“Awakening to” a new meaning of being at-risk for arrhythmogenic right ventricular cardiomyopathy: a grounded theory study

April Manuel · Fern Brunger

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Abstract Efforts of social scientists to understand how individuals living in a family at risk for a genetically linked condition make health care decisions, having brought to the forefront the contextual nature of risk perception. Using a grounded theory approach, this study examines the experiences of 29 individuals living in families at risk for arrhythmogenic right ventricular cardiomyopathy (ARVC). Attention is paid to how individuals (re)construct the meaning of being at risk in relation to the developing science of gene discovery. Findings highlight that individuals living in a family at risk for ARVC juxtapose existing scientific knowledge against experiential knowledge as they “awaken to” the fact that they or a family member are at risk. This process is pragmatic and fluid and contingent upon whether and how symptoms are aligned with the constructed image of the *at-risk relative*.

Keywords Risk perception · Arrhythmogenic right ventricular cardiomyopathy · At-risk relative · Predictive genetic testing · Grounded theory

Introduction

Social scientists have started to examine how the availability of genetic testing shapes the lived experiences of at-risk people and, in turn, their health care decisions. Researchers have proposed the “experiential paradigm” (knowledge gained through social relationships or by living through the phenomenon) as a complementary framework to traditional biomedical models of science in order to understand how risk is conceptualized (d’Agincourt-Canning 2005). Although research does address the notion of being at-risk in a genetic context, little attention has been paid to how the embodiment of risk is shaped and reshaped alongside evolving genetic discovery.

The purpose of this article is to examine how individuals living in families at risk for arrhythmogenic right ventricular cardiomyopathy (ARVC) in the province of Newfoundland and Labrador (NL) juxtapose scientific knowledge against experiential knowledge as they “awaken” to the fact that they or a family member are potentially at risk for this fatal heart condition. This work is unique in that it moves beyond isolated descriptions of risk perception and examines how ideas and meanings about risk emerge and shift alongside the discovery of causative gene for ARVC—a period starting with clinical testing in the early 1980s and concluding in 2007 with a definitive predictive genetic test for ARVC (Memer et al. 2008). Given the intense focus on genetic research and discovery in NL (Rahman et al. 2003), an understanding of how individuals become aware and cope with their risk is critical in order to develop health care resources reflective of the needs of this population.

Our approach to analysing the concept of risk follows that of Lupton (1999) and other critical scholars of risk perception. We take risk to be a socially constructed concept with multiple meanings ascribed by both the one “at” risk and the one measuring risk, with those meanings continually in flux and potentially contradictory even when held by the same individual

A. Manuel
Memorial University, St. John’s, Newfoundland and Labrador,
Canada

A. Manuel (✉)
Memorial University of Newfoundland School of Nursing, 300
Prince Phillip Drive, St. John’s, Newfoundland and Labrador A1B
3V6, Canada
e-mail: admanuel@mun.ca

F. Brunger
Division of Community Health and Humanities, Faculty of
Medicine, Memorial University, St. John’s, Newfoundland and
Labrador, Canada

at one given point in time. The meaning of risk is developed in relation to historical, social and cultural contexts. The phrase “at-risk” throughout this article is used to indicate an increased risk for a condition identified by ones’ family history or through genetic testing.

Arrhythmogenic right ventricular cardiomyopathy

To date, there are 13 ARVC causative genes known (teRijdt et al. 2014). ARVC5 is a fully penetrant autosomal dominant heart disease in NL that results in a sudden cardiac death chiefly in young males (Hodgkinson et al. 2009). The gene for ARVC is located on 3p25 and is the result of a missense mutation in TMEM43 p.S358L found in cardiac tissue (Merner et al. 2008). The function of the TMEM43 gene is not entirely understood; however, it is thought to be part of the adipogenic pathway regulated by PPAR γ . A dysregulation in this pathway leads to the replacement of the myocardium with fibro-fatty material on the ventricle wall that decreases the cardiac myocytes’ ability to function properly, predisposing individuals to fatal ventricular arrhythmias (Hodgkinson et al. 2009; Marcus et al. 1982; Merner et al. 2008). Management of ARVC includes the insertion of an implantable cardioverter defibrillator, pharmaceutical interventions, restricted physical activity (Gollob et al. 2011) and, in severe cases, a heart transplant.

