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**Being Genetically At-risk for Hereditary
Breast and Ovarian Cancer:
How Women “At-risk” Deal with the
Paradox of Certainty and Uncertainty**

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Being Genetically At-risk for Hereditary Breast and Ovarian Cancer: How Women “At-Risk” Deal With The Paradox Of Certainty And Uncertainty

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Abstract

In modern societies there is a broad consensus about the double structure of new technologies. As a tool of progress they are constructed to help solving problems, such as reducing risks. On a second hand, more often than not they produce new risks. In parallel, the popular meaning of risk is changing from a more fatalistic perspective of risk as a kind of danger, to a more individual concept. More and more risk transforms into something that can be controlled. A right and responsible individual decision may avoid danger. And inside this possibility is an appeal to do so.

This fact focuses on a new class of individuals: women, who use a modern biotechnological tool by undergoing a predictive genetic test and in the following are marked as “genetically at risk” for hereditary breast and ovarian cancer have to deal with this experience of risk. The genetic knowledge spends security in form of “knowing the devil”, but at the same time new uncertainties rise. How sure feels knowing a risk? Which decision may lead to what kind of future? How can the uncertainty be handled?

In this paper I ask for the implications of being “genetically at risk” in everyday life. How are affected women reading the diagnosis? Which role plays prevention in banning the new uncertainties? The empirical data will be interviews with women being “genetically at risk” for breast and ovarian cancer. On the basis of quotes, drawn from the ongoing survey “Genetic discrimination in Germany”, I will present a variation of strategies, used by the women, to integrate the probabilistic diagnosis in their daily lives.

At last I will discuss the meaning of the shown strategies in a wider range. Which factors may influence the women’s feelings and behavior? In what degree is the social category “at-risk” a new sort of a social problem?

Key Words:

Acknowledgement:

Introduction

In modern societies there is a broad consensus about the double structure of new technologies. As a tool of progress they are constructed to help solving problems such as reducing risks. On the other hand they tend to produce new risks. This is especially reflected by a new class of individuals: women, who use a modern biotechnological tool by undergoing a predictive genetic test for hereditary breast and ovarian cancer. In the literature, persons with an identified mutation are classified with a newly founded category: because they are not ill and do not show any symptoms, and because they are not healthy either, they are stuck in-between the poles of health and illness. That is why they are called the “pre-symptomatics”. According to Billings, this category designates people who have a genetic diagnosis but who are asymptomatic or will never become significantly impaired“ (Billings et al, 1992: 476). As “pre-symptomatics” they enter a new stage. The test result makes this stage observable. They are not “healthy” any longer. Now they are “genetically at-risk” (Kollek/ Lemke, 2008).

Our survey shows, the genetic knowledge spends security in a way of “knowing the enemy”, but at the same time new uncertainties rise. From the very moment they get to know their risk-status, life changes. Their behavior and feelings are now influenced by fears of an oncoming disease and they need to invent strategies to deal with this fear. Affected women ask themselves: What does the future hold? In everyday life persons concerned have to integrate the probabilistic character of the test result. Living with the genetic knowledge means to develop a personal relationship to the general risk calculation of the disease. Although no symptoms occur yet, the disease gets an increasing relevance in the daily life of the person concerned. Now the affected are labeled as “pre-symptomatics”. Health becomes a project that will never be completed.

In this paper I will talk about the distinctive quality of a risk diagnosis for hereditary breast and ovarian cancer for one person’s self and ask for the role of prevention in this special context. Our empirical material shows experiences of women who underwent a predictive genetic test and received a positive result.

It is based on a questionnaire survey and subsequently conducted semi-structured interviews with eighteen women “genetically at risk”. They all have been tested positive on a mutation in the so called Breast-Cancer-Genes BRCA1 or BRCA 2. I will show that the meaning of prevention goes far beyond avoiding a disease. For the affected women, prevention has the function to help managing the uncertainty which occurs in cause of the risk diagnosis. But like the German sociologist Ulrich Bröckling has already stated, all measures can only help for the present and the prevention process is eternally open to be optimized (Bröckling, 2008).

I am going to illustrate how risk diagnosis and prevention options keep uncertainty latent and contribute to a permanently optimizing self.

