The Embodiment of Genetic Risk:

Women's Experiences of Genetic Testing for Breast Cancer Susceptibility Mutations

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Abstract

This paper tries to reconstruct if and how people who are medically defined as 'genetically at risk' actually embody this risk assessment and act accordingly by risk management. The specific case analysed by the use of qualitative interviews and discursive analysis was genetic testing for breast and ovarian cancer (BRCA), which informs women about a possibly inherited higher risk for these cancers. As possibilities for better prevention are few and disputed in the case of breast cancer, the problem arises of what women are to do with this kind of information. After outlining the discursive model that propagates a pro-active behaviour and a personal responsibility for one's health, connected to the ideal of 'informed decision', I will focus on the narratives of women who actually took the test. It will be shown that there are two different ways of dealing with the information. The two types of women will serve to show which contradictions and impositions are implied in the request to become the subject of one's genes and to manage one's health on the basis of a risk estimate. Also the model of 'informed decision' appears to be too simple.

Introduction

Talk of 'genetic risks' or 'gene tests for illnesses' is now omnipresent. Commercial providers such as the US company '23andMe' advertise a $399 offer to 'explore your genome', for finding out about the personal risk of developing Parkinson's disease or certain kinds of cancer, as well as the risk of 'alcohol dependence' (23andMe 2008). But institutionalised medicine has also established genetic tests as part of regular health care for such diseases as colon cancer or breast and ovarian cancer.

Drawing here on my fieldwork carried out in German breast cancer and genetic counselling centres, I want to concentrate on the latter case. This paper takes a closer look especially at the narratives of women who

have undergone the process of genetic counselling and testing. The intention is to reconstruct, if and how women actually embody genetic risk, how they define themselves as persons 'at risk'. This also means to question the model of 'informed decision' that is put forward in the informative material. In reconstructing how women take 'decisions', this model appears to be too simple.

Relying on population studies, medicine has already proposed earlier that 5 to 10% of breast cancer has a genetic component. In the mid-1990s, the BRCA-1 and BRCA-2 genes were decoded and carriers of certain mutations in those genes are said to be exposed to a higher risk for breast and ovarian cancer. (The numbers quoted vary in different publications: the risk for breast cancer is said to be between 45 and 80% compared to 10% in the average female population, and 10 to 60% for ovarian cancer compared to 1.2%.)

Women considered to be 'at risk' because of their family history of many cases of breast and / or ovarian cancer are encouraged to undergo genetic testing in combination with genetic and gynaecological consultation. During the time of my fieldwork, genetic tests were carried out in the course of a multi-centre clinical trial financed by the German Cancer Aid. About 2500 families were tested in the course of the trial. It has now become part of regular health care for so-called 'high-risk families' covered by national insurance.

Study design

In my research I analysed medical publications and guidelines, hand-outs of the clinical study and the genetic counselling sessions. In addition, I carried out in-depth interviews with genetic counsellors and other staff as well as with women who had undergone genetic testing. The interviews were held at least one year after they received the test result. Some had positive test results (mutation was found), some negative (mutation was found in the family, but they were non-carriers) and some indeterminate results ('unclassified variant' or no mutation in the family, which leaves the risk of having a mutation in another gene not yet found). None of the women had cancer themselves.

Before I focus on the stories of the women I want to outline briefly the procedure of the trial: Women go to one of the twelve specialised breast cancer centres through their own initiative or through referral by their gynaecologist. If a woman gets in touch with one of the clinical centres and meets the study criteria, she is invited to three consultations: a gynaecological, a psychological and a genetic one. The genetic counselling sessions inform women about the genetics of cancer in general and the BRCA genes in particular. They also suggest options if the result is positive: the options range from intensified screening (basically mammography and ultrasound) to taking part in other clinical trials with anti-hormones (Tamoxifen) to prophylactic surgery of breast and ovaries. Additionally, general life-style advices thought to be protective of cancer are given, e.g.: 'eat five portions of fruit and vegetables every day', 'do modest sports and avoid smoking', and after all: 'try to maintain the joy of life'. It is stressed that because all the options have (sometimes massive) side effects and none of the options will completely abolish the risk, the woman herself has to make the choice which is right for *her* – concerning the decision about taking a test at all, but also concerning the follow-up measures.

