

Why genomics will not revolutionise public health

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Abstract

The integration of genome-based information into public health is a challenging task. In this context, some emphasise a supposed fundamental difference of genome-based information compared to other health information. This view is contradicted by the author by arguing that genome-based information is determined by characteristics which also apply to other non-genome-based health information. Rather genome-based information integrates itself into the range of personal health information. Genome-based information deserves no “status apart” just because it was gained with the help of molecular diagnostic techniques. Thus there will be no revolution in public health, rather it can be discussed how genome-based information could be utilised to be part of public health approaches making public health more target-oriented and efficient.

Key words: public health genomics, genome-based information, exceptionalism, scientific revolutions, discrimination

Introduction

In recent years some medical and bioethical literature, predicted the arrival of a genetic revolution for health and health care [1-3]. The metaphor of a revolution was used to welcome the advent of better disease prevention due to a better predictability of health. People using the revolution metaphor are in general in favour of genetic advances and as such thought positively about the upcoming use of genomics for public health.

However, the flip side of a revolution is that revolutions are brutal and may claim many victims. Or if, in accordance with Thomas Kuhn's concept of scientific revolutions, one uses the metaphor more in the sense of a philosophy of science, revolutions bring about new paradigms. These are discontinuous and in many ways inconsistent with the old paradigms upheld before the shift. This would mean that new paradigms have new protagonists: the pioneers and heroes of the revolutions may become leaders of the new paradigm. [4]

Using the revolution metaphor came back to its optimistic users as a backlash. The use of this metaphor rather promoted the thesis that genome-based information – here defined as information obtained from the molecular analysis of the genome – is absolutely special, has a unique significance and fundamentally differs from other health information; in other words it is seen as being exceptional in its health-relevant characteristics.

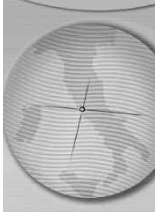
Proponents of a so-called “genetic exceptionalism” stressed the predictive power and potential for discrimination and stigmatisation of genome-based information. This assumption furthered a debate in which terms like “Gene-Angst” or “Genes are us” [5] were coined, giving genes a symbolic, almost metaphysical meaning [6,7]. This assumption bears an acceptance of genetic reductionism or genetic determinism. Genetic reductionism is the oversimplistic supposition that phenotypic traits of human beings are a linear consequence of genetic factors. Genetic determinism is the notion that our genes exclusively make us the persons we are.

In public health now, demands are being made for due consideration to be given to genome-based information. In this context, there are good reasons to argue that genome-based information is not exceptional and as such will not revolutionise public health despite a health benefit that can be expected. [8,9] The reason is: Genome-based information fits into the spectrum of health information and will add on to the approaches already taken in health. It is rather a logical continuation of research and practice. [10]

In the following chapter I will give reasons that support this anti-exceptionalist assumption.

Arguments for and against “genetic exceptionalism”

Many of the arguments in support of genetic exceptionalism underestimate the extremely complex interaction of the genetic endowment of persons and other factors contributing to health



(i.e. lifestyle and environment). Tom Murray has made this obvious by developing a “two bucket”-model of disease. In this model, one bucket contains genetic diseases and the other non-genetic diseases. Huntington’s Disease¹ belongs in the “genetic” bucket; “to get run over by a truck” in the “non-genetic” bucket. These would be the two most extreme versions of diseases with regards to genetic factors. Most diseases – and especially those common diseases Public Health Genomics focuses on – do not “neatly fit into either bucket”. [12] For example diabetes, schizophrenia or breast cancer are known to have genetic components, however, it is not exactly known how big the influence of genes is compared to the environment. Even “to get run over by a truck” presupposes a living human being and humans have genes that contribute to the possibility of living and walking on streets. So there is hardly a condition to which genes do not contribute – and on the other hand, Huntington’s Disease does not solely depend on having the Huntingtin gene. To suffer from Huntington’s Disease also depends on other causes that we are not in a position to determine or quantify yet, (otherwise the disease would always be expressed in the same way with all persons bearing the very same gene mutation). In this sense it is even misleading to talk of “genetic diseases” at all – and I will continue to use this term in brackets only.

