Living with the BRCA1 and BRCA2 genetic mutation: Learning how to adapt to a virtual chronic illness

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Published online: 23 Apr 2013.

To cite this article: A. Samson, J. DiMillo, A. Thériault, S. Lowry, L. Corsini, S. Verma & E. Tomiak (2014) Living with the BRCA1 and BRCA2 genetic mutation: Learning how to adapt to a virtual chronic illness, Psychology, Health & Medicine, 19:1, 103-114, DOI: 10.1080/13548506.2013.779729

To link to this article: http://dx.doi.org/10.1080/13548506.2013.779729

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The objective of this study was to understand how women living with the BRCA1 and BRCA2 genetic mutation adapt to this life transition and to identify the main adaptive tasks. A qualitative inquiry inspired by grounded theory revealed that participants cognitively appraised their test result in the same manner as women who have been diagnosed with breast cancer. Consequently, participants had to adapt to a condition that they perceived as a chronic illness. The following three main tasks were identified: Physical Task: Attempting to Limit the Impact of the Test Result, Psychological Task: Living with Uncertainty, and Social Task: Finding Effective Support. In conclusion, although these women live with the possibility of developing breast cancer, their experiences mirror those of individuals living with a chronic illness, and they must therefore adapt accordingly in a physical, psychological, and social manner.

Keywords: breast cancer; cancer prevention and screening; genetics; health behavior; qualitative methods

Introduction

Women who carry genetic mutations in BRCA1 or BRCA2 are at increased risk of developing breast and ovarian cancer with an up to 80% chance of developing breast cancer throughout their lifetime (American Cancer Society, 2011).

Although genetic testing can provide knowledge to a woman about her risk of developing breast cancer, it is nevertheless a debated subject due to the potential negative psychological implications of such a diagnosis (Beran et al., 2008; Di Prospero et al., 2001).

Previous research on the psychosocial implications of testing for mutations in BRCA1 and BRCA2 suggests that those who learn they carry mutations in these genes may experience increased anxiety and depression (Lerman, Seay, Balshem, & Audrain, 1995; van Oostrom et al., 2003), anger and distress (Croyele, Smith, Botkin, Baty, & Nash, 1997; Dorval et al., 2000), affected quality of life, an altered self-concept,
vulnerability, stigma, and mastery (Esplen et al., 2009), increases in cancer perception risk, as well as worry about cancer (Di Prospero et al., 2001).

Taking into account the negative psychosocial impact of receiving this test result, which has been identified by previous research, the primary goal of this study is to examine how women living with the BRCA1/BRCA2 genetic mutations adapt to this life transition.

A comprehensive understanding of the adaptation process may help guide women carrying mutations in BRCA1/BRCA2 and their relatives through the many uncertainties they face and help them to find a way to stabilize the sudden disruption they have experienced. Furthermore, health care professionals may be better equipped to understand their patients’ efforts to adapt.

According to theoretical models that describe adaptation to major life transitions in a medical context, this specific process is based around the perception of the stressor (i.e. the test result), and its psychosocial impact on the individual (Cohen & Lazurus, 1979; Corr, Nabe, & Corr, 2003; Moos & Tsu, 1977; Samson & Siam, 2008). Research suggests that women typically perceive receiving a test result indicating they carry the BRCA1 or BRCA2 genetic mutation as a threat (DiMillo, 2011). Perceiving the source of stress as threatening triggers psychological responses such as denial, fear, anxiety, resentment, or even anger. These reactions are usually indicators that the source of stress has taxed resources to the extent that reality is perceived as being hopeless (Cohen & Lazurus, 1979).

According to the same theoretical models that describe the adaptation to major life transitions in a medical context, these primary psychological reactions tend to diminish with time. The stressor (i.e. test result) gradually becomes a part of daily reality and the perceived threat diminishes in intensity. This evolution engenders a secondary appraisal, which indicates that patients have begun to perceive their new reality as a challenge that requires undertaking adaptive tasks (Cohen & Lazurus, 1979).