Globally, the prevalence of ARVC is approximately between 1:1000 and 1:5000 (Gollob et al. 2011; Thiene et al. 2007), with NL being closer to the former ((Hodgkinson, personal communication, October 29, 2012). Merner et al.’s (2008) research on ARVC in NL reported that the median age of phenotypic expression of ARVC was 32 years of age in males and 44 years in females, with full penetrance in males by age 63 and females at age 76. The median life expectancy of affected males was 41 and 71 years in affected females. The relative risk of dying was 6.8 times greater in ARVC-affected males by comparison to ARVC-affected females. Up until 1998, when DNA haplotype analysis for ARVC gene became available in the USA (Ahmad et al. 1998), diagnosis of ARVC was based on clinical diagnostics. It was not until the causative gene for ARVC was isolated 2007 by a NL research team (Merner et al. 2008) that health care practitioners could diagnose ARVC with certainty using a predictive genetic testing. This paper describes how individuals (re)construct the meaning of being at risk alongside the developing science of gene discovery.

Laypersons’ construction of risk

The literature on patient and client perceptions of risk in relation to clinical genetics illustrates that risk is an evolving social process (Sivell et al. 2008). The literature emphasizes that risk perception, being dependent on the components of one’s experiential knowledge at a

particular point in time, is both transient and contextual. Laypersons construct their ideas about risk in reference to multidimensional factors outside the realm of numerical labels, drawing on the subjective nature of risk (Binedell et al. 1998; Brunger and Bassett 1998; Cameron et al. 2009; Cox 2003; Cox and McKellin 1999; d’Agincourt-Canning 2005; Etchegary 2006a; McAllister 2002, 2003; Norris et al. 2009; Shiloh and Saxe 1989; Smith et al. 2002). An individual’s awareness of being “at-risk” is shaped by a multitude of factors including past experiences, commonsense practical knowledge, personal values, personal theories of inheritance, disease patterns, growing up in an at-risk family and stories of what constitutes someone at risk (Brorsson et al. 1995; Cameron et al. 2009; Cox and McKellin 1999; d’Agincourt-Canning 2005; Davison et al. 1991, 1992; Etchegary 2006a, 2006b, 2010; Etchegary and Perrier 2007; Finkler 2001, 2005; Hall et al. 2007; Hallowell et al. 2006; Hunt et al. 2000, 2001; Katapodi et al. 2004; Kenen et al. 2003; Marteau et al. 1995; McAllister 2002, 2003; Senior et al. 2002; Shedlosky-Shoemaker et al. 2010; Sivell et al. 2008; Weiner and Durrington 2008). Research has also shown that the meanings assigned to being at risk are transient in nature, oscillating between something non-existent to a heightened state of awareness or relevancy depending on the significance of a critical event (e.g. onset of symptoms) to the individual at risk (Cox and McKellin 1999; Etchegary 2006a, 2006b).

While most of the literature does not attend specifically to the social construction of risk amongst patients or families living with cardiac disease, messages about “risk” that have been constructed within the medical model of cardiac disease are clear. In particular, the notion of a “coronary candidate” (Davison et al. 1991)—that is, the typical individual that develops cardiovascular disease, who is obese, who is under stress, a smoker, and inactive—illustrates how specific meanings attributed to being at risk for and managing heart disease are conveyed (Davison et al. 1989, 1991, 1992; Emslie et al. 2001; Hunt et al. 2000; Walter and Emery 2005; Weiner 2009). Several studies have highlighted how individuals juxtapose the notion of the coronary candidate against biomedical models of heart disease and their own life experience to understand their risk status, make lifestyle changes, and/or dismiss ideas about risk (Angus et al. 2005; Chan et al. 2011; Farrimond et al. 2010; Frich et al. 2006; Hunt et al. 2000; Marteau et al. 1995; Walter et al. 2004; Walter and Emery 2005). Not surprisingly, perception of risk for heart disease is also shaped in part by the existence of visible physical or measureable factors associated with heart disease (e.g. age, chest pain and cholesterol) (Angus et al. 2005; Chan et al. 2011; Emslie et al. 2001; Farrimond et al. 2010; Frich et al. 2006; Marteau et al. 1995; Weiner and Durrington 2008).

