HHS Public Access

Author manuscript

Soc Sci Med. Author manuscript; available in PMC 2023 May 01.

Published in final edited form as:

Soc Sci Med. 2022 May ; 301: 114905. doi:10.1016/j.socscimed.2022.114905.

Embodied risk for families with Li-Fraumeni syndrome: Like electricity through my body

Allison Werner-Lin, PhD, LCSW[a,b,*],

Rowan Forbes Shepherd, PhD[b],

Jennifer L. Young, PhD, AMFT[c],

Catherine Wilsnack, MSW, LMSW[d],

Shana L. Merrill, MS, LCGC[a,e],

Mark H. Greene, MD, MPH[b],

Payal P. Khincha, MBBS, MSHS[b]

[a]School of Social Policy and Practice, University of Pennsylvania, Philadelphia, PA, USA

[b]Clinical Genetics Branch, Division of Cancer Epidemiology and Genetics, National Cancer Institute, Rockville, MD, USA

[c]Stanford Center for Biomedical Ethics, Stanford University, Stanford, CA, USA

[d]Steve Hicks School of Social Work, The University of Texas at Austin, Austin, TX, USA

[e]Hospital of the University of Pennsylvania, Philadelphia, PA, USA

Abstract

Introduction: Experiences of illness change the physical body and embodiments, or the ways in which the world and the self are known through the body. When illness is anticipated, such as with inherited cancer predisposition syndromes, risk becomes embodied and shared in family groups. Embodied risk is experienced whether or not symptoms have manifested. To examine how individuals and families with genetic risk experience the world and understand their disease through their bodies, we employ Li-Fraumeni syndrome (LFS) as an exemplar. LFS is a rare, genetic, cancer predisposition syndrome with nearly 100% lifetime cancer risk starting from birth, limited opportunities for prevention, rigorous screening protocols, and early mortality.

Methods: Forty-five families, including 117 individuals aged 13–81 years, enrolled in the National Cancer Insitute's LFS study (NCT01443468) completed 66 open-ended interviews

[*]Corresponding author. School of Social Policy and Practice, University of Pennsylvania, 3701 Locust Walk, Philadelphia, PA, 19104, USA. awer@upenn.edu (A. Werner-Lin).

Credit statement

Allison Werner-Lin: Conceptualization, Formal analysis, writing original draft, Supervision, Methodology. Rowan Forbes Shepherd: Conceptualization, Formal analysis, writing original draft. Jennifer L. Young: Investigation, writing review & editing, Methodology. Catherine Wilsnack: Formal analysis, Project administration. Shana L. Merrill: Formal analysis. Mark H. Greene: Investigation, Resources, writing review & editing. Payal P. Khincha: Investigation, Resources, writing review & editing, Funding acquisition, Methodology.

Declaration of competing interest

The authors have no conflicts of interest to declare.

regarding LFS experiences. An interdisciplinary team used modified grounded theory to explore physical aspects of living with LFS in psychosocial contexts.

Findings: The physicality of living with LFS included constant monitoring of LFS bodies across the family to identify physical change that might indicate carcinogenesis. Cancer screening, risk reduction, and treatment acted as dually protective and invasive, and as an unavoidable features of LFS. Connections between family members with similar embodiments normalized aesthetic changes and supported coping with visible markers of difference. In some circumstances, participants objectified the body to preserve the self and important relationships. In others, intense pain or loss created thresholds beyond which the self could no longer be separated from the body to support coping.

Discussion: This paper focuses on Li-Fraumeni syndrome, a familial condition with a well-established genetic identity in which the body-self is experienced in relation to important others, to medical imaging, and to historical experiences with cancer. We expand on theories of embodied risk and inter-embodiment to describe experiences across disease trajectories, with attention to division and union between body, self, and other.

Keywords

Li-Fraumeni syndrome; *TP53*; Hereditary cancer; Cancer; Family; Embodiment; Dis-embodiment

1. Introduction

Embodiment is the process of knowing the world through one's body and how the body shapes and is shaped by social, cultural, political, and historic forces (Merleau-Ponty, 1962; Wilde, 2003). Theories of embodiment assume the mind and body are not separate—we do not 'have' bodies, we 'are' our bodies—and they provide a framework to understand how the ordinary, taken-for-granted practices of everyday living are experienced through and with the body (Leder, 1984).

Recognizing humans as both having and being a body (*i.e.,* 'bodyselves') (Ellingson and Borofka, 2020) means that serious illness, such as cancer, changes not only our physical bodies but also our embodiments, particularly 'movement in space, time, language, sexuality, emotions, and perception' (Wilde, 1999: 27). Our bodies tell stories about experiences of illness and well-being that allow us to constitute certain identities (*e.g.,* being seen as a cancer patient due to hair loss from treatment) or restrict us from other identities (*e.g.,* being seen as *dis*-abled due to paraplegia), and we read stories in the bodies of others that help us constitute their identities (Field-Springer and Margavio Striley, 2018). Thus, embodied knowledge is constituted and negotiated through engagement with the bodies of others, or, as Springgay (2008) argues, 'how we come to know ourselves and the world around us, our subjectivity, is performed, constructed, and mediated in relation with other beings' (p.22).

1.1. Embodiment of genetic risk

In late modernity, risk has become central to health discourse in Western society, especially in public health with its emphasis on disease prevention and health promotion (Lupton,

1995; Petersen, 1996). In turn, health is increasingly described at the level of the individual, in terms of individual responsibility and agency to control health risks (Hallowell and Lawton, 2002: 424). Under these conditions, the body is problematized as a clinical object over which individuals have autonomous control (Fitzpatrick, 2000; Howson, 1998b). Individuals are personally responsible for achieving a 'healthy body' and reducing disease risk by restraint, surveillance, and lifestyle modification (Aronowitz, 1998; Fitzpatrick, 2000; Williams, 1998), 'for the sake of his or her own health as well as the greater good of society' (Lupton, 1993: 429).

Environmental health risks (*e.g.,* pollution) and health risks consequent of lifestyle options (*e.g.,* smoking or poor diet) are considered external to the body and avoidable or modifiable by the individual (Kavanagh and Broom, 1998; Lupton, 1993). In contrast, heritable or genetic disease risk 'is located in the body of a person said to be at risk' (Kavanagh and Broom, 1998: 437). Risk is part of the individual's DNA and is, thus, constitutional to the body from birth to death. Genetic risk information, however, 'is predictive not prophetic' (Werner-Lin, 2007: 335). Asymptomatic individuals with a known genetic predisposition to cancer, for example, may have no bodily evidence of growing disease and may never develop cancer; yet, being 'at risk' means they live in anticipation of a diagnosis and may undertake drastic modification (*e.g.,* risk-reducing surgery) and surveillance protocols (*e.g.,* cancer screening) to stay healthy (Werner-Lin et al., 2020).

