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Public Health Genomics

Abstract:

Die zunehmende Veröffentlichung anwendungsnahen genomischen Wissens stellt auch Public Health vor neue Herausforderungen. Public Health Genomics (PHG) ist die Wissenschaft, die sich der Übersetzung von genom-basiertem Wissen in neue Handlungskonzepte und Politiken widmet. Sie greift dabei auf bestehende und neue methodische Konzepte zurück. Eine internationale Arbeitsgruppe hat hierzu mit Unterstützung der Rockefeller Foundation das Bellagio Statement on Public Health Genomics erarbeitet. In Europa hat die Generaldirektion Gesundheit diese Entwicklung zum Anlass genommen ein neues europäisches Forschungsnetzwerk zu unterstützen. Im Jahr 2006 hat das Public Health Genomics European Network (PHGEN) seine Tätigkeit aufgenommen. In Arbeitsgruppen auf europäischer Ebene, aber auch durch PHGEN National Task Forces in den Mitgliedsstaaten arbeitet das Netzwerk an neuen Konzepten und Empfehlungen, die langfristig einen Paradigmenwechsel in Public Health begleiten. Durch die Möglichkeiten der genetischen Diagnostik und die Fortschritte im Bereich der Systembiologie werden Mediziner und Public Health Experten zunehmend in die Lage versetzt, die Verlaufspfade von genetisch beeinflussten Erkrankungen zu verstehen. Dies bedeutet, dass Medizin und Public Health sich stärker den Kausalitätsketten zuwenden können, ohne das Entstehen von Symptomen abwarten zu müssen. Die Möglichkeiten der Gesundheitsüberwachung und der Gesundheitsberichterstattung verbessern sich dadurch erheblich. Bisher sind die Gesundheitssysteme nur unzureichend auf diese Schritte vorbereitet. Public Health Genomics, und hierbei speziell das europäische Netzwerk, entwickeln die Konzepte, die es den Beteiligten ermöglichen werden frühzeitig von genom-basiertem Wissen zum Wohle der Gesundheit aller zu profitieren.

The Human Genome Project, the development of new genetic tests, DNA chip technologies and related technologies all offer new opportunities for the promotion of population health which will lead to fundamental challenges in the healthcare delivery systems. Medicine and public health are given an increasing insight into the biological factors which drive disease mechanisms, in particular in the field of cancer [1]. This emerging genome-based knowledge calls for a paradigm shift in public health as we can see a clear need to adjust concepts of prevention and health service delivery. As a consequence we can describe a dichotomy: genomics needs to understand how it can include public health aspects in its work programme while public health needs to analyse how genomics changes the concepts of public health. The second approach is seen as the core task of Public Health Genomics. Still there is an interdependency between the two disciplines; e.g., as Public Health Genomics is also concerned about the organisation of genetic services and the genetic health literacy of the population.

A comprehensive health care system which regards genetic determinants as well as environmental, social and life style factors, will become essential as it creates new opportunities for individualised strategies in preventive medicine and early detection of

illnesses. The integration of genome-based knowledge will change primary, secondary and tertiary prevention. *Inter alia*, disease prevention programmes and clinical interventions will be specifically targeted at susceptible individuals and sub-entities of populations based on their genomic risk profile. So far health care systems, policy makers and industries are not prepared for the conceptual change and all stakeholders are struggling to transfer the emerging knowledge into clinical and technological applications. Public Health Genomics advocates the interdisciplinary discourse on and the understanding of genomics; it fosters progress in translational research and supports the introduction of new concepts of risk stratification and prevention.

The Bellagio Model of Public Health Genomics

Public Health Genomics (PHG) is an emerging multidisciplinary scientific approach which aims to integrate genome-based knowledge in a **responsible** and **effective** way into public health (Fig. 1).

