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Recommendations on the implementation of PM in regions based on case studies

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1.0 About the SAPHIRe-project

SAPHIRe, the Consortium for Securing the Adoption of Personalised Health (PH) in Regions, funded by the EU, aims to structure the application of personalised health in all European regions, including sparsely populated and remote regions, which will drive the transition towards sustainable healthcare and personalised health to the next level.

The project supports the agenda of the International Consortium for PM (ICPerMed), which was formally established in November 2016 at the initiative of the European Commission.

SAPHIRe is coordinated by the Department Economy, Science and Innovation (EWI) of the Flemish Government. Other consortium members include the Public Health Agency (PHA) from Northern Ireland, Hungary-based EIT Health InnoStars and the EuroBioForum Foundation (The Netherlands).

1.1 About the report – Deliverable 5.5

Case studies on best practice of Personalised Medicine (PM) implementation (M12 –M36): Case studies will be used to produce a set of best practice procedures for a fruitful implementation of PM research into real-world applications. These case studies will be accessible for all target groups via the website and presented within task 5.6. These case studies summarise the most important results derived from the discussions and work within the Work Packages.

2.0 Introduction

Although research and development in PM is widespread, its adoption into practice remains challenging. Best practice case studies can provide useful reference points to those interested in implementing PM in their regions. This paper will identify best practice examples in PM as well as some of the most common challenges and barriers to the implementation of PM, along with recommendations for improvement. The challenges, barriers and recommendations for improvement have been gathered from SAPHIRe stakeholders during workshops, interviews and roundtables. Desk research of the current literature has also been included.

During our various discussions with stakeholders there have been a number of recurring themes on how best practice can be achieved in PM and various barriers and challenges that need to be overcome. These themes include:

- Data use for PM
- The use of eHealth apps in PM
- Skills training for the future
- Cross collaboration
- Engagement of citizens
- Policy considerations

These themes will be discussed below in more detail.

3.0 Data use for personalised medicine

The use of data for PM allows us to understand health and disease, and it allows for improved strategic planning and decision making. The Department of Health and Social Care (UK) policy paper recognises "the potential of cutting-edge technologies to support preventative, predictive and personalised care is huge"[1].There are many examples of how data has been used to this effect, for example: the 100,000 Genomes Project has been able to identify underlying genetic changes responsible for rare disease[2]; and using machine learning to identify early signs of eye disease[3]

In previous SAPHIRe workshops and round table events our stakeholders agreed that the large scale integration of data will create opportunities to improve healthcare. There are still however a number of issues of concern around the data landscape.

SAPHIRe Stakeholders agreed that in order to further develop the data landscape and improve its integration with health and social care systems:

- Personal data needs to be managed sensitively and appropriately taking privacy issues into account– this includes ethical, legal, security and governance aspects. Proper data governance procedures are needed to build trust and ensure management of data across platforms.
- Data can only be useful if it is available and of sufficient quality; it also needs to be well characterised and compatible with healthcare systems - Across healthcare, interoperability of data remains one of the greatest challenges.
- Clinical decision-making needs complex data to be presented in a quick and easily interpretable form.
- Fragmentation of data in healthcare systems throughout Europe remains a problem.
- Data needs to be FAIR (findable, accessible, interoperable, reusable).
- There needs to be widespread acceptance and adoption of artificial intelligence in healthcare.

3.1 Data security

Across EU Member States and regions there is a varied and rich collection of medical and health data, such as electronic health records (EHRs), prescribing data, diagnostic and laboratory data and disease registry data. This data holds huge potential to support medical and health research and public health interventions. Open and trusted health data systems can help Europe respond to the variety of different challenges currently facing society and the economy today, and European health data ecosystems are maturing to support secondary use of health data[4].