Materials and methods

A grounded theory approach (Glaser and Strauss 1967) was used to guide the study. A detailed overview of the method is provided in previous work (see Manuel and Brunger 2014). Inclusion criteria included individuals who were able to communicate in English, were able to understand the purpose of the study, were living in a family at risk for ARVC (e.g. spouse, sibling), and had engaged in clinical or genetic testing for ARVC. Theoretical sampling guided the recruitment of 29 individuals (Glaser 1978, p. 37). Participants were recruited through: (1) a genetic counsellor at the NL Provincial Medical Genetics Programme and (2) snow ball sampling, whereby individuals heard about the study from existing participants and self-referred to participate. Interviews included nine individual interviews, three focus groups ($n=4$; $n=12$; $n=5$), and five follow-up interviews for a total of 34 data sources (see Table 1 for sociodemographics). Eight spouses of ARVC-positive individuals were included in the focus group interviews. Taped interviews lasted 45 to 90 min in duration.

The sample was comprised of 29 participants who had experienced the genetic testing process for ARVC (including the eight spouses). Of these 29 individuals, eight participants had engaged in clinical investigations for heart disease in the 1980s and continued with haplotype testing in 1998, up to the discovery of the ARVC gene in 2007. Four participants became involved with clinical testing between 1994 and 1997. Three participants experienced the genetic testing process in 1998, a time when haplotype analysis was only offered outside Canada. Ten participated in genetic testing in 2005, after haplotype analysis had become available in NL. Three participants had genetic testing post-discovery of the ARVC gene in 2007, and one participant was waiting to have testing.

Compliance with ethics guidelines

All procedures followed were in accordance with the ethical standards of the Newfoundland and Labrador Provincial Health Research Ethics Authority (2010), Canada's Tri Council Policy Statement: Ethical Conduct for Research Involving Humans (TCPS2 2010) and the Helsinki Declaration of 1975, as revised in 2000(5). Informed consent was obtained from all participants included in the study. Ethics approval was obtained from the provincial Health Research Ethics Board and the Research Proposals Approval Committee of the regional health authority. Confidentiality of participants was ensured by removing any identifying information in the transcribed verbatim.

Results

The theoretical construct *Awakening to a New Meaning of Being At-Risk* examines participants' earliest experiences of living in a family at risk for ARVC prior to being offered genetic testing. It describes the process by which at-risk individuals become aware of their risk (expressed by one participant as an "awakening"), the factors that impact this awareness, and the psychosocial and behavioural responses to the new meaning of risk with respect to clinical testing (1980s), post haplotype testing (1998) and the development of a definitive predictive genetic test for ARVC (2007). Embedded within this discussion is the experiences of the eight spouses; their experiences were similar to those affected individuals, solidifying the fact that the awakening process is not something that exists in silo. That is, it does not only impact the lives of those who are being tested for a genetic condition but the entire family unit. Two categories, (a) *making sense of numerous losses* and (b) *struggling to break the cycle of uncertainty*, capture the process of awakening to being at-risk.

Category 1: making sense of numerous losses

For the majority of participants in this study, including the spouses, the awareness of being at risk emerged at a time when the science of genetics had not yet emerged as an explanation for sudden cardiac death. Many participants knew from an early age that "heart problems ran in the family": "We've lived with it all our lives, knowing that there was something wrong". This sense of knowing that something was wrong was precipitated by the numerous accounts of illness and loss in the family that appeared to follow the same pattern. Watching history repeat itself over and over, participants worried about their own health at an early age: "My favorite uncle died in his 30s. Then my cousin's brother died at 26. From that time on, I worried that it was going to happen to me". Often, it was the sheer number of deaths in the family that sparked an awareness of being at risk: "I have three brothers, and three of them have passed away".

For others, the awareness that something was "going on" in the family was sparked by the news of a relative's death at such a young age: "The first indication that there was any sickness was my uncle. I knew he was having some trouble ... at Christmas, he died. He was 42". The circumstances surrounding the death often left a lasting impression on participants' sense of being at-risk: "It came to light when my brother died. My sister got married ... my brother walked her down the aisle; he dropped dead on the floor at the wedding". Incidences such as this would cause participants to discuss their families' history with each other and their spouses in effort to re-trace their family history of loss, as the idea that they were dealing with something more than ischemic heart disease emerged.

