Knowing, relationships and trust—citizens' perceptions of whole genome sequencing for the Genetics Clinic of the Future

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Abstract

The objective of this article is to present various views from different groups of citizens on the topic of whole genome sequencing (WGS). Sixteen focus groups were carried out in Italy and Austria which aimed at reflecting on the question of how to ensure that the implementation of WGS into the clinic is relevant and responsive to the needs of all members of society. In the qualitative analysis of the focus groups, three key themes (knowing, relationships and trust) were investigated. Although the majority of the participants favoured a person-centred care approach, we also found more radical perspectives in the relationship theme. This includes a self-centred orientation in which health care institutions should be prepared to integrate self-interpretation efforts of citizens and develop strategies to deal with them. Different attitudes towards getting to know genetic information (knowing) and varied approaches to decision-making for or against the use of WGS were observed. Personal capacities, in particular those to handle medical information, were emphasized as key factors. This means that it is important not to connect the desire not to know with a rejection of the technology per se but rather to support information and consultancy processes that effectively involve citizens. Concerning the third theme, we have underlined the important role of mistrust in addition to trust because it mostly points to areas or conditions considered problematic. Thus, mistrust is also a way to articulate critique, for example, of the profit-making with patient data, that has to be taken seriously by governance.

Keywords Whole genome sequencing · Public engagement · Focus groups · Public perceptions

Introduction

Over the last few years, a rapid scientific and technological development in the field of DNA sequencing has been observed. Whole genome sequencing (WGS)—the ability to determine the entire DNA sequence of an organism—has quickly grown from a very expensive scientific sensation to an affordable application, and sequencing whole human genomes has already started entering the clinical setting (Cirino et al. 2017; Holm et al. 2018; Vassy et al. 2014; Vrijenhoek et al. 2015). New sequencing technologies and the corresponding analyses and interpretations pose advances for the diagnostic and therapeutic process, such as the identification of causes for genetic diseases. Despite this potential,

WGS is still at an early stage of implementation, and further evidence is needed for its clinical utility, in particular regarding the connection between genetic variants and disease risks or treatment options (Krier et al. 2016). At the moment, WGS might provide results for which the implications are not yet known (e.g. variants of unknown significance).

WGS poses also a number of social interrogatives (Klein 2004), such as the handling of huge amounts of produced data, a shift of responsibility to the patient in decision-making, questions of distributive justice and access for vulnerable and less advantaged groups, to name just a few. Hence, it can be described as a “radical innovation” (Stirling et al. 2015) that deserves particular attention and regulation. This diagnosis is highly related to a change of deeply entrenched practices, roles and responsibilities of and the relations between different stakeholders such as health care policy, medicine, research, patients, consumers and companies (Borry et al. 2018).

Aiming at socially robust decisions in the implementation phase of WGS, it is advisable to involve diverse relevant actors (Nowotny et al. 2001) and to include the views of

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different stakeholders including those not dealing with WGS in a professional way. This might resolve the “implementation problem” (Zuiderent-Jerak 2007) that could occur if psychological and social dimensions or, in other words, not strictly scientific criteria are not adequately integrated in decision-making concerning complex issues such as innovations in genetics.

Borry et al. (2018) thoroughly identified a variety of challenges in the concerned relationships (e.g. between doctors and patients or family members) to be respected in the light of these developments, and the authors recognize the need to take the “views of the public” into account. To date, there is limited information about these views and the interest of the public in WGS and in knowing about one’s genetic constitution. As the technology is entering the clinical practice, there have concurrently been studies characterizing participants of genome sequencing studies (Genetti et al. 2018; Kohler et al. 2017; Lewis et al. 2015; Taber et al. 2015) and their interests and reasons for participation. As Lewis et al. (2015) pointed out, participants in the ClinSeq project resembled early adopters of other technologies, showing high levels of optimism and resilience. Kohler et al. (2017) studied personal utility, which had been defined as perceived benefits that are distinct from clinical utility among adult participants of the same study. The elements to be found most relevant were “increased self-knowledge, knowledge of the condition, altruism, and anticipated coping” (Kohler et al. 2017, p.290). Genetti et al. (2018), on the other hand, analysed arguments of parents declining participation in a newborn genetic screening study which had been conducted in the USA. Among those who provided other specific reasons than a lack of interest in research, some mentioned doubts specifically regarding genomic screening, for example, not being interested in or not being comfortable with genomic screening (13%); concerns about privacy or discrimination (13%); or concerns about the possible psychological impact of unfavourable or uncertain results (12%).

Less is known about the general population’s interests regarding genomic sequencing. Etchegary (2014) published a review on public attitudes towards genetic risk testing. She concluded that attitudes were generally positive towards genomics and that there was interest in genetic testing for predisposition to disorders. However, the issue of data sharing seems to be more controversial, and people are hesitant especially when it comes to sharing genomic data internationally (Majumder et al. 2016).

Nevertheless, data on public views regarding WGS are still limited, and this paper thus aims to add insights regarding concerns about WGS as well as the potential opportunities and challenges citizens perceive accompanying its implementation. The study at hand focused on the views of people not (yet) professionally involved in genetics, genetic counselling or whole genome sequencing, but who might encounter it in

the future—as patients, family members, customers or in their professional lives. In this article, we present results from focus groups which took place at the beginning of a larger public engagement activity. This study was part of the international project “A Stepping Stone Approach towards the Genetics Clinic of the Future” (GCOF) which aimed at reflecting on the question of how to ensure that the implementation of WGS into the clinic is relevant and responsive to the needs of all members of society from different (trans)disciplinary angles as well as at exploring possible solutions.

Methods

In this section, we describe the design of the GCOF-PE model and, in particular, the material and methods used for this article.

The underlying aim of the engagement process was to establish mechanisms, structures and spaces to enable mutual learning and understanding between different participants. Therefore, a variety of social groups (such as patient support groups, senior citizens, communicators, young people and health professionals) was involved, and a mutual learning process between these groups and the participating professionals (geneticists, ethicists, bioinformaticians, scholars of future studies and biobanking) was tested. As a consequence of a mutual knowledge exchange, both professional and “lay” understandings of the issue should proliferate. The method was based on some sensitizing concepts (Blumer 1954), such as “mutual learning” itself. The mutual learning concept draws from Argyris and Schön’s work (1996) in organizational learning theory. Scholz (2000) transferred mutual learning to the field of science studies, applying it in a case study on responsible soil use. Mutual learning as implemented in the process being presented reflects recent developments in scientific governance, culture and practice, aiming at moving from research for society to research with society. It is hence characterized by new ways of knowledge production (Gibbons et al. 1994), in which all parts of society benefit from new input and ideas, learning is maximized, and problems are managed jointly.

The public engagement design process

The GCOF-PE process consisted of three main phases which were realized in Austria and Italy in parallel—two national contexts that were expected to differ in terms of public views towards biotechnology. Austria has been known for its broad public scepticism towards the collection of health data, whereas the general public in Italy is more willing to participate in genetic research (Gaskell et al. 2010). Involving participants from those countries allowed for a wide range of views within the limits of the available resources.