The European General Data Protection Regulation (GDPR) remains one of the main challenges around the reuse of data (secondary use). There is a need for the development of governance tools to allow data reuse and to understand secondary data use; this includes for example: codes of

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[1] Department of Health and Social Care, 2018. *The future of healthcare: our vision for digital, data and technology in health and care*.
[2] 100,000 Genomes Project | Genomics England
[3] Breakthrough in AI technology to improve care for patients | Moorfields Eye Hospital NHS Foundation Trust
[4] Open Data Institute, 2021. *Secondary use of health data in Europe*. [online] Available at: <http://theodi.org/wp-content/uploads/2021/09/Secondary-use-of-Health-Data-In-Europe-ODI-Roche-Report-2021-5.pdf> [Accessed 25 January 2022].

conduct, ethics committees, infrastructure for real-world data and real-world evidence, stronger data institutions, and a clearer legal frameworks[5].

In response to this the European Medicines Agency (EMA) in collaboration with the European Commission (EC) initiated a project "EHR: access, share, expand"[6] [7] under the Health Policy Agencies Collaboration (HPAC) programme. As part of the project, a set of Questions and Answers on GDPR and the secondary use of data for use in medicine and public health are being developed in order to improve and facilitate compliance with data protection rules, as well as helping patients and the public understand their rights and the safeguards that are in place to protect personal data[8].

3.2 Data quality and interoperability

Data interoperability still remains a challenge in healthcare. Interoperability refers to the ability of data to flow from one location to another, without effort when it is needed. Fragmentation of healthcare systems means that interoperability is still an issue and significant effort will be required to establish strong health-data ecosystems and the infrastructure required for reusing data[9].

3.3 The European Health Data Space

The TEHDAS project, involving 25 countries, has been set up to develop joint European principles for the secondary use of health data. The goal of THEDAS is to ensure that in the future European citizens, communities and companies will benefit from secure and seamless access to health data regardless of where it is stored. Results from the project to date can be accessed here. THEDAS proposed the creation of a European Health Data Space. The creation of a European Health Data Space is one of the priorities of the Commission for 2019-2025, including the health sector. €77 million has been allocated to its establishment. A common European Health Data Space will promote better exchange and access to different types of health data (EHRs, genomics data, registry data), not only to support healthcare delivery (primary use of data) but also to support health research and health policy making purposes (secondary use of data).

To exploit the full potential of health data exchange, the data must be of good quality, and the various sources of health data (e.g. EHRs, registries, IT or digital tools) are able to interact with each other. This requires interoperability between the different infrastructures and IT systems. It is also crucial that health data are findable, accessible, interoperable and reusable (FAIR). The Commission is supporting mapping and "FAIR-ification" of existing health data registries and other data sources in order to establish common data sets for exchange for health research and policy uses.

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[5] Open Data Institute, 2021. *Secondary use of health data in Europe*. [online] Available at: <http://theodi.org/wp-content/uploads/2021/09/Secondary-use-of-Health-Data-In-Europe-ODI-Roche-Report-2021-5.pdf> [Accessed 25 January 2022].

[6] Brosch, S., 2020. *Electronic Health Record: Access, Share, Expand Project: Project objectives*

[7] Eotvos, O. and Brosch, S., 2020. *Electronic Health Record: Access, Share, Expand Project Secondary use of healthcare data; groundwork for Q&A*.

[8] https://www.encepp.eu/events/documents/Discussionpaper.pdf

[9] Open Data Institute, 2021. *Secondary use of health data in Europe*. [online] Available at: <http://theodi.org/wp-content/uploads/2021/09/Secondary-use-of-Health-Data-In-Europe-ODI-Roche-Report-2021-5.pdf> [Accessed 25 January 2022].

The European Health Data Space will be built on 3 main pillars:

- Strong data governance systems and rules for data exchange
- Data quality
- Strong infrastructure and interoperability

3.4 Best practice examples of data use in healthcare:

1. Belgium's series of data portals are good examples of how data can be linked in a structured way:
   a. Brusafe - A computer server that permits the electronic and secure sharing of encrypted and structured health data.
   b. CoZo: The Collaborative Care Platform (CoZo) is a digital collaboration platform that allows patients, carers and healthcare institutions to swiftly and securely exchange and share medical data
   c. nexuzhealth (mynexuz.be)

2. Since 2003 French health insurance databases are organised into a huge digital data warehouse, the Système national d'information inter-régime de l'assurance maladie (SNIIR-AM). It covers the entire French population[10].