Being 'at risk' of disease, therefore, becomes a unique disease state. Awareness of risk 'forces an awareness of the body as separate from self' (Garro, 1994, 782); the body becomes an object to be regulated, modified, and scrutinized to maintain health (Lupton, 1995; Petersen, 1996). Embodied experiences of the 'at-risk consciousness' are well-documented (Cox and McKellin, 1999; Howson, 1998b; Polzer et al., 2002; Robertson, 2000). Several investigators have described at-risk bodies as 'treacherous' (Robertson, 2000), 'risky' (Nettleton, 1997; Rees, 2018), or 'panic bodies' (Lupton et al., 1995a, 1995b), and, notably, for women undergoing genetic testing for hereditary breast and ovarian cancer risk, 'dangerous' bodies (Hallowell et al., 1998). Methods for containing, or managing, disease risk also bear implications for embodied experience. For example, women with a pathogenic *BRCA1/2* variant who elect risk-reducing oophorectomy (surgical removal of ovaries) experience the procedure as a threat to self-identity (Hallowell, 2000). Altering the materiality of their bodies to substantially reduce cancer risk meant becoming not just a woman with a 'disciplined body,' but a new person who looked, felt, and acted differently and who was seen as different by others, including loved ones (Hallowell and Lawton, 2002).

1.2. Shared embodiment in families with genetic risk

Intergenerational experiences of inherited cancer risk, diagnosis, and loss within families also bear significant implications for historically situated embodiment. What is learned about and by the body through multigenerational experiences with cancer may suggest unspoken, shared, and common expectations about the body or about ways of relating body, mind, and identity (Ogle and Ullstrup, 2006; Peile, 1998). In a sample of women and men from families with a pathogenic *BRCA1/2* variant, d'Agincourt-Canning (2005)

distinguished *embodied knowledge*, developed through a personal cancer diagnosis, from *empathic knowledge*, developed through witnessing loved ones' diagnosis and treatment course. Both forms of knowing contributed to how participants understood their present and future embodiments of cancer risk. Similar forms of knowing have since been reported as important contextual factors for women making prenatal screening and testing decisions (Boardman, 2017; Etchegary et al., 2008).

More recently, Jenkins et al. (2013) described 'inter-embodiment' as an 'interdependent embodied identity' that is shared among family members with familial hypercholesterolemia, a heritable genetic disease (p.537). Each body in the bloodline is genetically and historically linked; these linkages suggest reference points for individuals to interpret their own genetic test results as forms of either biographical re-enforcement or disruption, depending on the result. Experiences of inter-embodiment may have particular salience among families with hereditary cancer syndromes where early detection, diagnosis, and treatment are deeply physical and risk of cancer is a life-long threat.

Amongst hereditary cancer syndromes, embodied risk may involve: 1) the simultaneous possibility, or presence, of cancer now and the chance of disease and even premature death in the future (Kavanagh and Broom, 1998); 2) the ways we know and what we do to our bodies in the service of cancer prevention (*e.g.,* risk-reducing surgery), early detection (*e.g.,* screening), and treatment (Hallowell and Lawton, 2002); and, 3) what is learned about and by the body through a family's historical experiences and stories of cancer (*i.e.,* inter-embodiment) (Jenkins et al., 2013). Since the completion of the human genome project, research exploring embodiment as it relates to these components remains nascent, resulting in limited discourse through which to interpret and address embodied risk in clinical and empirical settings (Kavanagh and Broom, 1998). For rare hereditary cancer syndromes, like Li-Fraumeni syndrome (LFS), the focus of this study, embodied experiences of cancer and cancer risk for individuals and in family groups are yet to be explored.

1.3. The case of Li-Fraumeni syndrome

LFS is a rare, inherited cancer predisposition syndrome caused by pathogenic variants in the tumor suppressor gene, *TP53* (Malkin et al., 1990; Srivastava et al., 1990). 'Classic LFS' confers a lifetime cancer risk of nearly 100%, with syndrome-related cancers occurring at all ages, and high risk of developing multiple primary malignancies. LFS affects multiple organ systems, most frequently soft-tissue, bone, female breast, brain, and adrenal cancers, though others have been reported (Mai et al., 2016). By age 31 for women and age 46 for men, approximately 50% of individuals with classic LFS will develop at least one cancer (Mai et al., 2016). While most *TP53* variants are passed on from parent to child, approximately 7–20% are '*de novo*': *i.e.*, they spontaneously occur during embryonic development and are new to the affected family member and their descendants (Gonzalez et al., 2009; Renaux-Petel et al., 2018).

Since LFS-associated cancers are heterogeneous, optimal cancer risk management involves a comprehensive regimen of biochemical and imaging surveillance. Individuals are recommended to engage in intensive assessment to detect early-stage cancer and improve treatment outcomes. Widely accepted protocols include regular whole-body magnetic

resonance imaging (WB-MRI) in addition to brain, breast, and abdominal imaging from early ages (Kratz et al., 2017; Villani et al., 2016). Effective prevention is largely unavailable; risk-reducing mastectomy is recommended for adult women, but of modest potential utility to reduce syndrome-related morbidity and mortality given the wide spectrum of cancers that characterize the LFS phenotype[1] (Amadou et al., 2018). Notably, aggressive bone cancers associated with LFS may require amputation, sometimes at young ages (Evans et al., 2020).

Individuals with LFS, therefore, live with an almost certain and inescapable risk of cancer. Consequently, at-risk individuals, their providers, and their loved ones, may promote the necessity of ongoing cancer screening, surveillance, and risk-reducing surgeries (Forbes Shepherd et al., 2020, 2021). It is not uncommon for multiple family members across generations to have, or have had, cancer, undergo treatment, and face end-of-life simultaneously or with limited respite between one episode and the next (Forbes Shepherd, 2020; Oppenheim et al., 2001). These challenges have direct consequences for lived experiences of cancer risk.

Study aims.—To date, the 'body' of psychosocial literature on LFS is nascent and the study of embodiment absent. Despite recognizing the impact of cancer on embodiment, we have not taken advantage of theoretical understandings of embodiment to explore the intersection of genetic risk and the body in managing families with hereditary cancer. LFS presents an ideal setting to explore how individuals and families with significantly elevated genetic risk of cancer across multiple organ systems experience risk in their bodies and in their family groups. This article focuses specifically on physical and aesthetic aspects of living with LFS, how these are interpreted in extended family kindreds and environments, and what their impacts are on individual experiences and embodied identity.

2. Methods

This analysis was nested in the National Cancer Insitute's LFS study (NCT01443468); our broader investigation of the clinical, behavioral, epidemiological, and genetic etiology of LFS to inform global screening standards. The primary investigational screening tool was MRI because it avoids potentially carcinogenic ionizing radiation exposure from x-ray and mammography. MRI can image the entire body, head-to-toe, creating an opportunity to identify LFS-related cancers for which no alternate screening modality exists. Study participants and their first-degree relatives were offered genetic risk assessment, genetic counseling, genetic testing, and disclosure of genetic test results to inform clinical decision-making.

