

Governance in genomics: a conceptual challenge for public health genomics law

Tobias Schulte in den Bäumen

Public Health Genomics European Network (PHGEN)

Correspondence to: Tobias Schulte in den Bäumen, Public Health Genomics European Network (PHGEN), c/o Deutsches Zentrum für Public Health Genomics (DZPHG), Kurt-Schumacher-Str. 6, 33615 Bielefeld, Germany. E-mail: tsidb@fh-bielefeld.de

Abstract:

Increasing levels of genomic knowledge has led to awareness that new governance issues need to be taken into consideration. While some countries have created new statutory laws in the last 10 years, science supports the idea that genomic data should be treated like other medical data. In this article we discuss the three core models of governance in medical law on a conceptual level. The three models, the Medical, Public Health and Fundamental Rights Model stress different values, or in legal terms serve different principles. The Medical Model stands for expert knowledge and the standardisation of quality in healthcare. The Public Health Model fosters a social point of view as it advocates distribution justice in healthcare and an awareness of healthcare as a broader concept. The Fundamental Rights Model focuses on individual rights such as the right to privacy and autonomy. We argue that none of the models can be used in a purist fashion as governance in genomics should enable society and individuals to protect individual rights, to strive for a distribution justice and to ensure the quality of genomic services in one coherent process. Thus, genomic governance in genomics requires procedural law and a set of applicable principles. The principle which underlies all three models is the principle of medical beneficence. Therefore genomic governance should refer to it as a key principle when conflicting rights of individuals or communities need to be balanced.

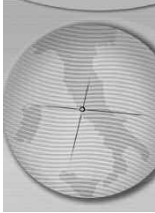
Key words: genomics, models of governance, medical model, public health model, fundamental rights model, rules and principles, procedural law, indication, information justice

Introduction

The avalanche of technical changes in information technology and the increasing understanding of genomic influences on human health have created new governance challenges that policymakers and scientists have to face when they discuss the future application of genomics in an ethically and religiously stratified society. The ethical and religious stratification and the special need in genomics for a balance between individual rights and the social-good pose new governance challenges. Governance as a term is nowadays widely used in many different contexts from corporate to global governance. This paper aims to reflect upon the interrelation between governance and regulation in genomics. The idea of governance goes much further as it encompasses institutions, structures of authority and even collaboration to allocate resources and coordinate or control activity in society or the economy. Thus, governance as an idea can be described as an umbrella which needs to be transferred by using different concepts [1]. There has been a tradition in the regulation oriented governance to use top-down approaches but the rising complexity and the need for the integration of diverse interests has led to a new concept which we can call a network

governance idea [2,3]. Governance in genomics requires more than a set of rules as there is a clear need to *govern* the different interactions in the everyday application of genomics which goes beyond the scope of regulation. Still, regulation and even more the models used in regulation are cornerstones of governance and within this paper we will focus on what Kooiman has called the general rules of governance [4]. This focus on regulation and governance also needs to reflect upon the different concepts of governance, e.g. if we think of ethics committees and the idea of a governance without government [5]. The normative scope of the paper is chosen as many European countries still discuss the need for special laws on genetic services and one may argue that there is an increased need for conceptual thinking in this discourse.

Still, no matter what governance idea a society may follow governance in genomics remains an issue which needs to be revised continuously as the genomic avalanche has just started. In its study "Information and Biological Revolutions" RAND [6] called this phenomenon a dual revolution with two sciences stimulating each other. With the emerging influence of globalisation a third force needs to be considered in addition to these two scientific ones.



