'Cancer doesn't have an age': Genetic testing and cancer risk management in BRCA1/2 mutation-positive women aged 18–24

Allison Werner-Lin, Lindsey M. Hoskins, Maya H. Doyle and Mark H. Greene

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'Cancer doesn't have an age': Genetic testing and cancer risk management in *BRCA1/2* mutation-positive women aged 18–24

Allison Werner-Lin
New York University, USA

Lindsey M. Hoskins
National Cancer Institute, USA

Maya H. Doyle
New York University, USA

Mark H. Greene
National Cancer Institute, USA

Abstract

Increasingly, 18–24-year-old women from hereditary breast/ovarian cancer (HBOC) families are pursuing genetic testing, despite their low absolute risks of breast and ovarian cancer and the fact that evidence-based management options used with older high-risk women are not generally available. Difficult clinical decisions in older carriers take on substantially more complexity and value-laden import in very young carriers. As a result, many of the latter receive highly personal and emotionally charged cancer risk information in a life context where management strategies are not well defined. We analyzed 32 in-depth interviews with *BRCA1/2* mutation-positive women aged 18–24 using techniques of grounded theory and interpretive description. Participants described feeling vulnerable to a cancer diagnosis but in a quandary regarding their care because evidence-based approaches to management have not been developed

Corresponding author:
Allison Werner-Lin, Silver School of Social Work, New York University, 1 Washington Square North, New York, NY 10003, USA
Email: awernerlin@nyu.edu

and clinical trials have not been undertaken. Our participants demonstrated a wide range of genetic and health literacy. Inconsistent recommendations, surveillance fatigue, and the unpredictability of their having health insurance coverage for surgical risk-reducing procedures led several to contemplate risk-reducing mastectomy before age 25. Parents remained a primary source of emotional and financial support, slowing age-appropriate independence and complicating patient privacy. Our findings suggest that, for 18–24-year-olds, readiness to autonomously elect genetic testing, to fully understand and act on genetic information, and to confidently make decisions with life-long implications are all evolving processes. We comment on the tensions between informed consent, privacy, and the unique developmental needs of *BRCA1/2* mutation-positive women just emerging into their adult years.

Keywords
BRCA1/2 genetic mutations, family relations, human development, risk management, risk perceptions

Hereditary breast and ovarian cancer risk and prevention for women aged 18–24

Inheriting a deleterious mutation in the *BRCA1* or *BRCA2* genes dramatically increases a woman's lifetime risk of developing hereditary breast and ovarian cancer (HBOC). By age 70, an estimated 60–70 percent of *BRCA1* mutation carriers and 45–55 percent of *BRCA2* mutation carriers will develop breast cancer, and 40 percent of *BRCA1* mutation carriers and 20 percent of *BRCA2* mutation carriers will develop ovarian cancer (Clark and Domchek, 2011). Much of this risk occurs before age 50. Results of a large clinic- and population-based study and a meta-analysis suggest by age 30, 3.4 percent of *BRCA1* mutation carriers and 1.5 percent of *BRCA2* mutation carriers will develop breast cancer and between 1–2 percent of *BRCA1* and *BRCA2* mutation carriers will develop ovarian cancer (Chen and Parmigiani, 2007; Chen et al., 2006; Evans et al., 2005/2006).

Strategies for risk management that evolved to care for older mutation carriers may not be adequate or appropriate for this new population of high-risk women. For example, protocols for early detection are not generally indicated for women aged 18–24, and some present conflicting information (Samuel and Ollila, 2005/2006). Breast self-examination (BSE) is recommended for high-risk women starting at either 18 or 20 years (Pruthi et al., 2010). Yet BSE has never been proven effective in detecting early breast cancer or in reducing breast cancer mortality in either the general population or high-risk settings (Humphrey et al., 2002). Frequent biopsies may result in increased anxiety, or BSE may result in a false sense of security due to low sensitivity to palpable breast abnormalities in young women (Newcomb et al., 1991). In the context of HBOC, expert opinion suggests starting clinical breast examinations between 20 and 25, and mammography and/or MRI between ages 25 and 30, or 5–10 years earlier than the earliest age at first breast cancer diagnosis in the family (Pruthi et al., 2010). X-ray-based mammograms, however, increase exposure to ionizing radiation, a known cause of breast cancer (Hendrick, 2010; Land, 1995). *BRCA1/2* genes are both involved in repairing DNA

damage of the kind caused by radiation exposure (Campeau et al., 2008). Further, the high density of young women's breast tissue often makes mammograms diagnostically inconclusive (Mandelson et al., 2000).