2.1. Recruitment and sample

In 2011, the Clinical Genetics Branch of the National Cancer Institute (NCI) began nationwide recruitment of patients and their families via the National Institutes of Health

[1]A phenotype is the observable trait produced by single or multiple genes in interaction with environments. Phenotypic variability refers to the range of visible or measurable symptoms associated with a specific gene or set of genes linked to a diagnostic category (https://www.genome.gov/genetics-glossary).

(NIH) and other healthcare providers, the NCI Cancer Information Service, professional healthcare organizations, interested laypersons and patient advocacy groups, plus family members themselves. Consented participants completed telephone screening to define the family's medical history. Each enrolled family member completed a rigorous, multispecialty baseline evaluation that included medical and mental health history assessments.

2.2. Data collection: multigenerational family group interviews

Participating families enrolled in the screening arm of the study visited the NIH Clinical Center annually to complete whole-body, brain, and breast MRI (for female participants with breast tissue), physical examinations, and psychosocial interviews. During center visits, a genetic counselor completed psychosocial interviews with each family member individually, and a marriage and family therapist conducted an interview with all presenting family members together as a group. The present study focused on the family as the unit of data collection and analysis. Recruitment efforts encouraged participants to define 'family' and identify who constituted 'family members' as they saw fit. This paper reports findings from the family group interviews.

Multiple members of the inter-professional LFS study team collaborated to build the family group interview guide, including a family therapist, genetic counselor, social work consultant, behavioral/psychosocial investigators, and oncological physicians. The guide included a semi-structured protocol focused on within-family communication issues, reproductive decision-making, couple relationships, cancer prevention behaviors, and choices regarding genetic testing for adults and children.

We invited all family members aged 13 and older attending the NIH Clinical Center to complete the family group interview. Interviews were audio-recorded with participants' permission. Researchers de-identified transcripts and replaced names with participant-selected pseudonyms prior to analysis.

2.3. Data analysis

We employed the tenets of grounded theory (Charmaz, 2006; LaRossa, 2005) and interpretive description (Thorne et al., 1997) for analysis. Grounded theory is an iterative research methodology in which data collection and analysis occur simultaneously. Interpretive description shares foundational methods of grounded theory (*e.g.*, concurrent data collection and anaylsis), while contextualizing biomedical and psychosocial data in pre-existing empirical and practice knowledge.

Over five years, 45 family groups completed 66 interviews in multiple configurations, including sibling groups, parent/child dyads, partners, and extended family units. Families completed one to five interviews during this period, totaling 66 interviews. The constellation of family members who attended the NIH Clinical Center varied annually based on study funding parameters and the family's priorities. Researchers created a family-based case file that included all transcribed interviews and the family pedigree.

The coding team was comprised of four researchers experienced in working with medical transcript data and trained in qualitative methods, a cancer genetic counselor, and the study

interviewer. To establish a codebook, the four researchers independently conducted open coding on the same transcript, identifying *in vivo* codes that emerged organically during interviews (Denzin and Lincoln, 2008) and *a priori* codes originating from sensitizing concepts (Bowen, 2006). They met to discuss codes, resolve discrepancies, and identify avenues for further inquiry. They then selected three other case files for maximum variation, and the same four investigators coded them all in round-robin style. The four researchers then met to examine the set of coded documents, compile a working list of codes, define decision rules for their application, and cluster codes into thematic categories. All transcripts were then loaded into Dedoose™ software and two of the four investigators double-coded all 66 interviews. In the final analysis phase, the coding team met to discuss interpretations and to synthesize findings into recurring patterns.

2.4. Data quality and rigor

The study team met weekly to discuss recent and upcoming families scheduled for annual screening visits. These meetings provided a forum to discuss concerns regarding participants' mental health, follow-up on unresolved questions, and cross-check information gathered from multiple sources. To ensure the codebook was applied consistently, two researchers coded transcripts in tandem to check each other's work. An experienced qualitative researcher and PhD-prepared social worker with expertise in hereditary cancer genetics provided feedback regarding thematic findings. The team employed several other methods of assuring rigor, including prolonged exposure (Roy et al., 2015), inter-professional collaboration, and triangulation with pedigree data.

3. Findings

3.1. Overview

Families presented in one of four combinations, including 26 couples, 19 parent-child dyads, 11 sibling groups, and 10 mixed-extended family groups (*e.g.,* grandfather and grandson dyad). A total of 117 participants aged 13–81 years participated, including 78 participants with a confirmed pathogenic *TP53* variant and 42 participants who attended in a supportive capacity. Interviews ranged from 21–81 minutes (mean 50 mins) and participants were predominantly white and educated (*i.e.,* college or post-graduate degrees).

Every family group discussed how LFS altered their individual and shared sensory experiences. The physicality of living with LFS included constant monitoring of one's own LFS body and the bodies of loved ones with LFS, especially underage children, to identify physical change that might indicate cancer growth. High-impact screening and limited possibilities for surgical risk reduction left participants and their caregivers in a liminal space regarding what was knowable in the body and what was preventable given the constraints of modern medicine. Participants described witnessing reduced physical functioning across family kindreds following cancer treatment. These changes impaired identity and incited profound grief. For many family groups, this grief connected loved ones with similar embodiments of LFS: for example, some family groups normalized aesthetic changes, such as amputation or scarring, to cope with disrupted physical functioning and the social impact of visible markers of difference. Intense physical impairment and pain,

however, also created thresholds beyond which the self could no longer be separated from the body to preserve identity and support coping.

3.2. A new awareness of the material body

Following genetic testing for LFS, the body was manifest as a repository of risk. Living 'at risk' meant cancer was a persistent threat to the body-self. The phenotypic variability of LFS meant constructions regarding where in the body cancer might develop remained ambiguous, fluid, and uncertain. Many patients and supportive family members discussed a preoccupation with checking the body and attending to waxing and/or waning physical sensations as signs of emerging disease, inciting fear and distress; any and all bodily symptoms (including any that might have been previously ignored) suddenly became potential omens for cancer.

> Talking to my oncologist, I said, 'There's certain things people shouldn't know, and one of them should be that you're almost certainly going to die of cancer.' Because then every little pain you get in your side, you're like, 'Oh, this must be cancer.' (Olsen)

> I found a lump when I was taking a shower. I knew it was bad. It was hard, it was big, and it scared me. (Evans)

Through the lens of embodiment, physical change was interpreted as a threat, provoked strong emotional reactions, and carried a clear physical impact.

As the guardians of underage children with pathogenic *TP53* variants, parents sought, tracked, and responded to children's physical changes or reports, carrying their embodiments for and with them. Like many parents and caregivers articulated, the following participant kept her own body and her children's bodies under strict surveillance and interpreted change as symptoms of disease. Whether or not parents also had LFS, they experienced their children's risk in their own corporeal form, suggesting a powerful type of empathic embodiment.

