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The lived genome

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Abstract

For medical professionals, the genome is primarily a source of health information that can be used for diagnoses and disease risk assessment; in some cases, gene therapy is an option. For scientists, the genome is the DNA, which can be sequenced and used to explain heredity and individual development. But what is the genome for those who have it and live it? A systematic exploration of the lifeworld from a first person perspective, while focusing on practices of personal meaning-making, can help to answer this question.
This chapter explains how. How can the genome be investigated if it is not just a scientifico-medical thing but an interpreted part of the lived body? What is the genome from the perspective of those living a body, this ‘lived genome’? The genome is also addressed in a first-person-perspective. Here we see the necessity of critical contributions from phenomenology of the body to a critical understanding of genetic implications of all genetically related diseases. Examples will be taken from a project on chronic inflammatory bowel diseases where currently a ‘geneticisation’ is going on. How do patients and their families make sense of their genomes?

From a medical perspective, the genome can today primarily be used as a source of health information for diagnoses and prospective disease risk management. Gene therapy may be an option in the future. For scientists, the genome is the sum of an organism’s DNA molecules, which can be sequenced and used to explain heredity and development. What is a genome for those who have it in their bodies and who *live* it? How do they make sense of it? What meanings are associated with the genome in their lifeworlds, where identities are formed and decisions taken in personal, family and cultural contexts? It is a matter of perspectives. We all live a genome, but the questions that arise as people who *live* a genome are different from those raised by doctors and scientists who look at the genome as a functional part in cells. In the perspective of their own embodiment people act as interpreters of their own ‘lived’ genome, of both its knowns and its unknowns.

Studying these acts of interpretation is an emerging area within the interdisciplinary

field of Medical Humanities, combining qualitative research approaches, empirical ethics, philosophy and cultural studies. It could be regarded as 'critical' in the sense of aiming to defend the first-person perspective as a space of active interpretative work, against an exclusively third-person biomedical view. Lay people are considered experts in their lifeworlds and they are 'moral pioneers', as anthropologist Rayna Rapp[1] has put it in her study of the moral dilemmas of prenatal testing. In a broader sense, they are pioneers of sense-making in the course of a geneticisation of body knowledge.

We can specify two levels of questions that need to be raised in studies of the 'lived genome'. The first of these relates to how information about genes and mutations affects the self and the identity of individuals and families, for instance those with monogenic conditions such as hereditary cancer risks, Huntington's disease or cystic fibrosis. How do people communicate genetic risks to each other? How do they decide whether or not to know their genetic status? How do they narrate predictive genetics in regard to a specific condition?[2] How do they individualize the probabilities? The second level is the framework of more general genomic information, which is developed in genomic medicine and biology. This provides background concepts to interpret what becomes accessible individually through single nucleotide polymorphisms (SNP) based personal genome scans,[3] or whole genome sequencing.

The genome, which is investigated by the burgeoning fields of molecular biology and genetic medicine in its amazingly complex functions, is a part of our bodies – a part of which humans were ignorant before the advent of modern genetics. Being invisible and insensible, (other than arms, legs or muscles), the genome is not naturally part of what phenomenologists have described as the body scheme, i.e. within the work of Shaun Gallagher.[4] But nevertheless it is creatively imagined and integrated in a culturally and individually negotiated and narrated corporeal identity, within diverse local accounts of intersubjective relatedness. As such it is part of the lifeworld. By systematically exploring the lifeworld from a first-person perspective of those living and shaping it, and by focusing on practices of personal meaning-making, phenomenology and qualitative research can approach the question of what a genome *is* by asking : What does it mean to live as somebody with this genome?

In the first section of this chapter we introduce our theoretical perspective more

broadly by distinguishing between two perspectives on the genome, which appears as the 'biomedical genome' and the 'lived genome'. We will also introduce the way in which we are mobilising the concept of the 'lifeworld' in the context of our research. The genome is something peculiar if conceived from a first-person perspective. Those living a genome do not only *have* it and get the information about the body they *are* but they do many things in order to make sense of their genome. There is interpretative work done in social and practical contexts, we propose to speak of 'reflexive embodiment'. Reflexive embodiment is an active process rather than something in which people are just 'affected' by the implications of genomics. The second section is dedicated to the guiding questions and methods for investigating this 'lived genome' and the social epistemology of genomics. In our final section we discuss examples from a study on Crohn's disease and ulcerative colitis, both inflammatory bowel diseases. These conditions were selected because while they have been treated as paradigmatic 'psychosomatic' diseases,,genetic factors have recently been found to be involved and these findings are currently transforming the medical understanding of the disease aetiology. The 'geneticisation' of these diseases can be observed in real time, affecting both medical practice and disease experience.