These tasks encompass the principal aspects of human functioning that allow the process of adaptation to occur. The adaptive tasks are physical, psychological, social, spiritual, and vocational in nature (see Figure 1). The physical tasks consist primarily of meeting all medical requirements as prescribed by health care professionals. The psychological tasks involve maintaining satisfactory emotional equilibrium and regaining a sense of control over one’s life. The social tasks aim at gaining effective social support from significant others, friends, and family. The spiritual tasks of adaptation are efforts at giving meaning to the onset of the new medical situation and all the consequences it engenders. The vocational tasks include work that is both paid and unpaid, as patients may resume their professional occupations, give a new orientation to their career, or get involved in volunteer work.

If adaptation tasks are seen as the general domains of adjustment, then coping skills may be understood as the specific means used to accomplish these tasks (Moos & Tsu, 1977). Furthermore, these skills, which embody both a mental and behavioral aspect, are likely to be used in conjunction with one another, rather than individually.

According to Moos and Tsu (1977), there are two possible outcomes: (a) positive and (b) negative. While the former is indicative of a new state of psychosocial equilibrium, highlighted by a re-established sense of normalcy, the latter alludes to a certain degree of psychological deterioration and decline. In other words, a positive outcome is achieved when the illness is seen as less of a disruption and becomes an accepted aspect, among others, of patients’ lives. Patients regain a certain sense of control over the course of their existence (Samson, 2006).
The goal of this article is to describe how women with the BRCA1 and BRCA2 genetic mutations adapt to their new medical reality. More specifically, our aim is to identify and better comprehend the adaptive tasks associated with carrying these genetic mutations.

Methods
The methodology for this qualitative research is also described in detail elsewhere (DiMillo et al., 2013).

Participants
Participants were French or English-speaking women who had undergone genetic testing for hereditary breast/ovarian cancer (HBOC) and who had received a positive test result indicating that they carried a deleterious mutation in BRCA1 or BRCA2.

For the purposes of this study, only women who had received positive test results at least six months prior to the interviews, and who had elected to undergo close monitoring by their physicians and specialists were invited to participate. In other words, women solely undergoing screening for breast cancer due to a BRCA1 or BRCA2 genetic mutation were included, while women undergoing screening for ovarian cancer due to an HBOC genetic mutation were not included.

Demographics
All six participants were married, and each had at least one child (see Table 1). None of the participants reported having taken part in any drug trials for the treatment of...
breast cancer. In addition, only one woman had a prophylactic surgery (oophorectomy) following the receipt of her test result.

All six participants had a family history of breast and/or ovarian cancer. In addition, all six participants had at least one family member who had also been identified as having a mutation in BRCA1 or BRCA2, and most of the participants (four) reported that their mothers and/or sisters had also been identified as carrying the genetic mutation.

**Methodology**

A qualitative study utilizing procedures inspired by grounded theory methodology was conducted. The goal of this study was to obtain a rich description of categories and an overarching theme that described the experiences of these women, and hence, analysis involved the conceptual organization of data (Strauss & Corbin, 1998). Methodological rigor for this study was established utilizing the criteria brought forth by Lincoln and Guba (1989), Glaser and Strauss (1967), and Sandelowski (1986).

**Procedures: recruitment**

Participants fitting inclusion criteria were recruited at a specialized risk assessment clinic through invitation letters disseminated by a designated nurse. Once a patient agreed to participate, semi-structured interviews that each lasted approximately 1 hour and 10 min were conducted.

Consistent with the grounded theory method, data collection and analysis was undertaken concurrently (Glaser & Strauss, 1967; Rennie, 1998). In other words, the information gathered from the initial interviews, as well as its following analysis, helped to shape future interview questions and guide further data collection in order to better understand the participants’ experiences.