Some countries like Austria, Switzerland and Estonia have reacted to this challenge by setting up new regulatory tools such as laws on genetic research or genetic diagnostics. These special statutory laws primarily focus on the use of DNA tests in certain fields such as employment or insurance matters. The approach used by the first countries to act has been supported by policymakers in many other states, as it seems evident that the new risks being derived from genomics need a special legal watchdog. Recently, more and more policymakers have drawn a different conclusion: special statutory regulations on DNA testing foster a scientifically unjustified public perception which is often characterised by the term “genetic exceptionalism”. With the transfer of genomic knowledge in every-day medical applications, genome-based knowledge becomes an integral and “normal” (but still rapidly progressing) part of medicine. The revolutionary development of the past and the upcoming integration of genome-based knowledge in health care systems poses a challenge to any regulatory attempt which acknowledges genomics as a part of a coherent regulatory framework. Due to the current uncertainty and the often criticised potential to abuse genomic knowledge, any governance model must perceive the shattered state of genomics in the societal discourse [7] [8] while enabling patients and consumers to profit from the transfer of genome-based knowledge into health care. Taking a look at the hopes and fears which exist due to the progress in genomics, policymakers are facing a matrix of potential regulatory conflicts. Some of them like the “right to know” versus the “right not to know” are well known but this individual conflict between family members depicts only one layer of the whole regulatory issue [9]. Policymakers need to create an integrated governance concept which focuses on public health as a public good just as much as on constitutionally guaranteed individual rights. Thus it needs to be reflected upon whether the existing governance models are appropriate with regards to the necessary process of balancing potentially conflicting rights. In the first stage of this analysis we will describe three models which can be applied both in law and medical sciences, then we will take a look at the principles of and the principles for regulation in the field of Public Health Genomics.

The medical model as a traditional governance model

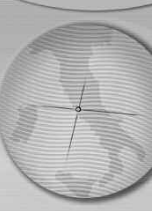
The Medical Model has been the standard for centuries in North America and Europe, as the physician has been noted as an expert who serves

as a gatekeeper to medical knowledge [10]. The relationship between patient and physician was based upon an isolated upstream and downstream, that is to say, the patient would describe the problem and the physician would act solely on his expert knowledge without seeking any consent of the patient. The old Medical Model, which refers to a hierarchy between doctor and patient, has come under pressure in most western countries due to the emergence of the idea of autonomy and self-determination, but some scientists argue that the concept of informed consent has rarely been seen in practice [11].

Despite the inflation in medical malpractice cases, where the information policy of physicians is at stake, the Medical Model still has a strong influence on health systems around the globe and currently progress in medical fields like genetics has changed little [12]. The key to understanding the importance of this model is the analysis of the medical standard as a concept. Even modern techniques of assessing the applicable medical standard, such as HTA, focus on the scientific basis rather than the patient view and the patients' needs. As a consequence, the Medical Model still has a vast conceptual influence on standards in medicine, as experts are called to assess the advances in modern health care from their expert point of view. Even in genomics, where the information and counselling of consumers is at the centre of the debate, the degree of disclosure is predominantly influenced by the professional standard [13]. The Medical Model is ethically based on the quality of genetic services, the medical benefit for patients and the morality of physicians. It also meets to a high degree the modern attitude towards medicine as a service which has to “function” in order to satisfy the needs of customers. The ongoing standardisation of health care services in Europe lobbies for the revival of the Medical Model, even if it has turned into a scientific model which relies on scientific data instead of the sole will of the physician. The governance idea behind the model remains the same: there is a hierarchy between experts and (political) decision-makers; as only experts are able to assess the developments in health care technologies.

The public health model – governance from the population oriented point of view

The Public Health Model has been transferred into the health care system due to rising concerns regarding diseases which are likely to spread amongst the population if no interventions on a population oriented level are carried out. These



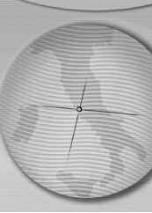
interventions include education, vaccination and screening programmes and the participation by large parts of the population in order to stop the further spread of a disease, e.g. in the fight against HIV or, in former times, the fight against syphilis. The idea behind the Public Health Model still is the idea of causation which makes it rather illogical to apply the Public Health Model on human genomics. The (involuntary) participation of the population in traditional screening and information programmes may be ethically justified in cases when a substantial risk exists that each member of the designated target group might transmit the disease due to voluntary contact with infected and healthy persons or in cases where large population groups set necessary conditions for the "infection" of other, healthy groups (e.g. smoking) [14]. The dimension of public health aspects, for the further development of genomic services, is an underdeveloped issue in many European countries such as Germany, while US-scientists, like Khoury [15], refer to genomics as an ultimate tool for the preservation of public health.