Only the main steps were planned beforehand, leaving enough possibilities to adapt the process in a flexible way. Every step took the experiences from the previous steps into account, considering the participants' wishes and/or concerns. Thus, the operational details varied within the two case settings, Austria and Italy.

The first phase focused on opening up the topic of whole genome sequencing by engaging different social groups in homogenous discussion groups. These groups were a basis for participants to decide whether they wanted to engage with the topic further or to remain within the project and cooperate with the consortium partners in the subsequent development of stepping stones for a Genetics Clinic of the Future.

In the second phase of the process, a new, heterogeneous collective was formed. To achieve continuity in citizen participation, interested participants (16 in Italy and 17 in Austria) volunteered at the end of each group discussion to be more actively involved in the further course of the project. These participants had the role of “ambassadors”, with the task of initiating and facilitating bi-directional communication, and acted as an interface between the GCOF project and their social environment. Ambassador engagement involved a mix of different methods (e.g. workshops, notebook for field notes, exchange of questions and information via email or telephone) and deviated according to the needs and interests of the ambassadors in the two countries. Some of the methods, such as the notebook for field notes, were developed and provided by the project team, aiming at a balanced mix of methods and ways of engagement. Others, such as a workshop with a medical geneticist in Austria, had been suggested by the ambassadors themselves and organized by the project team.

Finally, to enable face-to-face mutual learning between local ambassadors and scientists of the project consortium, two meetings were organized on two consecutive days in November 2016 (one in Austria and one in Italy). The major aim of these four-hour events was to merge the insights that consortium members and ambassadors had gathered during the previous year and take this basis to work together on very specific issues and questions, such as a web platform for the management of dynamic consent (Budin-Ljøsne et al. 2017) and a data management tool (Woolley et al. 2018).

Discussion groups: selection, composition and framework

Group selection was mainly aimed at diversity with regard to professional background and age, but it also considered a certain connection to or interest in the issue in order to enlist participants for a long-term and continuous engagement with the project. Homogeneous groups provided the space and environment for discussions with people sharing similar characteristics or backgrounds and a first step in opinion forming, shaping and expression. Homogeneous had been defined as

people sharing particular aspects of their lives which might be professional, demographical, based on interest in or connection to the issue, etc. Involving homogenous groups provided certain advantages, such as easier access to participants due to already existing collectives and their respective contact person, as well as minimizing problems in entering the discussion through a shared experiential background, collective patterns of understanding and a rather familiar surrounding (Bohnsack 2003).

After agreeing on the groups (patient support group members, senior citizens, high school science teachers, high school students, science communicators, general practitioners, social sciences university students) in the project team, the participants were approached via different and specific channels, such as professional associations, senior associations, patient support groups and mailing lists. The invitation letter included a short description of the overall project, the issue of WGS, the course and purpose of the discussion groups and the contact details.

Between October 2015 and March 2016, 16 discussion groups were conducted (seven in Austria and nine in Italy) (Table 1). In Austria, a double interview was held with two general practitioners instead of a group discussion. Despite expressed interest of the professional association and its members, there were finally not enough participants registered for a discussion group.

Issues and questions to be discussed in the groups were identified via literature research, additionally taking into account the research and innovation topics within the project team that resulted in a comprehensive discussion manual. The manual included the methods to be used in the discussion groups, including free association, a video as stimulus material, a topic guide, eight scenarios and two short questionnaires. The input material and methods were used in a flexible way according to the setting and the previous knowledge of the participants.

One of the respective national project team members moderated the groups. An additional observer was present and took notes. Each group discussion lasted about two hours. The discussions were recorded (audio only) and transcribed after obtaining consent from all participants.

Data analysis

The analysis of the study at hand was based on the transcripts of 16 homogenous focus group discussions (seven in Austria and nine in Italy). A thematic analysis (Braun and Clarke 2006) was carried out using an inductive approach, moving from the data to empirical generalization (Glaser 1978, 1992; Bulmer 1984). In a first coding procedure, three main themes and related questions (see Table 1) were identified. These three themes were knowing, relationships and trust. For the

Table 1 Overview of the discussion groups held in Austria and Italy

Date	Group	Number of participants	Location
10.10.2015	Science communicators	12	Padua/Italy
21.10.2015	Senior citizens	11	Conegliano/Italy
9.12.2015	High school science teachers	7	Vienna/Austria
11.12.2015	High school students	9	Vienna/Austria
12.1.2016	Patient support group	6	Vienna/Austria
13.1.2016	Social sciences university students	19	Vienna/Austria
21.1.2016	General practitioners	8	San Pietro di Feletto/Italy
11.2.2016	Science communicators	7	Vienna/Austria
2.2.2016	High school science teachers	9	Conegliano/Italy
16.2.2016	High school students	12	Conegliano/Italy
23.2.2016	High school students	14	Padua/Italy
26.2.2016	High school students	11	Padua/Italy
25.3.2016	Senior citizens	9	Klosterneuburg/Austria
31.3.2016	General practitioners	2	Vienna/Austria
16.4.2016	Patient support group	4	Pordenone/Italy
19.4.2016	Patient support group	6	Venice/Italy

further analysis, they were used to extract specific subthemes (dimensions) (see Table 1).

In all stages of the analysis, an iterative process of exchange and adjustment took place between the authors. The consistency of the dimensions with the themes and guiding questions was discussed in several sessions.

They are presented similarly to Weberian ideal types (Emanuel and Emanuel 1992) and thus do not describe in detail the complete focus group interaction. The construction and the use of an ideal type, in a Weberian sense, is a heuristic way of investigation. The ideal type is not an average type nor a simple description of real-world phenomena; it is a conceptual tool used to systematically characterize, combine and order an indefinite number of phenomena into unified, concise analytical constructs. The results are presented and discussed in the following chapter.

Empirical results

Knowing

In all discussion groups, participants assumed that knowing about one's genetic profile could have concrete consequences. Some of them might be ethically problematic, such as regarding family planning (in particular: decision against a child) and prenatal diagnosis (in particular: abortion). Personal disadvantages, for instance, with insurances or employment, were also mentioned as possible negative consequences of

knowing, especially when it comes to severe diseases or stigmatized traits. Coming out with one's knowledge would, moreover, have consequences for social relationships. People might cut off contact because they cannot cope with the problems this issue triggers (e.g. job loss). Hence, knowing is, in the view of the participants, not merely a personal endeavour, but it has concrete consequences for the whole family as well as for the relationships between family members who might, for example, have to cope with feeling guilty for passing on a gene and thus suffer mentally. Looking for a partner, especially when one plans to have children, might as well become more difficult, although it was also argued that knowledge could lead to a better planning of reproduction, i.e. with reproductive technologies. Finally, knowing might cause changes in behaviour, lifestyle and one's attitude towards life. On the one hand, it bears the danger to isolate oneself and to permanently think about the illness and the possibility of dying. On the other hand, knowing could be the impetus to make the best of the rest of one's life and do what one always wanted to do beforehand. One could make provisions and change one's lifestyle to counteract a disease. This was described as an advantage. Participants, however, objected to the notion that many people would automatically change their routines if they knew. But they saw the risk that some might restrict themselves too much to avoid everything that is related to causing, for example, cancer, and hence only live in fear of the disease.