For *BRCA1/2* mutation-positive women, concurrent transvaginal ultrasound with color Doppler, CA-125 serum marker, and pelvic exam are recommended every six months starting either at age 30 or 5–10 years earlier than the earliest age of first diagnosis in the family (Petrucelli et al., 2010). Although these are currently the only available methods of ovarian screening, they are not proven to reduce morbidity or mortality from ovarian cancer. Risk-reducing salpingo-oophorectomy (RRSO) lowers ovarian cancer risk by approximately 85 percent for women without a previous breast cancer diagnosis. This procedure induces surgical menopause, halting ovulation and estrogen production (Lynch et al., 2009; Pruthi et al., 2010). RRSO substantially lowers lifetime risk of breast cancer for premenopausal women (Narod, 2010), yet increases lifetime risk of osteoporosis and heart disease (Kauff et al., 2008). Since ovarian cancer risk is nearly zero before age 30 (Stratton et al., 1999), surgeons are generally wary of removing healthy ovarian tissue from women in this age range. In addition, the typical primary care provider or gynecologist, through whom many women in the United States access genetic testing for *BRCA1/2*, is not likely equipped to bring a thoughtful, balanced, authoritative perspective to the extraordinary life problems faced by young mutation carriers. As a result, these young women may experience significant challenges in adjusting to their mutation status, leading to greater distress (Van Oostrom et al., 2003; Watson et al., 2004) than that experienced by older mutation-positive women.

Two frameworks: Life cycle perspective and ecological systems theory

Two conceptual pillars support this project: the family life cycle perspective on human growth and development and ecological systems theory. A family life cycle perspective suggests human development progresses through normative, sequential stages with anticipated psychosocial challenges and changes (Borysenko, 1996; Carter and McGoldrick, 1999). The ecosystems perspective argues that development occurs within important familial and environmental contexts that support or constrain growth and change. Systems theory addresses the reciprocal influence of dynamic, interacting systems (e.g. family, community, work, society) on human development through the life course (Bronfenbrenner, 1979, 2005). By integrating family life cycle and systems theories, this project investigated the critical developmental, familial, cultural, and medical influences on service delivery to and implications of genetic testing for emerging adult women.

Genetic testing for a BRCA1/2 *mutation during emerging adulthood*

Arnett (2000) was the first to describe individuals aged approximately 18–25 as *emerging adults*. He argues emerging adulthood is theoretically distinct from adolescence, marked by the rapid physiological and relational changes of the high school years, and from early adulthood, marked by durable responsibilities to family and work. Rather,

emerging adulthood is characterized by instability, exploration, change, and possibility. Emerging adults are less constrained by the normative roles and responsibilities of older adults. As a cohort, they are delaying marriage and childbearing (US Census, 2009), they may experience residential variability, inconsistency in education and career paths (Arnett and Galambos, 2003), and potent concerns about body image (Tiggemann and Pennington, 1990). Exploration during emerging adulthood supports identity development and continued socialization into and preparation for adult roles in work and family life. Exploration also involves experimentation with risk behaviors at higher rates than adolescents and adults. Yet, by the end of emerging adulthood, most individuals have made significant decisions that will have life-long ramifications.

Current guidelines suggest genetic testing to identify a *BRCA1/2* mutation may be offered once a woman reaches age 18 (Trepanier et al., 2004). This recommendation is based on the premise that women of this age can make autonomous, informed choices about genetic testing and risk management. Yet, independent decision making is a developmental milestone not particularly well established by this time in life (Arnett, 2000). Ironically, just as emerging adults achieve a measure of independence from their families, moving out of the home and into professional and social worlds, those pursuing genetic counseling find themselves in need of expert guidance (Hamilton et al., 2009) and emotional support to facilitate informed decision making. This generation of young adults is increasingly technologically savvy and has unprecedented access to information, yet may lack both the education to fully understand genetic concepts of illness (Kapingst et al., 2009) and the autonomy to act (or not act) on genetic information. Emerging adults from families with confirmed *BRCA1/2* mutations may contemplate normative life cycle transitions against the backdrop of an expected illness timeline, disease-related anxiety, or chronic grief (Werner-Lin, 2007). Renegotiating dependence and autonomy may be further complicated if a parent is actively ill and in treatment, amplifying the emotional and medical ramifications of genetic testing and risk management.

Geneticization and informed choice in complex systems

Bronfenbrenner (1979, 2005) suggests social and cultural systems are characterized by unique norms and rules that shape what individuals and families are exposed to and what they learn about the world. Within the context of these nested systems, personal and social factors (i.e. risk perceptions, social support, exposure to family illness) intersect with the larger environment (illness and gender norms, medical and reproductive technologies, public policy) over time to predispose *BRCA1/2* mutation carriers to varied risk constructions, as well as risk management and family life trajectories. Emerging adults are coming of age during an era of increasing cultural *geneticization*, in which public understandings of conditions and traits rely on genetic concepts (Ten Have, 2001) to a greater extent than for previous generations. Highly medicalized explanations for disease expression may privilege causal links between genetic variation and illness, as well as between health behaviors and long-term morbidity. Yet, we have limited understanding about the variety of risk factors that scatter

BRCA1/2 mutation carriers across the extraordinarily wide risk spectrum. To facilitate autonomous, informed engagement with systems of care, potential patients must consider both the goals that can be accomplished with knowledge of one's mutation status at this point in the life cycle, as well as the benefits and harms that may be generated by knowing (Ten Have, 2001). Women aged 18–24 may experience greater harm and fewer benefits, or be led toward fewer concrete goals than their older counterparts, as a result of learning their *BRCA1/2* mutation status. Thus, the decision-making process in which they engage (with or without the support of family members and healthcare professionals) is necessarily distinct from the larger population of women in HBOC families.