Table 1 Participant sociodemographics

Characteristic	Number of participants, <i>n</i> =29
Age	
15–20	3
21–30	0
31–40	8
41–50	8
51–60	7
61–70	2
71–80	1
Marital status	
Single	5
Married	20
Widow	3
Divorced	1
Employment status	
Employed	14
Unemployed	4
Retired	8
Students	3
ARVC status	
ARVC positive (males)	8
ARVC negative (males)	1
ARVC positive (females)	7
ARVC negative (females)	4
Pending testing (males)	1
Pending testing (females)	0
Inconclusive test	
Negative Spouses	8
Treatments	
ICD	14
Heart transplant	1
None	14
Location	
Smaller rural centre (health centre)	17
Larger rural centre	7
Urban centre	5
Education	
In high school	3
Completed some high school	17
Post-secondary school	9

Description of populations: Small rural centre (less than 400); Larger rural centre (between 9000 and 15,000); Urban centre (200,000)

Several participants emphasized the process by which they tried to normalize the fact that people die young in their family. As the number of deaths experienced in the family increased, it became clear that the deaths were not a coincidence and might have a genetic basis; however, during those years prior to identification of the causative gene, the technological

means to substantiate this belief did not yet exist. In response, participants reported, they struggled to normalize something that they knew intuitively was not normal:

You're kind of wondering about it [reason for deaths], but you don't acknowledge it.

Over time, participants' ideas about risk begin to shift from being a suspicion to a belief that they were at risk for a genetic heart condition. Based on their experiential knowledge, and in the absence of a scientific genetic explanation, participants spent considerable time trying to understand the meaning of being at risk for themselves and others in order to cope and make health care decisions.

Age, gender, physical signs of the disease, family history, modifiable lifestyle factors and the concern felt for children in the family were the most influential in constructing the sense of being at risk for all participants. Together, this list of factors led to the creation of the *at-risk relative*, a mental representation of the individual at risk for ARVC. This mental image of the at-risk relative was used as a reference point to construct ideas about risk, to make health care decisions, and to predict the likelihood of a cardiac event. Participants drew upon the factors of the at-risk relative that were most relevant and were aligned with their own beliefs about heart disease (and, later, about the science of genetics) to understand risk and to make health care decisions. Depending on the context, new factors deemed more relevant or "risky" emerged while other factors assigned a status of being "less risky" were ignored or dismissed and diminished in significance.

Age and gender were key components in developing a framework for assigning meaning to risk. The fact that young males were the target of this unknown condition was so apparent that it could not be ignored:

My first recollection was hearing that my grandfather had passed away at 49 As time went on, my uncle passed away. Their son passed away at 27. Another uncle went out fishing and drowned. My brother was 26 when he died with a heart attack.

As participants approached the age of 40, they experienced a shift from the abstract sense of risk to an embodiment of risk, causing a heightened sense of risk anxiety. This concern was precipitated by memories of observing their older relatives anguish as their own age of "being at-risk" drew closer:

I remember my father was always worried. I remember seeing him sitting on a chair with his head down on his arm. I think it was always there in his mind that something was going to happen to him; because at that time his second brother had died; and they were young.

For most participants, it is the onset of physical signs and symptoms thought to be cardiac in nature that often marks the entry into the illness experience and causes significant stress, “The first time I was aware that I probably had this heart condition was that syncope incident in the fall of 1993, when I almost blacked out”. In the case of the spouses, being notified that their significant other was ill was distressing but it was not overly surprising, as they were aware of realities of living in a family with a strong history of heart disease. This awakening to the idea that they were at-risk was something expected given the strong family history of loss. For others, this was not the case, as they continued to struggle to understand why so many family members were dying.

Substantial psychological effort went into reconfiguring the mental representation of the at-risk relative to encompass the asymptomatic individual who did not meet the at-risk relative profile, as many relatives did not have any symptoms prior to death. Several participants spent considerable time monitoring themselves and others for any signs and symptoms that bore a resemblance to those experienced by the at-risk relative (e.g. chest pain, shortness of breath). The presence of signs and symptoms that were perceived as risky caused a heightened sense of risk, resulted in psychological distress, and marked the onset of the illness experience and a sense of urgency around symptom management, as captured in this statement by a spouse: “I am so afraid to take a chance that it’s just chest congestion and not something else, because [my son] said he feels tightness in his chest”.