These possible consequences of knowing were basically shared in all groups, but there were very different ways participants thought they could be personally handled. Therefore, three main discursive argumentation lines were identified

Table 2 Themes and dimensions regarding WGS

Themes	Dimensions	Leading question(s)
Knowing	<ul style="list-style-type: none"> • Knowing as a burden • Knowing as empowerment to act • Knowing as a game 	<p>Do participants want to know about their genome?</p> <p>If yes, what, when and why do they want to know and under which circumstances?</p> <p>If no, what, when and why do they not want to know and under which circumstances?</p> <p>What is the basis for one's decision-making?</p> <p>Which consequences could knowing have for an individual, his or her closer environment, and for society as a whole?</p>
Relationships	<ul style="list-style-type: none"> • Informative-technical • Dialogic • Self-centred 	Which kind of relationships between health care/research and patient do participants imagine if WGS is implemented in health care practice and research?
Trust	<ul style="list-style-type: none"> • General scepticism • “Healthy” scepticism • Selective mistrust 	How do participants assess the roles that different professionals, stakeholders and institutions (research, state, companies, doctors, insurances, patients, etc.) play in the process of WGS?

which represent major dimensions of the knowing theme (see Table 1). In the next three paragraphs, we provide a detailed analysis of these three dimensions.

Knowing as a burden

In the first dimension, a passive/defensive position to WGS prevails. Participants claimed that receiving predictive information for certain diseases would lead to constant worries and anxiety which would reduce their room for manoeuvre in life. This argument was based on the perceived uncertainty of one's own mental capacities to handle unpleasant information. Hence, participants who characterized themselves as easily worried immediately refused to know about their genetic status. Moreover, they assumed that their relatives (partner, children) would feel the same and that informing them about possible diseases would burden them as well. Thus, for the participants, how a person handles medical information played a major role in the decision-making process. In this regard, it was argued that for a person who might have tendencies for depression, going through the process of WGS “is perhaps not a good idea and is self-destructive” (FG high school students, Austria). Another aspect of this argument was that you would have to be mentally ready to take the personal (e.g. lifelong medication), social and institutional consequences of knowing. Concerning the latter, it was stated that in order to be able to argue in a self-confident way with health representatives, you need to be a strong, competent and informed person. Against this background, it was considered better to ponder long and intensively over the question of whether you want to know. This was argued with a view of knowledge as something “you cannot get rid of”, as being like a “tattoo”, “for eternity” which “you can only try to suppress” (FG high school students, Austria). Another argument against knowing was the possible consequence of needless fear—

needless, as a disease does not necessarily have to occur. In discussion, this was connected with a “wait-and-see” attitude and a concept of illness being fate that should be dealt with not before the point of time when the person becomes ill: “If an illness comes, it comes. Then there is still time to cope with it and accept it” (FG senior citizens, Austria). Thus, not knowing allows for a more easy-going and carefree way of life, in which one should think positively: “Everything will work out fine” (FG senior citizens, Austria). Participants characterized people who do not want to know as having a more fatalistic attitude towards life who take and accept things as they come.

However, the perception of knowing as being a burden was related to the kind of information people would get, especially with regard to the certainty of the results: “What is frightening to me is not the knowledge of too many things, but knowing something that might happen but also might not” (FG high school students, Italy). The burden of knowing would not weigh that heavily if it entailed clear and accurate information instead of probabilities, as one participant put it bluntly: “That they just tell you that you might get this and that, and in the next three years you have a 70% probability of dying – who wants something like that?” (FG patient support group, Austria). In this regard, science was described as not yet being ready to give adequate certainty. Thus, the actual purpose of WGS would currently be only collecting data for research. Therefore, predictive genetic medicine was seen by some as superfluous, because most diseases would have multifactorial reasons and percentage rates would be too vague. Others suggested that a certain grade of “tolerance of ambiguity” (FG science communicators, Austria) was a major prerequisite for handling genetic knowledge. People who decide to have their genome sequenced have to be aware to live with knowledge about probabilities which may not be well founded, as well as to have no place to go for further clarification. It was assumed, accordingly, that people who have some kind of knowledge in

genetics could handle their genetic health risks more easily. Scientists were characterized in this regard as inherently much more curious and as having more experience in dealing with uncertainty. Lay people, on the other hand, would be in greater danger of being concerned when confronted with probabilities. So, different grades of relation to the issue would lead to different perceptions of it.

Knowing as empowerment to act

In the second dimension, an empowerment argument prevails that WGS was seen as a possibility to gain more knowledge about oneself, which allows for action and thus opens up room for manoeuvre. Timely knowledge would allow for timely treatment, leading to better chances for a cure. Regarding self-perception, it was explicitly argued that knowing about one's genome would not change oneself as a person; it just provides you with an information basis for adequate and personalized action: "The clock is ticking anyway, but then I would know how it ticks" (FG high school science teachers, Austria). In this regard, a participant of the science communicator group referred to the prevailing inherent human will to survive and to prolong one's own life: "Nobody says: 'I'm not going for a medical check-up because I'm scared about having a higher risk of breast cancer'. People want to know to take countermeasures" (FG science communicators, Austria).

Similar to the first dimension, there was ambivalence related to the uncertainty of predictions, and probabilities were seen critically. This reflects a common dilemma: the possibility for precaution versus discomfort with probabilities. In the science communicator group, this dilemma was compared with "a threat of living with the sword of Damocles" (FG science communicators, Italy) and described as a difficult act of balancing the fear of living with a genetic predisposition and the chance to counteract a possible disease at an early point of time. The solutions for this dilemma were the frequently used "you never know" or "there are no 100% guarantees" sayings. This was a strong rhetorical figure in the discussion of nearly every group, both against and for WGS. It was based on the conviction that there is no certainty to stay healthy and that one has to let go of guarantees anyway. Examples were having a predisposition for Alzheimer's but dying beforehand in a car accident and the story of a person with a "very good" genetic constitution travelling to Brazil or Africa, getting Ebola and dying.

Another important resource to escape this dilemma was selective knowing—what and how people want to know about their genome. Our data show that most of the participants would prefer to know only certain aspects in contrast to knowing everything about their genome. A particularly frequent argument was that they were only interested in knowing about conditions that are not lethal and are treatable through lifestyle change/adaptation or medical therapy, which was illustrated

with drinking less alcohol when you have a predisposition for liver cancer and eating less sugar in the case of diabetes. So, treatable diseases received a significant status and were differentiated from non-treatable ones. Knowing could, in this respect, take away fear and open room for counteraction. Knowing one's probabilities related to Alzheimer's or Huntington's disease (both not treatable) was seen as scary. A counterargument against selective knowing was that with one's data, research has the possibility to develop drugs against such incurable diseases. So, the more data research has, the better. In this regard, the argument of not wanting to know because of problems with handling the information of getting a fatal disease was presented as a very egoistic one and given added weight with a killer argument: "I would do it the other way around, do it when they are not treatable, because you have to think about your children. Do you want that your children die of a major inherited disease?" (FG high school students, Austria).