> Anytime one of my kids threw up it was *like electricity through my body*. It was like, 'Okay, maybe it's a brain tumor' … the shock went through my system. It would just bring it all back [the child's diagnosis]—every time the kids get sick it freaks me out. (Cason, italics added)

Participants, including parents like Cason experienced emotions regarding LFS-related cancer risk as physical sensations. Some interpreted these physical sensations as purely emotional, and they sought tools, like exercise, diet, mindfulness or spiritual practice, and social connection, to manage them appropriately. These tools strengthened the mind to support coping and strengthened the body to support a future cancer diagnosis:

> I think it was exercise, during all the surgeries that I had to have, getting right back on my bike or back to the gym or whatever has kept me more mobile, more flexible, and stronger. Keeping your body prepared for whatever might be next is important. (Walker)

By maintaining vigilance, participants stayed in communication with their bodies, and the bodies of loved ones with LFS, as discrete and full entities. Maintaining a dialogue with an LFS body did not/could not fully mitigate risk, but for these participants, such mind-body dialogue enabled management of emotional risk factors, such as distress, fear, and worry about self and other. Several participants simultaneously defined vigilance over physical sensation as a way to give voice to the body. As one participant noted, '*You need to be an advocate for your own body' (Elkhorn).* They 'listened' to their bodies, and to the bodies of their loved ones with LFS, came to know these bodies in intimate ways, and found undiscovered strength. This action of close listening united the body and self to empower participants in their own care.

> Trying to manage stress levels with prayer, and friends, and deep breathing, and simple things like that. Just trying to keep life from being a constant up and down, which just can't be good for your body, that kind of thing, and then just vigilance. I do my breast exams. I do my skin exams. I check the kids. (Lewis)

For this participant, body vigilance remained balanced, or in tension, with efforts to bridge the mind-body divide through faith-based activities, important relationships, and behavioral monitoring to reduce physical (dis)stress.

Socialization into body awareness. Most participants lived with inherited versus *de novo* pathogenic variants. Consequently, they witnessed at least one loved one's diagnosis, treatment, and death. Many families normalized visible differences in physicality resulting from cancer treatment, particularly those that shifted functioning (*e.g.,* amputation). This enabled a sense of connection within and across generations. Normalizing divergence from typical social models helped to set expectations for younger generations with LFS as they grew into the physicality of their bodies and adapted to risk and disease.

> Growing up and seeing mom's [reconstructed] breasts, I don't remember her not having any. In my mind, what she had was what everybody else had. Like, I didn't know what real breasts looked like. So, I'm not as disappointed in my reconstruction because this is what I grew up looking at. (Leigh)

Expectations for one's own experiences of the physical world were strengthened when they were shared within family groups. Yet, divergence from cultural norms incurred social risk. In response, families created cross-generational boundaries around those with shared embodiments to protect those who experienced their body and the physical world in vastly different ways. For the following family, this relational connection was tested by vicarious experiences of being ostracized or judged.

> He lost an arm, and she lost a leg. You guys were super connected because you both had suffered from amputations as children … and you can't forget, it's so visible. I'm sure, with Charles, because he lost his arm, you had to explain it a lot … I always hated that or that people treat you differently or would call you out on that or, like, even that picture you have when you were little and you were doing a self-portrait in school and he started drawing himself with two arms and one of his friends was like, 'Charles, you only have one arm,' so he erased it and you can see the outline of the arm in the photo. (Voga)

3.3. The body-self in cancer screening, surveillance, and treatment

Short interval screening and surveillance, or '*aggressive screening'* as many participants called it, in combination with vigilance over bodily sensations, led participants to distinguish and separate the self from the body. The former could retain holistic integrity and create meaning to cope with distress while the other was subject to often unpredictable acts of invasive clinical evaluation, dissection via imaging and biopsy, new cancer discovery, and excision.

WB-MRI surveillance introduced a new and permanent way of relating to the LFS body. Participants reported that WB-MRI was time-consuming, uncomfortable, and an ongoing reminder of persistent, embodied risk. More critically, WB-MRI marked a technological form of knowing that exceeded, and even supplanted, human knowing; only WB-MRI could effectively monitor and fully know the material body. This shifted the locus of control away from the self and into the hands of technology, provoking experiences of disembodiment. Thus, the body was manifest as an agent of deceit, potentially concealing 'danger' (*i.e.*, cancer) that had to be managed by constant surveillance.

> Steve: Is it that you're afraid of the possible results or the sheer being in the machine for that amount of time?
>
> Anna: It's a bit of both because the MRI takes forever but there is an element like you don't know what you are going to find. (Smithman)

Since families often scheduled scans together, they witnessed each other's subjugation to this technological knowing. While waiting their own turn, family members watched as loved ones were dissected. This disempowered family members and caregivers wanting to protect their loved ones, particularly children, from physical and emotional distress.

> And then [my family] has seen the situation where just completely out of the blue, I'm here for a whole-body MRI, I feel great, and boom, 'Jane, you have lung cancer.' So, it's anxiety-provoking. These visits and tests always make [my son] very nervous because my last cancer was diagnosed as a result of the whole-body MRI three years ago. (Murphy)

The juxtaposition of embodiment and disembodiment in the single setting of WB-MRI drove a wedge between self and body in an unanticipated fashion. Despite the perceived power of WB-MRI to identify disease, WB-MRI could only provide temporary relief that one's body was healthy before the cycle of screening would soon begin again. Leigh described relief from negative scans as a fallacy.

> I was here two years ago and was tested with MRI imaging. I was given a clean bill of health and several months later came up with more cancer. So, to put all of your eggs in one basket and think, 'Oh wow, I passed that test, I'm fine … ' You can never be that sure. (Leigh)

Considering actionability.—Participants recognized their best tools for survival relied on early detection. They lamented that opportunities for cancer prevention were non-existent for men and limited to risk-reducing mastectomy for women (Kratz et al., 2017).

Consideration of surgical risk reduction, and its limitations for cancer prevention given the wide spectrum of cancer risk that characterizes LFS, incurred the dual sense of protection and loss for women and their loved ones. With either course of action (risk-reducing mastectomy or not) participants reported anticipating physical and emotional pain alongside high-stakes uncertainty.

> It's not like I can get a double mastectomy and be like, 'Well, I just fixed my 50% chance of having cancer.' No one can remove my brain, my skin, my liver, all of these body parts are all soft tissue. I can't eliminate every risk. (Voga)

Participants described risk-reducing mastectomy as woefully inadequate to address the broad spectrum of full-body cancer risk. Thus, participants described *actionability* as severely limited and limiting, not in the clinical sense of available interventions for individuals with known pathogenic variants, but as the belief that disease expression could not be mitigated even if genetic risks were known. One participant noted, '*there's a limit to prevention'* compared with other well-understood hereditary cancer syndromes (*e.g.,* hereditary breast and ovarian cancer and Lynch syndrome).