Despite witnessing and listening to numerous stories of people who had succumbed to heart disease, many of the older participants took a pragmatic approach to understanding their risk, given the lack of available scientific technology that had been available in the 1980s to explain what was happening. Given that science did not hold any answers, many took the stance that there was no point in dwelling on it, “It’s like being in a house and you don’t know if this place is haunted, but there’s weird stuff going on so I won’t look”.

Others normalized their own risk (“[I felt that the symptom] was just once or twice a year and could happen to anybody”) or normalized their families’ risk, arguing that they were not dissimilar from many other normal families and thus required no immediate intervention:

We had no sickness like heart, no more than anybody else, probably less than some. We used to say we thought we were pretty good; pretty well off because we didn’t have any major problems or anything. There was nothing wrong with us.

Through the interviews, it was clear that as participants struggled to understand and generate an explanatory framework for their risk, most engaged in what Davison et al. (1991, 1992) describes as the work of a lay epidemiologist,

gathering information through observations or reports about the factors that predispose one to be at risk for heart disease. The explanatory models created by the lay epidemiologist are juxtaposed against existing beliefs about the factors that influence risk (e.g. age of onset, gender) and the at-risk relative profile. Participants made mental notes as to the number of cases of heart disease; they searched for predisposing events and they collected data on the array of health outcomes within the family:

I knew something was going on; but not until my brother really sat down and talked about it did I start to make connections. It was always men that were involved. Mom’s brothers had all passed away young. She had a sister who died suddenly at 49; her two brothers were on their way back from a funeral, had an accident, and died. We wondered if the one who was driving had a heart attack.

What was particularly stressful for participants was the knowledge that they too (or their significant other, in the case of spouses) could have a fate similar to that of their relatives, “I knew that I could drop down dead having this condition [ARVC] ... my father had died with this same condition, so I knew it was a possibility”.

Based on the conclusions of the lay epidemiologist, participants made the choice to modify their lifestyle or not. Depending on the decision made, they would either live as they had before, now with the knowledge that they may have a fatal condition, or they would alter their lifestyle in reaction to the new risk knowledge. For several participants who chose the “normality” path, the knowledge of being at-risk was stressful: “The stress that I’d gone through trying to go out and live life knowing that I had something that could kill me where I could drop dead was more stressful”.

Of those who recognized the potential for modifying lifestyle as a means of managing risk status, a healthy diet and regular exercise were cited as being critical to avoid the fate of the at-risk relative: “I gave up drinking, I gave up coffee, and I tried to be physically fit”. However, physical activity had two different connotations in relation to modifying risk status. On one hand, regular physical activity was associated with lessening risk:

Growing up, you think to yourself, there is heart disease in the family so you take care of yourself. I eat lots of oily fish; exercise, play basketball, volleyball, and try to stay in shape.

On the other hand, when participants became aware of individuals who had died during physical activity, the mental representation of the at-risk relative shifted to include exercise as something deemed to be a risky behaviour that must be stopped. This new understanding of risk was hard for many

to assimilate into their existing framework, given that physical activity traditionally is an activity synonymous with heart disease prevention.

As participants awaken to the meaning of being at-risk themselves, they also experience an awakening to the fact that their children are at-risk: “Every time one of the youngsters said they felt weak or sick I wondered, have they got this heart disease?” This concern results in diligent monitoring of the children for physical signs and symptoms of ARVC: “Every morning I got up, opened the door, and went in [to the children’s room]. As long as I saw the chest rising or they were breathing, I was fine”.

Thus, making sense of numerous losses—drawing on the explanatory models created through “lay epidemiology” and juxtaposing that knowledge against the factors that influence one’s own risk (age of onset, gender and so on) in relation to the at-risk relative profile—meant that the meaning of being at-risk was constantly fluctuating with variations in observations about family members and changing individual risk factors. For participants who awakened to the meaning of being at risk at a time when the science of genetics had advanced, the (re)construction of risk status was far more complex.

Category 2: struggling to break the cycle of uncertainty

While receiving a professional’s suggestion that they may have a genetic-linked disease in fact merely added credence to what participants already knew, many struggled with trying to give new meaning to being at-risk from a *genetic* stand point, particularly in the absence of a definitive diagnosis, consistent treatment or visible signs of the disease. Participant experiences varied, influenced by experts’ knowledge and care, available technology and the meanings assigned to the at-risk relative.

