New challenges of public health: bringing the future of personalised healthcare into focus

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The greater personalization of healthcare represents a driver of innovation for research, and for the healthcare systems and industries as a whole. Still policy-makers, healthcare professionals, citizens and private companies need to take some steps to realize the potential for such a radical shift. In this paper, we illustrate the challenges, the benefits and consequences that might accompany the implementation of personalized healthcare, and the steps that policy-makers and practitioners would need to take to realise its potential. Six main prerequisites for radical change in healthcare are presented, that include achieving better genetic literacy for professionals and for the public; engaging citizen in the discourse; improved governance, consent and trust in healthcare; feeding and harnessing the data–knowledge cycle for better health; adopting and adapting the Health Technology Assessment framework for the evaluation of the new technologies; and retaining humanity and community in health and care. Some of these concepts originate from a discussion on the future of health and healthcare, looking at least 15–20 years into the future, that we had at the end of 2016 at Ickworth with an international group of experts, under the aegis of the PHG Foundation.

The scenario

Healthcare around the world is at a crossroads, with financial pressures undermining the sustainability of health systems. As highlighted by the European Steering Group on Sustainable Healthcare, a sustainable healthcare requires a shift from treatment of established disease to disease prevention and early diagnosis, and it relies on the need to engage citizens to take greater responsibility for their health in order to establish a more participatory healthcare model, instead of a paternalistic one.1 In order to keep healthcare sustainable, few elements appear to be a consistent feature of all reports that seek to address these issues: placing the individual citizen at the centre of health systems; increasing the emphasis on prevention; a radical reorganization of services in which care is moved from a separated model (hospital/community) to an integrative one in which hospital and community care work together; and a systematic evaluation of the new and existing technologies using the Health Technology Assessment (HTA) and Health Outcomes Research approaches to invest or procure based on real value.

A great challenge to a sustainable healthcare is currently represented by the advancement of biomedical and digital science and technologies, that jointly with the demographic trends, and the expectation of citizens, are suspected to act to widen the gap between the available resources and the requirements for healthcare. In imaging future healthcare, in fact, some core technology-based components of a more personalized approach to health can be envisaged: the availability of comprehensive electronic health records; the use of biomarkers assays including whole genome sequencing at key points in life course; and the continuous self-monitoring of lifestyle parameters using mobile connectivity systems. These components can be considered as a number of inputs and outputs comprising a system that aims to achieve better health for individuals, and populations, through delivering more effective preventive and therapeutic interventions. The inputs are the data themselves from multiple sources and knowledge abstracted from these that will be accrued by both healthcare providers and citizens themselves. The outputs of such Big Data will be the stratification of populations on the basis of their risk of particular diseases and/or their responsiveness to particular therapeutic interventions.2

The hypothesis underlying the construction of such a system relies on the assumption that targeting interventions to groups or individuals who are most likely to be responsive to those interventions could enable significant improvements in health compared with current ‘one size fits all’ approach to preventive and clinical healthcare. This transformation in the vision and practice of medicine has been variably called personalized medicine, precision medicine and ultimately precision health.3,4 For most clinicians, till now, the tangible benefits of personalized medicine have been in cancer care, either in the prevention, diagnosis and treatment; in the diagnosis of monogenic and syndromic conditions; and in the field of reproductive health.5,6 The implementation of precision health, in fact, remains contingent on significant data acquisition and timely analysis to determine the most appropriate basis on which to tailor health optimization for individual in the prevention, diagnosis and disease treatment. A personalized approach to healthcare also raises unique ethical issues because of the informed consent process, ownership of data (including relatives’ implications), return of results, privacy and confidentiality, and extent of current uncertainties with respect to genomic data interpretations and disease associations. Achieving effective and proportionate governance of health and health-related data will be essential for the future healthcare systems.

In 2011, the EC engaged an interdisciplinary and independent group to provide non-binding advice on matters related to effective, accessible and resilient health systems. This expert panel on effective ways of investing in health, has been called to deliver opinions in support of health policy-making in the EU in some areas, among which the implications of disruptive innovation for health and healthcare in Europe.7 In 2016, the panel published a report where some strategic areas for disruptive innovation were identified, including personalized healthcare. In the meanwhile, we had published the results of a survey on the policies in the field of genomics in the EU Member States countries, by interviewing...
the Chief Medical Officers. Although the results were quite encouraging, the situation was very heterogeneous, thus requiring the need for a common policy framework in this context. In the meanwhile, an EU roadmap has been designed around the topic of personalized medicine, starting from the Personalized Medicine (PerMed) initiative launched in 2014 from the European Commission. This project will be continuing in the next few years under the umbrella of the International Consortium for PerMed (ICPerMed) that aims to foster and coordinate research and innovation activities to deliver personalized medicine approaches for EU citizens. In the UK, the NHS has embarked on the 100 000 Genomes Project, a bold multiyear initiative to transform UK biomedical research and clinical care to a precision-health oriented model. Outside Europe, currently large public and private organizations are investing heavily in developing precision health, including, ‘All of Us’ of the NIH (formally Precision Medicine Initiative, https://allofus.nih.gov/), and the large initiative of data collection from the system catchment area of the Kaiser Permanente in California. Similarly, China, Iceland and Japan have major, long-term precision health initiatives.