If a woman decides to take the test, a so-called index patient in the family who has already suffered from cancer must first be tested. Only if a mutation is found in that person may healthy women also be tested. In 50 % of the families tested in Germany, no mutation was found. Clinicians believe cancer could still be inherited within these families, as other genes not known at present may also be involved. Despite this the women involved mostly understand the result as 'negative'. In many other cases so-called 'unclassified variants' are found, meaning the particular mutation is not in the international databases and cannot be clearly identified as being cancerous or not.

Generally, the possibilities of genetic diagnosis outweigh therapeutic progress. In the case of BRCA testing, options are either not evaluated sufficiently or have massive side effects, like the prophylactic removal of those organs that are to be protected. This raises the question of how women deal with such diagnostic results that are *indeterminate* in a two-fold way: on the one hand, unlike symptomatic diagnostics, they do not

tell anything about the current status of the body, but about a possible future. On the other hand, there are no clear-cut therapeutic options following the diagnosis.

A short history of creating 'at risk' patients

The problem of this new tendency to create 'patients without symptoms' has been addressed by others previously (Feuerstein & Kollek 2001; Lemke 2004; Nelkin & Lindee 1995). Instead of healing, medicine tries to act preventively, which relocates interventions to an earlier point in time. This is to be seen in the context of a general intensification of preventive medicine that aims to prevent illness trough the identification of 'health risks'. Such strategies for prevention and insurance of the 'provision state' (Ewald 1986) have also expanded into the sphere of public health since the hygienic movement in the 19th century. One set of such strategies is the identification of 'high-risk groups' associated with a higher susceptibility to illness because of epidemiological and statistical analysis (Rose 2001, 7).

Historically, these strategies have been linked to different modes of regulation. A closer look at the history of cancer prevention and early detection, which was already a major issue during the National Socialist period in Germany,[1] shows how a tenor of obligation to the state changed to an increasing appeal to the self-determination of the individual to regulate his or her health independently (Weymayr & Koch 2003). This is congruent with a development in cancer research: to concentrate on genetic and life-style factors, which can be individualised, instead of environmental or workplace factors like pollution or dangerous substances at work, which would have to be negotiated in the political sphere (Wolf 2000).

That health is increasingly seen as a matter of competence, as an outcome of one's own will and actions (Greco 1993), also manifests itself in the development of genetically based prevention. Instead of a genetic determinism that was feared earlier, the concentration on genetic factors for illnesses affiliates with an appeal to the individual to 'manage' its genes. This management means to identify oneself as 'genetically at risk'

and to initiate appropriate action to prevent the disease (Rose & Novas 2000). Human genetics thereby takes its place in the logic of increasing control over the body, that determines the history of civilisation, according to Max Horkheimer and Theodor W. Adorno. These representatives of the so-called 'Frankfurt School' have described in the 'Dialectic of Enlightenment' (Horkheimer & Adorno 1979) how the relationship between mind and body becomes a subject-object relationship in which the mind (or 'spirit') dominates the body. Modern medicine can be seen as one of the means to control and objectify the body and – in the long run – to abolish natural perishability.

The subject that manages the object 'body' in the contemporary world does this in the mode of self-determination:

> Medicine, including medical genetics, notwithstanding its resolutely somatic understanding of the mechanisms of disease, has been one of the key sites for the fabrication of the contemporary self – free yet responsible, enterprising, prudent, encouraging the conduct of life in a calculative manner by acts of choice with an eye to the future and to increasing self well-being and that of the family (Rose & Novas 2000, 490).

With regard to the relationship to doctors, concepts such as 'patient autonomy' are propagated. This is what we encounter in the case of genetic testing for breast cancer in Germany: women (just as men in other cases) are encouraged to become active patient-subjects, to inform themselves, to find out about their so-called risk status and to manage it using practices of risk management. They are encouraged to make up their own mind about it: the 'woman's own decision' is emphasised throughout the informative material and stressed many times during the counselling sessions as essential for the question of taking the test as well as the question of which follow-up measures to take. This is a tribute to the concept of 'informed consent' that has been established in human genetics and also in medicine in general. It is not only a protection for patients, but also a discharge of responsibility for doctors.

Ways of subjectification

The question is if and how this demand to become an active subject of one's genes can be met. How is a genetic risk actually *embodied?* In the following, by analysing the narratives of women, the focus will be on ways of subjectification. The point is to reconstruct how one becomes an at-risk person or identifies as such.