In the following, the arguments for and against genetic exceptionalism will be categorised and discussed. [5, 12-19]

Concern for kin

People share parts of their set of genes with their parents, children, brothers and sisters. There is concern that with the knowledge about his or her genome, a person will receive health information that is also relevant to others. As such one obtains knowledge on aspects of the future of one’s kin that the respective person of the family does not yet have or would prefer to keep to him- or herself. This raises issues of privacy, confidentiality and the right (not) to know. Furthermore, genome-based information can have an impact on family planning. These concerns for one’s kin show proponents of genetic exceptionalism that genome-based information is unique.

However, a family can be most seriously affected by non-genome-based information as well, for example by tuberculosis or sexually transmitted

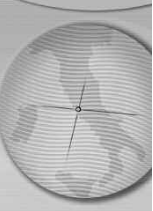
diseases. This information also concerns privacy, confidentiality and the right (not) to know issues. Hence, concern for one’s kin is not exclusive to genome-based information. The mere fact that genome-based information has an effect on one’s family does not distinguish it from other health information. Compared to the passing on of genes, which can only happen vertically from parents to children, infectious diseases – such as HIV/AIDS – can also be passed on horizontally to one’s partner or as with tuberculosis to the whole family.

Potential for stigmatisation and discrimination

Advocates of genetic exceptionalism are afraid that genome-based information provides potential for stigmatising and discriminating persons or risk groups. Isolated cases of discrimination are reported in discussions and literature. One paper that is most often cited is a case study by Lisa Geller et al [20]. However, it was reported from a large-scale international study that there is “little solid evidence of refusals of insurance or employment on genetic grounds.” Rather, studies like the one of Geller “claiming high penetrance of genetic discrimination are anecdotal, they consist of collections of cases without the use of accepted survey methods” [5, cf. 21].

Nevertheless, even though health information might provide grounds for stigmatisation and discrimination, its potential is not exclusive to genome-based information. Leprosy has long been a disease to stigmatise persons, mental diseases often lead to stigmatisation, information on positive HIV status has led to cases of discrimination, while information that migrants from a certain country have a high rate of newborn mortality might also lead to their stigmatisation. It is a demand of justice that a fair health and social policy counteracts these cases of discrimination and stigmatisation [13]. But there is no need for genome-based information to be specifically protected in this context. On the contrary: To give genome-based information a special protection status may lead to discrimination of those who have no “genetic diseases” but have gained their knowledge from conventional medicine because the assumption that genome-based information “should be subject to more comprehensive protection compared with non-genetic information – leads to unjustified unequal treatment of persons affected.” [17]. For Lemke there is no good reason “why a person whose higher risk of contracting a specific type of cancer,

¹ The rare Huntington’s Disease (in Germany about 1/100,000) is a fatal neurological late-onset monogenetic disease. Bearing the Huntingtin gene is indeed predictive of an almost 100% certainty of developing Huntington’s Disease. However, the point in time of the onset and also the exact course of the disease cannot be predicted. As such, Huntington’s Disease is a paradigm disease for genetic counselling in the medical setting. [11]



say, is attributable to genetic factors, should enjoy greater protection than someone whose health is threatened by environmental factors such as poor working conditions or polluted air" [17].

A special status of genome-based information would be the positive discrimination of people obtaining genome-based information and a negative discrimination of persons with conventional health information.

Furthermore, supporters of genetic exceptionalism provoke an unwanted effect. Murray argues that "the more we repeat that genetic information is fundamentally unlike other kinds of medical information, the more we implicitly provide for genetic determinism, for the notion that genetics exerts special power over our lives" [12]. Paradoxically, this rather *enhances* stigmatisation and discrimination - quite the opposite of what proponents of genetic exceptionalism want.

Ubiquitous information

The proponents of genetic exceptionalism claim that access to human cells is easily possible via, for example, saliva or hair can be refuted with the following arguments: Cells from saliva, hair or skin are indeed easily accessible. However, what is ubiquitous is rather the sample containing the DNA, not the information itself. Other health information, however, can easily be obtained (not to mention the genome related information via seeing a person whether he or she is male or female that might also be health relevant - e.g. to insurers). First steps into this direction are infrared thermal cameras at airports to check the body temperature of passengers [22]. This can of course also be done without people recognizing that they are being screened for fever.

