European Journal of Public Health, Vol. 24, No. 3, 349–350

doi:10.1093/eurpub/cku030

Editorial

Why is personalized medicine relevant to public health?

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Advances in genomics promise a new era of personalized medicine in health care. A major promise of the 'omics' research is that of delivering new information that can transform health care through earlier diagnosis, more effective prevention programs and a higher precision in the treatment of disease. Almost 10 years after the definition of the term public health genomics (PHG) as 'the responsible and effective translation of genome-based knowledge and technologies into public policy and health services for the benefit of population health', we are still facing the dilemma of how to implement genomics medicine into public health practice.[1]

Five main reasons can explain why progress was so slow. Firstly, from a philosophical point of view, the incorporation of the recent genome discoveries into public health practice deals with an apparent paradox. Although the mission of public health is to improve health from a population perspective, with its unit of intervention being the population, the individual approach of personalized medicine appears to be at odds. Secondly, over a decade after the sequencing of the human genome, few applications showed that the introduction of some genetic tests at the population level led to improved health, except for the newborn screening programs. Thirdly, in times of diminishing resources, new technologies have the potential to divert much needed resources away from what can be done in delivering basic public health services, with well-established evidence of effectiveness and cost-effectiveness. Fourthly, most of the public health practitioners do not have the knowledge to integrate rapidly emerging genomic information into their programs.[2] Finally, behavioural interventions based on the knowledge of inherited risk have mostly failed to demonstrate their effectiveness to a large extent.

I believe that the promise of a rapid integration of genomic discoveries into public health practice was partly unattended also because the promise of genomic science and technology was oversold.[3] If we agree that the paradigm shift in public health service provision, which takes into account the 'omics' data in the risk stratification, is not coming tomorrow, we can see where the path is already moving forward. In a commentary published in 2013, James Evans et al.[4] suggest for a new opportunity to expand the focus of PHG in a way in which its promise can be realized. Rapid and inexpensive sequencing of genes can currently identify individuals carrying individually rare mutations that confer dramatic predisposition to preventable diseases. This concerns, for example, the use of *BRCA* 1 and 2 gene testing for the hereditary breast cancer: the combined prevalence of these mutations is ~0.2–0.3% of the general population, but they confer a >70% lifetime risk for breast and ovarian cancer. Also, the four Lynch-associated genes are present in 0.2% of subjects and confer >80% risk for colon cancer. Even though the public health benefit of screening for rare diseases might seem a paradox, early detection of carriers can lead to a large benefit in terms of mortality reduction from cancer diseases that so far are identified by waiting until these individuals, or some of their family members, develop such diseases. For breast cancer, there are algorithms, based on family history, to assess whether a woman should receive genetic counseling, and if indicated, genetic testing; these tools, however, are not applied systematically in the primary care setting.

Even when the clinical utility of a genomic application has been documented, as in the examples above, there is hardly any research to evaluate the implementation of such applications. This requires substantial investments and a multidisciplinary approach. Resources in health, however, are more devoted to discovery research than to implementation research that aims to assess the decision-making process, the educational needs of providers, patients and public, the cost-effectiveness evaluation and the evaluation of population health outcomes.[5] What we need now, from a public health perspective, is to speed up the implementation of the evidence-based measures that use genome applications to identify high-risk individuals who would benefit from these preventive interventions.

Recently, Ron Zimmern, the founder of the PHG Foundation in the UK and public health specialist, advocated for a fundamental challenge in public health practice that takes into account the two main drivers of genomic science and societal challenges that place the individual autonomy at the centre of the process. If public health practitioners fail to recognize the interaction of the environmental and social factors with genomic and other biological determinants, or to understand the opportunities and challenges of stratifying populations, the practice of public health in the coming decades will be much impoverished, he said.[6] I cannot agree more, and in this sense the newly published call of Horizon 2020 on personalized health care aims to support research that pilots a new model of health care organization within the personalized medicine approach that can be used by policy and decision makers.

In conclusion, I hope readers are convinced that personalized medicine is indeed relevant to public health. The unresolved question is whether public health is ready to meet the challenge. Should we continue being the honest broker of the emerging new technology to inform health practitioners on the evidence-based applications, or should we take a more active role in carrying out implementation research that can assist decision-making processes and inform providers, patients and the general public on the genome-based technology? In a situation where there is a profound gap between our ability of interrogating the human

genome and the ability to use that information to improve health, public health practitioners should take a more active role and embrace the changes by welcoming the innovation and the personalization of health care to ensure that it works for the benefit of population health.

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