Dialogue regarding surgical risk reduction evoked a range of complex reactions, including from family members who did not have LFS, about what is phenomenologically essential, or expendable, to the corporeal form to constitute a self. In particular, family members often disagreed about the merits of preventive surgical risk reduction, viewing such action as extreme and potentially incurring added existential and physical challenges. In the Olsen family, two sisters with pathogenic variants discussed one sister's consideration of risk-reducing mastectomy. The other sister said to her during the interview:

> I never said you're crazy, but I also said with our disease, you would cut off your bones, your brain, your blood, your everything. Where do you draw the line? (Olsen)

Notably, several female participants underwent full surgical excision of their reproductive organs, extending injury to the body beyond current risk management recommendations[2] to minimize decisional regret. This pushed loved ones to consider the long-range impacts and caregiving needs.

> My sister said I was going to be like an eyeball on the bar next to her, 'All that's going to be left [of you] is an eyeball to drink a beer with me later.' Because they were like, 'We should get rid of this, we should get rid of that.' Just start getting rid of stuff that could cause additional problems down the road. (Kairis)

Thresholds of embodiment, disembodiment, and quality of life.—Participants often implied views of the body as a clinical entity separate from the self when considering cancer treatment.

> I've had several little tumor-like growths in my body that don't belong there. About every five years, I get something taken out. (Bray)

[2]Ovarian and uterine cancers are not uncommon in individuals with LFS; however, these risks have not been sufficiently quantified to result in formal recommendations for risk-reducing surgery (lfs.cancer.gov).

Separation of the physical self, however, was incomplete. Clearly visible or sensory reminders such as amputations, scars, and reduced functioning, rendered these changes visible, and therefore knowable, to the self, loved ones, and others in each family's social world. One parent said of her daughter's reaction, *'When she'd see me in the bathroom, she started asking about scars' (Davis).* Another spoke of a well sibling's reaction, *'Coming home, you could see the shock on his face of seeing his older brother without a leg' (Hill).* This well sibling saw how his brother's body had changed, and he had expectations about what this might mean for his brother's life, his own cancer risk, and their fraternity. Some participants discussed accepting and adjusting to loved ones (risk of) reduced physical functioning following cancer treatment alongside their own risk. Participants held this in tension with the desire to remain present (and alive) for important relationships. The following participant discussed the choice between his relationship with his body versus his relationship with loved ones:

> I knew what I was facing. I mean, it feels like the guy that had his arm caught in the rocks when he was mountain climbing. His only other choice was to cut it off himself, to live, and that's kind of how I felt. You have two choices. You go through the pain of cutting something off just to live. I've got grandbabies I want to see grow up. (Brown)

Participants like Brown recognized that his largely compromised quality of life (versus succumbing to disease) permitted him to remain engaged in family relationships; these participants partialized and objectified affected body parts as separate from the self for the sake of survival, when possible.

Several others struggled with thresholds at which the body could no longer be separated from identity, notions of self, and quality of life.

> Like, holy crud, he can't live without his flute. What are you going to do if he loses his arm? (Evans)

> [Cancer] grew into his brain stem and then there was nothing after that. And he knew it. He lost the use of his right side, and started to lose his … it was just, it was bad. He had a great attitude right up until he lost his right side, and then he was like I can't live like this. (Swenson)

Evans described his child's reaction to a younger sibling's cancer treatment. In this family, the child knew how constitutional the flute was to his brother's livelihood; he identified his brother passing a critical threshold because the absence of his arm would be intolerable for his brother's selfhood. His treatment demanded not only the loss of physical functioning via amputation of his arm but also excision of his identity as flute player. Swenson discussed her son's ongoing efforts to cope with reduced physical functioning before reaching his own threshold, below which he could not retain enough of his self to continue fighting this disease.

4. Discussion

Participants across family groups discussed experiences of LFS bodies as perpetually at risk. They articulated how screening, risk-reducing surgery, and cancer treatment acted in tension as dually protective and invasive, while also as core, unavoidable features of living with LFS. Furthermore, participants discussed the ways the physical aspects of experiencing LFS were shared in family kindreds, both within and across generations, as powerful forms of shared, or inter-, embodiment. With the increasing geneticization of medicine and of life, a fitting discourse is needed to support an ever-growing population encountering inter-embodiments of chronic risk or experiencing embodied risk for the first time.

4.1. Re-visiting embodiment and genetic cancer risk

Participants in this study were highly attuned to their bodies and to the bodies of loved ones, specifically regarding vigilance over physical signs or symptoms as possible manifestations of undiagnosed carcinogenesis. Simultaneously, they relied on technology as a powerful form of knowing to discover not-yet-palpable disease states before they become otherwise evident. Consequently, participants and families struggled with a liminal space regarding their own and their loved one's diagnostic status (Little et al., 1998). Many believed vigilance could protect against late-stage diagnosis. While this offered hope, such approaches to monitoring the body left participants and caregiver/parents responsible for identifying and heeding early warning signs, reinforcing notions of individual patient, and of genetic, responsibility (Hallowell and Lawton, 2002; Howson, 1998a; Polzer et al., 2002).

Medical imaging-based disease surveillance established technological knowing of the body that was, perhaps inappropriately, given more credence than the patient's own lived experiences. This created a fragmented, potentially conflicted sense of body and self (Giammarra, 2008; Green et al., 2002; Gunderman, 2005). For other well-understood hereditary cancer predisposition syndromes, this fragmentation has been framed relative to specific body parts (*e.g.,* the breast) (Griffiths et al., 2010). Due to the remarkably heterogeneous cancer risks related to LFS, however, the entire LFS body became an object of detailed, expert scrutiny (Reventlow et al., 2006). Consigning such assessment to the hands of technology and its experts may have undermined patients' sense of autonomy and confidence relative to their own health assessment. This situated patients and families as responsible for identifying early warning signs of disease while undermining the ability of the self to fully know the body and the bodies of those they care for.

Fusion and fragmentation in experiences of embodied risk.—Considering the separate impacts of familial, or genetic, risk on the body and mind, and how ill versus well family members experience these impacts, is not new (Allen, 2011). In this sample, the conditions of pain, discomfort, and loss forced the mind and body apart to support emotional coping and adaptation to novel or evolving disease states. By considering mind/self and body separately, participants engaged with, and embodied, agency to prepare for surveillance of, or changes to, their physical form (Caiata-Zufferey, 2015; Henry et al., 2014; Slatman et al., 2015). This approach to coping may signal physical and emotional risks as distinct components of the LFS experience.

