Public Health Genomics and Personalized Healthcare: a pipeline from cell to society

Truly personalized healthcare facilitating the management of personal health drives a fundamental change not just in what is known but also in how we think of ourselves and the way we are living, thus redefining our society (1). We have to prepare now – just in time! – for all the various organizational changes ahead of us. The political will is there. However, the real paradigm shift depends on the willingness to restructure our current policies, to support knowledge transfer to maximize benefit to public health, and – most important – to change our minds. So far, all stakeholders including policy-makers and the private sector are struggling to translate the emerging knowledge into public health. Public Health Genomics (PHG) is the area of public health ensuring that scientific advances in genomics (“from cell...”) triggered by innovative technologies are timely, effectively and responsibly translated into health policies and practice for the benefit of population health (“...to society”).

But, what evidence emerging from basic sciences needs to be translated?

New insights are being obtained from genomics, proteomics, transcriptomics, metabolomics, epigenomics, microbiomics, and other “omics” technologies. As these data are integrated through the use of information and communication technologies (ICT), we are at the edge of achieving an understanding of the systems biology and systems biomedicine of human health and disease that also incorporates environmental contributions such as lifestyle, toxic agents, social and economic factors, as well as health systems determinants. In this way, we can begin to envisage new approaches to the promotion and management of human health across the entire life course of an individual. Indeed, we can now consider a future involving truly personalized healthcare in which technological advances are placed at the service of population health. The evidence we now require to demonstrate the benefit of new technologies will need to follow a new paradigm, however. What is required is an assessment of individual benefit rather than overall effects in large populations or even subpopulations of patients. Thus, public health assessment and evaluation tools must now address concepts such as “personal utility” rather than clinical utility.

These developments and the involvement of the patient brought forward the concept of P4 (predictive, preventive, personalized and participatory) medicine serving already as a blueprint for Public Health Genomics to prepare healthcare systems and policy-makers for this shift in our approach to healthcare. The P4 medicine is no longer a vision, it is a mission!

We can and should go beyond the P4 medicine and recognize in the light of a “systems approach to public health”, that (2):

- Common complex diseases can be considered in terms of a constellation of “rare” diseases, each of which reflects a complex biological system.
- We are moving away from a traditional classification of disease and towards groups of shared pathology that can be described as “diseasomes” or disease nodes.
- We are moving away from a focus on risk factors within biostatistical models of populations and towards an emphasis of individual pathways or networks.
- It is time to emphasize personal rather than clinical utility.

Thus, as the user or patient become aware of knowledge through the influx of information present on the internet and (social) media and related online tools and digital libraries, the user demands personalized or preferentially individualized approaches towards his/her health interventions and wants to be more involved not only in the decision-making process but also in managing their own personal health. Therefore, individuals should have the lifelong skills to find and assess the relevant and reliable information on the internet, which is also related to the concept of health literacy. In Europe, the consortium of the European Health Literacy Project (HLS-EU) defined four dimensions of health literacy (3). Using this definition, health literacy can be used as a catalyst to accelerate the accessibility, understandability, appraisal, and application of genome-based information matching the needs within different population groups. In this...
context, examples such as the internet can provide a powerful health literacy-friendly electronic environment facilitating self-management, simplicity and user-centered applications. Thus, the future patient may turn from a passive consumer of health interventions into a proactive consumer or “prosumer”.

Until now we still see just incremental progress and changes leading to personalized (stratified) medicine and precision medicine. There is nothing new here. However, a radically new vision for healthcare is knocking at our doors. What is different here, what is visionary, what is disruptive?

Combined genomic and phenotypic analysis has become possible owing to the increasing role of ICT in healthcare driven by improved technological options and the interoperability of various technologies. The complexity of the task when applied to diagnosis and therapy, however, demands algorithms and mathematical models to reduce uncertainties. As a result, efforts are now being made to generate computational models of individual persons (“virtual twins”). Such models can be used to follow individuals throughout their lifetime and enable health professionals to virtually simulate and optimize treatments, as well as all types of interventions. Traditional medical decision-making may turn into in silico decision-making. In this way, it becomes possible to improve the safety, quality, effectiveness, and efficiency of healthcare services. In addition, by following individuals rather than remaining tied to a given healthcare system, it will enable citizens to handle and access personal health-related data whenever needed.

