Public health genomics – Relevance of genomics for individual health information management, health policy development and effective health services

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Abstract

Healthcare delivery systems are facing fundamental challenges. New ways of organising these systems based on the different needs of stakeholders are required to meet these challenges. While medicine is currently undergoing remarkable developments from its morphological and phenotype orientation to a molecular and genotype orientation, promoting the importance of prognosis and prediction, the discussion about the relevance of genome-based information and technologies for the health care system as a whole and especially for public health is still in its infancy. The following article discusses the relevance of genome-based information and technologies for individual health information management, health policy development and effective health services.

Key words: public health genomics, prediction, individual health information management, biobanks and surveillance systems, health policy development, health services

Background

Public health practice has to date concerned itself with environmental determinants of health and disease and has paid little attention to genomic variations within the population as well as between populations. The advances brought about by genomics is changing these perceptions [1]. Many predict that this knowledge will not only enable clinical interventions but also health promotion messages and disease prevention programmes to be specifically directed and targeted at susceptible individuals as well as subgroups of the population, based on their genomic profile contributing to risk stratification. Obviously, the integration of genome-based knowledge and technologies into public health research, policies and health services for the benefit of all will be one of the most important future challenges that our health care systems will face [2-6].

Already one of the key questions faced by all health care systems is the question whether “the right” interventions and services are provided by the various stakeholders: Are the current public health strategies evidence-based, i.e. are we assuring the “right” health interventions (based on combined concepts of health needs assessment and health technology assessment), in the “right” way (based on concepts of quality management and policy impact assessment), in the “right” order and at the “right” time (based on concepts of priority setting and health targets), and in the “right” place (based on concepts of integrated health care and health management)?

Since so far there has been almost no systematic integration of genome-based knowledge and technologies into all of these concepts, it increasingly apparent, that current public health strategies are not evidence-based at all.

Thus, the public health agenda now demands a vision that reaches beyond research to application and public health impact [9, 10].

Public health challenges: individual health information management, health policy development and effective health services

European and US public health institutions and platforms, like the Public Health Genetics Unit in Cambridge (PHGU), UK, the German Center for Public Health Genomics in Bielefeld (DZPHG), the Turkish Center for Public Health Genomics and Personalized Medicine (TOGEN) in Ankara or the US National Office of Public Health Genomics (NOPHG) at the Centers for Disease Control and Prevention in Atlanta (CDC), who work closely together with researchers from genetic and molecular science (“modern biology”) as well as those from population science, humanities and social science, are much more optimistic and clear about the relevance of genomics in public health than others [11, 14-16]. Interestingly enough, they
all have strong links with or are even part of their respective national genome research projects and are translating genome-based knowledge, supported by biotechnology, bioinformatics and biobanks through (genetic) epidemiology into public health ("translational research"). By using methods like horizon scanning, fact finding and monitoring to identify research trends as early as possible, they are already doing a prospective evidence-based evaluation, i.e. an evaluation that is already carried out in the process of basic research and not just in the (retrospective) implementation process of public health strategies and policies [17], which will always tend to lag behind.

In the past twenty years, advances in genome-based research have revolutionised knowledge regarding the role of inheritance in health and disease [18].

In this context, previously there was a narrow focus which concentrated mainly on the role of inheritance in monogenic diseases and the genetic testing of more than 1000 diseases. At present, the role of genetic susceptibility and other biomarkers in biological systems and complex diseases has already been discussed (medical, community health as well as in the public health setting). In the future the focus will be even broader, and include analysing the role of genomic variants together with other determinants of health problems (public health setting).

For example, nowadays, it is known that the DNA determines not only the cause of single-gene disorders, which affect millions of people worldwide, but also predispositions for common diseases ("susceptibilities") [19], which are based on genotype and haplotype variants [20, 21]. New technologies will allow researchers to examine genetic mutations at the functional genomic unit level [22], and to better understand the significance of environmental factors such as chemical agents, nutrition or personal behaviour [23] in relation to the causation not only of diseases like cancer, diabetes, osteoporosis, cardiovascular and cerebrovascular diseases [24], which accounted for 86% of all deaths and 77% of the burden of disease in Europe in 2005, but also of psychiatric disorders, allergies and infectious diseases [25, 26].