Similar to risk-reducing mastectomy for LFS, however, the success of fragmentation as a coping strategy was limited; late-stage disease and treatment provoked profound changes to the body-self. The nature of these changes (loss of balance, agility/mobility, independence) often heralded the encroaching end of life and forced integration of body and self. Consequently, compartmentalization, or objectification, of the physical body as separate from the self could only exist below individually defined thresholds of suffering. Once this threshold was reached, injury to the self-identity and to quality of life required fusion of body and self. These data suggest that for the 'at-risk' disease state, at least for LFS, body-mind integration and separation must exist as companion states of being, with degrees of physical suffering acting as catalysts between the two states. Holding both possibilities, that the body-self can be separated and interconnected, introduces flexibility and facilitates a disease process that supports holistic self-preservation for as long as possible.

4.2. Inter-embodiment in multigenerational family narratives of disease risk

In contrast to sporadic cancer, cancers that result from heritable pathogenic variants are shared in bloodlines and their impact is felt throughout family networks. Though research on more commonly studied conditions has identified shared narratives as a core feature of genetic disease (Werner-Lin and Gardner, 2009), research exploring embodiment, particularly with respect to how embodiments of risk are shared in family kindreds, remains nascent (Hallowell, 2000). This has resulted in the a lack of empirical discourse or practical language through which to interpret and respond to families with such conditions (Kavanagh and Broom, 1998).

Our findings align with the notion of inter-embodiment among families with genetic disease (Jenkins et al., 2013). We extend this work beyond the event of genetic testing to describe unique experiences of inter-embodiment over disease trajectories for families with LFS. In this context, the lived-bodies of family members become a fertile substrate for shared identity and ways of knowing in the face of severe genetic disease. Embodiments of LFS may be passed from generation to generation in families alongside the pathogenic *TP53* variant through explicit and implicit teaching of how to keep one's body healthy, how to manage an oncologic event, and how to continue living in a compromised body after such an event. Embodiments that are taught or communicated within family groups contribute to shared identities (Wilsnack et al., 2021) and become opportunities for normalizing, or bonding over, living in and as 'at-risk' bodies. For example, in this study, participants discussed connection with loved ones who shared illness experiences, or felt their connections were under threat when experiences were not shared or due to early mortality.

For these participants, embodiments were born of concern, compassion, caregiving, empathic connection, or common diagnostic trajectories. The notion of shared embodiments was most notable in parents 'feeling' their children's embodiments of LFS, experiencing the physicality of their own emotions regarding their children's risk ('*lightning in my body*'), and in sharing disease states and treatment outcomes within family groups (*e.g.,* amputations). These circumstances suggest the need to further expand the mind-body paradigm to include a *'we'* perspective. From this stance, a body-self framing is expanded

to consider multiple bodies as jointly situated in an historical family narrative of illness and loss (Wilde, 2003). The family narrative grows in the context of modern genetic medicine, with genetic testing as a mechanism for identifying new actors.

Agency and actionability in family groups.—The notion of shared embodiments is particularly critical for how individuals and families consider available courses of action to manage heritable, genetic disease. This perspective introduces questions beyond how we come to know our own bodies. These new questions include how loved ones come to understand *each other's* bodies, to identify empathically with each other's bodies, and interpret their own and each other's personal and social identities and disease states based on what they see or experience. Such questions are relevant to families with LFS since, from a patient-centered perspective, actionability in LFS is severely limited and the likelihood of disease, with early mortality, very high. Families in this study understood that the high, multi-organ cancer risk characterizing LFS meant none of their actions would guarantee or maintain a healthy LFS body. Yet, supporting a recent report (Forbes Shepherd et al., 2021), families in this study engaged in all available methods of risk management, despite their limited utility, to do all they could to stay alive.

4.3. Implications for future inquiry

The LiFE Consortium is an international collaboration of LFS researchers that is aggregating data from over 2000 individuals to understand rates of cancer expression, effective mechanisms for early detection, prevention, and treatment, and aspects of LFS that are yet unknown (Mai et al., 2020). Collaboration with this network to conduct psychosocial research is imperative. Qualitative methodologies that leverage personal and communal meaning, understanding, and action in the LFS context are critical to identifying pathways for improved medical, behavioral, and psychosocial care. For example, findings from this study illuminate the need for greater understanding of how individuals and families with LFS balance agentic relationships with their corporeal form over time with loss felt at physical and relational levels.

Specifically, research is needed to understand the range of ways individuals and families with LFS, and other conditions with multi-organ involvement, experience the body-self when multiple body parts that cannot be excised or effectively screened are considered a threat to survival. Does such an experience heighten the body-self division or do individuals find adaptive ways to engage with integration? What supports agency and prevents, minimizes, or helps individuals heal from loss, or achieve integration? How might this inquiry normalize aesthetic differences to inform ableist notions of the healthy body? For LFS, the separation of body and self as a coping mechanism, and the thresholds beyond which this separation was no longer feasible, suggests an underappreciated consequence of living with LFS that may call for explicit psychosocial research and behavioral support (Peters et al., 2016). Research is needed to understand how people with LFS prioritize and as needed, reconstitute, bodies to achieve activities of daily living that serve as coping strategies to help the body feel good, remain connected to the 'self' as appropriate, and permit both to remain strong over time (Lashbrook et al., 2018). Though these activities

do not mitigate cancer risk, they may strengthen the mind and the body to endure cancer treatment.

4.4. Strengths and limitations

Despite genetic information having implications for family groups, family groups are rarely designated as the unit of study, and even fewer studies address multigenerational family groups. Using such a strategy in this study permitted us to analyze shared family experiences with embodiment and intergenerational transmission of beliefs about mind-body (dis)connections.

Study participants were largely a homogenous population: white, highly educated, employed, and insured. As an NIH study, enrollment clearly cannot be representative of all U.S., or international, families living with LFS. Most probands joined the study following diagnosis of an LFS cancer and were aware of their pathogenic variant upon enrollment in the study. Although, many at-risk family members were untested at the time of enrollment and were only subsequently found to harbor the familial variant. Those diagnosed with cancer were temporarily ineligible for the screening arm of the study and, thus, ineligible for family interviews, until treatment was completed. Understanding the perspectives of individuals undergoing active treatment for LFS-related cancer will add to this dialogue about embodied risk. Individuals without access to regular cancer screening or who made decisions about risk management that may not align with study procedures were not represented in these interviews.

5. Conclusion

This study demonstrates that relationships between self, body, family, and others are far more complex than the risk discourse of genetic medicine would suggest. Risk discourse problematizes the body as a clinical object, and, empowered through the language of personalized and technology-based medicine, may neglect meaningful notion of shared embodiment and disembodiment. We have shown that shared embodiment is a powerful lens through which to explore the intersection of genetic risk and the body in families with hereditary cancer. Unlike organ-specific inherited cancer syndromes (*e.g.,* hereditary breast and ovarian cancer), LFS confers an all-consuming, whole-body cancer risk, introducing new ways of relating to, knowing, and separating from the body. By expanding empirical attention to experiences of individuals *and families* living with inherited cancer risk through the lens of embodiment, genetic health and mental health providers may better understand the everyday lives of people with a genetic predisposition to cancer and the unique meanings they ascribe to their illness to provide sensitive and knowledgeable care.