Knowing as a game

The playful character of knowing was raised as a more marginal position in the discussion with science teachers. The process of interpretation was described as fun, exciting and interesting, "a game with yourself" (FG high school science teachers, Austria). This game metaphor was used also in other groups, especially by participants who had some kind of professional or scientific background. Scientists were, in this regard, characterized as inherently curious: "So, when I said curiosity, it was only because I have a background in science and therefore am curious by nature" (FG patient support group, Austria). Family doctors also presented themselves as generally curious and interested in their genome, but for them the question remained about what to do with the information. Like other professionals in the groups, they would not automatically trust the results. Instead, they would play with them, analyse them or do additional research: "I could not get the information and just leave it at that" (FG general practitioners, Austria).

In the seniors group, on the other hand, the game metaphor was used differently. Self-experimentation with data was explicitly rejected. The group expressed that WGS would only make sense if you got out very concrete data on treatments for the individual, and that medicine had to have answers for therapy. The argument here was that "insight alone is not enough" (FG senior citizens, Austria).

Relationships

In this section the expectations and arguments expressed by different focus group participants regarding the interactions between different actors in the health system—such as doctors, researchers or patients—are described. This involves the

goals of doctor-patient interaction, the use of information and data, the doctors' obligations, the patients' values and level of autonomy and the level of power that structures the relationship.

There is extensive literature on the patient-doctor relationship (Clayman et al. 2016; Emanuel and Emanuel 1992; Frank et al. 2014; Kelley et al. 2014), but there is no detailed study of the changes it might undergo with the proliferation of genetic sequencing. The study at hand took into consideration how the participants perceived the role of actors, addressing not only the patient-doctor relationship but also the perception of those actors who do not directly interact with patients but are involved in analytical and diagnostic steps of the WGS process. In what follows, we identify three different dimensions of relationships, which are characterized through aspects such as the density of relationships, the directions of communication and the main actors involved in the relationships.

Informative-technical

In this dimension, a unidirectional flow of information from doctor to patient prevails, and the interaction is dominated by information transfer rather than by shared decision-making. Physicians use their role as gatekeepers for information more than in other dimensions of relationships. They know about the spectrum of genetic diseases, and they communicate and translate the sensitive information for patients. But they stress also their supportive function, for example, when it comes to clarifying media hype, such as in the case of Angelina Jolie,¹ and to answering questions that consequently emerge. As an Austrian doctor put it in a nutshell: "There will be a lot of information and appeasement effort. But that's what I am doing now anyhow" (FG general practitioners, Austria). Hence, they play a decisive intermediary role and are a first personal contact point for the numerous questions which arise regarding genetics on the side of patients or citizens.

In this informative-technical relationship, doctors and the health system have the duty to protect and control the information they present to patients. Doctors who emphasize this particular dimension refer to the importance of their professional autonomy. Although they certainly integrate external expertise in their decision-making, this process remains more or less opaque for and undiscussed with patients. Regarding the management of the process of WGS and data processing, participants demonstrate a high degree of trust in medicine and the contribution of scientific research. This indicates that these actors are ascribed the primary responsibility for information and that their competence is not doubted. Doctors, in turn, present themselves as competent and technical experts who are able to guarantee anonymity and data protection: "I

see the concept of privacy, and it will never change, because it is a closed circle between the subject, the laboratory that performs the analysis and the doctor on whom he relies. [...] Patients will have a trusted person as the family doctor to perform the tests and the diagnostic procedure" (FG general practitioners, Italy).

However, in such forms of interaction, there is hardly any room for the discussion of social or psychological issues, which can lead to the patients' impression that information is disconnected from themselves. Thus, it is perceived as informative-technical. Down's syndrome was used as an example in this regard. Participants described it as a well-known and researched genetic condition but claimed that, still, serious communication between doctors and patients after giving the diagnosis is lacking. Communication would often stop at this point of time, and patients would be left alone with their questions, especially related to the impacts for their everyday life: "I know mothers who drove through red traffic lights afterwards. They just let them go home without psychological support. Without asking them: 'What can we do? Who can support you? Is there anybody to pick you up?' Or anything. And this is so dangerous..." (FG patient support group, Austria). Hence, the strong concern was raised that if comprehensive counselling is not implemented for known genetic conditions, such as Down's syndrome, how should it work for WGS where so little is known?

Participants furthermore criticized official decisions on financial support relying on persons with limited insight in the cases.

Participant 2: "Yes, the medical assessor claims that she knows, and she says: 'No, the disease will be assessed differently now'. And I say: 'The disease? Have you ever seen my daughter? Do you know her history?'". Participant 5: "And that's just it, that in spite of genetics, the human is still a human and not only genetics" (FG patient support group, Austria).

Such bureaucratic handling of individual cases would reinforce a rather technical relationship between patients/families and doctors or other professionals in the health care system.

Dialogic

This dimension is characterized by a strong network character in order to provide sound information and support for all involved actors. Exchange happens between professionals and patients, between professionals in an interdisciplinary way and among patients. Communication and interaction are institutionalized and not only dependent on informal, arbitrary means and ways.

Family doctors and patients interact with each other on different levels. Doctors stressed to consider the context in

¹ Borzekowski et al. (2014) investigated the so-called Angelina Effect on the public.

which patients live, with particular attention to the family environment: “We are lucky in our work, perhaps it’s luck, that we got to know our patients when they were healthy, not after they had fallen sick, so we have been with them for some time, and we have also learned to manage the interpersonal relationship” (FG general practitioners, Italy). Participants adopted a person-centred approach, which, in their view, might contribute to a more accurate diagnosis and prognosis through genetic sequencing data. They also maintained that the dialogical relationship allows them to have a certain serenity: “Dialoguing and passing on information can, in any case, make it easier to reassure me about certain things” (FG patient support group, Italy).

Hence, the dialogic dimension offers an approach where the genetic results are discussed and communicated with a strong focus on capturing the consequences on patients’ daily life, avoiding purely medical one-way information processes. Nevertheless, such person-centred relationships were not seen as realized yet. Regarding the doctor–patient interaction, participants mentioned shortcomings in the communication of delivering sensitive issues to the patient. In order to enter a real and empathic dialogue, participants suggested more training in this regard for medical practitioners. This would be particularly important when it comes to transmitting sensitive genetic information to patients.

A particular vision of the health system was raised in the Austrian patient support group: an institutionalized network of different professionals who consult each other if necessary. This implies different ways of actively involving diverse actors in the process of managing genetic data and patient care, including expertise in non-strictly medical aspects such as psychological, ethical and social issues. Hence, in this dialogic dimension, continuous multi-level communication takes place, and a whole network of relationships collaborates to serve the individual needs of the patient.

In a dialogic perspective, providing data for research purposes was seen as positive and sometimes even extraordinary—something that people are normally unable to do. Participants perceived it as a possibility to contribute to medical progress: “You give yourself, and that’s the thing that sets you apart from any other on the face of the earth” (FG high school students, Italy). The Italian patient support group signalled the need to give recognition, not only monetary but also symbolic, to those who participate actively in the process of sequencing and in disseminating data. Remuneration would, in this sense, contribute to a balanced partnership of equals.

Self-centred

This dimension of relationships particularly includes aspects concerning decisions to autonomously use genetic tests, methods of data management/interpretation and offers of the

health system. Personal relations are reduced to a minimum here, and technology seems to replace the communication between humans, such as in doctor–patient interactions.