1. Two 'genomes'

Actively and passively, more and more people take part in cultures of genetic knowledge.[5] On many levels, they are immersed and involved in communication about genetics. People in industrialized countries are increasingly encouraged to make decisions about predictive or diagnostic genetic tests when starting a family; before[6], during and after pregnancy; and before, during and after illness. Medicalisation is followed by geneticisation. Foetal DNA testing is now accessible non-invasively and with minimal risk to the pregnancy through a few drops of mother's blood, with the result that more pregnancies may be turned into 'risk pregnancies'.[7] More and more tests are included in newborn screening. The cost of a complete personal genome sequence has been tumbling; currently, it has fallen below the magic sum of USD 1000. Large biobanks and information repositories have been established, such as the UK Biobank which holds data on more than half a million patients, or the 100'000 Genomes Project constituting unique research

infrastructures. The media provide ample (selective and always interpreted) information about the latest breakthroughs in genomic research. Increasingly large parts of whole patient populations are transformed into genomic study samples followed by systems medicine (see also the chapter by Will Viney in this volume).[8] The omnipresence of the availability of genetic knowledge has changed the cultural 'frames' for disease, health and responsibility and new private and public duties seem to emerge: a possible duty of the individual to know his or her own genes[9] and a possible duty of healthcare professionals to tell people about their genetic risks.[10]

Much has been written about potential changes in self-image, body schema, and the possible implications of genetic risks for individual visions of future life.[11] Now new, further questions concern the meanings of genomic knowledge for those who have to deal with it and integrate it, transform and translate it into their everyday lives. We need a better understanding, as Barbara Prainsack and Jenny Reardon have put it, of how

> 'whole-genome information is used by, and what it means to, a wide range of users. […] An understanding of what a broader range of users hope to learn from this type of whole-genome information, and whether it would lead to actual life and behaviour changes, would help in assessing whether personal-genomics services are likely to be adopted in large numbers.'[12]

This knowledge about the user's (an non-users') hopes, fears and subjective understandings in regard to genetic knowledge must be based on an adequate kind of evidence. Such evidence would be needed for planning a good governance of genomics. Questions such as: 'What does my genetic make-up mean for myself and for my family?' or: 'In what sense "am I my genes"?'[13] should therefore be occasions not only for theoretical speculation but also for empirical research, applying qualitative, hermeneutic and phenomenological methodologies. We need to study the ongoing 'reflexive embodiment'[14] of genetic knowledge.

In the first place a theoretical framework is needed that can integrate these processes of the individual translation and management of genetic information and its integration into personal lives. By explaining the genome as something that is both investigated and used in biomedical contexts but also 'lived' individually, in families and in societies, we hope to contribute to such a framework.

The reflexively embodied genome is thus charged with basically two sets of meanings that both differ and interact. One is the set of meanings that are attached to the genetic in biomedical research and in clinical contexts. For the sake of simplicity, we call this perspective, and what is seen in it, ‘genome 1’. The genome, however, is translated and transformed into a related but dramatically different figuration that we call here ‘genome 2’, which is the genome seen within the lifeworlds of concerned individuals.

Scientists who are socialised into the frames of genome 1 may think that their genome is the only true one while the people’s view on the genome is just a subjective translation. They may find many elements of genome 2 (in lay people’s understandings) imprecise, even incorrect. Their view on ‘reality’ is the sober world of mathematical models, of physics and chemistry, and of the complex charts of cellular systems with which they work. However, some users of genomic knowledge (other than scientists and healthcare professionals, who may themselves be personal users of genomic information) may equally well find genome 1 too abstract and lacking clear sense for the practical decisions they need to make.

We do not think that either genome 1 or genome 2 is necessarily simpler but rather that they are related to different complexities. Similarly we do not think that either genome 1 or genome 2 is wrong or biased but that they have different truth criteria. Furthermore, genome 1 is not only the raw material for an simplification, or application into genome 2. Both are valuable, and their interrelation is interesting to study. Both are concrete for people, and both are in some way necessary, however they carry different phenomenological features of concreteness. The process of reflexive embodiment can hence be seen as an activity of mutual translations between different meaning contexts.

A linear ‘deficit’ model of the popularization of scientific knowledge from medical experts to patients, which had been assumed for decades, has become largely obsolete within Science and Technology Studies.[15] It is certainly not helpful for elucidating the process of reflexive embodiment of genetic knowledge. Both sides have advantages and deficits, and both sides need to tell each other what they know and how they know it, how their knowledge produces evidence. A deficit model does not allow the user perspective to be taken as seriously as the provider perspective, since users are considered to be at the receiving end of the communication

cascades. We prefer a model that assumes active contributions from both sides. The terminological symmetry between ‘genome 1’ and ‘genome 2’ should signify this. Meaning-making in the field of genetics and genomics is a joint enterprise between producers and users of genetic knowledge, between science and society. The meanings on the two sides, however intertwined, differ considerably – and sometimes they clash.