Evidently, these rapid advances in genomics and accompanying technologies are triggering a shift in our understanding of health and disease as well as providing insight into new prevention and therapy approaches [27-29]. Which outcomes can be drawn from this knowledge, how can it be translated into policies [17] and practice in a responsible and timely manner?

Clarifying the general conditions under which genome-based knowledge and technologies can be put to best practise in the field of public health, paying particular consideration to the public health specific ethical, legal and social implications (ELSI) [11, 15, 30], is currently the most pressing task within this emerging public health field, also known as public health genetics or public health genomics (PHG). Integral to the process of steering the application of genetic and molecular science towards the promotion of health and disease prevention through the organised efforts of society, is dialogue with all of the societal stakeholders, including industry, governments, health professionals and the general public [16]. Thus, the integration of genome-based knowledge and technologies into public health research, policy and practice will be one of the most important future challenges faced by all health care systems [31, 32]. Expertise is already available and can be clustered and evaluated for a socially accountable use.

For example, in a condition like coronary heart disease, to be a heterozygote for the LDL receptor gene confers an increased risk for developing this condition. But, as it is also true for all other risk factors (e.g., social factors, diet, smoking, physical activity), which have been identified by epidemiologists in this context over the past decades, the presence of the genomic biomarker is not 100% predictive, and those with it may not develop the disease, while those without it may end up with the disease [26] for other reasons. Obviously, the scenario is very much like that of coronary heart disease in the presence of raised blood pressure or cholesterol levels: the increased risk implies "only" a (high or low) probability, and the genomic biomarker is "just" another modifier in the causality of the disease and therefore not extraordinary [11, 12, 33]. Nevertheless, the ethical question is how we handle these susceptibilities. To answer this question, as a first step, population-based large-scale epidemiologic studies are needed to measure associations between specific genomic variants and environmental factors and the risk of coronary heart disease [34]. For translating such discoveries into interventions it is necessary not only to quantify the impact of genomic variations on the risk of the condition, but also to quantify the effect of modifiable factors that interact with genomic variations [35]. Based on the knowledge of these attributable risks, sound policies and effective interventions can be made in the future [11, 34].

Regarding infectious diseases, research is being expanded to include family histories as well as host genomic factors that influence the susceptibility to
certain infectious diseases or even the severity of the condition, and that also affect responsiveness to vaccines and therapies. The identification of several genome-disease associations for parasitic (e.g., malaria), viral (e.g., HIV or hepatitis) and bacterial (e.g., tuberculosis or cholera) infections provide critical clues to control these infectious diseases. By this, public health strategies will be more effective and efficient.

Policymakers must be aware of the current challenge to improve consumer protection, to monitor the implications of genome-based knowledge and technologies for health, social, and environmental policy goals, and to ensure that genomic advances will be tailored not only to treat medical conditions, but also to prevent disease and improve health [31]. Sound and well reflected genomics policies and programs require a timely and coordinated process for evidence-based policy making that relies on scientific research and ongoing community consultation [36]. An acceptable, and maybe delicate balance between providing strong protection of individuals' interests [37, 38] and enabling society to benefit from the genomic advancements at the same time must be found [11, 31, 39, 40].

Thereby it is essential to:

• identify needs for “genetic tests” as well as for genome-based information and technologies [41] (e.g., by using the method of Health Needs Assessment);
• weigh the benefits and risks of “predictive genetic tests” and genetic screening interventions [28, 42-45] (e.g., by using well established public health methods such as Health Technology Assessment (HTA));
• assess the benefits of preventive strategies; analyse complex new problems such as “genetic inequalities” [32];
• analyse genome-based technologies such as microarrays (e.g., Affymetrix versus Illumina) or the use of mass spectrometry for the analysis of genomic imprinting by DNA methylation and posttranslational protein modification.

On the one hand, even if, in terms of genetic susceptibilities and genomic variants, it turns out that “we are all at risk of something”, there is potential for social inequalities in health as well as for social exclusion: if “genetic tests” are not to be covered by sickness funds, access to genome-based knowledge and thus, to individual health information management and stratified prevention, diagnostics and therapy will lead to a two-tire system. On the other hand, even if “genetic tests” become reimbursable in most healthcare systems, there will be another ethical and social problems, which may be much more discriminating: since genomics is triggering the complexity of knowledge, public health professionals will have the task of empowering and enabling people not only to understand this new information, but also to make them capable of making sound decisions regarding the use of “genetic tests” [46] and genome-based information, and therefore to assure a fair equality of opportunities. Otherwise, the gap between people being able to handle this complex issue and those who are not, will potentially create a new kind of social inequality [47].