Potential benefits and challenges of personalized healthcare

In October 2016, together with international experts in health, public policy, public health, law and ethics from EU, USA and Canada, we had convened in Ickworth, UK, under the aegis of the PHG Foundation, to discuss on the future of health and healthcare, looking at least 15–20 years into the future. The discussion had already started in 2015, but this time the meeting addressed specifically the benefits and consequences that might accompany a greater personalization of healthcare, and some of the steps that policy-makers and practitioners would need to take to realize its potential. During the meeting, the experts had listed the potential benefits and challenges of personalized healthcare, which are reported in table 1.

Prerequisites for radical change in healthcare

For personalized approaches to be successful, the experts convened in Ickworth identified a radical set of actions. Firstly, ‘achieving better genetic literacy for professionals and for publics’, through increased education and training of professionals. Some initiatives at EU level are already going in this direction, such as the platform for primary care physicians developed by the Gen- Equip project (https://www.primarycaregenetics.org/?page_id=1098&lang=en). Other initiatives at national level include an online decision aid for genetic testing used in the Netherlands (http://www.keuzehulp-dnaonderzoek.nl), an online distance learning platform for educating physicians in the field of genomics in Italy (https://www.eduiss.it/course/info.php?id=208). At the global level, the JAMA journal has just launched an initiative called ‘JAMA Insights: Genomic and Precision Health’, consisting of a series of educational articles intending to help non-geneticist clinicians to overcome knowledge barriers.

Secondly, ‘engaging citizen in the discourse’. The placing of the individual citizen or patient at the centre of our healthcare systems will also challenge the current paradigm of healthcare provision. The eventual success of precision health depends on the well-informed decisions made by patients and the healthcare professionals who provide their treatment. The empowerment of citizens to take responsibility for their own health will need to become a central task for those with responsibility for health services. Personalization implies an approach to health that truly takes account of personal values and preferences, in order to establish a more participatory healthcare model, instead of a paternalistic one. While these attributes of a future health system are desirable, it remains to be determined how they can be developed in ways that are sustainable, equitable and effective. The necessary knowledge and understanding, however, will require explicit programmes of public engagement.

Thirdly, ‘improved governance, consent and trust in healthcare’. From both clinical and research perspectives, it is clear that effective use of these sciences will demand a greater sharing of personal biological data and biological samples. However, this raises some challenges given the scale, detail and personal nature of this information, and concerns about privacy and the extent to which and how patient and research participant data should be collected, stored and used. Currently, the European General Data Protection Regulation allows for Data Protection Boards to authorise Codes of Conduct, which might facilitate more harmonized approaches to be developed. It is probably time to move away from an assumption of ‘hypothetical privacy’ in which individuals ‘own’ their data towards a model in which healthcare systems are regarded as stewards of data. Formalizing reciprocity via novel intellectual property approaches also ensures that benefit flows into the system and may be advantageous for all.

Fourthly, ‘feeding and harnessing the data–knowledge cycle for better health’. The development of personalized healthcare remains contingent on significant data acquisition and timely analysis to determine the most appropriate basis to tailor health optimization for individuals. Beside the enormous technical challenge given the diversity of information systems used to generate and record this data and their often poor interoperability, there are regulatory, ethical and legal challenge to disentangle ownership and exploitation rights over this data. This issue is immediately linked to the former, as the move to engage citizens and patients will mean that they will require access to information about themselves. This will have implications for systems that collate and store information such as electronic health records and biobanks, as well as mechanisms developed for storing and sharing a diverse range of data.

