

Genperso study

Is Family Medicine ready to implement personalised medicine?

Marie S. Boyer^a, Daniel Widmer^{a,b}, Christine Cohidon^a, Béatrice Desvergne^c, Jacques Cornuz^a, Idris Guessous^d, Daniela Cerqui^e

^a Centre universitaire de médecine générale et santé publique (Unisanté), Université de Lausanne; ^b Médecin généraliste FMH, Lausanne; ^c Centre intégratif de génomique (CIG), Université de Lausanne; ^d Service de médecine de premier recours, Hôpitaux universitaires de Genève; ^e Institut des sciences sociales, Faculté des sciences politiques et sociales, Université de Lausanne

Background

The concept of personalised medicine, also known as «individualised healthcare», «stratified medicine» or «precision medicine» in literature, promises to change the face of traditional evidence-based medicine from the standardised «one-size-fits-all» model to tailored care. It aspires to provide individualised and optimised diagnostic, therapeutic as well as preventive strategies by exploiting the advances in the «omics» platform such as genomics (DNA) and transcriptomics (RNA) [1].

However, it has been demonstrated that primary care professionals are relatively unfamiliar with genomics and the overall concept of personalised medicine [2, 3]. In Switzerland, there is limited visibility of the perceptions and expectations of the patients and general practitioners (GPs). Besides three recent publications [4–6] from the research project named Genperso, part of the initiative «Personalised medicine and Society» [7] promoted by Leenaards Foundation in 2017, the other Swiss studies that addressed personalised medicine had no specific focus in family medicine to our knowledge [8–10]. Genperso [7] aimed to explore the perceptions, attitudes and expectations of GPs and patients in Romandy (the French-speaking part of Switzerland) with regards to personalised medicine in the

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prevention of chronic diseases. To address the objective of Genperso study, a mixed methods research approach [11] was used. A qualitative exploratory phase was conducted first to allow the preparation of a questionnaire for patients and a Delphi with GPs. These two independent but complementary axes (patients and GPs) reflected a global vision for family medicine. The

methodology is further described in the respective articles [4–6].

Some important results of the Genperso study

During the interviews, when no definition was given, both GPs and patients showed confusion over the term «personalised medicine». They described a bio-psychosocial type of medicine, an approach that is holistic and focused on the person. Some patients even compared the concept to an integrative type of medicine, where alternative and complementary methods add to traditional medicine. But when a working definition was given, based on direct-to-consumer genetic tests (DTCGTs) and related scenarios such as the general consent for research on biological samples, GPs agreed on potential benefits, namely, targeted therapy. They remained unconvinced as to the contribution of personalised medicine in preventive medicine but acknowledged they would sustain a central role in patient management and support despite advances in new technologies. However, they raised concerns over increased anxiety, risks of over-medicalisation, data management, patient support and counselling and highlighted the need for interpersonal skills training and further education to understand and adapt to this new development in medicine.

The perceived barriers identified were those of data confidentiality (who has access to these data?), of financial costs incurred (should it be reimbursed by compulsory health insurance?) and of the need for regulatory measures in order to reduce the possibility of two-tiered medicine.

In the quantitative study following the exploratory qualitative phase, 929 patients, from 28 GP practices, completed a questionnaire [4]:

- Approximately 40% had prior knowledge of DTCGTs and disease risk profiling.



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- About 43% of patients reported being interested in obtaining their genetic risk profile whereas 17% would have the test only if the prediction of risk for specific diseases were possible.
- Depending on the disease (type 2 diabetes, colorectal cancer and Alzheimer's disease were given as examples in this study), 65–80% would be willing to make lifestyle and behavioural changes.
- Nearly all patients (about 97%) would discuss the results of their genomic profile with their GP.

The second part of Genperso study was a Delphi study where GPs, as experts, expressed their opinions on different statements [5]. The role of the GP is not questioned in the emergence of DTCGT in primary care. On the contrary, many believed they could provide resources and patient guidance throughout the procedure. They underlined the need for further training, namely in the interpretation of genetic data, in managing the potential risk of over-medicalisation and ethical issues. An appropriate legal framework, professional code of ethics and organisation of the increased administrative workload would be necessary. Nevertheless, a few non-consensus viewpoints were also identified in the study. Doctors are not in complete agreement with their proactive role in innovative concepts, partly because they remain doubtful of the added value of genomics in comparison with existing and validated tools and guidelines. Some GPs believe that changes in preventive strategies should remain in public health.

