



Genetically at-risk status and individual agency. A qualitative study on asymptomatic women living with genetic risk of breast/ovarian cancer



Maria Caiata-Zufferey ^{a, b, *}

^a University of Applied Sciences and Arts of Southern Switzerland, Via Violino 11, 6928 Manno, Switzerland

^b University of Geneva, Department of Sociology, Bd du Pont-d'Arve 40, 1205 Genève, Switzerland

ARTICLE INFO

Article history:

Available online 19 March 2015

Keywords:

Switzerland
Preventive medicine
Genetic risk
At-risk status
Life-course perspective
Health project
Grounded theory

ABSTRACT

For the last 20 years, genetic tests have allowed unaffected women to determine whether they are predisposed to developing breast/ovarian cancer due to BRCA1/2 gene mutations. In the event of adverse results, women receive a specific label associated with a set of medical recommendations: the genetically at-risk status. This qualitative study adopted a life-course perspective to understand the impact of this status on women's agency. Following a grounded theory design, retrospective biographical interviews were conducted in Switzerland between 2011 and 2013 with 32 unaffected women at risk of developing genetic breast/ovarian cancer and aware of their predisposition for at least three years. The results show that the genetically at-risk status conveys an invitation to transform health into a project, i.e., into a set of planned activities realized in collaboration with the medical system in order to reduce the risk of developing cancer. This health project shapes women's agency in three ways: it enhances, constrains and questions it, thus creating a sense of disorientation about what is considered rational and appropriate in terms of genetic risk management. Based on these findings, the paper concludes by stressing the paradoxes of the genetically at-risk status and the limits of the medical system in managing women designated with it. The paper also suggests that because of the disorientation intrinsic to their situation, genetically at-risk women have to reflexively construct their own health project from a range of available options in ways that are coherent and viable for themselves and their significant others. This process of reflexive construction may be called legitimization.

© 2015 Elsevier Ltd. All rights reserved.

1. Introduction

On May 14th 2013, Angelina Jolie announced in the *New York Times* that she had undergone a double mastectomy followed by breast-reconstruction surgery because she possessed specific mutations that increased her risk of developing breast/ovarian cancer: "I decided to be proactive and to minimize the risk as much as I could (...) Cancer is still a word that strikes fear into people's hearts, producing a deep sense of powerlessness. But today it is possible to find out through a blood test whether you are highly susceptible to breast and ovarian cancer, and then take action". Jolie belongs to that group of unaffected women who have undergone genetic testing to verify whether they carry BRCA1/2 gene mutations. It has been estimated that these mutations are related to 5 to 10 percent

of all breast/ovarian cancers (Chen and Parmigiani, 2007). If a woman is found to carry them, she knows she has up to an 80 percent chance of developing breast cancer and up to a 60 percent chance of developing ovarian cancer throughout her lifetime (Easton et al., 2004). She thus receives an enigmatic status between health and illness: the status of a genetically at-risk woman.

According to Jolie, knowing that she is genetically at risk of developing breast/ovarian cancer has provided her with new choices for managing her health. This appraisal confirms the rhetoric of human genetics, according to which genetic knowledge enhances the individual's sense of agency (Petersen, 1998). Concretely, BRCA1/2 testing provides women with information about risk. In the event that the woman is found to carry the mutation, she is offered the possibility of adopting measures to minimize it. According to international recommendations (Balmana et al., 2011), these measures consist of intensive surveillance (regular and frequent clinical examination, imaging and laboratory evaluation) or prophylactic surgery (bilateral mastectomy, oophorectomy). Genetic testing and associated medical care are supposed

* University of Applied Sciences and Arts of Southern Switzerland, Via Violino 11, 6928 Manno, Switzerland.

E-mail address: maria.caiata@supsi.ch.

to be optional, and self-determination remains a central concept in human genetics (Clarke, 1997). Indeed, many countries have introduced laws that, beyond protecting the individual against genetic discrimination, also ensure their right to refuse genetic knowledge or opt for no further medical intervention in the event that a mutation is found (Lemke, 2013).

Nonetheless, the impact of the genetically at-risk status on individual agency raises dilemmas. According to Kenen (1996), the individual who is discovered to be genetically at risk of developing serious diseases receives a particular health status. This status can be considered a social position because it results from a process of negotiation and acceptance between the individual and the medical system and is associated with some role obligations: in particular, at-risk individuals are expected to try to avoid becoming ill. This imperative is implicit in preventive medicine, for which health is a precarious state that needs to be worked toward (Petersen and Bunton, 2002), and the same principle applies in the case of genetic risk (Löwy and Gaudillière, 2008). From this perspective, the international guidelines cited above may not indicate a set of options so much as a set of social constraints.