Study aims

This study sought a patient-centered perspective on the dilemmas faced by 18–24-year-olds considering genetic education, counseling and testing to identify a *BRCA1/2* gene mutation. Given the ambiguous nature of recommendations for early detection and prevention, as well as the incomplete readiness to make decisions with life-long implications, women aged 18–24 pursuing genetic testing to identify a *BRCA1/2* mutation may receive highly personal and emotionally charged cancer risk information before they have any definitive way to manage this risk. Much of the extant literature on the psychosocial aspects of *BRCA1/2* mutation-related cancer risk aggregates participants across the life span in recruitment, data analysis, and dissemination. Such an approach does not permit evidence-based services tailored to the unique developmental needs of the youngest consumers of genetic testing. Data for the current study are drawn from three separate qualitative studies, each of which used a developmental frame to investigate the experiences of *BRCA1/2* mutation carriers in their reproductive years. The present research aims to build on and enhance findings of these investigations by focusing on those challenges specific to 18- to 24-year-olds, a uniquely vulnerable group.

Design

This project used the tools of grounded theory and interpretive description to synthesize selected raw data from three separate qualitative studies of *BRCA1/2* mutation-positive women of reproductive age. *Grounded theory* offers a systematic approach to analyzing qualitative data using an iterative, '*constant comparative*' method (Strauss and Corbin, 1998). *Interpretive description* actively integrates existing knowledge into data management schema to develop increasingly complex formulations and interpretations of experience (Thorne et al., 1997). Interpretive description assumes the researcher brings to the endeavor both theoretical and practical knowledge which inform the study questions, execution, and data management. Investigators, thus, use a combination of inductive, or *in vivo*, and immersive techniques in data management. The purpose of integrating methods was to produce applied knowledge that could inform practice decisions and protocols, and develop hypotheses for future testing.

Secondary data analysis across two studies

Secondary qualitative analysis of multiple data sets is appealing and efficient, since collecting qualitative data is resource-intensive, and existing data sets are rarely fully exploited (Hinds et al., 1997; Thorne et al., 2004). Synthesizing raw data across studies permits development of more complex and persuasive policy and practice recommendations than are yielded by a single qualitative study alone. The two initial contributing studies were optimal for synthesis based on the appropriate fit between study inclusion criteria and recruitment, the ability of each data set to speak to the present study aims, and access to raw data at the individual participant level. Congruence in epistemologically constructivist frames, methodological approaches, and interview protocols enabled the two investigators to synthesize data and to collaborate in data analysis. In their original form, these data were subsumed as a part of each study cohort, in which participants aged 18–35, were pooled and analyzed together. In the current analysis, eligible participants were selected, re-coded and re-analyzed, with a specific interest in whether their issues and concerns were similar to or different from those of the entire study cohort.

Initial sample. The first (AWL) and second (LMH) authors independently recruited national samples of *BRCA1/2* mutation-positive women aged 18–35 from urban medical centers and nationally through online organizations. Participants across both data sets underwent genetic counseling and testing over a 13-year period (1997–2010), and all data were collected over a six-year period (2004–2010). Interview guides from both studies elicited extensive and data-rich reports of personal and family experiences with cancer and genetic testing; the impact of these experiences on relationships with families of origin, peers, and romantic partners; beliefs about how cancer risk influences individual development, family formation decisions; and attitudes toward risk reduction. Participants who were (1) younger than 25 when they completed genetic testing; (2) aged 18–27 when they were interviewed; and (3) female, were eligible for secondary analysis. Alphanumeric identifiers linking each text to the original study were given to 26 eligible transcripts. All eligible participants were unaffected with breast and ovarian cancer.

Novel sample. During the 2011 annual meeting of a North American consumer group focused on the information and support needs of *BRCA1/2*-positive women and men, investigators jointly recruited additional participants for a focus group on location, as well as follow-up, in-depth family history interviews. Institutional review boards at both investigators' institutions approved the focus group protocol. Eligibility criteria for the focus group included: (a) women aged 18–24 who had considered or completed *BRCA1/2* mutation testing; (b) women aged 25–27 who considered or completed *BRCA1/2* mutation testing prior to their 25th birthday; (c) English speaking; and (d) unaffected with breast and ovarian cancer. Within eight weeks of the conference, all six focus group participants completed an in-depth follow-up phone interview lasting between 25 and 90 minutes. Focus group and family history interviews were audio recorded and transcribed verbatim by a professional transcription service. Transcripts were checked against audio recordings for accuracy and deviations in meaning based on tone not captured in the transcription, such as the use of sarcasm or humor. During analysis of focus group and

family interview data, researchers read for both *a priori* codes generated during earlier analyses, and *in vivo* codes unique to this data set.