In the course of research and also in clinical practice, situations currently emerge where people typically need to decide about seeing large parts of their own genomic information. Genetic counselling normally deals with the complexities of genetic information about individual diseases or impairments. Now decisions need to be taken about knowing a whole genome, an exome, a wide range of SNPs. The people concerned can not know about all conditions they potentially will get predictive information about; too long lists of diseases may be potentially included. In such situations the capacity for voluntary decision-making about the disclosure of information is seriously challenged. This is not unique but intrinsically related to whole genome studies: too many heterogeneous possibilities that are virtually impossible to grasp, and therefore a ‘mission impossible’ for genetic counselling. The meaning of potential knowledge (in both the genome 1 and 2 perspectives) is tightly bound to the concrete conditions. Genetic risks for breast cancer pose different questions for decision-making than the risk for macular degeneration, or for Alzheimer’s.[16] The genome, as an object for informed decision-making about comprehensive disclosure, is therefore a moral conundrum. Decisions are virtually impossible to take ‘voluntarily’, in the classical sense of properly understanding all relevant implications of a decision.[17] In order to make better-informed decisions, ideally, decision-makers would need to understand the lifeworld meaning of the genetics of each of the conditions which are potentially covered by testing. If somebody is uneasy with just an a priori decision to be as informed as possible about all risks to health and asks for more detailed knowledge, this would need to include both genome 1 and 2 perspectives on all conditions possibly involved.

While genome 1 is actually studied condition by condition – this is the aim of all big research programmes in current systems medicine, genome 2 knowledge is not yet gathered systematically. A similarly progressive condition-by-condition analysis of genome 2 would be needed.

Reflexive embodiment is a basic human capability, connecting the individual and the social. As social philosophers Margaret Archer and Nick Crossley explain, it is undertaken through dialogues, internal conversations and conversations with others.[18] The individual person is always woven into social contexts and their self-image can be perpetually re-configured. The question of what we 'embody' is in the foreground here. The body has inherited older key terms of existence. Issues of genetics are issues of corporeality.[19] We can ask: How and on what levels do we internalize, process and reflect the highly complex and partially uncertain knowledge about our own genome when it is disclosed to us (and potentially to others) and interpreted by geneticists? How do we translate their interpretations into our own identities, into lifeworlds, when we, as culturally and socially embedded people, are confronted, so to speak, with our biochemical self? A special feature of genomic knowledge *is* that it is about our bodies and our selves. By definition it has 'anthropological' significance,[20] but this significance is not explicitly contained in the content of biomedical information. In order to understand this anthropological significance we need to explore and 'decode' genome 2 as well.

The first human genome sequence was completed by 2003, and, perhaps paradoxically, during the same historical period the biological role of the genome was contested and re-interpreted. These semantic transformations that took place for genome 1 are also important for genome 2. In more detail: The old conception of the genome as the 'genetic program' was devised by eminent biologists around 1960,[21] long before research in developmental molecular genetics had begun and details of the developmental functioning of genes were discovered. However, in the context of systems biology the genetic program view has lost its theoretical plausibility, while still being socially active in the context of the genome 2. It has now been replaced or complemented by less gene-centric, deterministic and essentialist images, such as the molecular orchestra of the cell, whose music (the life of the cell) is produced by an interplay of diverse acting components, of which the genome is one part; or a library, whose actualized information content depends on those who use it by checking out and creatively reading a selection of books.[22] Hence, the biomedical significance of the genome, while being significantly expanded, is also undergoing critical transformation.

We suggest using and developing 'genome 1' – the biomedical genome – and 'genome 2' – the lived genome – analytically, as two hermeneutic perspectives. They

represent two different but interrelated interpretative contexts of the genome, and at the same time two different levels of interpretation. Genome 2 is seen in lifeworld contexts by those who know the bio-psycho-social implications of genetic susceptibilities and diseases at first hand, that is, by people 'living' a condition, having had or not having had a test, being directly involved as a patient or indirectly by being a member of an affected family. Also healthcare professionals are (in part) concerned with genome 2. In their professional work, which combines the biomedical and the patient centred views, they are crossing the interface between the two perspectives. Genome 1 contains all testable genetic variations, SNPs, sequences, and genomic data, together with the corresponding medical interpretation given by doctors, scientists and genetic counsellors. It includes explanations of genetics risks, of inheritable factors etc., and explanations of the functioning of the genome and its variations in the cellular metabolism. Genome 2 is the genome in the understanding of those who embody the genome, who 'live' it, who are affected by it, who narrate it and understand their relationships to others by using elements of genetic knowledge, who make life plans accordingly (choosing a partner, planning a family and so on). The genome is imagined and continuously re-conceptualized in the lifeworld of those individuals and families who live the genome. This 'lived genome' interprets[23] the biomedical construct of a physico-chemical entity that is called 'genome', which – in contrast to other parts of the body like the beating heart – is not accessible to direct experience. It thereby integrates culturally mediated symbols and metaphors of genetics (such as the genome as a 'language', a 'text', a 'program', a 'mosaic' and the like) and combines them with personal understandings into a partially comprehensible and partially mysterious text.