**Results**

After receiving their test results, the participants experienced a variety of emotional reactions such as fear, anger, and anxiety, which led to self-perceptions of powerlessness and vulnerability. In fact, participants described experiences that closely resembled those described by women who have been diagnosed with breast cancer (Liao, Chen, Chen, & Chen, 2008; Warren, 2010). Hence, this qualitative analysis showed that receiving such a test result is perceived as a life-threatening condition that triggers an initial crisis.
Our results suggest that over time participants reappraised their test result and started to reconstruct different aspects of their lives, which had been altered. This qualitative analysis suggests that participants’ lives were affected in three specific ways: the way they functioned cognitively; the manner in which they interacted with others; as well as how they faced their new medical condition. In the theoretical context of the adaptive task model, this research identified three main tasks that were undertaken by participants: Physical tasks, psychological tasks, and social tasks (see Appendix).

**Physical task: attempting to limit the impact of the test result**

Results suggest that participants experienced a great deal of stress after receiving their test result. As previously mentioned, participants described their reaction similarly to women who have been diagnosed with breast cancer. This explains why they focused great energy on physical tasks. In other words, participants aimed to protect themselves, at least as much as they could, from this inner enemy that they perceive as having the potential to destroy their lives.

In achieving this physical task, participants mostly utilized active coping skills that gave them the impression they had a greater sense of control over their life. Active coping skills consisted of seeking additional information by learning more about their options and preventative methods. Learning about these preventative methods allowed participants to make personally relevant and appropriate choices, such as prophylactic surgeries, hormone therapy, medical follow-up, and lifestyle changes.

In summary, receiving the news of carrying a mutation of the BRCA1/BRCA2 genes was perceived as a potential threat to participants’ health and even their lives. Therefore, for participants, the physical task was one of the major undertakings necessary to achieve their adaptation process. By doing so, participants were attempting to limit the impact of the test result, and were consequently able to regain a certain sense of control over their lives.

**Psychological task: living with uncertainty**

As it was previously mentioned, following the receipt of their test result, participants reported feeling angry, shocked, overwhelmed, sad, and confused. For participants, it was therefore imperative to regain their emotional equilibrium. In order to achieve this task, participants modified their cognitive appraisal of their genetic predisposition.

From what was perceived to be completely negative news, over time participants reported being thankful for having received this test result. Their awareness facilitated the medical care necessary to help prevent, or quickly detect, the development of cancer in the future. Furthermore, others described that having the knowledge of carrying the genetic mutation has helped them to make the appropriate decisions, whereas not having received their positive test result may have led them to make choices that were not based on knowledge, and which may have been faulty.

Other cognitive skills that were helpful to participants included comparing themselves to other individuals they knew who had breast cancer and have survived. Some participants decided to develop a positive outlook, in that they consciously tried not to become too worried when they were screened for breast cancer. In addition, living in the moment rather than the uncertain future appeared to help some participants regain some sense of emotional equilibrium.

Minimizing the situation was another helpful cognitive skill utilized by participants. For example, some participants sheltered themselves by repressing the fact that they
carried the BRCA1/BRCA2 mutation. In the same vein, some participants used avoidance. Avoiding entailed that a participant was evading a situation, such as testing or prophylactic surgery that was strongly recommended as a way to reduce risk of cancer occurrence, even though she was aware of the potential benefits.

In summary, in order to adapt to their new medical realities, participants needed to engage in a psychological task to function and continue to live their lives. Hence, participants’ psychological challenge was to reappraise the test result, allowing them to redefine their sense of normality. Mainly, what allowed participants to reestablish their emotional equilibrium was to perceive their diagnoses as a chance to take charge of their health, regain a certain sense of control over their lives, and develop a new sense of normality that took into account their genetic predispositions. Nevertheless, from time to time, avoidance and minimizing appeared to shelter participants from the impending threat of cancer.

**Social task: finding effective support**

Following the receipt of their test results, participants perceived themselves as different from others. They had to face the reactions of their social environment concerning their predisposition to developing breast cancer. They reported feeling misunderstood, judged, and sometimes rejected. In this difficult context, participants were compelled to find meaningful social support to limit the impact of rejection, and find a source of comfort. This source of social support mainly came from family members, and other BRCA1/BRCA2 genetic mutation carriers.