The nature of the measures recommended by international guidelines raises additional concerns about the impact of the genetically at-risk status on individual agency. Such measures may be questioned in terms of necessity, efficacy and expediency. Necessity may be doubted because genetic testing provides only probabilistic information. Nobody knows if the disease is going to develop, as a result of which the measures could be superfluous. Concerning efficacy, early screening does not guarantee the best rate of survival in high-risk groups (Oei et al., 2006), and prophylactic surgery does not eliminate all risk of genetic breast/ovarian cancer (Finch et al., 2006). With respect to expediency, the cost-benefit relationship of the recommended measures may be considered disadvantageous because they strongly affect women's everyday lives: surveillance makes them permanently dependent on the medical system, while surgery modifies their bodies. These considerations suggest that at-risk women exercise agency under conditions of great uncertainty: *normative uncertainty*, because the norms that regulate their behaviors are multiple and contradictory, and *rational uncertainty*, because it is difficult for them to determine the best course to manage risk, due to the probabilistic nature of both genetic risk and the measures that are supposed to minimize it. In the event of BRCA1/2 mutation, the impact of the genetically at-risk status on individual agency is thus complex, as confirmed by the literature. Sometimes, genetically at-risk individuals have been found to feel a greater sense of control over their health (Scott et al., 2005). In other studies, they experience their agency as limited by a sense of "genetic responsibility", an obligation to take steps to minimize their risk (Hallowell, 1999). This literature, however, captures people's experience at a single moment, without focusing on the complex interaction between the individual and their context over time.

This study develops a comprehensive understanding of the impact of the genetically at-risk status on individual agency. In particular, it explores the advantages, drawbacks and challenges that women who are designated as genetically at risk of developing breast/ovarian cancer experience in terms of agency, as well as the social processes behind this experience. Because it investigates the consequences of a medical label, the study contributes to the sociology of diagnosis (Jutel, 2009) and simultaneously it widens it, because here the individual is not, as is usually the case, diagnosed with a disease, but with a predisposition to it.

Drawing on retrospective biographical interviews conducted in Switzerland with 32 unaffected women at risk of developing genetic breast/ovarian cancer and aware of their predisposition for at least three years, this qualitative study shows that the genetically

at-risk status shapes individual agency in three ways: it enhances, constrains and questions it. The paper concludes that the main challenge for at-risk women is to reflexively construct their own way of acting towards genetic risk.

2. Methods

This article is part of a larger study conducted in Switzerland between 2011 and 2013 on trajectories and lifestyles in the presence of genetic breast/ovarian cancer risk. A grounded theory design (Strauss and Corbin, 1990) was considered suitable for investigating this underexplored process and developing a theoretical understanding of it.

2.1. Theoretical framework

This study adopted a life-course perspective to understand the impact of genetic risk on women's agency. In this approach, the phenomenon under study is conceptualized as the result of the life course of the individual. Its *dynamic* and *interactive* dimensions are thus stressed. In particular, this study was informed by the concept of "chronic risk trajectory" (Kenen et al., 2003), which was, in turn, inspired by Corbin and Strauss's (1991) "chronic illness trajectory". A chronic illness trajectory is the course of an illness over time but additionally involves the actions taken by agents to manage or shape this course and the consequences of these actions for the agents. Based on this theoretical framework and adopting an interpretive paradigm, this study started from the perspectives of the women interviewed to discover the *events* delineating their chronic risk trajectory, the *activities* they have adopted to manage genetic risk in their everyday lives, the *social forces* that have constrained and enabled these activities and the *meanings* women have attributed to all these elements.

2.2. Context

The study was conducted in French- and Italian-speaking Switzerland. Genetic testing for breast/ovarian cancer has existed in Switzerland since 1995 and is usually done in genetic-counseling centers in hospitals. The increasing number of private companies offering direct-to-consumer genetic testing services led to the introduction of the Law on Human Genetic Analysis in 2007, according to which genetic tests can only be carried out by a medical doctor after the provision of sufficient information concerning the nature, meaning and consequences of the test, and only with the consent of the person concerned.

The management of breast/ovarian cancer risk is not a distinct subspecialty in Switzerland: once the mutation has been found, at-risk women are encouraged to return to their gynecologist. Women are free to decide what to do in the event of adverse results (Lemke, 2013); however, they are recommended to collaborate with their healthcare provider to implement surveillance and prevention measures. These measures are theoretically easy to adopt: all persons residing in Switzerland are required to purchase basic health insurance, which covers treatments detailed in the Swiss Federal Law on Health Insurance; for women found to carry the BRCA1/2 mutation, surveillance and prevention measures are covered by their health-insurance policies.

2.3. Recruitment

Four of the five genetic-counseling centers that operate in French- and Italian-speaking Switzerland agreed to contact the participants for recruitment. Before starting the recruitment process, we presented a detailed document to local ethics committees

Download English Version:

<https://daneshyari.com/en/article/7332812>

Download Persian Version:

<https://daneshyari.com/article/7332812>

[Daneshyari.com](https://daneshyari.com)