As participants engaged in diagnostic clinical testing (e.g. electrocardiograms) in the early 1980s, they found themselves looking to biomedical models of disease causation and the opinions of experts to help them (re)construct their “reality” of their risk status. Unfortunately, experts in the field did not have all the answers: “I remember asking the cardiologist, ‘What do you think I should do?’ He said ‘I honestly don’t know’”. Participants reported feeling disappointed and emotionally drained when, despite engaging in a myriad of clinical tests, results were inconclusive and they felt no closer to understanding their risk:

I had seven tests done [...] because they could not stimulate an arrhythmia [...] [the cardiologist] told me he didn’t see any need of putting me on medication or doing electrophysiological studies [...] ‘Just go on home and live your life as normal’.

A sense of relief was described for some participants who did receive a negative result, as it implied that everything was fine; however, for others, this was not true: “Still in the back of my mind I was thinking that if this could happen to my brother then it could happen to me”. This sense of uncertainty was also shared by the spouses. This response is not surprising in a context of repeated losses of family members, at a time when diagnostic tests were under development, and given the lack of clinical knowledge and management of ARVC in the earlier days of genetic science, as captured in this narrative:

If you’re going to three or four doctors you are getting different things from all of them. You really don’t know which one to believe; you just kind of got to hope for the best, really. I think it’s all a luck thing.

Added to this sense of uncertainty over the significance of clinical or genetic testing findings was the feeling by many that they were not being taken seriously by clinicians despite their strong family history: “The first time I went to the hospital [...] I told them I was having a heart attack, the doctor laughed and said, ‘You’re too young for that’”. Participants consistently reported that they appreciated the fact that gene discovery does take a long time, that health care professionals were providing the best treatment they could given the current state of knowledge and that this rare cardiac condition does pose challenges to current biomedical models of heart disease. Nonetheless, they reported feeling great frustration with the inability of clinicians to confirm (or deny) the validity of their embodied sense of risk.

Participants’ beliefs and awareness about risk were (re)shaped in light of new technological advancements and research on ARVC. The experiences of participants who had become aware of their risk for heart disease post haplotype analysis (1998) were different from those who had been living with the sense of unknown risk for years prior to the 1980s, and from those who received testing after 2007 when a definitive predictive genetic test for ARVC became available. When participants became aware of the availability of a predictive genetic test, their beliefs about risk were no longer solely contingent on their experiential knowledge but developed in conjunction with scientific knowledge.

At times, scientific knowledge took precedence and experiential knowledge was ignored or dismissed:

I balled up the paper [consent for procedure] and threw it in the garbage and said, “I am not getting that done”. I was frightened to death. I just sat there and said, “well I have to get it done”, and so I took the paper out, smoothed it out, and signed my name.

By contrast, when science could not answer participants’ questions, or in times of distress regardless of genetic status or state of existing scientific knowledge

about ARVC, participants turned to their experiential knowledge to try and make sense of what was going on and to make choices about their health: “My father died at 44. My grandfather died at 42. As I start to inch up in age I told myself, ‘I’m going to go and get tested when I am 40’”.

Equipped with the framework of the at-risk relative, participants engaged in testing in hopes to confirm or discount their risk based on how similar they were to the criteria of the at-risk relative. What was disappointing was that despite engaging in a multitude of clinical tests, some were no closer to answering their questions: “They worked on ultrasounds and hooked me up to the heart machine and stuff like that, but they couldn’t pick anything up”. At times when participants perceived that science in its current state did not have the knowledge to provide the answers they wanted, they turned to their own experiential knowledge to cope and manage their risk, drawing on lay stories of loss and the at-risk relative to create their own interpretations of the meaning of risk and approach to disease management.

Family history and interactions with experts were significant factors in shaping and reshaping participants’ perceptions of being at-risk. As scientific advances with gene discovery were made, participants began to incorporate a genetic component into their conception of the at-risk relative, refracting their experiential knowledge through a genetic lens. A key moment in that process was when the genetic counsellor or cardiologist first approached a participant with the information that they may be at risk for a genetically linked heart condition. This confirmed what many of them had started to entertain as a logical rationale for the numerous losses within the family:

He [relative] did say at the time it could be genetic; and then a couple of years later I received the call from [genetics counsellor]. I think that’s really when I realized it could be genetic and there could be something to it.