For the future, this supports a conception of public health taking leadership by implementing an evidence-based mode of policymaking. This is the reason why in the US, in the UK, in Germany as well as in Turkey Public Health Genomics has already been seen as the integration of genome-based knowledge and technologies into public health research, policy and practice for the benefit of population health.

For the public health community it is important to stress, that “genetic determinism” as well as “genetic exceptionalism” is obsolete [33]. In addition, it has to be clarified, that public health genomics is not synonymous with genetic epidemiology in the same way as public health is not synonymous with epidemiology, and also that community genetics [48, 49] is not synonymous with public health genomics as community health is not synonymous with public health [50].

Medicine (and here in the past mainly human genetics, but increasingly also all other medical disciplines such as psychiatry, cardiology or oncology), community genetics and public health genomics can be understood as having complementary tasks. While in the medical setting the interest focuses on the use of genetic tests and other biomarkers in the clinical practice, in public health genomics it is on the use of genomic variants (“genetic determinants”) together with other determinants of health in the health care system. Community genetics is the bridge between both settings.

Furthermore, in terms of public health genomics the idea of integrating genome-based knowledge and technologies into the aims and tasks of public health should be understood and promoted.

**Issues and priorities of public health genomics**

During the past century, achievements in public health have led to enormous improvements and benefits in the health and life expectancy of people around the world. Immunization programs
and better sanitation practices resulted in the eradication or reduction of many infectious diseases as well as in safer food and water supplies. Advances in occupational safety considerably decreased the number of work-related injuries, illnesses and deaths. In the past 30 years, identification of behavioural risk factors, such as smoking, inactivity and poor dietary habits, have gave rise to educational interventions and resulted in decline in death rates from certain chronic diseases.

In the future, the National Office of Public Health Genomics at the CDC predicts “…perhaps because of these accomplishments, the determinants of disease and disability - whether natural or human made - are often perceived as originating outside the body. Although it has long been recognized that disease generally results from a constellation of host- and environment-specific factors, scientific and technologic limits have concentrated attention on the environment. Exogenous influences will continue to be vital for public health, but focusing solely on these influences may lead to diminishing rates of return compared to the triumphs of the past. To continue making significant strides, the effectiveness of public health interventions must be strengthened by more fully incorporating knowledge of internal, host-specific factors and their interactions with environmental exposures including the social environment and lifestyles…” (personal communication).

In the area of social policy making there is a need to create a clear strategy for assessing and translating this novel knowledge and application in a timely manner. Policymakers now have the opportunity to take action. Preconditions for immediate action are strategic planning across health programs, promoting genomics competencies among all health professionals, enhancing surveillance and epidemiologic capacity (e.g., by combining already existing DNA-based biobanks and integrating them into well-established surveillance systems) to support evidence-based policy-making, building partnerships such as public private partnerships and seeking input from stakeholders. Here, integrating genome-based information into health related communication will be an essential tool to generate distributed knowledge.

The likely benefits as well as potential risks of integrating genomics into public health interventions (assessment) should be identified. The framework (corridors) for effective, efficient and socially acceptable policies (policy development) should also be described. Furthermore, steps and means should be proposed to assure the implementation of these policies into public health practice (assurance). At the same time, these three steps (“public health trias”) describe the core functions of public health agencies at all levels of government [51].

One specific task of public health genomics is to rethink and systematically evaluate every public health task as well as every condition of public health interest [32].