To date, the idea of public health is most likely be detected in prenatal and newborn testing but further extension to other (multifactoriell) diseases like adipositas seems inevitable. This further extension is likely to be supported by the fact that only a few therapeutic options exist to cure or soften genetic conditions and therefore the public health related idea of prevention moves to the centre of genomic health policies [16]. The increasing importance of public health ideas for furthering development underlines the issue that a balance between the human goods of autonomy and health is needed if the Public Health Model is not to undermine the necessary recognition of self-determination in a diverse society. Some authors like Gostin have favoured "social contract" [17] in order to regulate privacy and health, but the execution of such a contract might also lead to a genomic expert oligarchy as indicated in the Medical Model. Supporters of the Public Health Model will argue that this criticism is unjustified and if we take a look at today's public health concepts we should agree with them. Ethical and social assessment has become an integral part of public health and the expert system as described in the Medical Model is hardly ever seen in a pure form. One may doubt whether it takes a new "social contract" in order to apply the Public Health Model in European health care systems. From a legal perspective health needs to be democratised and due to the new medical concepts in genomics (such as the risk assessment for multifactoriell

diseases) we should think of genomics as a model area for the integration of health into democracy and vice versa. In this sense the Public Health Model also advocates a network oriented governance structure which is integrating the knowledge driven hierarchy between experts and laypersons in a democratic process.

The fundamental rights model – the individualistic concept

The third model [18] has a reverse notion of health care services compared to the Public Health Model, with autonomy, self-determination and voluntariness being central to this approach. Health is not seen as a public but as an individual good, with the person being in charge to define his or her own desired health standard as well as selecting ethically valid measures to reach this individual standard [19]. A purist version of the Fundamental Rights Model would oppose any standardisation which has the potential to obstruct the individual self-determination of the patient or consumer. In this sense "any use of a particular health care service (...) must be voluntary and uncoerced" [20]. As any participation in genomic research or any use of a genomic service must be voluntary, no "discriminating" pressure must be put on patients or patient groups. Voluntariness also requires the existence of different options, and if possible that means more than just a yes/no-decision should be possible. Options need financial funding and therefore the Fundamental Rights Model also advocates the unpressurised public funding of genomic services in those cases when the creation of options contradicts the economic limits of health care plans and insurance policies. The Fundamental Rights Model requires financial resources which would contradict all of the budgetary constraints we see in Europe. However, financial resources are not the only issue, and Andrews states that a close connection between fundamental rights and information also exists. An ethically and legally valid execution of fundamental rights requires that competent persons have been informed to the highest possible level [21]. This subcategory of the Fundamental Rights Model, the concept of informed consent, is often equated with the Model as a whole due to the enforcement of informed consent in all European jurisdictions. But we need to see clearly that the informed consent is part of a broader and more complex concept (e.g. as the principle of informed consent is not linked to a concept which calls for free choice). Thus, the Fundamental Rights Model



poses a complex governance challenge as neither the old-fashioned hierarchy orientation nor the modern expert (committee) network model can guarantee the precedence of the individual. Governance in the sense of the third model must be seen as a framework for individual freedom whilst setting up, as a core task of governance, a process of balancing contrary individual rights.

Rules and principles as a governance tool

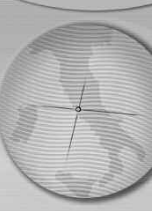
Now that we have depicted the three major models in a rather conceptional way we should also take into consideration how they might be technically transferred into regulation. Due to the particular impact of genomic information, there is a natural conflict between a negative ("the right not to know") and a positive right of claim ("the right to know" plus the obligation to assist) on an individual level. To date these individual conflicts, like the conflict between the two rights, are primarily discussed in most jurisdictions as constitutionally safeguarded rights that are at odds with each other. From a public health point of view the process of balancing the individual rights at stake is a necessary, but not a sufficient step, as public health is not subject to the parties' disposition. Still, public health recognises the individual rights in its conceptual framework and thus analysis is needed to determine how the tension between individual's rights and public health and between a pure Medical Model and a Public Health Model may be eased. We can only strive for concordance of all three models if we understand their specific individual value. From the Medical Model, the importance of a scientific basis for health care decision making can be derived, for example, the evidence-based decision making process in HTA. The Public Health Model reminds us that there is more at stake than individual rights, as the whole healthcare system should enable and empower patients to access a suitable, high quality medical service [22]. Finally, with the rise of autonomy and the loss of common values in a diverse society, the Fundamental Rights Model reminds us that neither the Medical nor the Public Health Model should obstruct the freedom of individuals unless there is an ethically valid reason to do so.