IT Future of Medicine (ITFoM) provides such a platform for Europe being one of six pilot projects in the European Future and Emerging Technologies Flagship scheme (4). It is a very ambitious project, which aims to harness the vast potential of ICT to revolutionize human healthcare and targets to ‘lead the way towards truly personalized healthcare’. Dealing with (in space and time) highly dynamic personal (health) information, moving from the understanding of statistical risks within groups to individualized evidence, and using virtual individual models as a tool is not only visionary, it is a radical new vision of healthcare – ICT for health and health for ICT! In a nutshell, stratified medicine will be replaced by truly individualized medicine. No groups exist, only individuals. Every test will be part of treatment. No test result can be transferred to another patient. Every therapy is unique, not reproducible. Furthermore, until now no method exists on how to evaluate this new type of technology: How can we fulfill the hierarchy of evidence, the golden standard of evidence-based medicine to prove the efficacy of a treatment? The patient is not only a consumer of the technology but also part of it. There is no boundary between patient and treatment anymore. The patient is a unique part of the technology itself.

We face a time when boundaries of disciplines are crossed and the understanding of diseases is changed as it happened before with the jump from the macroscopic view in anatomy to the microscopic view in cell structure!

How can we translate these disruptive innovations into healthcare systems?

There is no doubt that health systems across Europe and beyond must prepare for change arriving from the novel ICT solutions developed by highly visionary projects such as ITFoM to reach progress in treating complex diseases (5). To drive this change it has become clear that the future of healthcare depends upon major breakthroughs in science and technology, as well as to translate these breakthroughs in a timely, effective, and efficient manner. This includes proving evidence for the decision-making process, preparing the necessary ethical, legal, economic, and regulatory frameworks for the organizational changes ahead of us, and setting up sustainable tools for the constant technology and knowledge transfer. At present, there are no models or tools that facilitate the timely implementation of health innovations such as individualized diagnostics or drugs into the healthcare systems. Tools that are currently available either support the industry side (technology transfer principles) or the health policy side (public health assessment tools), which means there is a lack of tools that combine both ‘worlds’ and form a bridge between industry and health policy-related stakeholders.

The LAL model (Learning Adapting Leveling) has been recently developed (6) and is being piloted. It covers the whole process from the first idea of a product to its implementation in the healthcare system. The core of the LAL model revolves around a relative parallel initiation of the Technology Transfer (TT) activity and the Public Health Assessment Tools (PHATs), the latter of which include Health Needs Assessment (HNA), Health Technology Assessment (HTA), and Health Impact Assessment (HIA) (7). It is an overarching framework which focuses on the timely as well as real-time integration of relevant technologies into healthcare systems through early-on involvement of all stakeholders through crosstalk, bilateral communication, consultation, public-private partnerships, and collaboration. This will help to reduce the gap in the integration timeframe of health innovations. Furthermore, this will also help to come to an early-on strategic decision on the ongoing technology development through feedback on the direction in relation to its evidence and perceived value in healthcare, because it addresses the solutions around bottlenecks in healthcare implementation during the development of a product.
Thus, the LAL model is highly innovative and can be seen as a raw model or best practice for all industries in Europe and beyond, because it will accelerate and increase not only the likelihood of successful market introduction of personalized health interventions but also of their application in the healthcare system as a whole. Although challenging existing European frameworks for assessing effectiveness of healthcare interventions, it will provide solutions meeting the Europe 2020 goals of growth, innovation, and social inclusion, as well as contributing to the Innovation Union and also to the Horizon 2020 goals.

In a nutshell, we have to be prepared in time, and we have to define today what type of (policy) guidelines we need for tomorrow – the future is built today!

The Public Health Genomics European Network (PHGEN) has been asked by the European Commission to fulfill this task and to produce the first edition of “European Best Practice Guidelines for Quality Assurance, Provision and Use of Genome-based Information and Technologies” (8, 9) assisting all EU Member States, Applicant and EFTA-European Economic Area (EEA) countries with evidence-based guidance on the timely and responsible integration of genome-based information and technologies into healthcare systems for the benefit of population health.

On 19 and 20 April 2012, key European and national organizations and institutions from policy-making, academia, and private sector came together at the final PHGEN meeting in Rome – among them the European Society for Pharmacogenomics and Theranostics (ESPT) and the European Medicines Agency (EMA) – to discuss the future of public health genomics and to endorse the Declaration of Rome on 19 April 2012, a summary of the “European Best Practice Guidelines for Quality Assurance, Provision and Use of Genome-based Information and Technologies”, which is published in this issue (10).

The next steps for PHGEN will now be to ensure the implementation of the European best practice guidelines in the different European countries through the efforts of the PHGEN National Task Forces. PHGEN is therefore now working towards a Joint Action in 2013 with the support of the EU Member States. Collaborations between PHGEN and the European Regional Office of the World Health Organization are also being actively explored and links will continue to be maintained with the European Commission.

Indeed, personal health drives a fundamental change not just in what is known but also in how we think of ourselves and the way we are living. It is a change of view that changes everything!

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