Update after the Genperso study

The results of this Swiss project reflect similar findings to previous publications related to personalised medicine in primary care [3, 12]. Where do we now stand?

What are the next steps to consider? Instead of clear answers, the Genperso publications have raised even more questions that would have to be addressed, particularly if further education and training were to be available to GPs.

1. Certainly, personalised medicine is presently well integrated in oncology, where the technology of genome sequencing has allowed the prediction of prognosis and therapeutic response to specific drugs. Given the prevalence of other chronic diseases such as type 2 diabetes and cardiovascular diseases, mostly managed by GPs, **could the personalised medicine approach be an invaluable asset in family medicine and public health by using genomic profiling for disease risk assessment?**
2. **Would the public use direct-to-consumer genetic tests to learn about their disease risks?** In recent years, DTCGTs have become readily available to the general public via a simple purchase over the Internet. Offers from different companies vary from simple testing for ancestry and genealogy to assessing one's risks of certain health conditions and traits. Studies have shown that if patients choose to have a DTCGT, they seek help from their GPs for the interpretation of the results, as well as for counselling, support and medical recommendations. Hence, with the growing public interest and the ease of access to DTCGT, the involvement of GPs seems to be inevitable, as confirmed by Genperso studies.
3. **What would be the right term for this type of medicine?** The general confusion over the terminology and concept is also confirmed in this study [13,14]. The label "personalised" could be the cause of the misinterpretation, particularly in a field of medicine where the doctor-patient relationship is nurtured. Many have discussed the need for the right terminology, for example "precision medicine" as stated by Jaccard et al. [15]. To our mind, the term "genomic medicine" seems more fitting and clearer to doctors and patients.
4. **How to advance from a lack of knowledge and interest to commitment?** GPs avoid adhering unconditionally to new technologies even if they admit that genomics could provide better tools in preventive medicine and targeted therapies. They are rarely challenged with the monitoring of rare genetic diseases, but they should be able to provide support and accurate information to already informed patients. So, their central role in the healthcare system means that they would need to embrace the advent of innovative technologies to help them in everyday care. To help GPs commit to innovation, well-founded evidence-based research are required.

5. **How to prevent over-medicalisation?** Over-medicalisation often begins when existential issues are disregarded, and further unnecessary investigations and treatments are prescribed to respond to patients' demands. This is a recurring concern amongst GPs as genomic medicine takes after the biomedical model and overlooks the bio-psycho-social vision of today's medicine. This could potentially impact the doctor-patient relationship as more time is consumed in unwarranted tests. According to Vogt et al., the risk of over-diagnosis can be reduced through shared decision-making, hence affirming the importance of trusting doctor-patient relationships.

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6. **How to deal with ethical and social issues?** GPs interviewed have highlighted potential confidentiality, discrimination, and social issues (social insurance and costs) linked to personalised medicine. They advocate a legal framework. Medical continuing education should also provide appropriate training on how to address patients' questions and uncertainties regarding this innovative technology.
7. **What would be the role of GPs in evaluating new technologies?** We are convinced that GPs have an important role in the evaluation of new technologies, as shown by the Genperso research. It is unreasonable to validate and implement new technologies in a medical field without any concern about the professionals who would be involved in its use. GP's discourse must be heard, alongside that of their patients, technology promoters, payers, patient associations, philosophers and sociologists.

Conclusion

In short, the personalised medicine approach has piqued GPs' interest in many ways. It has also raised many concerns, namely pedagogical, socio-ethical, data protection, regulatory clarity and cost issues, which must be settled before implementing this approach in primary care. Despite the potential benefits of personalised medicine in the prevention of chronic diseases, the concerns raised are very pertinent and we must consider the possible consequences that such medicine could have in primary care and in society.

Disclosure statement

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Marie Sandy Boyer
Centre universitaire de
médecine générale et santé
publique (Unisanté) Université de Lausanne
Rue du Bugnon 44
CH-1011 Lausanne
boyer.mariesandy[at]
gmail.com