Acknowledgements

The authors thank the families who participated in this study and so willingly shared their experiences. This study was funded by the National Cancer Institute, Division of Cancer Epidemiology and Genetics Intramural Research Program, ZIA CP010144.

References

Allen K, 2011. Managing Prader-Willi syndrome in families: an embodied exploration. Soc. Sci. Med 72 (4), 460–464. [PubMed: 21216515]

Amadou A, Achatz MIW, Hainaut P, 2018. Revisiting tumor patterns and penetrance in germline TP53 mutation carriers: temporal phases of Li-Fraumeni syndrome. Curr. Opin. Oncol 30, 23–29. [PubMed: 29076966]

Aronowitz RA, 1998. Making Sense of Illness: Science, Society and Disease Cambridge University Press, Cambridge, UK.

Boardman FK, 2017. Experience as knowledge: disability, distillation and (reprogenetic) decision-making. Soc. Sci. Med 191, 186–193. [PubMed: 28926777]

Bowen GA, 2006. Grounded theory and sensitizing concepts. Int. J. Qual. Methods 5, 12–23.

Caiata-Zufferey M, 2015. Genetically at-risk status and individual agency. A qualitative study on asymptomatic women living with genetic risk of breast/ovarian cancer. Soc. Sci. Med 132, 141–148. [PubMed: 25813728]

Charmaz K, 2006. Constructing Grounded Theory: A Practical Guide through Qualitative Analysis Sage, Thousand Oaks, CA.

Cox SM, McKellin W, 1999. 'There's this thing in our family': predictive testing and the construction of risk for Huntington disease. Sociol. Health Illness 21, 622–646.

d'Agincourt-Canning L, 2005. The effect of experiential knowledge on construction of risk perception in hereditary breast/ovarian cancer. Journal of Genetic Counseling 14, 55–69. [PubMed: 15789156]

Denzin NK, Lincoln YS, 2008. Collecting and Interpreting Qualitative Materials SAGE, California.

Ellingson LL, Borofka KGE, 2020. Long-term cancer survivors' everyday embodiment. Health Commun 35, 180–191. [PubMed: 30466317]

Etchegary H, Potter B, Howley H, Cappelli M, Coyle D, Graham I, et al. , 2008. The influence of experiential knowledge on prenatal screening and testing decisions. Genet. Test 12, 115–124. [PubMed: 18307384]

Evans DR, Lazarides AL, Visgauss JD, Somarelli JA, Blazer DG 3rd, Brigman BE, et al. , 2020. Limb salvage versus amputation in patients with osteosarcoma of the extremities: an update in the modern era using the National Cancer Database. BMC Cancer 20, 995. [PubMed: 33054722]

Field-Springer K, Margavio Striley K, 2018. Managing meanings of embodied experiences theory: toward a discursive understanding of becoming healthier. Health Commun 33, 700–709. [PubMed: 28402137]

Fitzpatrick M, 2000. The Tyranny of Health: Doctors and the Regulation of Lifestyle Routledge, London.

Forbes Shepherd R, 2020. Coming of age with Li-Fraumeni syndrome: perspectives of young people and health professionals Sir Peter MacCallum Department of Oncology, The University of Melbourne, Melbourne, Australia.

Forbes Shepherd R, Keogh LA, Werner-Lin A, Delatycki MB, Forrest LE, 2020. Health professionals' practice for young people with, or at risk of, Li-Fraumeni syndrome: An Australasian survey. J. Genet. Couns 29, 737–747. [PubMed: 31872506]

Forbes Shepherd R, Keogh L, Werner-Lin A, Delatycki M, Forrest L, 2021. Risk management for young people with inherited cancer. Aust. J. Gen. Pract 50, 538–544. [PubMed: 34333565]

Garro LC, 1994. Narrative representations of chronic illness experience: cultural models of illness, mind, and body in stories concerning the temporomandibular joint (TMJ). Soc. Sci. Med 38, 775–788. [PubMed: 8184329]

Giummarra MJ, Gibson SJ, Georgiou-Karistianis N, Bradshaw JL, 2008. Mechanisms underlying embodiment, disembodiment and loss of embodiment. Neurosci. Biobehav. Rev 32, 143–160. [PubMed: 17707508]

Gonzalez KD, Buzin CH, Noltner KA, Gu D, Li W, Malkin D, et al. , 2009. High frequency of de novo mutations in Li-Fraumeni syndrome. J. Med. Genet 46, 689–693. [PubMed: 19556618]

Green EE, Thompson D, Griffiths F, 2002. Narratives of risk: women at midlife, medical 'experts' and health technologies. Health Risk Soc 4, 273–286.

Griffiths F, Bendelow G, Green E, Palmer J, 2010. Screening for breast cancer: medicalization, visualization and the embodied experience. Health 14, 653–668. [PubMed: 20974697]

Gunderman RB, 2005. The medical community's changing vision of the patient: the importance of radiology. Radiology 234, 339–342. [PubMed: 15670989]

Hallowell N, 2000. Reconstructing the body or reconstructing the woman? Perceptions of prophylactic mastectomy for hereditary breast cancer risk. In: Potts L (Ed.), Ideologies of Breast Cancer: Feminist Perspectives Macmillan Press/St Martin's Press, London.

Hallowell N, Lawton J, 2002. Negotiating present and future selves: managing the risk of hereditary ovarian cancer by prophylactic surgery. Health 6, 423–443.

Hallowell N, Statham H, Murton F, 1998. Women's understanding of their risk of developing breast/ovarian cancer before and after genetic counseling. J. Genet. Counsel 7, 345–364.

Henry M, Ho A, Lambert SD, Carnevale FA, Greenfield B, MacDonald C, et al. , 2014. Looking beyond disfigurement: the experience of patients with head and neck cancer. J. Palliat. Care 30, 5–15. [PubMed: 24826438]

Howson A, 1998a. Embodied obligation: the female body and health surveillance. In: Nettleton S, Watson JT (Eds.), The Body in Everyday Life Taylor & Francis Group.

Howson A, 1998b. Surveillance, knowledge and risk: the embodied experience of cervical screening. Health 2, 195–215.

Jenkins N, Lawton J, Douglas M, Hallowell N, 2013. Inter-embodiment and the experience of genetic testing for familial hypercholesterolaemia. Sociol. Health Illness 35, 529–543.

Kavanagh AM, Broom DH, 1998. Embodied risk: my body, myself? Soc. Sci. Med 46, 437–444. [PubMed: 9460824]

Kratz CP, Achatz MI, Brugières L, Frebourg T, Garber JE, Greer M-LC, et al. , 2017. Cancer screening recommendations for individuals with Li-Fraumeni syndrome. Clin. Cancer Res 23, e38–e45. [PubMed: 28572266]

LaRossa R, 2005. Grounded theory methods and qualitative family research. J. Marriage Fam 67, 837–857.