Edmund Husserl introduced the term 'lifeworld' in *The Crisis of European Sciences and Transcendental Phenomenology* of 1936.[24] The lifeworld exists in contrast to the objective knowledge of the physical sciences, and works as a collective inter-subjective pool of perceiving. It acts as a foundation for the scientific perception of reality, in which an idealized (mathematically abstract) 'nature' of scientific theory, in the perception of members of society, replaces the pre-scientific and concrete nature. As Husserl claimed, this process of implanting another reality started historically with Galileo, who successfully invented the method of geometrical idealization in physical research and believed, as he famously put it, that the book of nature is written in mathematical language – which is an ontological claim. This concept of the lifeworld

as a pre-scientific world was then expanded and developed by sociologists of knowledge such as Alfred Schütz and Thomas Luckmann.[25] They speak of reserves of knowledge that have certain structures and are composed of different types of knowledge with different levels of relevance. In order to make this concept useful for a social epistemology of genetic knowledge, the lifeworld needs to be understood in such a way that it can incorporate scientific knowledge as well. That is what we meant when we spoke of the genome 1 and genome 2 as 'interrelated'. But scientific knowledge is not incorporated into genome 2 in its 'raw' form, as produced by science and medicine. It is transformed and translated in order to meet the needs of everyday life. How this is done is an interesting empirical question. We need to ask the people who do this translation, since they are the experts on their lifeworld genetic knowledge.[26] This investigation needs to be sensitive to a transformation of the 'gaze' that is implied in the geneticisation of disease experiences. The gaze of the Galilean type[27] sees the limits on embodied human existence as issues for technical improvement, for the management of risks and the maximization of wellbeing, while a lifeworld 'gaze' would include a broader repertoire of meanings.

2. Research perspectives: methods for studying the lived genome

In order to study the lived genome we need to look at the processes by which people make sense of genomic information in lifeworld contexts. Geneticisation, a term introduced by Lippman in 1991, 'refers to an ongoing process by which differences between individuals are reduced to the DNA codes, with most disorders, behaviours and physiological variations defined, at least in part, as genetic in origin.'[28] Applying this to the study of genome 2 would mean monitoring which differences between individuals are actually re-defined as genetic, which aspects are lost by this reduction, and to what degree these variations are geneticised.

In order to understand, however, the ways in which genetic explanations are also seen as positively *meaningful*, we need to expand the concept of geneticisation. Instead of expecting genetic explanations to be reductionist in the first place, involving a loss of value and meaning, we first ask which questions are answered by genetic explanations. To what questions genetic information is providing answers? What are the hopes and fears that people direct towards genetic explanations, even before they know them? A lived genome studies poses questions such as: What kind

of evidence is produced by particular genetic explanations? In which context? By whom and for whom? What questions are hidden or silenced in social processes of ‘genetic reduction’? What is the benefit of a genetic explanation of a condition? And what are the losses? How are genetic explanations creatively integrated into or complemented by non-genetic explanations?

These questions inspire the guiding research perspective of qualitative empirical studies in different ways. Here are a few examples: Monica Konrad[29] asked what it actually means to be classified as a person with a predisposition (to Huntington’s disease), which local moralities guide the resolution of disclosure dilemmas, and how the life sciences create ‘pre-symptomatic persons’ as new forms of social identities. Scully, Porz and Rehmann-Sutter[30] asked how time functions as a factor in genetic decision-making. Britta Pelters[31] organized her study of breast cancer patients ‘doing health’ around the question of how genetic diseases are socialized and which roles the disease adopts in families. Martin Richards[32] made an autobiographical ethnography describing his use of personal genome scans from two direct-to-consumer test companies. Robert Klitzman[33] took a comparative approach and studied the testing and disclosure decisions of symptomatic and non-symptomatic individuals with genetic predispositions for Huntington’s disease, alpha-1 antitrypsin deficiency, and breast cancer. Andrea zur Nieden[34], in her recent study of the subjectification of breast cancer, took an approach via narrative identity, asking how the interviewees position themselves in the discourse as a particular type of personality.

Since meanings need to be explored openly and testable hypotheses are rare, qualitative approaches are the methods of choice for studying the ‘lived genome’. Genes carry meanings within the lifeworlds of persons who experience the biological, psychological and social implications of genetic diseases – whether as affected individuals, family members, scientists or health professionals. These meanings need to be captured and understood. In narratives (which can be collected in interviews, focus groups, participant observation, or documents) the intricacies of individual meaning-making and the individual interpretation of genomic knowledge (and other -omics data) can be explored in depth. By observing situations of communication we can explore how different explanations and coping approaches interact with each other. Breaks can occur within and between individual epistemologies; tensions can be expected, where new understanding emerges. A ‘lived genome’ study should

centre on the views and experiences of individuals, compare their local 'sense-making strategies' with those of others, and analyse both the narratives and (if a study includes focus groups or participant observations) the interactions.

For the interpretation of the data inductive research strategies can be used, such as Grounded Theory (GT)[35] or Interpretative Phenomenological Analysis (IPA)[36]. The IPA perspective sensitizes the researcher to the double hermeneutics of data analysis: the participants are searching for sense within their own experiences about life with a genetic disease or with the results of a genetic test, and in turn, the researcher is searching for sense within the narrated and transcribed sense-making strategies of the participants.[37] The researcher must reflect upon his or her preconceptions and pre-understandings of the data and the topics covered.[38] The interpreter should try adjust her/his own preconceptions as part of the hermeneutical process.[39] This methodology is not meant to be exclusive; it can be complemented by methods from cultural studies, history, literary analysis, or philosophical phenomenology.