At first, I will explain the double structure of a risk-diagnosis for hereditary breast and ovarian cancer that goes along with predictive genetic tests. Second, I would like to illustrate the meaning of prevention as a strategy to manage the balance between certainty and uncertainty - on the basis of quotations of women interviewed. I will show how the women affected give four meanings to prevention. Finally, I would like to discuss social implications of the circle of certainty and uncertainty – diagnosis and prevention – as a tool of self-regulation.

Hereditary Breast and Ovarian Cancer as a risk diagnosis

Experts assume that 5%-20% of all breast cancer diseases have a genetic background. Since the 1990ies genetic tests enable them to verify the assumption of a deviance in the so called breast cancer genes BRCA 1 or BRCA 2. This genetic mutation is associated with an up to 80% higher risk to develop breast and ovarian cancer in lifetime. The mortality rate is specified with approximately 14% within 5 years (Meindl et al, 2011).

In Germany, policy holders of statutory health insurance are able to avail the genetic test free of charge, if women document a defined number of breast cancer diseases in their family history. In case of a positive result, the health insurance provides prevention measures in selected hospitals. The offered medical prevention is a composition of efforts to reduce risk factors, known as primary prevention, and detect and manage supposed symptoms, known as secondary prevention (Kast et al., 2009). Women “at-risk” are invited to frequent medical examinations and additionally to prophylactic surgeries in certain cases. Although no one would deny that prevention is better than cure, no study has proved a significant impact on mortality rates yet. Aggressive tumors cannot be stopped by screenings. On the other side, experts discuss cases in which unnecessary treatments stress patients, who possibly would never have fallen ill.

Being genetically at-risk for the affected women is the knowledge of carrying a disorder in their individual genes. Looking at the whole body system, genetic mutations are a normal biological phenomenon, in the isolated perspective the test takes, a single genetic deviance is considered as a meaningful pathological condition. To explain the diagnosis medical professionals apply the terminology of cancer. The gene where the mutation is detected in, does not only relate to breast cancer, but is referred as “breast-cancer-gene”. A mutation is further associated with a higher risk for other types of cancer, for example skin cancer or brain tumors. Although these facts sound awesome, genetic knowledge is seen as a resource. Hamilton could show that genetic testing is valued by the women as a clarifying step, which helps them to know where they stood in relation to their family’s genetic risk (Hamilton, 2009). In public health discourses, genetic information is constructed as empowering, because it offers choices and promises the ability to control future health (Hallowell/Lawton, 2002). Our data reflects that.

Nearly all women of our sample, report that they are satisfied to know about their risk, because something got “clear” and the knowledge provides solutions.

But at the same time the diagnosis creates feelings of being ill. Particularly cancer is seen as an ultimate disease and is associated with physical degeneration and death. Additionally, the incomplete penetrance of the mutation allows no prognosis of possible individual physical manifestations, because the risk of developing a disease in the future is constructed on the average of a large population. In fact of this, Kenen et al. (2003) compare the at-risk status with the condition of a chronic disease. They show that people at-risk and people suffering from a chronic disease face similar issues, such as biographical interruption and an uncertain future. Both are continuously confronted with the probability of an occurring disease. Every day they undergo situations that make the risk salient and bring it back to their minds, accompanied by the uncertainties and the questions about their future. Both, people with a chronic disease and people being genetically at-risk, have to deal with this uncertainty which they might experience as a loss of control. Kenen et al. give a chronic perspective to risk because of the heightened awareness and introduce a concept of “chronic risk” (Kenen et al., 2003) which is also described by one woman of our sample with the phrase “sitting on the powder keg” (Schmitz A04, 562).

This combination of security and insecurity suggests that controlling the deviance by taking advantage of preventive measures may lead to a safer future. Like Hamilton could show before, prevention measures like prophylactic surgeries and a healthy lifestyle help regain a feeling of “normality” and control of the own health (Hamilton, 2009).

Risk factor and prevention options are a joint production. Together they represent a paradox: the diagnosis connects risk factor and therapy – cause and effect. But the risk status permanently recalls the unknown future, especially by controlling it with precautionary efforts. Our empirical material shows exactly this kind of problem: women with a positive test result for hereditary breast and ovarian cancer report us about their feelings of losing control over their lives and how they try to reestablish agency and the ability to make plans.