Quantitative data has shown that mutation carriers experience increased anxiety after receiving the test result, which declines to the level before the test after some time, while a negative test result leads to feelings of relief. There are in any case no studies of the long-term effects to date. (The review by Hopwood (2005) summarises major studies.) These studies measure psychological effects on standardised scales or by means of standardised questionnaires which asked women e.g. whether they would take the test again. In contrast to that, in my study I investigated *qualitatively* how women talk about becoming a risk-person *in their own narrative accounts*.[2] This also concerns the question of how women themselves experience an 'informed decision'. What is the 'information'? And how is a 'decision' taken?

The model that is put forward in informative material addressed to women, but also in the internal debate surrounding the genetic testing issue, proposes an original *problem*, that makes women first *inform* themselves and then *decide* by calculating the risks. In reality, as I will show through the analysis of my interviews, the *problem* only occurs to the women in the first place because of the discourse on genetically based prevention. A *decision* is only necessary because of the possibility of testing and the information about it. Of course, from the medical point of view, the problem already exists because of the 'high-risk families' that doctors actually want to inform about. But the aim in this paper is mainly to reconstruct the perspectives of the women for whom there is no problem to start with. Through these perspectives I will also contribute to raising awareness for the problematic consequences of this kind of information and what the setting demands of women. Taking the little possibilities of prevention in the breast cancer case into account, it might also serve to question this kind of diagnostics in general. Many

quantitative studies have revealed that women who decided to take the BRCA test had increased levels of cancer related fears *before* the test (Foster et al. 2002, Hopwood 2005). While in most of the literature only the psychological changes *after* the test are considered as consequences of the test, in my opinion, the increased anxiety *before* the test must also be considered as an effect of the entire setting of counselling and testing: women consider cases of cancer in their family as being relevant for their own health only as a contingency of the emphasis on hereditary cancer factors made in the medical discourse of today.

Additionally, the process of decision-making is not always structured in the way the discursive model proposes. In my interviews, I basically found two different ways of dealing with the test. The first type of women, the 'informed risk managers', present themselves very much like the ideal active patient-subject the discourse suggests. The second type, the 'care-seeking clients' actually don't perceive themselves as active agents in the process of testing and treatment following the test. Decisions are often not addressed as such, events just seem to 'happen' and only other people act. Through the hermeneutic analysis of the narration of 'agency' in the interviews it is possible to reconstruct these differences (Lucius-Hoene & Deppermann 2002, 59).

'Informed risk manager'

The first type of women that I referred to as 'informed risk-manager' match the ideal model of the discourse. These recount their history before the test as if they had always lived in the knowledge of 'being at-risk' because of many cases of cancer in the family. It is not clear how they found out about a potential hereditary factor, but asked how they got to take the test, they talk about these cases and often state that doctors sent them to mammography already at early age. These institutional practices transform into their own concern about cancer, as in the following quote:

> because of this I have already been dealing with the situation for quite a while, and my gynaecologist knows about it and has always made intensified check-ups, and has actually always tried to do that regularly.

> Of course, you go to the check-up in a different way as if there had been nothing, well, I got my first mammogram before the age of thirty.

For these women we find what has already been found in other studies (the quantitative studies mentioned above, but also in international qualitative studies, e.g. Gibbon 2006; Hallowell 1999; Robertson 2000; Scott et al. 2005): they are very alarmed before the test and have great cancer worries because they see the cancer cases in their family as being connected to genetic factors. The discourse on genetic prevention is so natural to them that they perceive themselves as risk persons for already quite a while and act as informed decision-makers, informing themselves and calculating the costs and benefits of the knowledge as well as the different options. The risk figures make sense to them and serve as a rule for making decisions, as may be seen in the following quote:

> I would recommend everybody who belongs to the risk population, to take the test, (...) simply because I appreciate being in the *know* about the things that are inside of me, and if there is a risk inside of me, then I am really the first person who should know about this, so that I can deal with it, no matter *how* I deal with it. Well, basically it is *my* problem how I deal with such a matter, (...) And I think the risk *not* to know and to be unnecessarily anxious is so high.