There is the potential for many more target-oriented and stratified prevention strategies [52] to finally replace the “one strategy for all”. Moreover, clearly there is potential to avoid ineffective or even “faulty” preventive strategies, for example, there is already the challenge to differentiate between persons, who will respond to certain vaccinations and those who will not. Why then should those who do not respond take the risk of experiencing side-effects from a vaccination if the vaccination will be ineffective and therefore be of no benefit on an individual as well as population level? In this specific situation, which is estimated to be true for at least 10% of the population, would one not consider this kind of primary prevention to be immoral? Obesity is another example of this; obesity is not only influenced by lifestyle habits such as inactivity or nutrition, but also (in more than 60%) by several genomic factors. Furthermore, it is triggered by many other factors such as infectious diseases and social factors. At least 2% of these 60% are solely due to mutations in the MC4R-gene. Individuals carrying the MC4R-mutation are almost “resistant” to any diet and physical activity. Therefore is it not a “faulty” preventive strategy to give advice to these individuals that “five a day” or “a low-fat diet” will be effective? Would it not be a “better” (preventive) strategy to give societal support by respecting them as they are? Of course, there are many more polymorphisms involved in obesity, and there are several polymorphisms that play an important role in the effectiveness of diet and sports. There are even polymorphisms that increase the risk of dying after physical activity. Thus, it should be kept in mind that one should be a little careful about the general public health message “prevention and health promotion is good for everybody”.

In this context, the “right not to know” and the “right to know” deserve the unbiased attention and must be mutually assured [11, 53]. So far this has not been considered in most of the European discussions about the regulation of genetic tests. Besides the questions of reimbursement and access to genetic tests and genome-based
information, restrictions in the provision of genetic tests such as physicians' proviso, which has already been considered in several European countries, seem to be sheer naïve in the era of e-health, globalisation and integrated health services. Instead of proclaiming (ineffective) restrictions, would it not be much more effective and efficient to promote health literacy in order to protect the consumer [53]? From a legal point of view, should not the use of information be regulated instead of the information itself? And from an ethical point of view, would it not perhaps be more appropriate to use the model of “informed contract” [54], which is based on the idea of “benefit sharing” between the consumer and the provider, instead of continuing to use the “informed consent” and “informed choice” models in the doctor-patient relationship?

New genome-based information and technologies will force health communities to enhance surveillance systems by integrating this knowledge (e.g., arriving from biobanks) as well as to enhance the epidemiological capacity for collecting and analysing information stemming from community-based assessments of genomic variation [55], providing evidence about the burden of various diseases. As with other fast-paced scientific and technological advancements, the intersection between genomics and public policy will continue to require close monitoring using public health methods like health technology assessment (HTA) [56-61], health needs assessment as well as health impact assessment and will also continue to require timely action. In this way, there will be a chance to ensure the appropriate and responsible use of genome-based information and new technologies [62].

Thus, in summary, the following eight public health genomics challenges as well as issues and priorities have been identified:

1. **Risk stratification, risk communication, and health communication**

   For example:
   - earlier & higher precision of risk strata (distinction between and identification of high, moderate and low risk groups; “genomes-based standardisation” in addition to age and sex standardisation of diseases)
   - differentiation between disease (sub)entities resulting in the same phenotype (e.g. breast cancer), that either follow mainly a mendelian trait (“inherited”) or a mainly genomic-environmental pattern
   - the concept of a genomic variant can be for different individuals a risk factor or a protective factor at the same time (e.g., role of ACE insertion-deletion polymorphism in stroke (increase of risk) and Alzheimer's disease (decrease of risk) [63])
   - pleiotropic effects of susceptibility genes in complex diseases being associated to more than one disease (e.g., the role of G-308A TNF alpha gene variant in obesity, asthma and non-Hodgkin lymphoma [64-66]; shift from disease-orientation to “risk orientation”, “disease clusters” and “health outcomes”
   - individual genomic profiling (simultaneous evaluation of multiple susceptibility genes) as the basis for individual health information management, where the individual genotype status on all genomic variants increases or decreases the risk for certain diseases
   - shift from carrier screening to a screening based on individual genomic profiling
   - risk stratification and categorisation based on “disease clusters” or on the total number of genomic risk variants in an individual
   - the role of genetic determinants not only within a group of other health determinants (e.g. social, behavioural, environmental, biological) but also as a modifier and triggering factor (e.g., epigenomic effects)
   - “genetic determinants” (i.e., genomic variants) as “necessary but not sufficient” determinants in the development of complex diseases and health problems
   - differentiation between predictive tests (0-100% probability; continuum; referring to monogenic as well as to complex diseases) and tests for diseases with high penetrance (referring to monogenic diseases as well to complex diseases following a mendelian trait) and low or moderate penetrance (most complex diseases)
   - a shift in the definitions: from “genetic test” to “genetic determinant” to “genomic variant” to “genome-based medical information” to “individual health information”
   - systematic analysis of risk perception (e.g., the higher degree of trust people have in governance, the higher the positive attitude regarding genome-based information and technologies and the higher the acceptance [67])