Participants explained that their spouses played a significant role in their support structure. For instance, spouses generally attended the appointment during which their wives were given the news that they carried the genetic mutation predisposing them to breast cancer. Similarly, participants tended to perceive other BRCA1 or BRCA2 genetic mutation carriers as providers of support. Participants generally described that these individuals were better able to understand the meaning behind carrying these genetic mutations.

Finally, for some participants, close friends were also a source of support. They described them as very understanding of the test result, and often present and empathic during times of need. Participants also perceived a great deal of support being provided to them through the medical team. They often described that their physicians, and at times nurse at the clinic, were a reliable source of support and able to provide care and a listening ear.

In summary, participants viewed others as a source of support or help when they perceived them as being able to help them cope and support them through this time of need. They found main sources of support in their inner social circle, such as family, their spouses, and close friends. They also extended this social support to other BRCA1 and BRCA2 genetic mutation carriers, as well as health care professionals.

**Discussion and conclusion**

**Discussion**

Two main findings emerged from this research. Firstly, participants in this study reacted to the receipt of their test result as though they were diagnosed with breast cancer, even though they have no clinical signs or symptoms associated with a breast cancer
diagnosis (DiMillo et al., 2013; Liao et al., 2008; Warren, 2010). This reaction led them to experience an initial crisis in which they perceived their genetic predisposition as a life threatening condition.

Secondly, this research identified that participants had to adapt to their new condition the same way that patients usually adapt to chronic illness (Samson & Siam, 2008). Specifically, participants had to reconstruct certain aspects of their lives that were affected by their test results: Medical/physical, emotional/psychological, and social/relational. In order to achieve this, participants used active coping skills, such as attending appointments or educating themselves; acknowledging the positive impact of having the knowledge that they carried the genetic mutation; and taking advantage of the social support offered to them through family, friends, and medical staff.

In other words, after receiving the positive test result indicating that they carried a mutation in the BRCA1 or BRCA2 genes, participants often engaged in three different tasks with the goal of regaining control and seeking to reclaim a sense of normality in their everyday lives. The qualitative analysis of the participants’ experiences suggested that participants underwent a genuine life transition that required an effort of adaptation to a completely new medical reality.

The virtual aspect of participants’ medical condition may explain why there were no particular effects on participants’ work or career, and henceforth no implication for the vocational task.

Finally, the qualitative analysis suggests that undertaking the tasks functioned in synergy. In other words, the completion of one task may facilitate the completion of another task. For example, undertaking the physical task helped participants to regain a certain sense of control over their lives, which in turn was helpful in returning to an emotional equilibrium.

Previous research suggests that women who have discovered that they carry a mutation in the BRCA1 or BRCA2 genes often report many negative psychological effects (Croyle et al., 1997; DiMillo et al., 2013; DiProspero et al., 2001; Dorval et al., 2000; Esplen et al., 2009; Lerman et al., 1995; van Oostrom et al., 2003). Although the participants in this study described similar negative effects from having received their test result, this research also found that participants were able to learn from this experience and overcome or learn to cope with many of the negative aspects of receiving this test result. More specifically, despite feeling negative emotions, participants were also eventually able to see a positive aspect in knowing they carried this genetic mutation (Croyle et al., 1997; Dorval et al., 2000; Lerman et al., 1995; van Oostrom et al., 2003). Furthermore, although participants reported feelings of rejection and vulnerability, they also described seeking support, and using resources, such as friends and family, to undertake the social task (Esplen et al., 2009).