Fifthly, with more genomic applications becoming available all the time, it is becoming increasingly important to have systems in place to timely assess and evaluate them, to distinguish between clinically useful and redundant tools, and to understand their possible impact on clinical practice. However, this requires not only investment in the discovery and development of new biomedical technologies, but also translational efforts including resources to reliably determine their benefits, within the ‘HTA framework’. As reported from the Chief Medical Officers of the EU Member States, some Countries including Austria, Belgium, Croatia, France, Hungary, Poland, Spain, the Netherlands and the UK already had in 2015 mechanisms in place for the HTA of new devices and drug, including genomic applications. For new diagnostics including genomics, however, a more comprehensive approach is needed to include proportionate evaluation criteria. In 2005, the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) working group was established in the USA to support the CDC in the development of a systematic process for assessing the available evidence regarding the validity and utility of rapidly emerging genetic tests for clinical practice. Several reports have been published since then (https://www.cdc.gov/egappreviews/), but as genome sequencing moves towards routine clinical practice, new approaches will also be required. As with data, discussed above, there is a fine balance to be drawn between over-regulation and the stifling of innovation and allowing a market with insufficient ethical and legal scrutiny.

Lastly, ‘retaining humanity and community in health and care’. There are concerns that such shift can lead to a depersonalization of the healthcare delivery, changing the nature of the relationship between health professionals and citizens. Personalized approaches, however, might enable citizens to have greater determination over the values that are expressed within value-based health systems even
Table 1 Potential benefits and challenges of moving towards a more personalized approach to achieving better health, identified from the experts convened in Ickworth

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<th>Potential benefits</th>
<th>A more effective and efficient health system</th>
<th>Social benefits</th>
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<td><strong>Clinical practice</strong></td>
<td><strong>Patient benefits</strong></td>
<td><strong>Normalization of genetics for citizens</strong></td>
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<td>Improved confidence in clinical decision-making</td>
<td>Improved health outcomes</td>
<td>Greater predictability of future health enabling more effective prevention</td>
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<td>Bridging the intuitive personalization of medical practice with evidence based medicine</td>
<td>Optimization of the benefit/harm ratio for interventions at an individual level by:</td>
<td>Increased effectiveness and efficiency of interventions through targeting to more responsive subpopulations/individuals</td>
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<td>Rational reclassification of diseases</td>
<td>- Reducing incorrect diagnosis or unnecessary interventions</td>
<td>Cost savings</td>
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<td></td>
<td>- Enhanced personal autonomy</td>
<td>Enabling more effective and efficient interactions between health systems and industries</td>
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<td>- Empowering informed decision-making</td>
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<td></td>
<td>- Greater customization of services for individuals</td>
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<table>
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<th>Potential challenges</th>
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<tr>
<td><strong>Gaps in knowledge and evidence base</strong></td>
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<tr>
<td>Lack of scientific knowledge required to truly ‘individualise’ risk rather than simply defining sub populations at risk</td>
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<td>Lack of professional knowledge and understanding could limit implementation</td>
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<td>Lack of knowledge and understanding could affect acceptability to policy-makers and citizens</td>
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if some aspects are depersonalized, but an underlying ‘community structure’ will be needed.

**Delivering change**

The profession of public health should have an important role to play in achieving these changes, but regrettably most of its practitioners have not yet embraced this agenda. In looking to the steps needed to deliver the fundamental changes in healthcare demanded by personalization, the summit participants in Ickworth found that public health values would be vital, notably an increased role for health systems and individuals in disease prevention and health management. We also envisaged a need for a ‘strongly radical and visionary leadership’ to achieve transformational change in health systems, and the need for these leaders to work effectively with citizens and patients as well as health professionals to deliver such disruptive innovation. In leading the change, public health professionals should continue being the defenders of public health values, particularly the commitment to social justice and equity, and the skills of public health practitioners in gathering and using data to drive quality, efficiency and equity in health systems. As long as the opportunities available to improve the health of a population are finite, there will be a role for policy-makers and public health in shaping how those resources are directed, and their decisions should be based on public health indicators. This would be particularly important as a way to minimize the health inequalities anticipated to arise from differences in access to consumer technologies and the knowledge that would, in the future, provide the cornerstone for much health management activity.

The implementation of a personalized approach to healthcare will require also a ‘change in the organization of health services’. On a wider basis, public health leaders have a responsibility to help catalysing the change in the organization of health services and public policy to ensure that genomic and other technologies are used to best effect. The demographics of health and disease suggest that services will have to cater for a greater number of elderly patients with more chronic diseases, in a context of more complex and often expensive interventions, yet in an environment of increasing financial stringency. Consequently, it is in the fields of disease prevention and chronic disease management where changes in health systems will be most needed and have the greatest impact.

In conclusion, we envisage an urgent need to engage current health system professionals, leaders and citizens in both grasping the opportunities and tackling the challenges of a personalized healthcare. The current economic, social and political forces skew most public discourse around health towards tackling short-term issues of sustainability and inequality facing all health systems. The way in which the public health system adapts to this evolving health ecosystem will be a significant determinant of whether personalization of healthcare can, in the end, lead to greater gains in overall population health.

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