The most self-centred behaviour was proposed by students when they expressed the desire to independently choose if and when to use genetic tests and how to use the results regardless of any medical check: “It’s my choice, I want to know what is possible” (FG high school students, Italy).

It is a radical position but indicative of a consumerism approach looking at genetic testing or WGS as something to buy as an object of investigation and knowledge, disconnected from or not necessarily connected to the health system. Moreover, young people usually do not have a frequent relationship with the health system and do not have regular meetings with family doctors: “I would not go to the family doctor [for WGS] because I rarely see him” (FG high school students, Italy).

Another group of people who argued in this way were those who have some kind of expertise in the field of genetics or at least know where to find information (e.g. papers, software) for interpretation (general practitioners, science communicators with a background in genetics, high school science teachers with scientific background). The following excerpt from one discussion group illustrates this position:

Moderator: “Where would you go with your hard drive (genetic data)?”

Teacher 1: “I would read papers”.

Teacher 5: “To the Internet. We would do that. If you know how science works, you do not trust one single study” (FG high school science teachers, Austria).

In the self-centred dimension, participants emphasized to not have mediators in the case of analysis and interpretation procedures, leaving free initiative to the individual. This orientation signals a limited consideration of the health system as a coordinated set of experts and skills, as well as a scepticism about scientific studies. It also corresponds with seeing the process of “knowing as a game”, which we described earlier in the knowing theme.

In line with these attitudes, the desire to make private use of data and a certain resistance to make them available or collect them for scientific research purposes is looming. Consequently, it is considered appropriate to prevent them from being made public: “All these discourses concerning DNA –it’s a personal thing because it’s unique, it’s my private life, I have to decide” (FG high school students, Italy).

Trust

The question of how participants assessed the roles that different professionals, stakeholders and institutions (research, state, companies, doctors, insurances, patients, etc.) play in

the process of WGS revealed particularly different aspects of trust. As Hall et al. (2001) have pointed out, there are numerous definitions of trust, but a common theme is the “optimistic acceptance of a vulnerable situation in which the truster believes the trustee will care for the truster’s interests” (p. 615). One can distinguish two levels of trust: the interpersonal relationships and, on a macro level, trust in institutions or systems. These two are, of course, intermingled, and the relationships with representatives of the system (e.g. doctors) influence the view of the system and vice versa. In the focus group material, the following main dimensions of trust have been identified.

General scepticism

Especially young discussion participants displayed a general scepticism of expert systems including the health care system. Their arguments and discussions were characterized by questioning every player in the system and by—to a greater or lesser degree expressed—distrust. Participants thought through a whole mistrust chain, where at the end they concluded not to be able to trust anyone but themselves. A strategy to cope is taking matters into one’s own hands through self-analysis and interpretation (see dimension “self-centred” in the relationship part). Here, the classical network of actors involved in genetic analyses and WGS is bypassed, and, accordingly, mistrust is reduced. A common rhetorical figure in arguing general mistrust was the use of the word “they”, designating a collective that is not further specified: “As soon as I give away my data, it is not in my hands anymore. Even if they tell me they will not do anything with my data and the interpretation is solely done by themselves – they can tell me anything” (FG university students, Austria). This statement furthermore illustrates the feeling of not being able to verify declarations, especially regarding data management and data security. One discussant put it very clearly: “It is certainly an illusion that a sequenced person has full control over his/her data” (FG university students, Austria).

Regarding the choice of who to confide genetic data to, it was argued that this “opportunity [was] gone” (FG university students, Austria) and that it was not even relevant to think about it as there were enough data available about everyone using the Internet which could be analysed similarly to genetic data.

“Healthy” scepticism

Another observable stance, or rather the absence of such, was characterized by being torn between trust and distrust. This became apparent in the discussions as participants argued in opposite directions, as, for example, regarding trust in doctors: It was argued that “they [the doctors] want to help” (FG senior citizens, Austria). But at the same time, a feeling of being at their mercy was expressed: “Again, being at the mercy of the

doctor because we cannot assess them [the results]” (FG senior citizens, Austria). The attitude of some participants towards pharmaceutical companies was characterized by being torn as well. On the one hand, companies were perceived as important regarding research and the developments of drugs. On the other hand, participants believed that companies have the power to and actually do influence the health care system (e.g. standards or thresholds) for their own benefit.

Selective mistrust

Selective mistrust designates mistrust addressed only to specific parts or professions of the health care system. An argumentation line often found in the discussions was the division between public institutions versus private, for-profit institutions or companies. While public institutions were perceived as acting for the common good, there were numerous instances where companies were assumed to act in a non-transparent way and might use patient or customer information abusively. As two participants put it: “And there is also the possibility that the researcher in a for-profit setting also uses data badly” (FG senior citizens, Italy), “Because private companies have always been looking for profit” (FG senior citizens, Italy).

A second, rather experience-based articulation of mistrust was a very critical attitude towards health authorities. This was especially expressed by members of the Austrian patient support group who had, due to their deep personal involvement, a lot of experience with the system. A perceived lack of knowledge and expertise on the side of health authorities had resulted in distrust among patients and their families. Science was seen as a trustworthy actor in this regard which should get more involved in clinical medicine and play a more active part, for example, in informing doctors, medical officers and health authorities. This distrust had arisen not merely out of having experienced doctors and health authorities as not knowing the medical facts but also out of having experienced them as not competent in dealing with patients.

Discussion

The analysis of the focus groups allowed us to deepen three key themes (knowing, relationships and trust) that emerged from the discussions among the participants. Identifying the main dimensions for which the participants proposed arguments, ideas, doubts and requests allowed the reconstruction of an articulate picture of elements that depicts the complexity of perceptions, attitudes, norms and values linked to WGS.

Regarding knowing, the consequences of a technological innovation, such as WGS, was debated on two levels: societal and personal. The ethical and social questions, such as data abuse or genetic modification of the human being, were seen

as very complex and hardly solvable. Participants reached the conclusion that the willingness to deliberately accept the potentially negative consequences seemed to be the most realistic strategy if society does not want to waive the benefits.

Although some of these consequences also apply for current practices of genetic testing, such as panel testing, WGS was perceived differently especially concerning the huge amount of (potentially predictive) data and information produced. In line with the notion of the GCOF project focussing on the “Genetics Clinic of the Future”, in the discussion groups, WGS was discussed in a broader context and imagined as a technology universally used in society. This raised new questions, such as responsibility, involvement and affectiveness, since the circle of tested persons would be extended from a risk group to all members of society.

The decision whether people would want to know about their genome was mainly based on their mental constitution, attitude to life and profession. People might take on different roles regarding their handling of the results. The “knowing as a game” argument highly corresponds with the characteristics of a hobbyist (Borry et al. 2018) or bricoleur (Levi-Strauss 1966), where the purpose of testing is curiosity and a passion to work or potter about with the results at hand in an unconventional and unexpected way. In this process, the bricoleur makes use of resources, which comprise artefacts and knowledge about methods as well as the procedures on how to use them, and creatively relates them to fit in the whole. Specific professions, such as teachers, doctors and science communicators, possess exclusive knowledge, such as how to find research papers or publicly available possibilities (e.g. websites) for self-interpretation, that is essential for the process of knowing in this more game-like way.