3. In Finland, citizens are able to add their own health data from apps and devices to their MyKanta portal.

4.0 eHealth

With the advent of new technologies and applications in recent years, ehealth has been changing the landscape of healthcare and has the potential to provide opportunities for patient-centered healthcare. Technologies such as EHRs can provide opportunities for disease monitoring and feedback from healthcare professionals[11], and for improved self-care through self-management interventions[12]. Similar challenges apply to ehealth as to data, for example data ownership and privacy, and interoperability between different types of e-health technologies. There has also been some concerns about data quality that can be accessed through apps and patients' health literacy [9].

4.1 Best practice examples: eHealth

The best practice examples below were presented at the SAPHIRe best practice in PM workshop (2019) by representatives from Extremadura, the Autonomous Province of Trento, Flanders and Podlaskie. Examples 1-3 have all integrated data from multiple sources in order to try and maximise the power and usefulness of the app (no.4 has not yet been integrated with EHRs); they are all also

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[10] Moulis, G., Lapeyre-Mestre, M., Palmaro, A., Pugnet, G., Montastruc, J. and Sailler, L., 2015. French health insurance databases: What interest for medical research? La Revue de Médecine Interne, 36(6), pp.411-417.

[11] Tang, P. C., et al. (2006). Personal health records: Definitions, benefits, and strategies for overcoming barriers to adoption. Journal of the American Medical Informatics Association, 13(2), 121–126.

[12] Al-Ubaydli, M. How social networks enable patients to be more involved in their healthcare. The Guardian April 17 2012; Available from: http://www.guardian.co.uk/healthcare-network/2012/apr/17/patients-social-networks-new-technologies.

empowering citizens to take control of their own health, promoting a move towards preventative healthcare.

1. The **electronic health record (EHR) in Extremadura** is a fully integrated medical record with 100% electronic prescription capability. There is a single medical record per patient and integration with the population database, departmental management systems, financial, human resources, procurements and contracts and management control systems. Medical appointments can be scheduled by phone/SMS (text message)/internet. The crucial aspect of this system is that it is fully integrated and that is why it works well. It was government mandated and all citizens had to adopt it.

2. As a result of the increasing elderly population, and a rise in chronic diseases with no sustainable costs to support either, the **Autonomous Province of Trento** has developed a prescription-based health app to help face these challenges. The **Trec app** is currently being used by over 170,000 citizens. The app integrates information from both the patients' clinical data / lifestyle data and the clinical professionals and healthcare service providers. The app provides a personalised means of healthcare for citizens. It allows users to access their EHR from their mobile phone in order to consult their own medical reports, and order and collect specialist prescriptions and medicines.

3. **Emma.health**, as presented by the Flanders' region is a preventative, PM app that aims to put people at the centre of their own health, and empowers people to take control of their own health needs. The app provides tailored health information, linked to the patients' GP record.

4. **IMAGENE.ME** was presented by the Podlaskie region. It is an app system consisting of a DNA test, Genetic Predisposition Report, teleconsultation with specialists and a PM application.

5. **The ReMIND project**: Robotic ePartner for Multitarget INnovative activation of people with Dementia - The ReMIND project aims to enhance the quality of life of patients with mild neuro-cognitive impairments by stimulating the cognitive and physical activity through music, pictures and physical exercises; to evoke positive moods and emotions and to support social interactions. The ReMIND ICT solution is an interactive combination of robot and tablet applications.

5.0 Skills training for the future

During the SAPHIRe workshops and round tables one common theme from stakeholders, when discussing the implementation of PM in regions, was that of skills training amongst healthcare professionals. It is clear that as the number of patients with co-morbidities increases and our ageing population expands a more holistic view of patients is required, which will require more interdisciplinary collaboration, communication and coordination between staff and departments. There has also been a surge in the use of new technologies in recent years within the healthcare sector and this requires new ways of working together with both colleagues and patients and their families.

5.1 Best practice examples: Skills training for the future

1. [The DISH project, as presented by Southern Denmark](#) demonstrated how they are addressing abovementioned issues. In order for PM to become embedded into the healthcare agenda it is:

- Necessary to work better and closer together
- A multidisciplinary approach and collaboration is required
- Essential to be being able to identify flexible solutions
- Interaction between the clinic, research, technologies, patient safety and ethics is crucial
- Important to empower the patient

2. **The PRECeDI and ExACT projects**

[The Personalized PREvention of Chronic DIseases consortium "PRECeDI](#)" generates high-quality, multidisciplinary collaboration through exchanging knowledge in research activities within the field of personalised prevention of chronic diseases.