Procedures of secondary data analysis. Investigators began with line-by-line coding of transcripts from each study in isolation and maintained a separate codebook for each data set. Investigators independently examined the same subset of transcripts from AWL's data to generate a set of provisional codes and categories. The Consensual Qualitative Research method (Hill et al., 1997), a set of procedures derived from grounded theory to guide analysis when a project employs multiple coders, was used to facilitate discussion toward agreement on a set of codes and categories that best represented this data subset. Investigators communicated about the data and about their hypotheses regularly by phone and email, and via memos embedded in the coded texts. Once a working list of codes was established for the data subset, all transcripts were reread and coded. During this process, codes were expanded, collapsed, and redefined as new data were examined. Once the coding was complete and the codebook established, it was compared with the codebook of the original contributing study to identify points of similarity and difference. After completing this process for the first data set, investigators repeated it with the second data set (LMH), followed by the third data set comprised of the focus group and family history interviews.

Data synthesis: Arriving at findings

After investigators coded all 32 transcripts, they compared and contrasted the three codebooks to refine and finalize each. As an organized list of codes emerged in each stage of data analysis, the investigators tested their applicability, added or collapsed codes, and refined code definitions to capture the range of participant experiences. Once investigators identified a fixed set of codes, reaching saturation (Strauss and Corbin, 1998), investigators began axial coding (Charmaz, 2000) across the three data sets. This process sought to identify conceptual relationships between codes and to group them into meaningful clusters (e.g. 'parents as navigators' and 'protective buffering' grouped into 'Family Process') regarding how 18–24-year-old women approached genetic counseling, constructed risk perceptions, and integrated them into beliefs about the utility of genetic testing. We approached the separate codebooks as three waves of data collection, following not individual participants over time, but rather how and whether the experience of genetic testing for 18–24-year-olds changed over time. We were concerned that changes in cancer prevention and risk management, public policy addressing insurance and discrimination concerns, and the social perceptions of cancer risk and genetic disease susceptibility during this time frame might have influenced our findings, and wanted to capture this in our analysis, if it existed (Beeson, 1997).

Research quality. Collaboration across professional specializations and with experts in qualitative data analysis helped to maintain rigor and transparency. An *audit trail* (method of tracking decisions and interpretations of data) was maintained documenting the steps, procedures, and decisions made throughout the data analytic process. Memos embedded in the coded text formed the backbone of the audit trail, chronicling the data management

process. 'Outliers', known as 'negative cases', were included and analyzed for unique contributions to the development of theory and practice recommendations (McPherson and Thorne, 2006).

Findings

Introduction to findings: Descriptive demographics

Data from a total of 32 women aged 21–27 (mean age = 23.2) were included in the current analysis. At the time of original data collection, 13 women reported being single, 15 were in relationships, and four were engaged or married. Two had children, 24 reported wanting to nurture children in the future, and six either did not want children or were undecided. None were personally affected by hereditary breast or ovarian cancer. One participant was in remission from cervical cancer at the time of her interview. No one was pregnant.

No participant in this cohort initiated genetic testing because she was peri-diagnostic (i.e. in the process of being evaluated for symptoms that later proved to be cancer-related), and none reported pursuing genetic testing to inform family planning. Thirteen first learned of their genetic risk because a loved one (mother, aunt, sibling) was peri-diagnostic, and were thus motivated to pursue genetic testing. Sixteen grew up familiar with their families' cancer profiles and presented for genetic testing once they were eligible for testing. Three were unaware of their HBOC risk while growing up and were told by a mutation-positive parent or by a healthcare provider once they were old enough to complete genetic testing. At least three were part of large family cohort studies and had genetic testing as part of those protocols. The majority had made active lifestyle choices to support healthy living since learning their mutation status. Five had completed risk-reducing bilateral mastectomy (RRBM) or had one scheduled in the months following the interview, and none had scheduled or completed risk-reducing salpingo-oophorectomy.

Still learning

Collectively, our study participants are still actively learning about their family histories of cancer, personal risk estimates, and risk management options. One said:

> I tested positive and I was like, OK, I don't know what that means. And she's [the subject's genetic counselor] like, 'I'm going to go ahead and refer you to an oncologist' and when she said that, I obviously knew what that was and kind of got scared, but, still, again, had no idea what the implications were, what my options were going to be or what it even really entailed at that point. (Linda)

Many sought out information online or in professional journals. Pam told us of her frustration at attempting to read a medical journal, 'You're reading it and you have no idea what it says. I mean every other word I'm like, "medical dictionary, what, what is this?" Need a little bit more layman's terms' (Pam). Other participants described acclimating to regular breast self-exams, 'there are so many lumps and bumps in there. I didn't know what was a good lump or a bad lump' (Sara).