2. **Prevention**

   For example:
   - evidence-based primary, secondary and tertiary prevention by integrating genome-based knowledge (example of osteoporosis or infectious diseases)
   - stratified prevention by identifying high, moderate as well as low risk groups instead of “one prevention strategy for all” (“prevention
paradox”: low genetic penetrance and high frequency of genetic susceptibilities with minor effects as the specific challenge for public health (genomics)

• earlier prevention based on genome-based knowledge (“individual genomic profiling”) minimising “faulty prevention” (examples of target-orientated vaccination or of sports and sudden death)
• antidiscrimination by higher target-orientation based on genomics (examples of obesity, drug and alcohol addiction)
• a shift from primary prevention to either health protection or to secondary prevention (concept of “healthy ill” in the past and genomic variants as probabilities at present): “end” of the concept of primary prevention?

3. Surveillance systems/biobanks
for example:
• integration of genome-based information and DNA-based biobanks into the many already existing population-based surveillance systems (e.g., into cancer registries, surveillance systems for infectious diseases, EUROCAT, ALSPAC, EPIC, ISAAC or even into health observatories) as well as into future surveillance systems covering health problems and linking individual information (record-linkage based surveillance) during the whole lifespan (example of Sweden and Denmark: social security number as a personal identifier in newborns)
• genome-based surveillance systems as a basis for individual genomic profiling and as a tool for individual health information management (example of Western Australia: Western Australian Genetic Epidemiology Resource)
• simultaneous surveillance of samples (DNA and other biomarkers as well as tissue) and data from records
• linkage of records (e.g. perinatal quality assurance programs, hospital discharge data) and data from registries (e.g. cancer registries) with data from (genome-based) samples in addition to population-based (mega)biobanks such as in Finland, Iceland, Estonia, UK, Norway
• recognition of the possibility that in most countries well established newborn screening could serve as an already existing nationwide DNA-based biobank (in the public sector, in private hand or in public private partnership) with the possibility of reanalysing up to 25-year-old Guthrie cards
• a shift from newborn screening of metabolic diseases to newborn DNA-based screening. In addition to metabolic diseases, which validated genomic variants should in the future be tested for? Should such a program include complex diseases, which have the highest burden of disease (e.g., for cardiovascular diseases, cerebrovascular diseases, diabetes, cancer and osteoporosis accounting for 77% of burden of disease in 2005 in Europe), or perhaps orphan diseases (accounting for 10% of all diseases in the whole population) for which a resequencing chip could be developed?
• implementation of long running cohort studies (starting as early as possible in life and including nested case-control studies at various ages and at various occasions)
• implementation of case-control studies of those in the population who are most elderly in order to generate hypotheses on genomic-environmental associations as well as on “disease clusters”

4. (Genomic) inequalities in health
for example:
• inequalities in genomic variants between individuals (“we are all at risk for something?”)
• inequalities in genomic variants within and between populations (stratified screening programmes instead of one population screening for all) (example of specific migrants’ needs of the newborn screening in the Netherlands)
• inequalities in access to genetic services
• inequalities in access to genome-based knowledge and technologies at the global level
• inequalities in reimbursement of “genetic tests” as well as of genome-based biomarkers and technologies (e.g., use of microarrays)
• inequalities in health literacy regarding the complexity of genome-based knowledge (“widening the gap”)”

5. Regulations, good governance & public health ethics
for example:
• the balance between self-responsibility and social welfare (example of reimbursement by sickness funds)
• the strong protection of individuals’ interests while enabling society to benefit from genome-based advances (example of employment and occupational health)
• the balance between “right to know” and “right not to know” (example of European national laws on genetic diagnostics as examples for ignoring and overriding these balance)
• the regulation of the use of genome-based information instead of the information itself
• rethinking the principles of social justice, solidarity and subsidiarity on individual and institutional levels
• PHELSI (Public Health ELSI) (e.g., analysing and assuring demands versus needs, analysing norms, values and preferences, assuring health literacy, applying criteria of public health ethics)