The aim of the PRECeDI consortium is to promote knowledge transfer between academic and non-academic entities that can lead to a proper integration of –omics information into public health interventions. The main goal of this platform is to cover an existing gap in the evidence-base use of the –omics approach in the prevention of chronic diseases, by sharing knowledge, building synergies and expertise and encouraging an exchange of best practice among top level institutions. In the long run, the results of the consortium activities will enhance the scientific basis for an appropriate implementation of the –omics applications into true benefits for population health.

[The European network staff eXchange for integrAting precision health in the health Care SysTems (ExACT)](#) is using a multidisciplinary approach to the integration of precision health in European health systems with the aim of educating professionals who can contribute to the future of health systems. The scientific topics include: the definition of a framework for the use of big data in healthcare; the definition of new curricula for the next generations of health professionals including leadership; the identification of new citizen engagement models; the adoption of innovative models of health technology assessment for 'omic technologies, and the related ethical, legal and organizational aspects

3. **Undergraduate and postgraduate degree training courses**

[Ulster University offers an undergraduate BSc degree in PM](#). Graduates of this degree course go on to various job roles including: Research Scientists, software developers and data analysts. The degree covers topics such as immunology, clinical decision making, data analytics, genomics, proteomics and metabolomics. There are also a number of Maters level degree courses available in the UK and Ireland in precision medicine[13] [14] [15] aimed at those wanting to pursue a career in industry or academic in precision medicine.

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[13] [Precision Medicine MSc | Prospective Students Graduate (ucl.ac.uk)](#)
[14] [MSc Precision Medicine - course details (2022 entry) | The University of Manchester](#)
[15] [MSc in Precision Medicine | UCD School of Medicine](#)

6.0 Cross collaboration

A common thread throughout discussions with stakeholders was that of cross collaboration. In order to achieve a successful outcome, a multidisciplinary cross-sectorial approach was required in most instances. The value of communication between clinicians, scientists, patients, public and private sector was highlighted. The importance of giving patients a voice was also noted.

6.1 Best practice examples: Cross collaboration

1. [The TOPFIT programme in East Netherlands](#) is an open innovation consortium that promotes the acceleration of innovations in health & wellbeing with a focus on creating a high level innovation ecosystem. There are both national and international cooperation programmes with the aim of creating new collaborations in health, food and tech expertise that serve social impact and citizen science, along with new business development and scale ups.
2. Another successful example of cross collaboration is Flanders' [VIB Grand Challenges programme](#), where the aim is to "Achieve global impact with global research partnerships". Projects selected for funding through the programme are able to collaborate with transdisciplinary external experts and are actively encourage to do so. The programme funds projects aimed at significant societal impact and has a reverse translation outlook, where they expect applicants to begin by looking at the problem in society and then work backwards towards a solution.
3. The requirement to foster collaboration between regions was highlighted as crucial by stakeholders, as addressing this would facilitate the spread of good practice and uptake of PM initiatives. One example where this has already worked well is in Sweden. Within Sweden there are issues such as data sharing between regions, heterogenous diagnostic standards, a plethora of EHR systems and little collaboration. Genomic Medicine Sweden set up [Genomic Medicine Centres in regions](#), in order to foster national collaboration within healthcare, and to harmonize care across regions, create unique national research opportunities, and further industry innovation and collaboration. The centres are organised and run by the host university and university hospital and are able to support specialised care. They offer a wide range of molecular diagnostic tools, and are part of a national coordinated effort.

7.0 Engagement of citizens

The engagement of patients and the public in initiatives in PM was an important theme in stakeholder discussions. It was clear that more needs to be done to involve and inform the public about the work and research that exists in order to gain their support. In order for PM initiatives to be successful in the long term it requires citizens who understand the potential benefits and risks,

who are willing to share their personal data and are able to provide opinions towards decision making[16]

A number of best practice examples exist that demonstrate the engagement of citizens in PM initiatives:

1. The VIB Grand Challenges programme aims to structurally support translational and/or disruptive research programs around a specific 'grand challenge'. One of the programme goals is to make the voice of the patients heard by working collaboratively with scientists, clinicians and patients to better understand unmet needs.