Participants demonstrated a wide range of medical and genetic literacy, and varied tremendously in their understandings of cancer risk estimates, as well as the etiology of inherited cancer risk. Two separate participants illustrated this range as they discussed genetic testing for sibling groups when one has already tested positive for a mutation:

> It's 50–50 for each person. It's not 50–50 for your offspring. If it was 50–50, there's a good chance that my sister would have it and then I wouldn't. But it's per each individual. So, that's why it shows up a little more. (Nichelle)

> The way the genetic counselor explained it to us, there's only a 25 percent chance of us both having it. Of course, it could still happen, but you know, it is less likely that she would be the second person that had it. (Hannah)

Transcripts were replete with errors in calculating personal cancer risk, misinterpretations of genetic counseling recommendations for risk management and risk reduction. One young woman said of her oncologist's recommendation to consider RRBM, 'My oncologist suggested 30, and I've also read it in research articles that the risk is greatest before you're 30' (Alysha). Another said, 'Statistics, studies have come out recently that say even if you detect ovarian cancer early, it has not decreased the morbidity rate. Detecting it early or detecting it late, your chances of surviving it are the same' (Cara). A few used sophisticated medical language in talking about themselves and their risk, while others became increasingly inarticulate when discussing plans (or the absence of plans) for addressing cancer risk.

Navigation

A significant feature that distinguished 18–24-year-old mutation-positive women from those who were older was reliance on parents for continued emotional, pragmatic, and financial support. In our prior work, when older mutation-positive women who were in committed, life-long partnerships and who were nurturing children discussed the impact of their mutation status on family life, they typically referred to partners and children (Hoskins et al., 2012; Werner-Lin, 2008a). The age group of 18–24-year-olds, however, more commonly referred to their parents, siblings, and affected relatives.

Many of our participants with living parents relied on them extensively to support navigation to and through genetic counseling and/or testing and risk management decision making. Parents introduced participants to the need for HBOC cancer risk assessment and the possibility of genetic testing, arranged appointments with providers, and accompanied participants to these appointments. This was particularly evident when participants' *parents* (a) were affected by or survivors of cancer; (b) had a parent of their own die of cancer during their childhood; or (c) had siblings who were affected or deceased. Several participants alluded to testing 'for' a parent, to either ease the parent's worry or because parents had asked directly. As a result, 20 participants sought consultation with their parent's providers. In these cases, the provider's relationship with the parent predated the provider's relationship with the participant. Generally, this pre-existing relationship was not problematic. Julia described the experience of having the family history

mapped out for her already: 'The (genetic counselor) already had an overall background about our family history and had worked with both my aunt and my mother and was just really, really exceptional.' For others, however, this relationship compromised the privacy of young adult women. For example, in six instances, providers shared the participant's mutation status with parents *before* sharing it with the adult child. One participant said: 'I had been diagnosed positive and my mom was waiting for the right moment to tell me' (Hannah). Another participant thought she should have been the one to tell her mother, not the genetic counselor. She said:

> My mom calls me and tells me that the doctor called her and told her my gene status. And that sort of upset me too. I didn't feel like that was something – I mean, obviously, it was something I was gonna share with my mom, but I didn't think she should be the first person to know. I guess because he [their shared care-giver] saw us as such a close-knit family, he thought it would be fine. (Alysha)

Five participants and their parents sought genetic counseling and testing together, generally when the parent or another close relative was peri-diagnostic. Many described open and fluid communication in conversation with parents and providers about testing results, risk management, hopes and fears. However, some participants using shared providers experienced a truncated process of genetic counseling; these participants perceived their mutation-positive parent as harboring guilt and self-blame about having passed the mutation to the adult child. As a result, during joint counseling sessions, participants hid worries, denied concerns, or refrained from asking critical questions about the implications of their mutation status, their cancer risk, and about various methods of risk reduction, in an attempt to protect their parents from additional burden and emotional distress. Alysha went on to discuss receiving her test results with her parents and her brother present:

> It was sort of uncomfortable because, you know, my dad and my mom have a pretty good relationship with the oncologist because you know she's been with him for over 10 years now, and so when we went back to do the counseling in the back room with the doctor and the nurse, it was me and my brother and my parents. And some of the questions I had I wouldn't feel free to ask in front of my parents, and sometimes it was a little uncomfortable talking about, you know, birth control and things like that in front of my younger brother and parents. (Alysha)

Although parents remained a significant source of emotional and pragmatic support for young mutation-positive women, parental involvement risked compromise of participant privacy and the loss of control over disclosure of their mutation status to loved ones. 'Protective buffering' (DeMarco et al., 2008) left some participants unable to fully explore risk management options or ask highly personal questions during genetic counseling sessions that included parents.