For some participants, awareness that they may have a genetically linked condition was the result of discussions with other relatives and friends under investigation for heart disease: “I found out about ARVC when my aunt started showing some kind of symptoms. She came to me and said, ‘You got to go for testing’”. For others, the reshaping of risk in light of a genetic cause was something that evolved with the news of relatives falling ill, in a pattern too obvious to ignore: “While mother was in the hospital, her cousin, and her nephew got admitted, and somebody had the foresight to say there might be something going on with genetics there”.

Discussion

The construct *Awakening to a New Meaning of Being At-Risk* captures how individuals living in a family at-risk become aware of and respond to an emerging sense of risk alongside novel gene discovery. Similar to the findings of other studies (Cox and McKellin 1999; d’Agincourt-Canning 2005; Etchegary 2006a; McAllister 2002, 2003; Taylor 2005), participants juxtaposed their experiential knowledge against their scientific knowledge in assigning meaning to their sense of risk. Key to this juxtaposition is what we refer to as the at-risk relative, which serves as a reference point to construct ideas about risk, to make health care decisions, and to animate discussions amongst family members as to who is at risk and why (similar to the construction of the coronary candidate depicted in the cardiovascular literature [Davison et al. 1989, 1991, 1992]). The characteristics of the at-risk relative are constantly reconfigured to reflect participants’ knowledge about heart disease and genetics specific to one’s personal life context (e.g. age, gender, family history of loss, physical signs of the disease) and in relation to the state of the science.

The components of the at-risk relative were acquired and gradually became more pronounced as participants engaged in the awakening process. It is the awakening to the presence of the at-risk relative that sparks a shift from what was once an abstract sense of risk to what Cox and McKellin (1999) refer to as an “intersubjective awareness” of risk. This process was not easy and caused a significant amount of psychosocial distress as participants tried to decipher and assign meanings to competing ideas about risk, similar to that experienced by participants in other studies (Braithwaite et al. 2004; Cox 2003; Cox and McKellin 1999; d’Agincourt-Canning 2005; Etchegary 2006a, 2006b, 2009, 2010; Frich et al. 2006; Hall et al. 2007; Hallowell et al. 2006; Hunt et al. 2000, 2001; Marteau et al. 1995; McAllister 2002, 2003; Ponder et al. 1996; Walter and Emery 2005; Walter et al. 2004; Weiner and Durrington 2008; Weiner 2009; van Maarle et al. 2003).

Knowing this, it is important that health care providers create a relational space (e.g. common meeting place) so that health care professionals and these at-risk individuals can discuss freely how their awakening to the notion of being at risk unfolds. Such a venue will provide an opportunity for individuals, family members and experts to develop relationships, discuss competing ideas about the at-risk relative, review existing treatment options and develop a collaborative approach to care. This plan needs to be fluid; it needs to account for how individuals’ ideas about risk shape and evolve, and it needs to be tailored to the needs of each individual and family. It must take into consideration how the awakening period may differ depending on the state of science and individuals’ everyday life experiences. Furthermore, it needs to reflect the fact that this “awakening period” does not happen spontaneously but is a process that emerges over time and is

facilitated by the fostering of a trusting therapeutic relationship with members of the interdisciplinary team (e.g. geneticists, genetic counsellors, cardiologists, nurses, social workers and psychologists).

Limitations

This study was based on participants' retrospective accounts of their experiences of awakening to being at-risk. Therefore, the full range of types of experiences may not be fully captured. Second, the sample was comprised of middle class, employed and educated individuals within one Canadian province; hence, the findings may not be fully generalizable, particularly in terms of socioeconomic status. Third, the sample did not have any participants between 21 and 30 years of age, a time when life decisions are often made.

Conclusion

This research demonstrates that risk perception is pragmatic and fluid. It is pragmatic in that participants assign meanings to factors in their everyday lives in order to understand and manage their risk. It is fluid in that risk perception is ever changing in response to one's experiences and in juxtaposition with the state of scientific knowledge. Knowing the impact that experiential knowledge has on the awakening process and the formulation of the at-risk relative, in the case of rare or new conditions such as ARVC, it is important that health care providers acknowledge the origins of existing competing explanatory frameworks in their provision of information and care to clients and patients. It may be useful to include lay persons in decision making around research design, care planning and policy development. The experiences of at-risk individuals are crucial to understanding the embodied experience of genetic risk.

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Compliance with ethical guidelines All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with Helsinki Declaration of 1975, as revised in 2000(5). Informed consent was obtained from all patients for being included in the study.

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