3. Workshop: Embodiment and socialization of genetics of chronic inflammatory bowel diseases

We are currently investigating the 'lived genome' in a project about people living with chronic inflammatory bowel diseases, such as Crohn's Disease and Ulcerative colitis, in Germany.[40] Within our understanding of the aetiology of these diseases there is a shift taking place – from a psychosomatic paradigm to one of genetic susceptibility. Our working hypothesis is that this ongoing 'geneticisation' has practical significance in patients' and their families' lives, and that it is possible to investigate how the patients embody, socialize and reshape genetic knowledge in their own ways.

Currently there are 69 persons participating in our study. The sample spread over all regions of Germany and all layers of society. It includes 37 cases of Crohn's disease, 14 cases of Ulcerative colitis and 18 relatives and friends of concerned persons. The majority of the participants are women who are suffering from one of the two diseases. Most of the participating men are spouses or close friends.[41] Yet we have conducted 40 semi-structured narrative interviews with patients and members of their families. Most of our study participants were recruited through a self-help

organization, the ‘Deutsche Morbus Crohn/Colitis ulcerosa Vereinigung (DCCV)’. They were interviewed by the second author of this chapter. The interviews were recorded and transcribed verbatim. Pseudonyms were chosen by the researchers. In the course of the interviews, we invite the participants to talk freely about their life history with respect to their disease. This helps us to obtain an insight into their own understanding of their lifeworlds and their experiences with their diseases. These narratives already contain independent interpretations, pictures of and attitudes towards genetics and heredity that are deeply intertwined with life circumstances in both individual and social terms. As soon as this background is deployed we invite the participants to talk about their attitudes, expectations and individual experiences of the genetic explanation of Crohn’s or colitis.

As described above, we use a mixed methodology for the analysis of the obtained material: An alloy of grounded theory strategies and interpretative phenomenological analysis. This mixture helps us to analyse the sense-making processes of our interlocutors within the rich context of their experiential worlds. Thereby we search for recurring patterns on the level of the narratives and compare them to those of other participants. Unlike Robert Klitzman, Andrea zur Nieden, Britta Pelters or even Monica Konrad we try to avoid an early psychologization in this process and take our material serious on the structural and narrative level. This kind of analysis revealed two basic narratives of coping with the (partially conflicting) genetic and psychosomatic explanations of the disease: A narrative which centres guilt and shame and a narrative that centers individual agency. In the following lines we will sketch out and illustrate them with some translated quotes from interviews.

Narrative 1: Guilt. A first group of narratives that participants gave oscillated about experiencing guilt. Both patients and relatives described guilt as a constantly present and sometimes overwhelming emotion. Crohn’s or colitis, which sometimes causes ‘terrible, debilitating diarrhoea’,[42] can severely affect everyday family life. Especially during sporadic exacerbations, phases of adverse food reactions or in the context of a difficult progression of the disease, many things become extremely difficult or almost impossible to plan: periods abroad for study or vacations, dinners with friends, birthday celebrations, professional appointments, or even visits to the hairdresser. For our participants, these issues were frequently associated with apologies,

remorse, and the implicit sense of disappointing somebody.[43] Ulrike, a 45-year-old Reiki master, reported that she sometimes feels guilty about poorly schedulable events – particularly in connection with her adult daughter.[44]:

> ‘I was off on a Reiki weekend with her – something we’ve done together for years – and I had eaten salad two days in a row. What I did not realize was that it was just the moment where I could not take it, and the scars in my gut worsened the situation. That means for a start that it was all closed off. And then I threw up all night, and I recognized and saw the signs […]. [T]hen I thought: “That poor girl, now she’s here alone with me, there’s no one else here to support her as well.” That was a situation where I have realized, very, very intensely, what a disease like this means or could mean for my children as well […].’[45]

Here, guilt is also felt because of the possibility that her daughter has inherited the disease susceptibility.

A second significant dimension of guilt is often imposed from the social environment and is verbalized as some kinds of accusation. Relatives, friends, colleagues, partners and sometimes even family doctors tell the patients they are exaggerating their condition, they should pull themselves together, or give themselves a treat to overcome phases of disease. In the eyes of many of our interview partners, such statements imply that they are not making enough effort to improve their condition, that they are using their illness to evade responsibility or exaggerate at least parts of their condition.[46] The disease experience is interpreted as a mental health issue.[47]

These negative reactions in the social environment are associated by many interviewees with psychosomatic explanatory models. Thus, those models are perceived rather negatively, whereas other explanations such as a genetic aetiology are perceived as a relief. In the words of 46-year-old Ute from Karlsruhe:

> ‘I did understand that there is this argument that there is a barrier defect [a microbiomic explanation of CIBDs which is compatible with genetic explanations], and I found this a very great relief, and I said: “Look, this explains my rheumatism as well. That simply explains the whole situation […].” And for me that was for a start a very great relief and an unburdening,

> because it is an incredible psychological pressure when you have the feeling that it's somehow your fault, I mean, if you put it on a psychosomatic track.'[48]

Even the saying 'a healthy mind in a healthy body', which is used in everyday life as well as in theories of illness, is hurtful in this context because it can be contradicted by lived experience. For example, Mechthild, a women in her late 50s, who is involved in various self-help networks, states:

> 'When I was a representative [of the DCCV] and doctors tried to show [other patients] that if they treat it properly they'll be as well as I am, then I always objected vehemently and said: "I think you're blaming us if you use me as a positive example, because I know many [other patients] who would like to live as well as I do, and I experience them as life-affirming as well. I mean, they're similar to me but they're still very ill." So I've always been very much against it, if they tried to push us into this "all-in-the-mind category", because I felt they [the doctors] wanted to abdicate responsibility, I mean, they didn't want to refer us to gastroenterologists as helpers, but to the psycho-people [psychotherapists].'[49]

The participant speaks about experiencing the way that health and personal attitude are often brought into a reciprocal confirmation context, by medical professionals – and she was used for this purpose in her function as the representative of a patient organization. Her observation emphasised, however, that people can have severe diseases despite having a similarly positive attitude as she herself did. This made her resistant to such causal explanations based on a psychosomatic model, because she recognizes it as a practice of blaming patients for their condition. Moreover, she interprets the references to psychosomatics and the corresponding therapies as a way of physicians avoiding responsibility. Central to these statements is Mechthild's perception of psychosomatic medicine as something that transfers the blame for the disease to the patient – which is challenged by her everyday experience. Such positions arise frequently in the broad and prolonged experience of chronic inflammatory bowel diseases.

Patients may become a part of the medical history of their own diseases – as some of the participants told us. Personal biographical experiences and epistemologies can connect and combine so that they turn into medical explanatory models. One example of this is the story of the 65-year-old retiree, Agathe. She first suffered from

severe diarrhoea and cramps in the 1970s. After a few weeks she consulted her family doctor and was diagnosed with ulcerative colitis – considered a psychosomatic disease within the prevailing medical paradigm of that time.[50] The doctor told her, as she recollects, to "[…] have some psychotherapy to feel healthy again soon".[51] But that did not work well and her psychotherapist sent her for further diagnoses to Lübeck – then a leading location for psychosomatic research on chronic inflammatory bowel diseases. There, she gained more experience of the psychosomatic model and was given encouragement within that framework. Although the psychosomatic explanation worked well in therapy, it was repeatedly used by 'the outside' (i.e. her social environment) as some kind of 'my fault theory'.[52] She felt the rise of genetic explanations in the 2000s to be a great relief, especially since it coincided with lived experiences within her family. She states:

> 'Well, my grandfather probably also had Crohn's disease. He was in hospital 100 000 times because of intestinal obstructions and of his four children, three of them had a chronic inflammatory bowel disease […]. So my aunt died of it during the war at the age of 19. She quite simply starved to death. [2] And my [1] father died after an operation on an abscess. It wasn't yet fully diagnosed, but he had an abscess in his bowel and he died after the operation and it's very likely that that was also something similar […].[53] Well, since I'd somehow been thinking for over 30 years that the predisposition is inherited, it [the genetic explanation of some aspects of CIBD] wasn't particularly sensational for me. Well, I just accepted it. [2] […] But I think it was a relief. It was a relief to know. Above all [1], because of this psychosomatic theory, I mean, for many people this is a "my-fault theory" of the type, "You just have to try hard enough and you'll be well again." I mean, that's actually not very funny'.[54]

Two aspects about these statements are particularly interesting. First, Agathe has pre-existing genealogical inheritance ideas, which in her eyes are only confirmed by the scientific molecular biological evidence. In other words, she creates her own concept of intrafamilial disease transmission (her own genome 2) and takes it as relevant experiential knowledge before genome 1 (the scientific and biomedical concept) became important in the explanation of CIBD in her life. Second, Agathe's statements exemplify something that is characteristic for all participants in our study who interpret their disease experience in the context of guilt narratives. She

welcomes the rise of genetic explanations and instrumentalizes them as a kind of tool with which to rebut the perceptions of the disease in her social environment. Through this, genes gain lifeworld significance as a defence against the attribution of irresponsibility (and individual guilt feelings resulting from this) by their relatives, friends, colleagues etc.[55]

Narrative 2: Agency. Other participants interpret the geneticization of chronic inflammatory bowel diseases through the use of a complementary narrative – mostly against the background of a social environment that turns more towards lived experience. Although they also experience occasional guilt about the impact of the disease on their relatives, they manage to use it positively by converting it into agency. This works particularly when they receive positive feedback or prove to be resilient against guilt narratives.[56] These participants prefer psychosomatic explanations over genetic models because they assume that they can deal with them more autonomously. A kind of agency that in their eyes is lost with genetic models.

A good example for this was provided by 45-year-old Sabine. In her view, self-responsibility is the key to living well while coping with chronic inflammatory bowel diseases, and the best way to achieve this is through various methods of psychotherapeutic treatment plus self-care. From the perspective of some affected persons like Sabine genetic explanations threaten this, because in her view the genetic explanation makes it easy for patients and their relatives to diffuse responsibility. Sabine explains:

> ‘I think general personal responsibility gets a very raw deal, and um, if our musicians [the scientists and doctors who advocate genetic explanations for CIBD] now start to sing its praises, um, then I’m, that’s my fault alone. So I would agree that, it’s perhaps ignorance, it’s definitely terribly frightening to say all at once that there’s something physically wrong and I’m responsible for it myself. […].’[57]

Against this background, the disease can be interpreted as a compass for or a companion in self-care. In the words of Sabine:

> ‘I am, um, very much convinced that Crohn’s is my buddy, that he’s saying inside me: “Now look, you’ve gone too far here.” Or, “hey, won’t you even stop […].”[58]

Sabine describes a way of dealing positively with the disease. But this agency is also interpreted as fragile and threatened by genetic explanations, particularly through the associated risk of loss of personal responsibility.