Perspectives on provider recommendations

Despite being given low absolute breast and ovarian cancer risk estimates for their age during genetic testing (and, for some participants whose loved ones were diagnosed in

their childhood, before genetic testing), participants felt vulnerable to an impending cancer diagnosis and felt pressured to take action before they crossed a threshold into territory they perceived to be unsafe. Feelings of vulnerability manifested as a sense of urgency in risk management, especially since the clinical utility of screening *and* prevention options are not yet well defined. For others, parents exerted pressure to pursue risk reduction. Yet for many, their young age erected a barrier to addressing aspects of cancer risk. These included: being too young for conventional screening tools, expressing interest in screening or prophylaxis only to have providers dismiss their concerns as premature, and being ineligible for clinical trials. A participant seeking screening in a mid-western city struggled to find a provider who would agree to screen her for breast cancer. She described this experience, saying:

> The first thing I did was actually to meet with a surgeon. I couldn't have an MRI done. They were having an awful time trying to get someone to do a mammogram on me. They said it would come back and because your breasts are dense at that age that it would be a waste of time to even do it. (Pam)[1]

The inability to actively and adequately screen breasts led participants to feel frustrated with the lack of clarity and specificity around provider recommendations. Nichelle shared: 'I'm 21, so I can't do mammograms[2] and MRIs until I turn 25. So, it's four years of just self-examinations and clinical examinations. And you know what? I need to know. I want to be proactive.' Some, like the participant below, felt dismissed by providers when they sought risk-reducing mastectomy:

> Twenty-five is a very young age, and so people are, like, well, you can wait a few years, why don't you wait and see, and wait until you have kids and you can breast feed your kids, and – and I would try to remind people that, well, it's a great idea, but also, you know, I didn't feel like I had enough time to play with. (Tracy)

In the absence of adequate screening tools, particularly for ovarian cancer, participants reported that their providers advised them to complete their family formation goals to enable risk-reducing salpingo-oophorectomy by age 35 or 40. Participants who believed a cancer diagnosis was imminent experienced this counsel as pressure to have children quickly. One participant living in a rural area shared:

> I actually had a doctor tell me a couple weeks ago that I should just do in vitro now as a single girl even though I'm living – like I just had a couple of surgeries, and so I'm living with my parents. And I'm unemployed, and I'm not feeling my best. And I really am not just doing super well. And she actually told me that I should just get pregnant with in vitro with a sperm donor and live at home, being unemployed, because my parents would fall in love with the kid and just take care of both of us. And I was just like, what!? (Theresa)

Although this provider's recommendation was extreme, Theresa was not the only participant to report similar experiences. Participants reported feeling frustrated with providers who they perceived to be advocating for early child bearing, and they felt inadequately prepared (emotionally, relationally, and financially) to become pregnant or to raise a

child. One participant said of her husband, 'There's no way in hell my husband is ready to have a child now. Maybe in three years. Who knows? And he's four years older than I am' (Ally). Yet the desire to experience a conventional pregnancy and nurture genetically linked children stood in the way of effectively minimizing this woman's cancer risk, particularly for ovarian cancer. This pitted life-long goals (to become a parent) against sustained health (achieved through risk-reducing surgery), further confounding these young women.

Resources

Participants were actively seeking readily accessible resources to clarify and facilitate risk management. Since existing screening protocols did not actively address their risk management needs, some intended to pursue, or had completed, risk-reducing surgery earlier than anticipated (or earlier than recommended by providers). Some participants who were able to find providers to conduct syndrome-appropriate breast cancer screening experienced distress produced by 'surveillance fatigue' (Hoskins and Greene, 2012). Regular and repetitive screening, the need for frequent biopsies, and the increased stress of anticipating screening and test results were emotionally taxing, even when psychosocial and fiscal supports were in place. One participant, discussing her experience of needing multiple biopsies and anticipating future screening, said:

> Even though (doctors) try to reassure me and say it's probably nothing, but we're just following up because of, you know, your *BRCA2* status, it definitely does take a toll. I do worry because I've had it happen a couple times now. Now when I go in to the test, whereas before I'd just see it as, oh, this is just a routine surveillance, now I see it as great, what are they going to find this time? (Charlotte)

Anticipating years, possibly decades, of screening – multiple times per year – increased distress and prevented many participants from developing and working toward life's goals. As a result, some pursued risk-reducing surgery both to decrease cancer risk and, of equal importance, to mitigate future/anticipated risk-specific distress.