Thus it seems favourable to consider a different governance model for genomics which integrates the existing models and which applies to both family conflicts in a family and those inside a particular genetic community. In principle this new governance model should fit into existing regulatory channels. One theory of regulation which might serve as a benchmark is the

positivism as described by H.L.A. Hart; according to Hart regulation is effected by primary and secondary rules [23]. Secondary rules are procedural in nature as they determine how the primary rules are formed, who is allowed to apply them and who is appointed by society to execute them. Now, in our concept of genomic governance primary rules seem to be decisive for the individual layer of the conflict as they either grant a right or impose an obligation on the person. Rules in this sense need to be fully enforceable, allowing the owner of "a right to know" (or not to know) to possibly consider legal action if need be. With regards to public health, the description of a primary rule seems to be more complex as the healthcare system and not the individual right of a person takes centre stage. A primary rule in this sense must serve a distributive justice rather than the individual interest and thus considerations should be given to a second concept of regulation in order to assess the concept of Hart from a public health perspective.

The theory of Hart (as the whole positivism) has been widely criticised; due to the scope of this article only the concept of Dworkin shall be presented as a counterpart. Within the concept of Dworkin it is necessary to focus on those functions of principles which help us to detect a different governance model behind the distinction between rules and principles. In his "Taking Rights Seriously" Dworkin describes the difference between rules and principles himself: "Principles have a dimension that rules do not – the dimension of weight or importance. When principles intersect (the policy of protecting automobile consumers intersecting with principles of freedom of contract, for example), one who must resolve the conflict has to take into account the relative weight of each" [24].

If we follow the concept of Dworkin, we need to clarify which layers of the conflict we want to assess. Recalling the conflict of the "right to know" versus the "right not to know" we may come, and I would argue that we should come to the conclusion that both rights have the same weight as they are both derived from the individuals autonomy. If we want to expand our investigation to the principles which represent the idea of public health, we must be aware that principles are assessed against the background of a preassigned benchmark. If we analyse a principle, we will already have a particular idea about the potential value of that principle as we include the "new" principle into the pre-existing set of principles which already structure our life.



This process which requires a reflection upon the importance of values takes place on both, the individual and the societal level.

As a consequence, it seems to be inevitable that we need to have an idea of the desired outcome before we start the process of attributing weights. Still, it is desirable to find a general principle which guides the decision making process. The relatively simple individual conflict between a "right to know" versus an individual "right not to know" challenges the person or institution in charge, as they would have to reflect all of the social and ethical consequences of a "rule of precedence". Due to the identical weight of the two rights the grounds of precedence must be found by applying a third principle, in this particular case one from outside the principles of autonomy and privacy. If we expand the concept to public health then analysis is required to determine whether and to which extend the communitarian principles of public health should be weighted against individual rights (principles). On a conceptual level the comparison of principles from different sources raises concerns, but with public health we may argue that public health guarantees every individual a fair share in healthcare. This fair share has two implications in terms of principles: public health ensures a distributive justice in society and it guarantees every individual the opportunity to use his autonomy, as health is a basic condition to do so. Thus, public health stands for more than just the aggregated interest of society; it also advocates the rights of individuals who can not represent themselves in a democratic decision-making process.

Principles in a procedural genomic governance model – a different solution or just a disguise of problems?

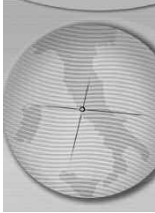
As a compromise, the growing knowledge regarding medical applications of genomic data might require "dynamic" governance which enables communities and family members to limit or prohibit the application of genomics while enabling patients and consumers to profit from new opportunities. Technically this may be done by either setting up a legal framework which allows state institutions such as ethics committees or courts to mediate genetic conflicts or by enabling families and communities to negotiate the weight of conflicting principles [25]. A "dynamic" approach depends on the ability to integrate rules ("claim rights") and principles in a structured process which regulates both the micro and macro level of decision making. Before we explore a model of governance which unifies the three models already addressed it is necessary

to reconsider the meaning of governance in this context; governance is described as the attempt to control, direct, shape or regulate human activities and the risks that are derived from it [26]. As this is not a self-executing process the question remains: who is going to set the (enforceable) rules concerning the application of genomic services and how should it be done? Inside the framework of Hart the State is the main regulator, defining rules which must be obeyed by citizens and courts. However, in the field of genomics and public health, any State will have difficulties in drafting stable rules which also anticipate the revolutionary development of genomics.