In most cases, the reaction towards genetic information corresponds with a specific personal view of how to handle medical information and thus amplifies already existing mental phenomena (fear, stress, worries, etc.), tendencies of self-improvement and self-care and curiosity and playfulness. Lewis et al. (2015) investigated in this relation the specific personality traits of participants of the first implementations of WGS, who were characterized as optimistic and resilient. This is a phenomenon often found in the group of early adopters of new technologies in general (Lewis et al., *ibidem*). As described in the dimension “knowing as empowerment to act”, for some people, the willingness to know is caused by an attitude of openness towards the unknown and the desire to open new scenarios that could activate new opportunities for research as well as for themselves. This perspective highly reflects the currently dominant societal goal of health prevention and the main health policy discourse and practice of precaution.

Our results, however, also reveal a deep rootedness of seeing knowledge as a burden, which definitely functions as a societal counter narrative. For Taber et al. (2015), this kind of

information avoidance is a defensive strategy due to a lack of threat management resources (self-affirmation or optimism). Participants in our study moreover argued that some individuals would lack the capacity (e.g. being strong, competent or informed) to be able to handle the personal, social and institutional consequences that knowing about a risk for disease might have. This was ascribed to those characterized as depressive or anxious as well as to disadvantaged and underprivileged people. It is important to note that some of those who claimed to not want to know did not match this characterization. They confidently presented their decision as a very conscious one and used a fatalistic attitude towards life and illness to justify their stance.

But which kind of support can be imagined on an institutional level for those who are uncertain and afraid? First of all, there has to be awareness among professionals working in the field of public health that a group of people exists who do not want to know and see knowledge as a burden. Knowing about their motivations would help to tailor counselling to their particular issues of concern. This has to be complemented with raising their health competence, especially concerning information about how to best manoeuvre through the health system. However, one of the most important measures suggested for the implementation of WGS would be to create a system which allows choosing what you want to know about your genome. Diseases would have to be distinguished in cohorts of actionable and non-actionable ones, because the first category allows for therapy, medication and lifestyle change, while the non-actionable ones could cause anxiety and thus might be excluded from the personal knowledge wish list.

The second point is related to the fact that participants articulated huge problems with dealing with probabilities and the consequential uncertainty of a result, which, in turn, might influence their intention to know and share results about their genome (Taber et al. 2015). While medical analyses are usually expected to reduce uncertainty, WGS results are likely to produce the opposite (Newson et al. 2016). Uncertainty is and will continue to be inherent to genomic medicine. It has been argued also in our study that science is not ready to, and maybe never will, deliver 100% true facts. Thus, people have to be equipped with a certain grade of tolerance of ambiguity and live with it. We moreover support putting into practice an “ethics of genomic uncertainty” (Newson et al. 2016), which emphasizes the importance that people should not get the impression that WGS will in any case reduce uncertainty, but that uncertainty should be communicated in a transparent and understandable way. As recent results in risk studies show (e.g. Renn 2008), uncertainty also occurs because most of the time scientific probabilities and percentages are understood wrongly, by both health professionals and citizens. Hence, besides installing genetic counselling that includes helping patients to understand specific issues of

uncertainty in WGS, we suggest taking measures that raise “risk literacy” (learning to deal with risk and uncertainty), which means equipping citizens with skills and principles that allow for a better assessment of ambiguous results.

The three dimensions analysed in the relationship theme reflect the literature concerned with different forms of power in doctor-patient relationships: from a more asymmetric relationship, in which the doctor acts in a paternalistic way referring to a rather mechanistic, technical approach (e.g. Freidson 1970), to trying to re-balance the relationship to achieve a partnership characterized through shared decision-making and dialogue (e.g. Epstein and Street 2011), to a shift of responsibility, judgement and power away from the doctor to the patient (e.g. Beck 1992).

The results of this study show that the sole authoritative-ness of the clinician is called into question and the desire for self-determination in the choices regarding the use of WGS is highly affirmed. Consequently, the transmission model of genetic information in a linear way from doctor to patient is strongly questioned. In the future, patients might come with their genetic results and data and discuss them with the doctor to jointly reach a diagnosis. Due to its complexity (medically, socially and ethically), WGS yet again calls for more inter- and transdisciplinary collaborations and network approaches such as those suggested decades ago, for example, in patient-centred care (Rogers 1961; Entralgo 1969). Close collaboration between different professionals regarding prevention, diagnosis and treatment as well as psychological and social support would serve the needs of the patient in a comprehensive way and simultaneously take away pressure and workload from the physician, who, in turn, would gain time to focus on important competences of the physician in the future—the communication with and long-term support of the patient. Against the background of big data, scholars (Fiske et al. 2019) even propose the creation of a new profession, the health information counsellor (HIC), with skills in interpreting health data, communicating medical information, handling legal and institutional aspects and advising further steps concerning medical issues.

The possibilities the Internet provides empower people to independently search for information, share experiences and question interpretations and therapies recommended by health authorities. Here, an interesting switch takes place: from the “patient researcher” (Prainsack 2017), who monitors him or herself and collects data that is then shared and discussed with a health professional, to the “self-analyst”, refusing to give away his or her data but nonetheless analysing and discussing it via the Internet to understand his or her own health status. An empowered handling of raw genetic data of non-professionals is, due to its complexity and dependence on highly specialized expertise, only at the beginning. Nevertheless, some participants in this study were in favour of going in this direction, and our results indicate that there are

target groups for websites offering analytical tools and information for the self-interpretation of results. The self-centred dimension reflects the growing vision of self-determination in the field of medicine with the intention of autonomously managing diagnostic procedures, health information and even self-interpretation of genetic data. In contrast to those searching for comprehensive counselling and consequently expanding their personal health-related relations, in the self-centred argument, relations are limited to a minimum and often pursued via the Internet.

Today, we live in a rather opaque regime of knowledge in which it is not possible to have definitive certainties. Especially when talking about WGS, the issue of trust is and has to be a crucial element. Our study reveals different dimensions of trust. The first, general scepticism, reflects a development over the past few decades in western societies, which is the decline of trust in authorities, experts and institutions with an increasing emphasis on individual strategies to judge risk and deal with uncertainties (Beck 1992). Self-analysis, exchange with and recommendations from non-professionals and searching for answers on the Internet represented ways of how to be independent from classical expertise, especially for the younger and higher-educated focus group participants. General mistrust is often not necessarily directed to the interpersonal relationship with a doctor or genetic counsellor but to the mainstream system, its imagined procedures and representatives. Mistrust is thus also a political statement, a critique of the non-transparent ways of how (genetic) data travel around the world for the profit of big companies. Young participants were highly aware that they are leaving traces on the Internet anyway and all the time, which leads them to a kind of resignation concerning data safety and protection. This generation, who grew up in the digital era, has been involved in social media and has learned or experienced that all of our digital interactions (use of credit cards, use of social media, etc.) leave “digital breadcrumbs, which, when pulled together, offer increasingly comprehensive pictures of both individuals and groups” (Lazer et al. 2009, p. 721). But trustful behaviour is not totally absent; it is shifting to other semi-professional, non-institutionalized communities and sources of information. In the future, trust might be built in other ways, towards other or new actors and through new forms of services of analysing and interpreting the genome (e.g. small local start-ups, community labs or open source software).