Associated with the "ubiquitous information" claim, is the view that the genome offers a unique chance to identify individuals. But the iris is even more unique than the set of one's genes: genetic twins, for example, have different irises.

Long-term storage and use for other purposes than originally consented

There is a concern that tissue containing human genomes can be stored for long times. Then the genome can be analysed and genome-based information can be obtained for reasons other than the original informed consent was given for. This is nevertheless also true for conventional health information. Bovenberg, for example reports on the Dutch Blood bank states that 14 years after samples had been taken they were searched for antibodies for other purposes than originally intended. [5]

Concern for genetic prophecy

Huntington's Disease has long been a paradigm of a "genetic disease". Yet, this incurable disease is not in the focus of Public Health Genomics that basically builds on preventive approaches for common diseases. Public health will basically deal with common diseases that have a high prevalence and are not so highly penetrant as e.g. Huntington's Disease. So rather diabetes or adipositas might become paradigm diseases of Public Health Genomics.

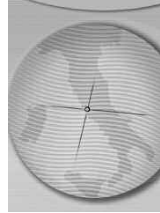
However, in general most genome-based information can, at the most, give a probabilistic estimation on the onset of a disease or its susceptibility. On the other hand, genetic prediction is not unique in offering probabilistic peeks into our future health: Information about a positive status of HIV, Hepatitis B or high cholesterol can also foretell the risks and probabilities of a person's future health.

The assumption on the predictive value of genes is also called "future diary". But a genome does not reveal much about a person's social and personal identity, character-forming experiences or his or her interests. The metaphor of a future diary presupposes and furthers a deterministic and reductionistic view. If using the diary metaphor at all, we should rather see it as a diary with empty pages that we will have to fill with words and stories ourselves. [12]

The origination of information

Health information might in some cases stem from the analysis of a person's genome but at the same time could also come from analyses at the phenotype level, for example, polycystic kidney disease (ADPKD). ADPKD is an autosomal dominant late onset disease with high penetrance. A person with the genetic predisposition for ADPKD will be born with small cysts in the kidneys. These cysts grow during their lifetime and will lead to kidney insufficiency later in life. The predisposition to developing ADPKD and the following kidney insufficiency can be detected in two ways in asymptomatic persons: on the genotype level by analysis of the DNA, or on the phenotype level by ultrasound or computer tomography. Furthermore, hypertonicity in persons at risk is a hint that they have inherited the disease. [23]

Thus, knowledge of this health information has the exact same meaning for the respective person - irrespective of whether he or she obtained this piece of information by DNA analysis or ultrasound. No matter how the information was generated, the relevant criteria of this information



are the implications of this predisposition for the presymptomatic person at stake.

Conclusions

It is plausible to argue that genome-based information is no exceptional health information with regards to its relevant characteristics, although some genome-based information bundles various characteristics. However, in general it seems irrelevant whether health information is genome-based or not. Morally relevant are the consequences that can follow from having the information with regard to its predictive power, implications for others, and potential for stigmatisation and discrimination. Special protection of genome-based information would only mean discriminating against conventional health information. In other words: it is always special to have predictive health information, relevant to a person and his family and potentially stigmatising (the speciality depending on the potential consequences that can follow), however, genome-based information is not per se more special than conventional health information. Rather, the potential consequences define the information handling that should follow, but – again – this is not a matter of the source from which the information was derived.

The argument that genome-based information is historically laden and that anxieties and prejudices are associated with genome-based information is no sufficient reason for treating genome-based information differently from other health information irrespective of its meaning. However, it might provide good reasons for considering anxieties and prejudices and thus for making sure that public information and education are well established in Public Health to respond to these anxieties and prejudices. It is a task of Public Health Genomics to *de-mystify* genome-based information and to discuss how we can make use of the benefits genome-based information will deliver.

To conclude, at least with regard to the character of health information, genomics will not revolutionise public health because genome-based information will *be integrated* into the spectrum of personal health information and will quantitatively add on to the approaches in health taken so far. To integrate genome-based information into the current paradigm of Public Health is more of an evolution than of a revolution. Integrating genome-based information into public health will be a consistent and logical continuation of public health approaches for more efficient and targeted prevention approaches.

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