Some participants also contemplated or completed risk-reducing mastectomy before age 25 due to their ability to have consultations, procedures, and follow-up care covered by their parents' established and robust insurance policies. A participant described the challenges presented by expectations of insufficient resources in the future:

> I am off my dad's insurance when I turn 24, which is December of this year, and he has rock star insurance. I'm planning on being self-employed, so my biggest pressure is like if I got this done before my 24th birthday I could get the rock star awesome mastectomy with the reconstruction exactly the way I want it and it would pay for everything . . . But money is really not the reason to make this decision. (Theresa)

One participant with her own insurance policy shared:

> Right now, I have a really steady job with really good health insurance and really good sick leave policy. And that is making me feel pressure to think about the surgery because I don't know when I will have that again. (Jennifer)

Another participant who lost a mother to cancer and father to heart disease between her 11th and 12th birthdays does not have the safety net of a parent's insurance coverage. She said of her college insurance plan:

> I was within a year or two of graduating, and I thought I might not have insurance for a while, and I want to get this all done while I have really good health insurance. And it would be paid for. And so I got it. (Cara)

Limitations

Secondary data analysis is always limited by the design and shortcomings of the original studies (Thorne et al., 2004). AWL's participants were interviewed before and LMH's participants were interviewed shortly after passage of the Genetic Information Nondiscrimination Act (GINA), signed into law on 21 May 2008, which provides US citizens with federal protection from health insurance or employment discrimination based on an individual's genetic profile. Participants discussed concerns about insurance discrimination, or revealed their confusion about the content of the law, while focus group participants – interviewed after the passage of GINA – were pre-occupied with pressures to pursue risk-reducing mastectomy before the age of 26, when they would no longer benefit from their parents' robust insurance policies. We maintained separate codebooks for each of the three studies to ensure these thematic differences were accounted for during data analysis.

Across all three data sets, we recruited women who completed genetic testing before the age of 21. However, none of the contributing studies actually recruited participants who were age 18–20 *at the time of data collection*. Also, none of the three studies enrolled any racial or ethnic minority women. The lack of minority representation in the young adult participants in both contributing studies and at the consumer conference reflected current populations presenting in high-risk clinics. Uptake of genetic counseling and testing remains low for women of African descent, in particular (Halbert et al., 2006), and is further complicated by their higher incidence of genetic variants of uncertain significance, a test result which is uninformative regarding risk stratification (Nanda et al., 2005).

Discussion

Eighteen to 24-year-olds are normatively at a stage of life in which they are acquiring knowledge about themselves and the world around them. They may or may not possess the maturity, the foundation of an established career or family trajectory, a realistic set of expectations about what genetic information will allow them to do, or even the health insurance to support risk management decision making. They may or may not fully understand the science behind *BRCA*-related cancer risk, penetrance, or prevention. But they do understand that their choices have both gravity (Lindenmeyer et al., 2010) and emotional implications for other family members (Hoskins and Greene, 2012; Werner-Lin, 2008b), who may variably respect young women's needs for privacy or sharing of genetic information, especially if a family member is actively ill or in treatment for a *BRCA1/2*-related cancer. Given their incomplete developmental

readiness to make independent and enduring life decisions, genetic education and risk management decision making for 18- to 24-year-olds must be understood as ongoing processes rather than as discrete events (Schwartz et al., 2005). The ability to more fully understand and act on genetic information will evolve as women's risk or family profiles change, as women mature through unfolding life circumstances, and as additional screening and/or prevention options become available (Hallowell and Lawton, 2002).

Reproductive-aged *BRCA1/2* mutation carriers experience social isolation (Werner-Lin, 2008a), anticipatory loss (Hoskins et al., 2012), distress traversing normative life cycle stages of partnering (Hoskins et al., 2008) and family planning (Hoskins and Greene, 2012; Werner-Lin, 2008a, 2010), as well as the possibility of long-term secondary illness concerns (e.g. osteoporosis risk is increased as a result of risk-reducing oophorectomy, Kauff et al., 2008). As Lippman (1999) argues, it is ironic that these choices and considerations might burden, or cause harm, to the populations they were developed to help.

Parental support and emerging autonomy

In genetic risk counseling, patients are presumed to understand and develop an action plan based on objective risk information. Such an emphasis privileges cognitive competence over relational obligation and developmentally appropriate dependence. Reliance on parents for emotional, financial, and pragmatic support may compromise the emerging adult's ability to act autonomously. The availability of genetic testing for *BRCA1/2* mutations and women's theoretically autonomous choice to undergo such testing are important medical advances; however, women do not choose to undergo such testing separate from the collective concerns and desires of their families (Lippman, 1999). Hallowell and Lawton (2002) found that *BRCA1/2* mutation-positive women across the life course make risk management decisions with important others in mind. While older mutation-positive women may have concerns about life partners and children, women aged 18–24 consider primarily the concerns of parents, physicians, and even hypothetical future partners (Hoskins and Greene, 2012; Hoskins et al., 2012). Parents may exert pressure on 18–24-year-old adult children, due to grief, guilt, or concern, to complete genetic testing (Hoskins and Greene, 2012; Werner-Lin, 2008b) and to make definitive risk management decisions (e.g. surgery rather than screening). The family's history with cancer and the young adult's understanding of that history become additional and critical lenses through which young women make meaning of and decisions about the results of genetic testing. Emerging adults and their parents are, thus, both beholden to obligations and beliefs that shape the ways they navigate genetic education, testing, and risk management, framing choice as a dynamic relational process rather that one that can be autonomously elected.