6. Consumer protection 
for example:
• marketing and sale of "genetic tests", genetic services and genome-based technologies
• the distinction between the “use” (referring to stakeholders’ responsibilities) and the “utility” (referring to quality assurance issues and outcome measurements) of genome-based information
• food directives on the safety and the quality of genetically modified food
• labelling of genetically modified food

7. Health protection
for example:
• nutrigenomics
• development of recombinant vaccines (which are safer, cheaper and more target-oriented regarding subpopulations)
• ecogenomics / bioremediation (e.g., water and soil pollution)
• toxicogenomics
• envirogenomics
• pharmacogenomics

8. Stakeholders’ responsibilities
for example:
• rethinking of stakeholders’ responsibilities in an integrated health care system(defining responsibilities of actors and institutions in the clinical setting, primary health care and public health sector)
• promoting public-private partnerships
• training all professionals from the entire health care system in genome-based knowledge and technologies
• counselling and empowerment of the public
• promoting relevant research in the field of public health genomics (e.g., promoting HTA in the context of genomics; instigating a shift from genetic epidemiology to a broader focus by integrating genomics into "classical" epidemiology and association studies)

The European perspective
Considering genomic variants (“genetic determinants”) as a factor which contributes to health then as a component of public health, their consideration is a necessary step to enable good health for everyone. Thus, “genetic determinants” have to play an eminent role in a new EU health strategy. To create sound genome-based policies and programmes, public health should get involved and what is more take the lead by applying the three core functions of public health (assessment, policy development, assurance) to the provision of not only genetic health care services but also all health care services.

The European Commission has in its report on “Life Sciences and Biotechnology” (COM(2004) 250, April 7th 2004) committed itself to achieve a high level of quality in genetic testing and to increase “co-operation and exchange of information in order to enhance coherence and disseminate best practice”. Furthermore, in the work plan 2005 of the “community action in the field of public health” the European Commission calls for the application of a “networking exercise … to lead to an inventory report on genetic determinants relevant to public health. This network will identify public health issues linked to current national practice in its application of genetic testing and on that basis it will contribute to developing best practice in applying genetic testing.”

Thus, from the beginning of 2006, the Public Health Genomics European Network (PHGEN) was funded by the European Union (www.phgen.nrw.de) (EU Project No 2005313), which is coordinated by the Institute of Public Health North Rhine-Westphalia (lögd) in Bielefeld, Germany. Associated partners are the Public Health Genetics Unit (PHGU) in Cambridge, UK, as well as the German Center for Public Health Genomics (DZPHG) at the University of Applied Sciences in Bielefeld, Germany. PHGEN involves experts as collaborating partners from the fields of public health and epidemiology, human genetics and molecular biology, social sciences, (public health) ethics, medicine, economics, political sciences and (European) law. From all EU member states, applicant countries and EFTA-EEA countries there is at least a representative from public health and genetics or genomics as well as from a relevant competent authority. Furthermore, representatives of other European networks (e.g. EuroGentest, Orphanet, EUnetHTA, PHOEBE or NuGO) as well as representatives of relevant initiatives and institutions on the European and international level such as WHO, WTO, OECD, STOA, AETMIS, CDC National Office of Public Health Genomics (NOPHG), GrPhInt, HumGen, TOGEN or UK DNA Banking Network are involved to ensure complementarity and the promotion of synergies [68, 69].

The aims of PHGEN are
• to conduct a networking exercise on PHG covering all EU Member States, Applicant Countries, and EFTA-EEA Countries
• to identify and list key experts and institutions relevant to PHG in these countries
• to provide an inventory of “genetic determinants” (genomic variants) relevant to public health
• to provide an inventory of PHG-issues and priorities in Europe
• to identify legal diversities and barriers in a cross-border market
• to analyse the relevance of EU treaties for PHG
• to contribute to the co-operation and exchange of information in order to enhance coherence and disseminate best practice in Europe
• to promote and stimulate the countries’ efforts in this emerging field by developing PHGEN and by supporting effective networking in order to reach sustainability (e.g. implementation of National Task Forces on PHG)
In the long run PHGEN will serve the European Commission as an “early detection unit” for horizon scanning, fact finding, and monitoring of the integration of genome-based knowledge and technologies into public health.
According to the already well-established public health trias, tasks of PHGEN include