Relationships of trust in the health care system are changing. Patients who trust blindly have been consigned to history. People question the doctor because they are aware that expert knowledge is contestable and they actively seek further treatments and second opinions. But trust-based relationships are still necessary (Rowe and Calnan 2006), especially in the area of medicine and health. Through a feeling of commitment and loyalty to a person or an institution, stability and continuity

could be reached which, in turn, would save costs for the health care system.

Nevertheless, some people experience difficulty in trusting or are torn between arguments for or against trusting certain actors in the health care system. This dichotomy is not necessarily connected to indecisiveness or a weakness in decision-making. As Abelson et al. (2009) described it referring to political sciences, distrust may be a “positive feature of relations of dependence and authority essential to democratic progress” (p. 64). In democratic systems, distrust is even institutionalized, providing “a kind of backup or insurance for those who would be ready to risk trust, a disincentive for those who would contemplate breaches of trust, as well as a corrective of actual violations of trust, if they occur” (Sztompka 1999, p. 140). People disputing in this direction, using arguments for and against trust at the same time, might thus be characterized by a kind of healthy scepticism, ready to trust but not blindly or unreservedly. As a result, healthy scepticism could be appreciated by the health care system as an impulse to strive for constant improvement. Selective mistrust, which is mistrust of particular fields or representatives of the health care system, always points to certain shortcomings in particular areas which should be carefully analysed and taken seriously.

Besides other factors, such as the practitioners’ competence and information sharing (Dibben and Lena 2003), our study shows that especially person centredness and transparency would encourage trustful relationships in health care. This reinforces other studies which conclude that the extent to which the health care provision is patient centred highly influences the level of trust (Calnan and Sanford 2004).

Conclusion

By analysing the concerns associated with WGS as well as the potential opportunities and challenges citizens perceive accompanying its implementation, we aimed at adding insights to the question of how to ensure that the implementation of WGS into the clinic is relevant and responsive to the needs of all members of society.

People have different perceptions of WGS and its imaginable integration into our health care system or our daily lives, and various approaches to decision-making regarding the potential use of WGS could be observed. The question of whether people want to know about their genetic information is not a new one but gains once again importance with a broader implementation or availability of WGS. Participants in our study emphasized aspects like personality type, mental constitution, social background, profession and attitude towards life as key factors for personal capacities to handle medical information. The preference not to know about one’s own genetic make-up should, in this regard, not be equated with a rejection of the

technology and its use per se. But it shows the necessity to ensure that enough resources and expertise for patient support are in place before implementing WGS in the medical system. This might include technical solutions such as the implementation of systems that allow people to define areas or characteristics of genetic information they want to get or to be disclosed for their physician.

Although the participants in general favoured a person-centred care approach, we also found more radical future-oriented perspectives that include patients or citizens using non-traditional approaches and resources for engaging with their health. Thus, besides a continuing focus on the further implementation of person-centred care, health care institutions should also be prepared for self-interpretation efforts on the side of citizens and develop strategies and practices to implement them to meet the needs of patients with what we describe as a self-centred perspective.

Dealing with uncertainties has long been another topic of discussion in connection with medical genetics; this discussion has been given a new perspective by WGS. Transparent communication about the—at least at the moment—inherent uncertain character of WGS data in science and health communication and education efforts might support people in coping with uncertainty and in developing tolerance of ambiguity.

As this and other studies have shown, trust lies at the core of nearly every discussion on scientific innovation in the field of health care. Like with yin and yang, there is no trust without mistrust. It is important to accept and communicate that arguing mistrust is part of a functioning and “healthy” democratic society made up of active, competent and informed citizens. Thus, enhancing trust in general cannot be the main goal of a health care system. It should rather focus on applying a more differentiated view on distrust to develop specifically aligned strategies to meet areas or issues identified as problematic.

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Ethical approval No ethics approval was necessary for our study since we were not working with sensitive or patient-related data. All project

activities were carried out in compliance with ethical principles as set out, for instance, in the European Code of Conduct for Research Integrity. Participants of the focus groups signed an informed consent form for being included in the study and a confirmed consent form for publication as part of the study consent form.

Informed consent Informed consent was obtained from all individual participants included in the study.