2. **Personal Genome Project UK (PGP-UK)**[17]: In this hybrid research and citizen science project participants agreed for their omics and associated trait, phenotype and health data to be deposited in public databases under open access. The main features that led to the success of the project were: transparency and interoperability at citizen and data levels which resulted in openness; and the introduction of an app (GenoME app) which allowed the public to engage with personal and medical genomics.

3. **Citizen forum on the use of genome information in healthcare**[18]: A forum commissioned by the Federal Ministry of Public Health and Social Affairs of Belgium. This forum demonstrated that citizens were able to contribute to complex discussions and participate in decisions that affect their lives.

8.0 Policy considerations

A lack of uptake of PM initiatives has been somewhat hindered by a lack of clear policy recommendations that would allow its widespread implementation. SAPHIRe stakeholders have agreed that policy is an important area that should be addressed for implementation of PM initiatives; a number of suggestions that were submitted include:

- Long-term success requires a 'whole system' view and working better together across the country/region(s).
- Decisions should be taken at policy level to help with the integration of health data as this is a key asset. It is vital there is a connected, inter-operable network if data is to be used in a meaningful way.
- It is important to fully utilise existing infrastructure investments for the adoption of new technologies to demonstrate the economic value of PM, the benefit to patients, and the potential to make savings for the health service.
- There is a need to enhance genomic medicine in (for example) the NHS, for diagnosis of rare diseases and to ensure that PM can be utilised in cancer treatment (and other chronic disease areas) as soon as possible.

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[16] Budin-Ljøsne I, Harris JR. Ask not what personalized medicine can do for you--ask what you can do for personalized medicine. Public Health Genomics 2015;**18**:131–8

[17] PGP-UK Consortium. Personal Genome Project UK (PGP-UK): a research and citizen science hybrid project in support of personalized medicine. *BMC Med Genomics* **11,** 108 (2018)
[18] citizens-reporten-executive-reportnl-fr.pdf (kuleuven.be)

8.1 Best practice examples: Precision medicine and policy

There has been some progress made in recent years in the policy area and a number of initiatives and working documents are available to support and guide interested parties in this area[19]

International Consortium for PM (ICPerMed) : Established in 2016 this consortium consists of over 30 European and international members representing a mix of research funders and policy-making organisations, together with the European Commission as an observer. ICPerMed works to:

    a. Establish Europe as a global leader in PM research
    b. Support the PM science base through a coordinated approach to research
    c. Provide evidence to demonstrate the benefit of PM to citizens and healthcare systems
    d. Pave the way for PM approaches for citizens

2. The European Commission have put forward a set of measures to increase the availability of data in the EU[20], building on previous initiatives to boost the free flow of non-personal data in the Digital Single Market.

9.0 Conclusion

There are a plethora of issues surrounding the widespread adoption of PM in regions, some of which have been touched on in this report. In order for a connected and interdisciplinary solution to current problems facing PM, regions need to communicate and interact in order to share policies, agendas and ideas; SAPHIRe aims to provide a platform to allow this to happen. By sharing best practice examples regions can learn from each other by identifying and filling knowledge gaps and generating creative and innovative ideas.

Throughout the SAPHIRe project our interviews, roundtables and workshops highlighted the many excellent examples of best practice in PM that are happening in regions across Europe. It also emphasised the need for more cross collaboration and communication, as most regions were not aware what was happening in other regions. The PM landscape across Europe is fragmented, with regions taking a somewhat individual approach. SAPHIRe will continue to work with their stakeholders to drive forward the implementation of PM in European regions.

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[19] Personalized Medicine in Europe, *Clinical and Translational Medicine*, E Nimmesgern,I Benediktsson,I Norstedt, 12 January 2017, Volume10, Issue2 March 2017, Pages 61-63
[20] Data in the EU: Commission steps up efforts to increase availability and boost healthcare data sharing (europa.eu)