PM we need to navigate and integrate clinical information (e.g. medical diagnosis, medical images, and patient histories) and biological data (e.g. genes, protein sequences, functions, and biological processes and pathways) that have diverse formats and are generated from different and heterogeneous sources. Data integration and making use of different data sources is at the core of PM. An important aspect to ensure data quality is data standardisation and terminologies with semantic mapping. A big challenge in ensuring data quality is understanding both syntactic and semantic differences in data sources and how they can be harmonised. It should summarise or abstract data in a meaningful way to translate data to information and knowledge. It still needs to be investigated to effectively translate large amounts of data to make use of it in decision-making. Health care data is continuously changing and evolving. These rapid changes in data pose a significant challenge in creating relevant domain models on-demand to be useful for searching, browsing, and analysis of real-time content. In turn, "this requires addressing the following issues: the ability to filter, prioritise, and rank the data (relevant to the domain or use case); the ability to process and ingest data quickly; and the ability

to cull, evolve, and hone in on relevant background knowledge" [9].

Health technology in connected and integrated care

Both electronic health (eHealth) and mobile health (mHealth) are becoming prominent components of health care. eHealth and mHealth encompass a vast spectrum of health care services, ranging from electronic prescribing and medical records, to text message prompts to remind patients to take their medicines. eHealth and mHealth are thus becoming prominent components of health care [10].

A main goal of this phase is the employment of medical data registered systems. Additionally, this phase aims to increase big data capacity to solve problems, such as the poor quality of collected medical data. For example, weak, insufficient, incomplete, incorrect data, or data saved in various formats. The other goal considered in this phase is to increase knowledge and to strengthen the involvement of citizens and communities in the monitoring system; measurable/inadequate use of ICT is the result of inadequate access to medical data and lack of trust in its quality [6]. Many pilot projects are being done worldwide and areas of opportunity are being identified on a global impact. Despite the potential of mHealth applications, the majority of initiatives fail in the pilot stage, thus limiting long-term impact. Barriers to large-scale adoption such as standards, security, and interoperability are also being identified [11].

Health industry (drive health care innovations)

The foundation for any personalised medical treatment is laid by a valid and precise diagnosis. For some diseases this can be a single biomarker, such as the identification of a genetic mutation, however for many diseases a more complex patient profile that moves far beyond "simple" genetics may be needed, including more phenotypical information.

In addition, precision diagnoses can be further optimised when coupled with new technologies, such as those which provide rapid and real-time results and those that can be used at the point of care. This key strategic area will be elaborated in the third workshop and will consider clinical studies, joint research, standardisation, Living Labs, training, technology transfer, and demonstration activities [6].

In this area several basic barriers can be mentioned. First, it can be difficult to get funding for innovative pharmaceutical ventures undertaken by start-up companies, as access to venture capital is severely limited across EU. Also, access to standardised data and biomaterial of sufficient quality is not yet developed to its full potential (see above, KA1), increasing development cost for individual companies. In addition, IP regulation is partially leading to an increasing use of trade secrets instead of patents blocking a free-flowing knowledge transfer. Another barrier is related to changes in EU regulations that alter the processes to be passed to obtain market access for novel diagnostics (Regulation 2017/745 and 2017/246) and pharmaceuticals (Regulation 536/2014), which have been updated by the EU, but which are not yet fully implemented, thus creating some degree of uncertainty for the industry. However, a major hurdle to the market entry of novel health approaches are the processes that need to be undergone for obtaining reimbursement within the national public health systems, as these are highly diverse and differ from country to country, making market introduction especially difficult for innovative start-up companies and putting large international incumbents at an advantage. For decades, big players of the pharmaceutical industry have relied on blockbuster approaches in product development. Increasing patients' stratification by introducing additional diagnostics can potentially reduce their market base instead of increasing it, making it less attractive for such companies to engage in pursuing personalised approaches. In contrast, market introduction of products that do not rely on reimbursement but rather address health-oriented consumers directly, is more straightforward making it more attractive for industries to address this private market instead of public health markets. This may lead to the effect that novel preventive approaches become more accessible to privileged EU citizens. The widespread use of novel monitoring devices such as arm-wrists, smart-watches, etc. is a clear indication of this development, whereas the use of continuous monitoring are rather the exception than the rule in public health settings.