The genetic risk – a statistical relationship that makes no statement about the individual – is reified to something inside oneself; and it is obviously perceived by this woman as something you can deal and calculate with; she can even weigh the costs and benefits of the test against the emotional risk *not* to know. This shows that she presupposes a situation in which women are aware of their risk status and therefore anxious anyway, which is illuminating: she already is constrained in her choices by the discourse on cancer heredity and the fact that she knows about the possibility of genetic testing, and in this situation of general awareness the choice to make the test seems the only rational one.

Another woman explains that before she made the test, and having calculated all the possible options, she decided to have prophylactic

breast surgery (mastectomy) in the event of her having a positive result (a situation which did not arise in her case):

> because if I know I have a risk of maximum eighty percent, that is really high; then it doesn't make sense to go to the check-up every three months. It makes much more sense if I take a really serious step.

Again, we see a strong agency. The probability has a clear-cut meaning to the woman and empowers her to act.

Concerning the *relationship to doctors*, I found that these women appear very self-confident, they actively ask for information and often decide against recommendations.

In the self-perception of these women, risk management really seems to make sense. The embodiment of risk, the notion that a statistical relationship between cancer and a set of bases in the DNA has something to do with their own body, and that this is a call for action, is working. Nevertheless, in the subtext of some interviews, it becomes obvious that the promise of more control is not fulfilled:

Interestingly, some of the 'risk managers' in particular are very preoccupied with thoughts of cancer, very anxious before the test and not totally relieved after a negative test result: the fear of cancer is still a central topic. One woman even asks herself how certain the test is. So the hope to have an instrument of control turned out to be illusionary. The 'reduction of risk' does not seem to be as satisfying as expected.

Genetic knowledge, just as other forms of modern knowledge, reveals its paranoid structure here: the more you know, the more you are haunted by the unknown, and a point of certainty, which was the original aim, is never reached. This is even more the case with a knowledge which can only be expressed in probabilities, because there are many factors interacting and there is no direct causation. This knowledge is uncertain in its inner structure. What makes it especially unpleasant in this case is that the unassertive knowledge concerns the future condition of one's own body.

In addition, the women's stories reflect how questionable the possibilities of individual risk management are in the BRCA case: there is no effective prevention except surgical removal of those parts of the body

that are at risk, such as the breasts or the ovaries, which, in the context of the uncertainty of the test, is not a very satisfying answer. That raises the question of what value the genetic testing has for women at all.

'Care-seeking clients'

In the narratives of the second type of women, they do not appear as active subjects informing themselves and then *deciding* what to do. When being asked how it came about that they made the test, they would say for example:

> that was completely by chance
>
> or:
>
> I didn't think much about it.

or describe only what the doctors suggested without talking about their own actions. Getting in touch with the clinic seems more like an automatic process than a conscious decision: most frequently women are informed about the test when visiting a near relative with cancer in hospital. The exact motives for the test remain unclear in their accounts.

A self-perception as 'being at risk' also only develops in the forefront of the genetic counselling sessions. The picture of a 'high risk family' emerges only through the counsellor's request to collect information for the pedigree. Typically, these women say:

> well of course, then I dealt with it, I didn't know this previously, because I didn't have any contact with these relatives. (…) Then it came to me that there is a lot of breast cancer in my family.

The awareness of having many cases of cancer in the family that might have something to do with their own health somehow develops during the process of participating in the clinical trial. Clearer than for type 1, it becomes obvious that for these women there is no *problem* to start with and nothing to *decide* about. It is only through getting in touch with

information about the genetic test that the problem arises of having a potentially higher risk and with it the necessity to decide. The various counselling sessions are therefore not only a help in making this decision an *informed* one, but also force the woman to make a decision.

Furthermore, this decision is not always narrated as such. Instead of an ideal process of will, plan and action, the agency of these women is often indirect, as they leave the agency to the doctors who thus appear to be the only persons acting in the process. The concept of 'informed decision' seems to be too simple to describe this: instead of a clear decision based on sound information, other forms of agency are involved which cannot be seen as mere passivity. It is a somehow indirect form of agency, as the doctors act and the women either follow their recommendations or when they do not trust the doctors, they stay away. Unlike the classic authority relationship with the 'white gods' of the medical profession, the condition for trust is to be acknowledged and also to be taken seriously as a *person*: doctors who treat people 'only as a number' are criticised.