Design

The presented study is part of the main study “Genetic discrimination in Germany“, that takes place between 2011 and 2014 at the Helmut-Schmidt-University in Hamburg and the Goethe-University in Frankfurt/Main. The study was qualitative designed and questioned people with a genetic risk for familial adenomatous polyposis (FAP), a hereditary colonic cancer, cystic fibrosis, hereditary haemochromatosis and hereditary breast and ovarian cancer, about their experiences with the genetic diagnosis and asked how they deal with the genetic knowledge. It starts with a questionnaire survey in the first phase. Then, follows a phase of semi-structured face-to-face interviews with affected people.

This paper presents results of that part of the study, in which women with a genetic risk for breast and ovarian cancer were talking about their special experiences.

Recruitment

The participants were recruited in self-help-organizations and support groups and in cancer clinics in Germany, where the questionnaire was handed out. Furthermore a website was designed, which informs about the study and also includes the questionnaire (www.genetischediskriminierung.de). The questionnaire contains the offer to contact the research group for an interview. The eligibility criteria for the women being interviewed were that they had a family history of cancer and had been to a genetic test where they got a positive result.

Data Collection and Analysis

89 women at-risk for breast and ovarian cancer answered the questionnaire and 18 of them were interviewed semi –structured. The interviews took place within six months. 16 interviews were conducted face-to-face at the women's home. Two were conducted over the telephone. Among other issues that focus on the management of the potentially stigmatizing genetic information, the women were asked about their feelings, when they got the positive results and how their lives had changed. The interviews lasted between 40 and 120 minutes. They were tape-recorded with consent, anonymized and transcribed.

The study was carried out according to the principles of grounded theory research (Glaser and Strauss, 1967).

The Participants

The ages of the women range from 24 - 64. 10 of them have children. 12 of them were free of symptoms yet (pre-symptomatic). All of them take advantage of intensified screenings, four underwent prophylactic surgeries additionally. In one woman, no suspicious gene was detected, but the medical experts confirm a high risk for cancer due to calculation software for family history of cancer.

Dealing with the Uncertainty: Four Meanings of Prevention

People at risk are confronted with the possibility of degeneration and death. In order to control their fears they use prevention as a tool that helps to reduce insecurity and increases a feeling of safety instead. The interviews reveal four meanings of prevention. The following quotes illustrate four different ways of “dealing with uncertainty”.

Prevention as a Possibility to Encourage Hope

The first meaning of prevention shows that prevention encourages feelings of hope by controlling risk. In one of the interviews, Mrs. Zeller, who has

known about her risk for three years before she finally fell ill, is convinced that,

“if it [the cancer symptom] is detected early, the chance to cure is really very very high” (Zeller K03 198f).

In cause of her risk status Mrs. Zeller underwent numerous examinations, every six month a palpation and an ultrasound - mammography and magnetic resonance tomography once a year. Now a suspicious node was found and treated.

Prevention promises that an intervention in a pre-symptomatic status increases the chance of cure and Mrs. Zeller wants to believe in this assumption. Believing in the promises of prevention means to give a chance to the opportunity of risk reduction. Doing the right thing now may be able to influence the future positively, even if it is a future with symptoms. This assumption she needs to prove now. Just in the moment after the examinations precautionary actions are safety-giving. But the risk of a potentially deadly disease in the future is still part of every precautionary effort.

Prevention as a Possibility to Keep Control

This leads to the second meaning of prevention: prevention is a possibility to keep control. To keep control is necessary to deal with the fear of death. Inherently risk contains various future scenarios that are related to current behaviour. With this, the incomplete penetrance of the Mutation offers a gap of hope. For women free of symptoms yet, as Mrs. Jahn, prevention is an important strategy to experience their selves less helpless and more active and influential. Mrs. Jahn tells us about her feelings of power:

“I want to do something and this is what I could do: making the test and actively work against it. This is why I decided to remove my breasts and maybe someday will take the option to remove my ovaries, too. The power to do something against it is with me. And this is the most important thing.” (Mrs. Jahn K1 214ff)

Prevention helps Mrs. Jahn not to stay victim of her fate. With the decision to remove her breasts she behaves pro-active. She did, what is considered as the right thing to reduce her risk and assume responsibility for her health. To act like this gives her feelings of power on the basis of the ability to act.