Facilitate the innovation flow in health care

The health care ecosystem faces multiple, complex challenges: increase in chronic diseases, population aging, emergence of new issues (health promotion, aging disability, social isolation, etc.), increase in social and territorial health inequalities, failing to seek medical treatment, increase in the cost of certain treatments, expectations for personalised approaches to care, etc., as well as the obvious financial constraints on the health care ecosystem. We observe a large spectrum of innovative responses to these challenges: technological, product and service innovations, organisational and managerial innovations, innovations in business models, renewal of R&D processes, innovations in governance, management and evaluation, public regulation often inspired by New Public Management, and finally innovations that renew the range of stakeholders in these movements [12]. The basic barrier is a provider-centric model of health care and yet the personal medicine must play a decisive role in the long-term sustainability of health systems. The fourth workshop will

invite leading organisations and experts with successful programmes and experience in the adoption of PM technologies by health care organisations. It will be organised around five subcategories:

a. Research and innovation infrastructures exploitation models to boost innovation.
b. Innovative Procurement Tools (PCP & PPI).
c. Screening and prevention programmes.
d. Procurement based on clinical outcomes from PM technologies.
e. Smart and future hospitals [4].

Socio-economic aspects

In order to guarantee the social and economic sustainability of health care, personalised health needs to produce changes in: A) training/education – new managers and professional figures need to be trained; B) facilitate a vertical integration between basic, translational research, technological development, and innovation processes; C) empower patients and citizens; D) guarantee interdisciplinarity [4].

One of the most relevant issues policy makers around the world have to deal with is the decision over whether or not to fund new health technologies when their uptake promise improved patient outcomes at an additional cost for the health care system compared to standard care [13].

PM is expected to have an impact on health care budgets, however, there is a widespread scepticism about the financial impact of PM. According to the report 56% of managed care executives feel that PM will increase cost of prescription medicines [14]. This is one of the main barriers for the introduction of PM.

PM is becoming one of the most debated topics on public and private health agendas worldwide. It has supporters among the industry, patient organisations, health care professionals, academics, funders, and politicians. Devoting energies and resources to pursue (and hopefully realize) the promises of person-centred health care would seem to be a win-win strategy for a number of stakeholders [13]. The scientific, economic, and societal barriers for these objectives are considerable; overcoming the hurdles will require new ways for scientists to engage with each other, new relations between patients, and industries and finally, will require new strategic partnerships among all stakeholders in the PM field [15].

Some regional and national systems have already created innovation tools, like Innovative Procurement, and screening programmes to facilitate the adoption of these technologies in routine hospital practices. Other health care organisations are creating and refining systems to increase and accelerate the innovation flow around PM in their facilities. Hospitals are also favouring links with the industry through their research and innovation infrastructures. Important lessons learned from all these experiences should certainly contribute to accelerating the adoption of PM technologies across Europe. They should also contribute to the definition of new policies and investment decisions at the European, national, and regional level. Considering the aims, scope, and national and international context of Regions4PerMed, one of the obstacles identified to the achievement of the project results is the Political Commitment. It should be worked on carefully by gathering, assessing, and providing response to the regional authorities in need of it. The use of appropriate dissemination and communication tools is essential to maintain a high level of interest and adequate level of response. Connected to the Political Commitment, another potential barrier may be conflicts between regional and national competencies. Especially in those countries where health care systems are managed at a territorial level, it should be very carefully assessed whether activities of Regions4PerMed Action are fully complementary and do not conflict with national competencies [4].