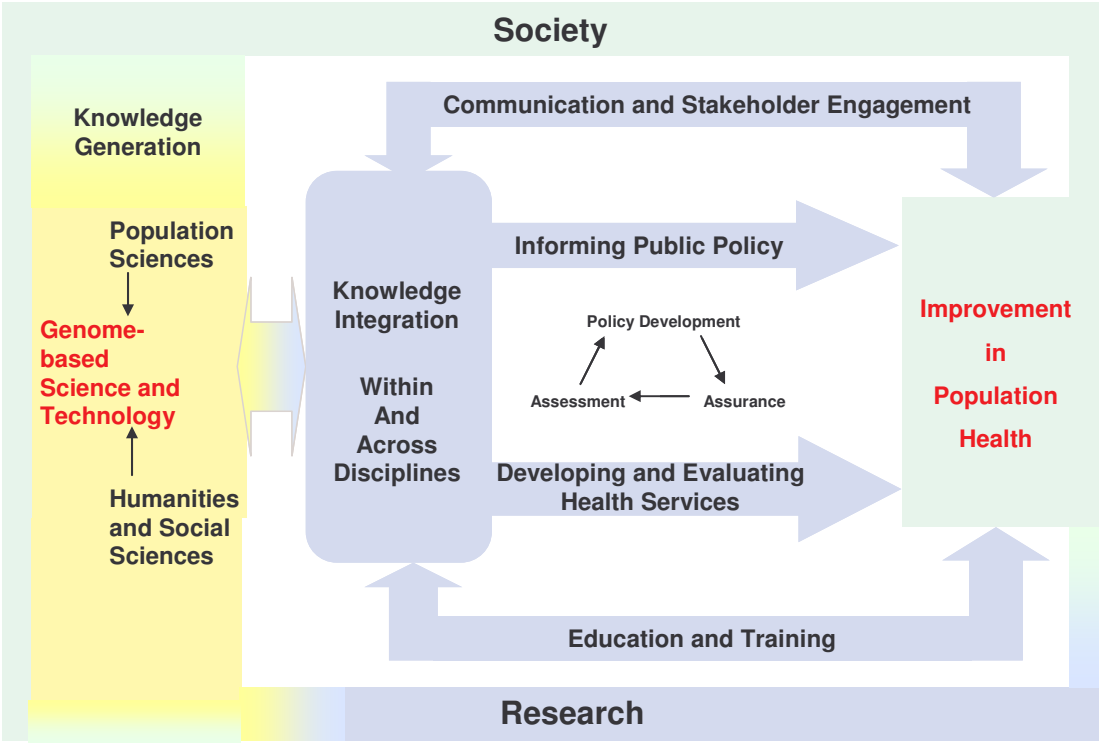

Figure 1 The Public Health Genomics Enterprise (Belagio Model). The figure displays the core tasks of PHG between the "Knowledge Generation" and the overall goal "Improvement in Population Health". (source: Bellagio Statement. Genome-based Research and Population Health. Report of an expert workshop held at the Rockefeller Foundation Study and Conference Center, Bellagio, Italy, 14-20 April 2005)

With the avalanche of emerging knowledge it is time to question whether we are offering the “right” health interventions (health promotion, prevention, therapy and rehabilitation) not only in the public health sector but also in the health care system as a whole for the benefit of population health. If we question the old concepts, we also have to ask whether our present and future public health strategies are evidence-based? [2]

In the past twenty years, the advances in genetic and genomic research have revolutionised knowledge of the role of inheritance in health and disease. Nowadays, we know that our DNA determines not only the cause of single-gene disorders, but also predispositions to common diseases, responses to therapies as well as the onset and progression of diseases.
Whereas medicine is presently taking a breathtaking development from its morphological and phenotype orientation to a molecular and genotype orientation promoting the importance of prognosis and prediction, public health practice has to date concerned itself with environmental determinants of health and disease and has paid scant attention to genomic variations between individuals, within populations and between populations. [2,3] In particular, the advances brought about by the Human Genome Project, HapMap or Systems Biology are changing these perceptions. [4,5] Many predict, that this knowledge will enable health promotion messages and disease prevention programmes to be specifically directed towards families and individuals at risk, or at subgroups of the population, based on a genomic risk stratification. Paradigmen shift in public health from subgroups to individual heath information management. This does not mean that we don’t need public health as an interdisciplinary and interinstitutional task for translating basic research in policy and practice. The opposite is true, the management of individual health information and guidance for major individual and collective health risks needs to be organised as our health care system is not prepared yet.