Prevention Provides Certainty of Action

The third meaning of prevention is that prevention provides certainty of action. The survey shows, prevention can be a benefit for people at risk, if they interpret the medical measures as solution for a functional disorder. Affected women report, how they feel “well looked after” by the doctors since they know about their higher risk. Mrs. Kruse, a woman who is free of symptoms, too, says in the interview:

“I know I’m in good hands now. They [the doctors] will identify it [the cancer symptom] in time and will be able to provide quick help. So you are going to be healed again.” (Mrs. Kruse A05, 80f)

Mrs. Kruse relies on the medical professionals, from whom she expects that they know what to do. For that, she betakes herself under continuous medical observation that started the medical treatment before a symptom occurs. She trusts in the early form of intervention and in the medical knowledge of her doctors. It makes her feel safer that there are some professionals who know what to do now to solve problems like hers. She believes in the causality of risk factor and disease. With that, she consolidates the correlation between knowledge and action that is part of the logic of prevention.

Prevention as a Survival Advantage

The fourth meaning of prevention is that prevention may serve as a survival advantage. Some women at risk give an opinion of advantage to prevention because they receive privileged access to medical treatment. Mrs. Michels, a young woman, tells how her sister - a medical professional – helps her after the shocking risk diagnosis to change her negative thoughts into positive:

“You are allowed to participate in the precaution system. See the positive side, no one else gets that. All the other women get a simple checkup at the gynecologist once a year and that’s it. And you are going to get a complete checkup twice a year. What more could you want?” (Mrs. Michels A09 150ff).

Mrs. Michels now interprets the genetic knowledge as an advantage against all the people who do not know about their possible risk status. The identified breast cancer risk turns into a risk that concerns many women. But only those with the detected gene have the luck to know about it. The extensive and frequent medical examinations for her are a privilege and may help to detect cancer and other disorders on an early stage. This particular surveillance helps her stay healthier than other people due to a better medical treatment. Knowledge can become an action advantage for women who believe in the logic of prevention. This advantage has the power to keep her more normal in a health perspective than other people.

The Extended Version of Risk and Prevention – A Demand for Self-Regulation

Women with a suspected predisposition for breast and ovarian cancer get to know about their genetic risk in a very concrete form when they receive a positive test result. With this definiteness new uncertainties go along with.

Women affected have to deal with the uncertainty of their future. They don't know if they will fall ill, nor when or how serious the illness will get. With the test result, their life changes. They have the definite diagnosis that they might fall ill in the future. With that in mind, they face a disease that they don't have. With the classification of a "pre-symptomatic", they enter a patient stage and become part of the medical precaution system. The survey shows that women with a genetic risk diagnosis find help in prevention in order to control fears. This follows from the conviction that medical treatments could only be good - the more and earlier the better. Depending on their specific needs, precaution offers a range of measures and of variants of interpretation that could help to influence the relation between certainty and uncertainty temporarily. Nevertheless, the risk of an uncertain future is always in the back. While prevention on one hand provides certain kinds of senses of security, on the other hand every act of risk management has the power to bring the uncertain future back to mind. With every appointment at the cancer clinic, the risk gets salient. By this, the fact is recalled that Prevention always has to be continued and needs to be adapted to every current situation. People at risk mark a gradual shift in the current medical diagnostic practice. They extend a continuum of "abnormity", where a genetic risk becomes the same relevance as a disease symptom. Aronowitz shows, that People at risk and people with a chronic disease face similar experiences and have to deal with similar problems (Aronowitz 2010). Both start a patient career and adapt illness in their identities. Both have to expect side effects of medical treatments and both are requested to watch themselves carefully and show responsibility for their own health. Like ill people, people at risk get part of a circle of risk production and risk reduction that keeps the uncertainty latent and asks them to regulate their selves permanently. Like Crawford pointed out, there is a nexus of control and anxiety that is generated by efforts for protection and that may escalate in a spiral of anxiety and control (Crawford, 2004). The risk diagnosis marks the starting point for efforts, that should keep the new categorized people as normal as possible. Now life is determined by a precaution schedule. The underlying concept of normativity is constructed under the special perspective on an isolated gene, but has implications for the whole personal and social life of people "at risk". Their "faulty" body becomes a new importance for their lives and there is always something to do. New biotechnological tools like predictive genetic tests once started with promising prospects for a better life. But for the increasing class of "pre-symptomatics", they work as a far reaching self-technology. Being responsible and keeping the standard are life determining instructions, driven by new fears of death and of the stigma of being "willfully ignorant".

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