Parents, privacy, and public policy

The familial implications of identifying genetic links to conditions and traits are shifting the existing biomedical paradigm that focuses on the rights of the individual patient to a broader focus on the rights of family members. Though guidelines exist to help families

navigate the ownership of information between parents and minor children, the boundaries for sharing between parents and *adult* children are less clear (Allen, 1999). Adult children may feel compelled to pursue and share genetic information with loved ones as a way to assuage guilt or provide reassurance, particularly if a parent has just completed testing for the expressed purpose of providing information and resources for an emerging adult child. As a result, the emerging adult may find themselves in situations where sharing of information with parents is assumed, even if privacy is preferred.

Public policy and genetic services. The Genetic Information Nondiscrimination Act (GINA) decreased public concern about the misuse of genetic information and reduced a meaningful barrier to genetic testing; yet, concerns about discrimination provided a credible and convenient excuse for reluctant individuals to decline genetic testing. As a result, it is possible that GINA reduced a barrier to testing on which young adults, developmentally in flux and without reliable and permanent employment or insurance plans, may have relied to justify their hesitance, or refusal, to pursue genetic testing.

The Affordable Care Act, signed into law in March of 2010 as part of broad healthcare reform, requires group health plans and health insurance companies to provide coverage of dependent children until their 26th birthday. This legislation created an opportunity for those considering genetic testing – who are often at an age at which they are uninsured – to be covered under their parents' insurance. As an unintended consequence, parents may gain access to financial records that detail medical tests and procedures, along with financial responsibility to pay for this care. This creates the potential for compromise of privacy for young adults pursuing genetic testing. Before adult dependants age out of their parents' insurance plans, and enter a period of being uninsured, they may decide independently or may be persuaded by loved ones or healthcare providers to pursue genetic testing or risk-reducing surgery so that these expenses are covered. These pragmatic constraints may force young female mutation carriers to make major cancer risk management decisions at a significantly earlier point in their life course than would otherwise be the case. Research has yet to examine the implications of GINA or the Affordable Care Act on the lives and decisions of women aged 18–24.

Future research directions

Research is needed to understand provider training and genetic service delivery, with particular attention to the implications of public policy shifts, specific to this young age group. We have implemented a separate protocol to study the needs and challenges faced by genetic counselors and oncology nurses in genetics as they work with very young mutation carriers. Genetic counselors, registered nurses, and, most recently, clinical social workers are at the forefront of managing the care needs of young mutation carriers; they are well positioned to meet periodically with 18–24-year-olds to provide genetic education, counseling, risk clarification, and help in talking through the difficult choices with which young, high-risk women are wrestling. A management strategy based upon ongoing rather than sporadic care will enable recommendations emerging from ongoing research studies to be grounded in both patient needs and the professional mandates of healthcare providers, yielding clinical suggestions that are both relevant

and feasible, tailored to the individual needs, values and life circumstances of a particularly vulnerable population of women at increased genetic risk of breast and ovarian cancer.

Notes

1 For high-risk women breast MRI is recommended to start between the ages of 25–30. Although some women are unable to have an MRI (if they are pregnant, claustrophobic, lack insurance coverage, or are too overweight to be accommodated by the MRI machine), the procedure was technically feasible for this participant. She had difficulty finding a provider to prescribe a breast MRI due to the perception that breast density common in women of her age would render an MRI inconclusive and that her objective breast cancer risk was too low to warrant an MRI despite the participant's *BRCA*-positive mutation status.

2 For high-risk women, mammograms are recommended to start between ages 25–30, or 10 years younger than the earliest age of diagnosis in the family (Pruthi et al., 2010).

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Author biographies

Allison Werner-Lin, PhD, LCSW, is Assistant Professor of Social Work at New York University. She received a PhD and Masters degree from the University of Chicago School of Social Service Administration, and a Masters degree in Human Development and Psychology from the Harvard Graduate School of Education. Dr Werner-Lin is a Licensed Clinical Social Worker in New York and in Illinois.

Lindsey M. Hoskins, PhD, LCFMT is a Post-Doctoral Fellow in the Division of Cancer Epidemiology and Genetics at the National Cancer Institute. Dr Hoskins received a PhD in Family Science and a Masters degree in Marriage and Family Therapy from the University of Maryland. Dr Hoskins is a Licensed Couple, Marriage and Family Therapist in Maryland.

Maya H. Doyle, LCSW, is a doctoral candidate at the Silver School of Social Work at New York University. She is a Licensed Clinical Social Worker in New York.

Mark H. Greene, MD, is Chief of the Clinical Genetics Branch of the Division of Cancer Epidemiology and Genetics at the National Cancer Institute. Dr Greene received an MD from Tufts University and completed his training in Internal Medicine at the Massachusetts General Hospital in Boston. He is board-certified in both Internal Medicine and Medical Oncology.