As we have seen Dworkin opts for a different concept leading us away from the individual conflict: "The origin of these as legal principles (the principles developed in the *Riggs* and *Henningsen* decisions / the author) lies not in a particular decision of some legislature or court, but in a sense of appropriateness developed in the profession and by the public over time. Their continued power depends upon this sense of appropriateness being sustained" [27].

If we draw a balance between these two concepts we might end up slightly puzzled due to the current state of genomic regulation and the ongoing public debate. Different solutions have been indicated within the three different models of governance as discussed above. The Medical Model seems to be an expert system, with medical experts setting the standards for access to as well as the feasibility of the genomic services [28]. Rules can be either set inside the professions (codes) or by a statutory legislator who may waive his political power in support of expert groups and expert committees. Principles depend on the appropriateness inside the medical sphere, replacing the public within the general concept of Dworkin through expert members of the profession. The Public Health Model combines medical (scientific) knowledge with ideas of political utilitarianism, setting genetic standards in a top-down process in accordance with public needs and financial limits. Rules and principles may be shaped inside the public health sphere, as the priority setting process requires the integration of expert knowledge from different professions into a patient and system oriented decision making process. The Fundamental Rights model follows a different approach as it is highly individualistic and without an inherent notion of medical knowledge.

The three different Models we mentioned, Medical, Public Health and Fundamental Rights Model, have substantially different implications on



a future model of governance in the field of law and human genomics. Yet we have to acknowledge that none of the three models is particularly suitable as the sole model of governance in genomics.

A solution is the creation of a more principle-driven procedural law which entitles the persons concerned to the right to be involved in a communication process, both on the level of individual conflicts as well as inside an affected community [29]. A procedural law, which guarantees the involvement of stakeholders as well as the deliberation of any ethical concerns if more than individual rights are at stake, is capable of integrating the main aspects from all of the models that we have analysed: medical knowledge, the input on healthcare systems, the ethical and social consequences and in particular the autonomy of the person affected. Still a principle oriented procedural law requires a "control point", or if we think of the process which leads to the balancing of conflicting principles, then we may call it a "master" principle which can be used as a benchmark in the process.

If we search for the least common denominator of all three models the principle of healing is included in all three and might therefore serve as a standard gauge for a "dynamic governance" [30]. In the Medical Model it is associated with the medical term of indication; an indication is necessary to justify any medical intervention. The same applies to the Public Health Model; if there is no medical indication then there is no requirement to spend public resources on the application of genomic services. Inside the Public Health Model there is a perspective shift resulting in the addition of a system-oriented and health economic view. Within the Fundamental Rights Model the medical indication of a genetic test must be seen as one decisive indicator for the precedence of a person's right in a conflicting situation.

Conclusions

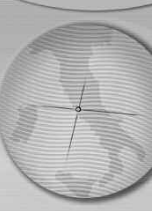
The principle of medical beneficence should be seen as a key reference point in the design of any genomic governance model which integrates principles into normative rules [31]. Medical beneficence as a *telos* should be assessed with regard to the scientific medical evidence and the cultural and ethical values which are present in the governed community. The concept of medical *indication*, which is a milestone within the assessment of the medical beneficence of a genomic technology, corresponds to the concept of the *purpose* in data protection law. Thus, the principle of medical beneficence serves to

establish concordance between the conflicting principles in both the medical and the privacy legal setting.

The governance idea behind the different applications remains the same: Genomics requires a new governance model which integrates the personal values of the people concerned, the medical knowledge necessary to define a genomic indication as well as the procedural law which enables those professions and families involved to make an ethically and legally acceptable prioritisation of dissenting interests in genomic services and data [32].

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