References

- Abelson J, Niller FA, Giacomini M (2009) What does it mean to trust a health system? A qualitative study of Canadian health care values. *Health Policy* 91(1):63–70. <https://doi.org/10.1016/j.healthpol.2008.11.006>
- Argyris C, Schön D (1996) *Organizational learning II: theory, method and practice*. Addison-Wesley, Reading, MA
- Beck U (1992) *Risk society*. Sage, London
- Blumer H (1954) What is wrong with social theory? *Am Sociol Rev* 18: 3–10. <https://doi.org/10.2307/2088165>
- Bohnsack R (2003) *Rekonstruktive Sozialforschung./ Einführung in qualitative Methoden*. VS Verlag für Sozialwissenschaften, Wiesbaden
- Borri P, Bentzen HB, Budin-Ljøsne I, Cornel MC, Howard HC, Feeney O, Jackson L (2018) The challenges of the expanded availability of genomic information: an agenda-setting paper. *J Commun Genet* 9: 103–116. <https://doi.org/10.1007/s12687-017-0331-7>
- Borzekowski DLG, Guan Y, Smith KC, Erby LH, Roter DL (2014) The Angelina Effect: immediate reach, grasp, and impact of going public. *Genet Med* 16(7):516–521. <https://doi.org/10.1038/gim.2013.181>
- Braun V, Clarke V (2006) Using thematic analysis in psychology. *Qual Res Psychol* 3(2):77–101. <https://doi.org/10.1191/1478088706qp0630a>
- Budin-Ljøsne I, Teare HJA, Kaye J et al (2017) Dynamic consent: a potential solution to some of the challenges of modern biomedical research. *BMC Med Ethics* 18:4. <https://doi.org/10.1186/s12910-016-0162-9>
- Bulmer M (1984) *Sociological Research Methods*. Macmillan, London
- Calnan MW, Sanford E (2004) Public trust in health care: the system or the doctor? *BMJ Qual Saf* 13(2):92–97. <https://doi.org/10.1136/qshc.2003.009001>
- Cirino AL, Lakdawala NK, McDonough B, Conner L, Adler D, Weinfeld M, O'Gara P, Rehm HL, Machini K, Lebo M, Blout C, Green RC, MacRae C, Seidman CE, Ho CY, MedSeq Project* (2017) A comparison of whole genome sequencing to multi-gene panel testing in hypertrophic cardiomyopathy patients. *Circ Cardiovasc Genet* 10(5):557–573. <https://doi.org/10.1161/CIRCGENETICS.117.001768>
- Clayman ML, Bylund CL, Chewning B, Makoul G (2016) The impact of patient participation in health decisions within medical encounters: a systematic review. *Med Decis Mak* 36:427–452. <https://doi.org/10.1177/0272989X15613530>
- Dibben M, Lena M (2003) Achieving compliance in chronic illness management: illustrations of trust relationships between physicians and nutrition clinic patients. *Health Risk Soc* 5(3):241–259. <https://doi.org/10.1080/13698570310001606950>
- Emanuel J, Emanuel L (1992) Four models of the physician-patient relationship. *JAMA* 267(16):2221–2226. <https://doi.org/10.1001/jama.1992.03480160079038>
- Enralgo PL (1969) *Doctor and Patient*. McGraw-Hill Book Company, New York
- Epstein RM, Street RL (2011) The values and value of patient-centered care. *Ann Fam Med* 9(2):100–103. <https://doi.org/10.1370/afm.1239>
- Etchegary H (2014) Public attitudes toward genetic risk testing and its role in healthcare. *Pers Med* 11(5):509–522. <https://doi.org/10.2217/pme.14.35>
- Fiske A, Buys A, Prainsack B (2019) Health information counselors: a new profession for the age of big data. *Acad Med* 94(1):37–41. <https://doi.org/10.1097/ACM.0000000000002395>
- Frank L, Forsythe LP, Ellis LE, Schrandt S, Sheridan S, Gerson J, Konopka K, Daugherty SE (2014) Conceptual and practical foundations of patient engagement in research at the patient-centered outcomes research institute. *Qual Life Res* 24(5):1033–1041. <https://doi.org/10.1007/s11136-014-0893-3>
- Freidson E (1970) *Profession of medicine: a study of the sociology of applied knowledge*. Dodd, Mead, New York
- Gaskell G, Stares S, Allansdottir A et al (2010) *Europeans and biotechnology in 2010 - Winds of change?* Publications Office of the European Union, Luxembourg. <https://doi.org/10.2777/23393>
- Genetti CA, Schwartz TS, Robinson JO et al (2018) Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. *Genet Med* 21:622–630. <https://doi.org/10.1038/s41436-018-0105-6>
- Gibbons M, Limoges C, Nowotny H, Schwartzman S, Scott P, Trow M (1994) *The new production of knowledge: the dynamics of science and research in contemporary societies*. Sage, London
- Glaser B (1978) *Theoretical sensitivity. Advances in the methodology of grounded theory*. Sociology Press, Mill Valley
- Glaser BG (1992) *Emergence vs. forcing, basics of grounded theory analysis*. Sociology Press, Mill Valley
- Hall MA, Dugan E, Zheng B, Mishra AK (2001) Trust in physicians and medical institutions: what is it, can it be measured, and does it matter? *Milbank Q* 79(4):613–639. <https://doi.org/10.1111/1468-0009.00223>
- Holm IA, Agrawal PB, Ceyhan-Birsoy O et al (2018) The BabySeq project: implementing genomic sequencing in newborns. *BMC Pediatr* 18:225. <https://doi.org/10.1186/s12887-018-1200-1>
- Kelley JM, Kraft-Todd G, Schapira L, Kossowsky J, Riess H (2014) The influence of the patient-clinician relationship on healthcare outcomes: a systematic review and meta-analysis of randomized controlled trials. *PLoS One* 9(4):e94207. <https://doi.org/10.1371/journal.pone.0094207>
- Klein J (2004) Interdisciplinarity and complexity: an evolving relationship. *E:CO Spec Double Issue* 6(1–2):2–10. <https://doi.org/10.17357/5b032d0fd094281a75e3ff2f998d161>
- Kohler JN, Turbitt E, Biesecker BB (2017) Personal utility in genomic testing: a systematic literature review. *Eur J Hum Genet* 25(6):662. <https://doi.org/10.1038/ejhg.2017.10>
- Krier JB, Kalia SS, Green RC (2016) Genomic sequencing in clinical practice: applications, challenges, and opportunities. *Dialogues Clin Neurosci* 18(3):299–312
- Lazer D, Brewer D, Christakis N, Fowler J, King G (2009) Life in the network: the coming age of computational social. *Science* 323(5915):721–723. <https://doi.org/10.1126/science.1167742>
- Levi-Strauss C (1966) *The savage mind*. University of Chicago Press, Chicago
- Lewis KL, Han PKJ, Hooker GW, Klein WMP, Biesecker LG, Biesecker BB (2015) Characterizing participants in the ClinSeq genome sequencing cohort as early adopters of a new health technology. *PLoS One* 10(7):e0132690. <https://doi.org/10.1371/journal.pone.0132690>
- Majumder MA, Cook-Deegan R, McGuire AL (2016) Beyond our borders? Public resistance to global genomic data sharing. *PLoS Biol* 14(1):e2000206. <https://doi.org/10.1371/journal.pbio.2000206>
- Newson AJ, Leonard SJ, Hall A, Gaff CL (2016) Known unknowns: building an ethics of uncertainty into genomic medicine. *BMC Med Genet* 9(57):1–8. <https://doi.org/10.1186/s12920-016-0219-0>

- Nowotny H, Scott P, Gibbons M (2001) *Re-thinking science: knowledge and the public in an age of uncertainty*. Polity, Cambridge
- Prainsack B (2017) *Personalized medicine: empowered patients in the 21st century?* NYU Press, New York
- Renn O (2008) *Risk governance: coping with uncertainty in a complex world*. Earthscan, London
- Rogers CR (1961) *On becoming a person: a therapist's view of psychotherapy*. Houghton Mifflin, Boston
- Rowe R, Calnan M (2006) Trust relations in health care—the new agenda. *Eur J Public Health* 16(1):4–6. <https://doi.org/10.1093/eurpub/ckl004>
- Scholz RW (2000) Mutual learning as a basic principle of transdisciplinarity. In: Scholz RW, Häberli R, Bill A, Welte M (eds) *Transdisciplinarity: joint problem-solving among science, technology and society*. Proceedings of the International Transdisciplinarity 2000 Conference. Workbook II: Mutual Learning Sessions. Haffman, Zürich, pp 13–17
- Stirling A, Glover D, Millstone E (2015) Regulating genetic engineering: the limits and politics of knowledge. *Issues Sci Technol* 31(4):23–26
- Sztompka P (1999) *Trust: a sociological theory*. Cambridge University Press, Cambridge
- Taber JM, Klein WMP, Ferrer RA et al (2015) Perceived ambiguity as a barrier to intentions to learn genome sequencing results. *J Behav Med* 38:715–726. <https://doi.org/10.1007/s10865-015-9642-5>
- Vassy JL, Lautenbach DM, McLaughlin HM et al (2014) The MedSeq Project: a randomized trial of integrating whole genome sequencing into clinical medicine. *Trials* 15:85. <https://doi.org/10.1186/1745-6215-15-85>
- Vrijenhoek T, Kraaijeveld K, Elferink M et al (2015) Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. *Eur J Hum Genet* 23: 1142–1150. <https://doi.org/10.1038/ejhg.2014.279>
- Woolley JP, Kirby E, Leslie J et al (2018) Responsible sharing of biomedical data and biospecimens via the "Automatable Discovery and Access Matrix" (ADA-M). *NPJ Genomic Med* 3(1):17. <https://doi.org/10.1038/s41525-018-0057-4>
- Zuiderent-Jerak T (2007) Preventing implementation: exploring interventions with standardization in healthcare. *Sci Cult* 16:311–329. <https://doi.org/10.1080/09505430701568719>

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