**Conclusion**

This is the first time that the task-based model approach has been applied to the experiences of women who carry mutations of the BRCA1 and BRCA2 genes. This will help to provide a comprehensive point of view of patients’ reality and the challenges that they face as a result of living with this genetic predisposition. In conclusion, this research has contributed to the understanding of these women’s experiences. Specifically, even though these women live with the possibility of developing breast cancer, their experiences mirror those of individuals living with a chronic illness, and they must therefore adapt accordingly in a physical, psychological, and social manner.
**Practice implications**

The findings of this research underline the importance of a proper follow-up for women who are given the news that they carry mutations of the BRCA1 or BRCA2 genes. More specifically, health care professionals should pay special attention to the psychological impact of such a test result. Also, the comprehensive task-based model is a very useful tool as it provides professionals with an overview of what aspect of these women’s lives are affected by living with this genetic mutation. Through utilizing this model, health care professionals can ensure that they will take into account all aspects of the patient’s experience, especially aspects that need to be reconstructed due to the impact of the genetic predisposition.

**Disclaimer**

I confirm all patient/personal identifiers have been removed or disguised so the patient/person(s) described are not identifiable and cannot be identified through the details of the story.

**References**


### Appendix

**Verbatim**

<table>
<thead>
<tr>
<th>Physical Task: Attempting to Limit the Impact of the Test Result</th>
<th>Psychological Task: Living with Uncertainty</th>
<th>Social Task: Finding Effective Support</th>
</tr>
</thead>
<tbody>
<tr>
<td>I basically went home, and we cleaned out the cleaners.</td>
<td>...as far as just knowing?</td>
<td>There could have been</td>
</tr>
<tr>
<td>My husband and I researched, uh the chemicals that are in, um, our products, to see, you know the; there's so many products, chemicals that imitate hormones, especially estrogen... I thought well, with the girls, they're [young], I should start now. 'Cause for me, in my case, I'm [older], whatever happened in the past, I can't change. But, I knew I could do something for them.</td>
<td>it empowered me more to instead of just, you know, floating along and thinking I'll be alive 'til I'm 80 something.</td>
<td>that day? Like I would wait, you know, waiting</td>
</tr>
</tbody>
</table>

*Um, there's a, Facebook has a, a bunch of different*... *Ummm, not... for*... *So I, as soon as she, as soon as she was diagnosed with*...
| groups where people blog, | awhile. Maybe not until I | breast cancer I actually felt |
| and they just talk about | was done having kids, | a bond with her. And then |
| their different experience, | because I didn’t want... I | to find out after; because |
| and so on... Um, I’ve read | didn’t want the pressure of | when she would talk to me I |
| some of them, and yeah, | like- should I have them | already knew all this stuff, |
| some of them, uh, I could | sooner? Closer together? | and of course she, she didn’t |
| see where we have | Should I... I didn’t want to | know I was BRCA2 positive. |
| similarities about, how we | be considering, you know, | Um, until pretty much when |
| want to change our lifestyle, | they’ve already started to | she was through all her |
| and, some of them I just | talk to me- you know, when | chemo and radiation and |
| don’t seem to, to get. You | you’re done having your | then, and then she started |
| know they...To relate to | kids, you should have your | talking about getting tested |
| them. Um, I would like to | ovaries removed, and that | and stuff and that's when I |
| meet other women who | sort of thing. And, I didn’t | told her; ’cause like I said I |
| have, um, this gene | want to deal with that? | don’t tell people that I’m |
| mutation, just to find out | (laughs); I don’t tell | people... Well I thought that |
| what they’re doing, you | | she could- yeah exactly. I |
| know- people that are local, | | thought she would |
| um, maybe if there’s a | | understand... |
| support group somehow | | |
| that we can discuss the | | |
| different, um...options | | |
| together, and decide, you | | |
know, just to have a support
group, but um, that's yeah.

So, yeah. I guess, I guess, I
think I've known about it
but it's been very much like
back burner stuff. And also
I was having children, I was
breastfeeding, and, I didn't
want to do any testing until
that [process]; that part of
my life was over. Because I-
there was nothing I was
going to be able to do in
terms of, like prophylactic
surgery or, or even I can't
even get, be properly,
couldn't be properly
screened when I was breast
feeding, so, I waited until I
was pretty much at the end
of that and then I've started
into the screening process.