Scientific contribution

Qualitative study - semi-structured qualitative interviews

Qualitative research focuses on understanding a research query as either a humanistic or idealistic approach. Qualitative method is used to understand people's beliefs, experiences, attitudes, behaviour, and interactions, which generates non-numerical data. The integration of qualitative research into studies is a research strategy that is gaining increased attention across disciplines. Although once viewed as philosophically incongruent with experimental research, qualitative research is now recognised for its ability to add a new dimension to interventional studies that cannot be obtained through measurement of variables alone [16]. Qualitative research gives voice to the participants in the study. Semi-structured in-depth interviews are commonly used in qualitative research and are the most frequent qualitative data source in health services research. This method typically consists of a dialogue between researcher and participant, guided by a flexible interview protocol and supplemented by follow-up questions, probes, and comments. The method allows the researcher to collect open-ended data, to explore participant thoughts, feelings, and beliefs about a particular topic and to delve deeply into personal and sometimes sensitive issues. Even with few resources, researchers can use semi-structured interviews. In contrast to i.e. surveys, researchers can conduct a highly meaningful project with interviews with as few as 8–12 participants. Semi-structured interviews can be conducted in multiple ways (i.e., face to face, telephone, text/email, individual, group, brief, or in-depth) [17].

A focus group, also known as a focus group interview, is a moderated conversation of several people on a designated area of interest. This qualitative method is one of the necessary tools. Focus research is mainly

aimed at identifying research problems and deepening quantitative interviews previously conducted.

Wroclaw Medical University, an academic partner of the consortium, would like to add a scientific aspect to the project. After each key action a focus group or semi-structured qualitative interview shall take place. The questions asked during the research will concern barriers and facilitators during PM implementation in the country of a subject. Questions will be asked to members of the project advisory board/representatives of the Interregional Committee from the region where the actions take place and chosen conference/workshop speakers. The question will concern every key action mentioned above: medical big data and electronic medical records; health technology in connected and integrated care; health industry; facilitation of the Innovation flow in health care; socio-economic aspects.

Expected outcome of the project

In order to create an environment in which PM can thrive for the patients' best outcomes, there is an urgent need for systematic actions to remove as many barriers as possible [18]. The focus group analyses along with semi-structured interviews are supposed to bring more qualitative data to the discussion. The qualitative study outcomes could be implemented into daily practice of the health care system's stakeholders through the best practices transferred to all five key strategic areas.

For KA1 in terms of Medical Big Data and Electronic Medical Records we suppose that qualitative study outcomes will help in creating a base to build an international cooperation for a continually learning health care infrastructure with real-time knowledge production.

For KA2 in the field of mHealth and eHealth it is desirable to create supra-regional cooperation and a network for building effective mHealth application solutions.

For KA3 in the health industry (drive health care innovations) it is expected, in connection with the participation in the project of the regional authorities, to generate ideas for more harmonised approaches for reimbursement decisions that would be sent, in the form of recommendations, to central authorities of European countries to support the market entry of PM solutions within the public health care systems.

KA4 in the area of facilitating the innovation flow in health care, it is expected to make health care authorities more aware of their innovation needs and more aware of how to acquire research and innovation products.

KA5 in terms of socio-economic aspects could benefit from the qualitative study by identifying the biggest mistakes in the creation of educational materials and approaches for the training of managers and other professionals in the field of PM.

Acknowledgements

The authors would like to thank Gianni D'Errico and Eva-Maria Stegemann for their comments and their insightful suggestions and careful reading of the manuscript.

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Word count: 3177 • Tables: – • Figures: – • References: 18

Sources of funding:
The Coordination and Support Action Regions4PerMed has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 825812.

Conflicts of interests:
The authors report that there were no conflicts of interest.

Cite this article as:
Stefanicka-Wojtas D, Duda-Sikuła M, Kurpas D.
Personalised medicine – best practices exchange and personal health implementation in European regions – a qualitative study concept under the Regions4PerMed (H2020) project.
MSP 2020; 14, 2: 64–69.

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Received: 12.06.2020
Reviewed: 19.06.2020
Accepted: 24.06.2020