The new technologies will allow researchers to examine genetic mutations at the level of the functional units of genes, and to better understand the significance of environmental factors such as noxious agents and of nutrition and personal behaviour for the cause of diseases such as cardiovascular diseases, psychiatric disorders or infectious diseases.
The novel knowledge such as knowledge about pleiotropic effects of susceptibility genes in complex diseases being associated to more than one disease (“disease clusters”); about the role of individual genomic profiling; about the role of a genomic variant being a protective factor for some diseases and a risk factor for others; about epigenomic effects will all require a modification of both clinical and public health strategies. The post-genomic era also calls for a new knowledge base, among other things, for the integration of genome-based biobanks into public health surveillance systems as a tool for generating evidence on

genome-environmental interactions as well as for understanding diseases. The challenges we currently face can only be tackled by an interdisciplinary and systematic approach regarding the evaluation of the future impact of genome-based knowledge and technologies on health care systems. [6] Public health follows the Trias which guides the assessment, policy development and assurance of Public Health actions.

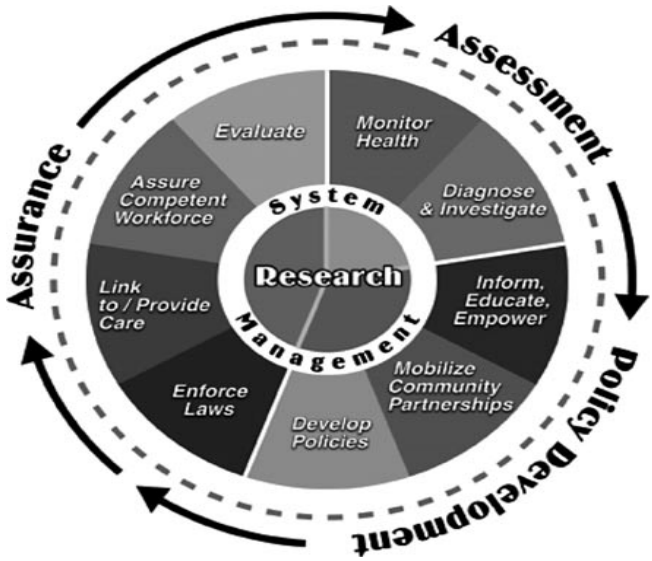

Figure 2 The integration of genomics into public health by using the Public Health Trias as an all-embracing methodology (adapted from IOM, 1998)

Obviously, the integration of genomics into public health research, policy and practice will be one of the most important future challenges for all health care systems. Clarifying the general conditions under which genomic knowledge can be best put to practise in the field of public health [7] and particularly considering the economic, ethical, legal and social implications, is presently the most pressing task of the emerging field of Public Health Genomics (PHG), [8] defined as the application of genetic and molecular science to the promotion of health and prevention of disease through the organised efforts of society. [9] Policymakers now have the opportunity to protect consumers, to monitor the implications of genomics for health, social, and environmental policy goals, and to assure that genomics advances will be tapped not only to treat medical conditions, but also to prevent disease and improve health. [10] Policy must find an acceptable balance between providing strong protection for individuals' interests while enabling society to benefit from genomics. [11]

The next decade will provide a window of opportunity to establish infrastructures both in the health care sector and on a policy level across Europe and globally, that will enable the scientific advances to be effectively, efficiently and socially acceptable and see them translated into evidence-based policies and interventions that improve population health.

The need for new products and processes is a major opportunity and a major risk at the same time. According to the Bellagio Model, Public Health Genomics wants to ensure the effective translation of genome-based knowledge. Health Technology Assessment (HTA) in combination with the concepts of Health Needs Assessment (HNA) and Health Impact Assessment (HIA) has the potential to assist Public Health Genomics fulfilling these tasks. On the other hand, these methodological approaches themselves are challenged by genome-based knowledge and technologies and have to be developed further.

The Public Health Genomics European Network

Experts from the different fields and stakeholders have urged the EC to set up a European network on Public Health Genomics. In 2006 the EU-funded Public Health Geomics European Network (PHGEN) started up and has successfully implemented both working groups on a European level and *National Task Forces* on Public Health Genomics. The network will develop a comprehensive set of policies which will serve as a framework for the future implementation of Public Health Genomics. [12] Together with partner networks and projects PHGEN will ensure the consistency and coherence of European policies.

Public Health Genomics has made substantial progress in recent years and is now seen as an integral part of public health. So far it has not fully reached its main goal, the transfer of genomic knowledge into all areas of public health. Still, the last two network meetings have proven the increasing acceptance of the challenge deriving from genomics. With the increasing genomic literacy of the population and the training of the workforces, Public Health Genomics will continue to diffuse into all public health actions.

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