Lashbrook MP, Valery PC, Knott V, Kirshbaum MN, Bernardes CM, 2018. Coping strategies used by breast, prostate, and colorectal cancer survivors: a literature review. Cancer Nurs 41 (5), E23–E39. [PubMed: 28723724]

Leder D, 1984. Medicine and paradigms of embodiment. J. Med. Philos 9, 29–44. [PubMed: 6726100]

Little M, Jordens CFC, Paul K, Montgomery K, Philipson B, 1998. Liminality: a major category of the experience of cancer illness. Soc. Sci. Med 47, 1485–1494. [PubMed: 9823044]

Lupton D, 1993. Risk as moral danger: the social and political functions of risk discourse in public health. Int. J. Health Serv 23, 425–435. [PubMed: 8375947]

Lupton D, 1995. The Imperative of Health: Public Health and the Regulated Body London.

Lupton D, McCarthy S, Chapman S, 1995a. 'Doing the right thing': the symbolic meanings and experiences of having an HIV antibody test. Soc. Sci. Med 41, 173–180. [PubMed: 7545314]

Lupton D, McCarthy S, Chapman S, 1995b. 'Panic bodies': discourses on risk and HIV antibody testing. Sociol. Health Illness 17, 89–108.

Mai PL, Best AF, Peters JA, DeCastro RM, Khincha PP, Loud JT, et al. , 2016. Risks of first and subsequent cancers among TP53 mutation carriers in the National Cancer Institute Li-Fraumeni syndrome cohort. Cancer 122, 3673–3681. [PubMed: 27496084]

Mai PL, Sand SR, Saha N, Oberti M, Dolafi T, DiGianni L, et al. , 2020. Lifraumeni exploration Consortium data coordinating center: building an interactive web-based resource for collaborative international cancer Epidemiology research for a rare condition. Cancer Epidemiol. Biomarkers Prev 29, 927–935. [PubMed: 32156722]

Malkin D, Li FP, Strong LC, Nelson CE, Kim DH, Kassel J, et al. , 1990. Germ line p53 mutations in a familial syndrome of breast cancer, sarcomas, and other neoplasms. Science 250, 1233–1233. [PubMed: 1978757]

Merleau-Ponty M, 1962. Phenomenology of Perception Routledge and Keagan Paul, London.

Nettleton S, 1997. Governing the risky self: how to become healthy, wealthy and wise. In: Petersen AR, Bunton R (Eds.), Foucault, Health and Medicine Routledge, London; New York.

Ogle JP, Ullstrup K, 2006. Breast cancer as an embodied life event: a synthesis of research and theory and directions for intervention and future work. Illness Crisis Loss 14, 223–244.

Oppenheim D, Brugieres L, Chompret A, Hartmann O, 2001. The psychological burden inflicted by multiple cancers in Li-Fraumeni families: five case studies. J. Genet. Counsel 10, 169.

Peile C, 1998. Emotional and embodied knowledge: implications for critical practice. J. Sociol. Soc. Welfare 25, 39.

Peters JA, Kenen R, Bremer R, Givens S, Savage SA, Mai PL, 2016. Easing the burden: describing the role of social, emotional and spiritual support in research families with Li-Fraumeni syndrome. J. Genet. Counsel 25, 529–542.

Petersen AR, 1996. Risk and the regulated self: the discourse of health promotion as politics of uncertainty. Aust. N. Z. J. Sociol 32, 44–57.

Polzer J, Mercer SL, Goel V, 2002. Blood is thicker than water: genetic testing as citizenship through familial obligation and the management of risk. Crit. Publ. Health 12, 153–168.

Rees S, 2018. No one scans you and says ‘you’re alright now’: the experience of embodied risk for young women living with a history of breast cancer. Health Risk Soc 20, 312–324.

Renaux-Petel M, Charbonnier F, Thèry JC, Fermey P, Lienard G, Bou J, et al. , 2018. Contribution of de novo and mosaic TP53 mutations to Li-Fraumeni syndrome. J. Med. Genet 55, 173–180. [PubMed: 29070607]

Reventlow SD, Hvas L, Malterud K, 2006. Making the invisible body visible. Bone scans, osteoporosis and women’s bodily experiences. Soc. Sci. Med 62, 2720–2731. [PubMed: 16356616]

Robertson A, 2000. Embodying risk, embodying political rationality: women’s accounts of risks for breast cancer. Health Risk Soc 2, 219–235.

Roy K, Zvonkovic A, Goldberg A, Sharp E, LaRossa R, 2015. Sampling richness and qualitative integrity: challenges for research with families. J. Marriage Fam 77, 243–260.

Slatman J, Halsema A, Meershoek A, 2015. Responding to scars after breast surgery. Qual. Health Res 26, 1614–1626. [PubMed: 26078328]

Springgay S, 2008. Body Knowledge and Curriculum: Pedagogies of Touch in Youth and Visual Culture Peter Lang, New York.

Srivastava S, Zou Z, Pirollo K, Blattner W, Chang EH, 1990. Germ-line transmission of a mutated p53 gene in a cancer-prone family with Li-Fraumeni syndrome. Nature 348, 747–749. [PubMed: 2259385]

Thorne S, Kirkham SR, MacDonald-Emes J, 1997. Interpretive description: a noncategorical qualitative alternative for developing nursing knowledge. Res. Nurs. Health 20, 169–177. [PubMed: 9100747]

Villani A, Shore A, Wasserman JD, Stephens D, Kim RH, Druker H, et al. , 2016. Biochemical and imaging surveillance in germline TP53 mutation carriers with Li-Fraumeni syndrome: 11 year follow-up of a prospective observational study. Lancet Oncol 17, 1295–1305. [PubMed: 27501770]

Werner-Lin A, 2007. Danger zones: Risk perceptions of young women from families with hereditary breast and ovarian cancer. Fam. Process 46, 335–349. [PubMed: 17899857]

Werner-Lin A, Gardner DS, 2009. Family illness narratives of inherited cancer risk: continuity and transformation. Fam. Syst. Health 27, 201–212. [PubMed: 19803615]

Werner-Lin A, Young JL, Wilsnack C, Merrill SL, Groner V, Greene MH, et al. , 2020. Waiting and “weighted down”: the challenge of anticipatory loss for individuals and families with Li-Fraumeni Syndrome. Fam. Cancer 19, 259–268. [PubMed: 32222840]

Wilde MH, 1999. Why embodiment now? Adv. Nurs. Sci 22, 25–38.

Wilde MH, 2003. Embodied knowledge in chronic illness and injury. Nurs. Inq 10, 170–176. [PubMed: 12940971]

Williams SJ, 1998. Health as moral performance: ritual, transgression and taboo. Health 2, 435–457.

Wilsnack C, Young JL, Merrill SL, Groner V, Loud JT, Bremer RC, et al. , 2021. Family Identity and Roles in the Context of Li-Fraumeni Syndrome: “No One’s Like Us Mutants. Health Soc. Work 16, 299–307.