In regard to their *relationship with the physicians*, these women do not see themselves as well-informed patients or lay experts having an equal partner relationship with the medical expert, but seek for the 'good doctor' that they can trust. A central topic is 'being in good hands'. They prefer to know a doctor personally and reject the anonymous atmosphere in some of the breast cancer centres, as the following quote illustrates:

> I don't go to the university hospital in town any more for all the examinations, because there is always a different assistant doctor. (...) I just need somebody I can trust, who knows me, who knows my case; my gynaecologist, she decides when I go to have a mammogram, she decides if I do it or if I *don't* do it, and now I go to the check-up every half a year.

Following her gynaecologist's advice, she also decided not to take part in the anti-hormone study which was offered to her at the university hospital after her positive test result. One woman explicitly rejects the position of being an informed decision-maker, narrating an anecdote where she was told about some changes found in her breast radiographs:

> then, you see, I ask a *doctor*, I *am* not a doctor myself: "so, what do you recommend" or "what shall I do now", and then he tells me: in my eight-month experience, women mostly react by doing this or that. And then I looked at him, and I said, ok, (...) I go to my gynaecologist.

Clearly the doctor's statement referring to a percentage of actions taken by others and leaving the decision to her, makes no sense to her. It also reveals another form of indirect agency: the call to be an active subject of one's health is rejected, the own decision is demonstrated in the *choice of the right doctors*.

A characteristic of this second type of women is also that they do not see a direct connection between knowledge and actions. The information the test gives cannot be translated into a demand for certain choices to take. As one woman says:

> that's actually why I really would like to know how it works, but probably nobody can tell you definitely. Well, I have that *gene*, it is true, but what is it that happens in order that the gene becomes *cancer*? (...) Is it because I have emotional problems, because it just happens, or is it because I don't eat healthily? You know, that's what I would like to know, but I think this is something they actually don't know themselves, otherwise everybody could do something against it, am I right?

It is clear from this that the gene mutation is not perceived as a calculable risk, but as an unpredictable one. The effect that one's own actions will have is thus completely uncertain. A risk management for gaining control over the future is not possible.

Nevertheless these women feel obliged to 'do something' about their health or feel guilty if they do not. The story one woman tells is illuminating in this context: she says that if she were to contract cancer, she would probably think: 'why didn't I stop smoking?' But then she recounts how her son got a rare coughing disease, although she and her husband never smoked in the house. If they had smoked in the house they would probably have blamed themselves for the cough. Her bottom line concerning cancer is:

> That's how it is. That's why I think to myself, perhaps I will stop smoking but get it anyway.

What she talks about here is the paradox of feeling responsible for the supposed consequences of your behaviour, although you cannot trace them back to it. In summary, the second type cannot be described as the informed decision-makers the discourse proposes.

Conclusion

The analysis of the narratives of women was meant to reconstruct what an 'informed decision' as part of genetic risk management means in reality. Although all of the women said they would take the test again, it became obvious how illusionary it is to be a risk-minimising active subject of one's own health. It was shown that the concept of a process of 'problem – information – decision' – is not realistic, instead the problem and the necessity to decide is imposed on the women only by the information (or: the alarming medical discourse). In this already anxious situation, taking the test appears to be the most rational course, because the outcome can only be that the risk becomes less (if the test result is negative).

Furthermore, it was shown that some women, as in type 2, don't match the ideal model of the discourse: they *do* engage and inform themselves, but they don't describe themselves as active decision-makers, don't see themselves as lay experts or equal partners of the doctors, they don't describe the risks and choices as calculable and don't feel they can control the future. But also for the first type of women taking part in the semantics of risk management it becomes obvious that this management often does not lead to a conciliation, but requires an infinite level of awareness. These 'side effects' of genetic testing should be taken into account. This is especially true when the possibilities of individually influencing the cancer risk are so limited: the most effective prevention is to remove the organs that are to be protected. This makes the concept of 'health as competence' and self-responsibilisation very questionable.

Thanks to all the women and counsellors for their time and willingness to tell me about their experiences.

Notes

1 Proctor's comprehensive study (Proctor 1999) shows how the war against cancer fitted ideologically into the general bio-political concept of National Socialism: 'Jews' and 'Cancer' were used as synonymous metaphors of degeneration.

2 Due to the qualitative approach, the interview results cannot be taken to be representative of all women in the same situation. But because certain frames appear repeatedly in the interview material it can be taken for granted that these results are not totally accidental. By using the qualitative approach it was possible to go into greater depth and to follow the women's priorities when talking about their experiences.

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