The two narratives presented above should give a brief impression of the complex exploration of genomic explanations through the eyes of concerned persons and also their social environment. They show how deeply scientific explanations and their interpretations can be interwoven with lifeworld experiences and their respective epistemologies: Genetic probabilities get interlaced with individual feelings about guilt or shame; Genetic explanatory models become conceptualised as a protective shield against blame by the patient's environment etc. They also show that the interpretations of genetic information in genome 2 cannot be separated from illness narratives and concrete disease experiences. The narratives of Ulrike, Ute, Mechthild, Agathe, Sabine and other participants in our study indicate that patients produce a certain kind knowledge which must be seen and integrated as a relevant element for the conceptualisation of medico-scientific concepts.

4. Conclusion

Genomics is about human bodies, as conceptualized by the life sciences. Those who are not professional geneticists are lay people in this respect. However, the lay side is nevertheless very active in interpreting and making sense of genomic information. Lay people are experts in their reflexive embodiment, which is interpretative work on their *lived* body. To study the genome as it appears in lifeworld contexts means to appreciate patients’ and families’ knowledge of the diseases. It contributes to a fuller understanding of genomic information. The reflexive embodiment of genomics is a cultural project. To understand genomic information is a real ‘translation’ (not an ‘application’) of information in biomedical frameworks into information in the frameworks of individual and social lives.

Endnotes

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[2] M. Konrad, *Narrating the New Predictive Genetics. Ethics, Ethnography and Science* (Cambridge: Cambridge University Press, 2005).

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[11] Still relevant is the collection edited by Marteau and Richards: T. Marteau and M. Richards, *The Troubled Helix: Social and Psychological Implications of the New Human Genetics* (Cambridge: Cambridge University Press, 1996).

[12] B. Prainsack and J. Reardon, 'Misdirected precaution', *Nature* 456, 2008, pp. 34-35.

[13] R. L. Klitzman, *Am I My Genes? Confronting Fate and Family Secrets in the Age of Genetic Testing* (Oxford: Oxford University Press, 2012).; C. F. Cranor (ed.), *Are Genes Us? The Social Consequences of the New Genetics* (New Brunswick: Rutgers University Press, 1994).

[14] This term has been introduced by N. Crossley. See: N. Crossley, *Reflexive Embodiment in Contemporary Society* (Maidenhead: Open University Press, 2006).

[15] G. M. Sinatra, D. Kienhues and B. K. Hofer, 'Addressing challenges to public understanding of science: Epistemic cognition, motivated reasoning and conceptual change', *Educational Psychologist* 49, 2014, pp. 123-138.; B. Nerlich, R. Elliott and B. Larson (eds), *Communicating Biological Sciences*. *Ethical and Metaphorical Dimensions* (Farnham: Ashgate, 2009).

[16] Robert L. Klitzman (*Am I My Genes?*) studied the local understanding of genetic information about Huntington's disease, Alpha-1 antitrypsin deficiency and breast cancer by interviewing individuals who did and did not have symptoms, and had and had not had genetic testing.

[17] R. R. Faden and T. L. Beauchamp, *A History and Theory of Informed Consent* (Oxford: Oxford University Press, 1986).; C. Rehmann-Sutter, 'Das ganze Genom. Ethische Überlegungen zur vollständigen Sequenzierung der individuellen 'NA', in C. R. Bartram, M. Bobbert, D. Dölling, T. Fuchs, G. Schwarzkopf and K. Tanner (eds), *Der (un)durchsichtige Mensch* (Heidelberg: Universitätsverlag, 2012), pp. 255-278.

[18] We refer to the work of social philosophers Margaret S. Archer (2007) and Nick Crossley (2006) who explore how humans develop their identities, aside from classical dualisms. They investigate the internal dialogue about experiences and new bodies of knowledge in the context of everyday practices and the journey through life.

[19] M. S. Archer, *Making our Way through the World. Human Reflexivity and Social Mobility* (Cambridge: Cambridge University Press, 2007), pp. 314-325; cf. N. Rose, 'Molecular biopolitics, somatic ethics and the spirit of biocapital', *Social Theory & Health* 5, 2007, pp. 3-29.

[20] R. P. George, 'Ethics, politics, and genetic knowledge', *Social Research* 73, 2006, pp. 1029-1032.; cf. C. Rehmann-Sutter, 'Genetics, a practical anthropology', in M. Düwell, C. Rehmann-Sutter and D. Mieth (eds), *The Contingent Nature of Life. Bioethics and the Limits of Human Existence* (Dordrecht: Springer, 2008), pp. 37-52.

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(Durham, N.C.: Duke University Press, 2006).; K. Schmidt, *Was sind Gene nicht? Über die Grenzen des biologischen Essentialismus* (Bielefeld: transcript, 2014).