1. **Assessment** (the systematic collection, assembly and analysis of genome-based information and technologies relevant to Public Health):
   • analysis of PHG concepts (e.g. definitions of PHG, genetic determinants, genomic variants, genome-based knowledge, risk stratification)
   • identification of PHG issues & priorities in research, policy and practice
   • identification and „best practice“ of PH methods relevant to PHG (e.g. HNA, HTA, HIA/PIA)
   • identification of networks and institutions relevant to PHG on national, European and global level

2. **Policy Development** (the development of European standards and guidelines which promote the responsible and effective use of genome-based information and technologies in European health systems):
   • analysis of legal diversities (e.g. conflicting laws) and barriers in a cross-border market
   • analysis of European minimal standards, guidelines & laws
   • analysis of economic implications and PHELSI
   • development of policies on education, information and empowerment

3. **Assurance** (the appropriate use of genome-based information and technologies in European health services):
   • critical proof of the need for enforcement of new laws and/or regulations (e.g. in most European countries there is already an overregulation)
   • assurance of stakeholders’ responsibilities in the application of genome-based information and technologies
   • assurance of a competent workforce
   • evaluation of health services (e.g. health promotion, disease prevention, therapy, rehabilitation)

By this network, across whole Europe, there will be the chance of scientific advances being timely, effectively, efficiently and socially acceptably translated into evidence-based policies and interventions that improve population health.

**Concluding remarks**
Which consequences can be drawn from genome-based knowledge and technologies and how can it be responsibly and timely translated into policies and practice for the benefit of population health [70]? The necessity of assessing health services as well analysing complex new problems, such as ‘genomic inequalities’ in health or the role of biobanks in surveillance systems, support the idea that public health should get involved and moreover take a leading role. Likely benefits as well as potential risks of the integration of genomics into public health interventions (assessment) should be identified. Systematically, the framework or corridors for effective, efficient and socially acceptable policies should be described (policy development) and steps and ways be proposed to assure these policies in public health practise (assurance) [71].

This is a feasible project [72], but it will require regional, European, as well as global coordination [73]. There is now an ethical obligation to prepare society to meet these challenges and to take up the opportunities provided by science in a medically useful, effective, efficient, socially desirable and ethically justifiable manner. Here, health literacy, health communication and empowerment for managing risks are the key for opening the doors to a truly beneficial public health genomics. All in all, this can be facilitated by implementing ethical benchmarks like respect for autonomy and social justice in the context of policy development.

By promoting communication about genomics, not only within the public health scientific community but also among other health professional groups, public health agencies and the public, there might be a return on public investment in the human genome research. There are already many more opportunities than risks to provide better health for the population [74].
Indeed, there is still a discussion about stigmatisation and discrimination due to genome-based information not only in the public but also in the scientific community. Nevertheless, whoever continues separating genome-based knowledge from other medical information by defining genome-based knowledge as exceptional, whoever continues promoting the obsolete idea of genetic determinism, and whoever continues claiming the “genetisation”, “molecularisation” and “medicalisation” of society, has not seriously tried to keep up with the genomic research in the past years. Explicitly, it should be emphasized at this point, that this accusation does not necessarily imply that public health professionals do not have the obligation to consider genome-based information as a highly sensitive factor in medical information.

Furthermore, it is not the question whether the combination of public health and genomics is dangerous [32]. The key question is whether not harm is done to people by omitting to integrate genome-based knowledge and technologies into public health interventions, and thus withholding the potential of stratified evidence-based prevention and policymaking.

The public health community will lose credibility, if on the one hand public health is promoting health literacy in a value-pluralistic and democratic society and enabling and empowering individuals for decision-making while on the other hand ignoring and withholding genome-based knowledge and technologies, and therefore still not providing evidence-based public health interventions. In terms of the individual’s “right to know” and in terms of best practice in public health, is this not a new form of discrimination?

The next decade will provide a window of opportunity to establish infrastructures, across Europe and the globe, that will enable the scientific advances to be effectively and efficiently translated into evidence-based policies and interventions that improve population health. Policymakers now have the opportunity to protect consumers, to monitor the implications of genomics for health services, and to assure that genomic advances will be tapped to prevent disease and improve health. We now have the chance to prepare public health professionals, the public and policymakers for the changes to come. The above presented examples show approaches for national, European and international institutionalisation of public health genomics that serve the aim to champion these challenges.

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