[23] Cf. the 'subject body' and the 'object body' in P. Ricoeur, *Freedom and Nature. The Voluntary and the Involuntary*, translated by E. V. Kohák (Evanston, Ill.: Northwestern University Press, 1966), p. 10.

[24] E. Husserl, *Die Krisis der europäischen Wissenschaften und die transzendentale Phänomenologie*, edited by E. Ströker (Hamburg: Meiner, 2012).

[25] A. Schütz and T. Luckmann, *Strukturen der Lebenswelt*, 2 volumes (Frankfurt am Main: Suhrkamp, 1979 and 1984).

[26] Felicity Callard's medical humanities article on ‚gene talk' and schizophrenia would be a useful reference here

http://www.palgrave-journals.com/biosoc/journal/v7/n3/full/biosoc201212a.html

[27] The contrast of two 'gazes' refers to Blumenberg's ‚Lebenswelt und Technisierung unter Aspekten der Phänomenologie' in his *Wirklichkeiten in denen wir leben* (Stuttgart: Reclam, 1981), pp. 7-54.

[28] A. Lippman, 'Prenatal genetic testing and screening: constructing needs and reinforcing inequalities', *American Journal of Law and Medicine* 17, 1991, p. 19.

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[32] Richards, *Reading the runes.*

[33] Klitzman, *Am I My Genes?*

[34] A. zur Nieden, *Zum Subjekt der Gene werden. Subjektivierungsweisen im Zeichen der Genetisierung von Brustkrebst* (Bielefeld: Transcript, 2013).

[35] Cf. J. Corbin and A. Strauss, 'Grounded Theory research: Procedures, canons, and evaluative criteria,' *Qualitative Sociology* 13, 1990, pp. 3-21, pp. 3–21.; K. C. Charmaz, '"Discovering" chronic illness: using grounded theory', *Sociology of Science and Medicine* 30, 1990, pp. 1161-1172.; K. C. Charmaz, *Constructing Grounded Theory: A Practical Guide through Qualitative Analysis* (Thousand Oaks: Sage Publications, 2006).

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[37] Smith and Osborn, *Interpretative phenomenological analysis*, p. 53.

[38] D. Mahr, 'Matthias Flacius Illyricus (1520-1575). Eine erste Theorie der Hermeneutik im Zeitalter der Reformation', in S. Luther and R. Zimmermann (eds), *Studienbuch Hermeneutik. Bibelauslegung durch die Jahrhunderte als Lernfeld der Textinterpretation. Portraits – Modelle – Quellentexte* (Gütersloh: Gütersloher Verlagshaus, 2014), pp. 158-165.

[39] Smith and Osborn, *Interpretative phenomenological analysis*, p. 53.

[40] See our project homepage:

http://www.imgwf.uni-luebeck.de/livedgenome/introduction.html (last visited: 13th April, 2015)

[41] This conspicuous gender distribution will be a subject at a later stage of our project.

[42] Anonymous, 'A patient's perspective: My battle with Crohn's Disease', *American Journal of Gastroenterology* 102, 1 March 2007, pp. 482.

[43] Interview with Nina and Jens.

[44] All citied interview passages were translated from German into English. We tried to preserve the linguistic style of the participants as much as possible. Square brackets tagged with ellipsis indicate omitted content that is not relevant for the argumentation of this chapter. Ellipsis without square brackets indicate that the interview partner paused her or his story at this point for more than one second. Square brackets also sometimes contain explanatory comments by the authors.

[45] Interview with the participant Ulrike. Timestamp: 00:17:40-1.

[46] To counter such attributions and accusations some participants report that they come up with excuses for their condition, absence or delay. See for example: Interview with the participating couple Hendrik and Melanie. Timestamp: 00:16:32-8.

[47] Typically, the 34-year-old Linda was told here: ‘It’s clear you’re just doing too much!’ See for example: Interview with the participant Linda. Timestamp: 01:16:16-3.

[48] Interview with the participant Ute. Timestamp: 00:22:48-7.

[49] Interview with the participant Mechthild. Timestamp: 00:54:55-3.

[50] Interview with the participant Agathe. Timestamp: 00:13:20.

[51] Ibid. Timestamp: 00:13:20.

[52] Ibid. Timestamp: 00:18:15.

[53] Ibid. Timestamp: 00:21:46.

[54] Ibid. Timestamp: 00:34:34.

[55] Interview with the participant Ute. Timestamp: 00:22:00-0.

[56] Cf. Interview with the participant Sabine.

[57] Interview with the participant Sabine. Timestamp: 00:28:53.

[58] Ibid. Timestamp: 00:18:07.

Recommended further reading

R. P. George, ‘Ethics, politics, and genetic knowledge’, *Social Research* 73, 2006, pp. 1029-1032.

R. L. Klitzman, *Am I My Genes? Confronting Fate and Family Secrets in the Age of Genetic Testing* (Oxford: Oxford University Press, 2012).

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J. L. Scully, R. Porz and C. Rehmann-Sutter, ‘“You don’t make genetic test decisions from one day to the next” – Using time to preserve moral space’, *Bioethics* 21, 2007, pp. 208-217.

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