Square Pegs, Round Hole? Ensuring Fit in the AYAO Spectrum for Adolescents and Young Adults with Genetic Risk for Cancer

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This paper presents a case example of a young woman at genetic risk for future cancer. We discuss psychosocial challenges that adolescents and young adults (AYAs) may share with their cancer survivor peers, and describe an example of psychosocial care. A scientific foundation denoting the needs of AYAs at risk for heritable cancers is lacking, and it is unknown if these AYAs receive adequate support services. This is a call to action for practitioners and researchers to engage in initiatives that assure these AYAs have access to valuable support and more clearly mark their place within the spectrum of AYA oncology.

Keywords: psychosocial, supportive care, BRCA mutation, genetic risk

RECENT RESEARCH HAS ADDED significantly to our knowledge of the unique psychosocial needs of adolescents and young adults (AYAs) diagnosed with cancer between the ages of 15 and 39.^{1,2} While accounting for a small percentage of cases, heritable forms of cancer such as those arising from BRCA mutations in breast cancer or familial adenomatous polyposis (FAP) in colorectal cancer are more common in AYAs.^{3,4} However, research examining the psychosocial aspects of genetic risk in currently unaffected AYAs or the late effects of associated prophylactic treatments in this population is minimal, and results are often mixed.⁵ Furthermore, the number of AYAs without cancer but with known genetic risk for future cancer may increase over time with continued advances in genetic medicine. It is unknown if these at-risk AYAs routinely receive adequate psychosocial, advocacy, or medical support services.

AYAs with cancer experience a multitude of psychosocial challenges that are distinct from their older counterparts.⁶ For example, younger women with breast cancer experience a greater level of stress, more depressive symptoms, and a lower quality of life than their older counterparts, and report concerns about weight gain, physical inactivity, infertility, and symptoms associated with premature menopause.⁷ Anecdotal, AYAs with genetic risk for cancer share common experiences with their affected peers diagnosed with cancer, including but not limited to: health-related anxiety, sexual dysfunction, relationship and/or career concerns, and infertility. Despite clinical observations, the literature base regarding at-risk individuals is in its infancy, with the vast

majority of studies focusing on those individuals at risk for hereditary breast and ovarian cancers. These studies emphasize psychological distress and often fail to differentiate persons under the age of 40, leaving many questions unanswered.^{5,8,9} For example, the broader psychosocial impact, potential differences among age groups (AYAs vs. older adults), and longitudinal trajectory of psychosocial concerns among at-risk individuals are unknown.

In addition to hereditary risk for breast and ovarian cancers, there is some emerging literature on the psychosocial impact of FAP. The diagnosis and management of FAP typically occurs during adolescence or young adulthood, but left untreated, it often progresses to colorectal cancer.¹⁰ A recent qualitative study of AYAs with FAP reported that cancer risk and associated surgical treatments acutely impact multiple aspects of life, including relationships, employment, and family planning.¹⁰ Also noted were participants' fears of developing cancer in the future and anxiety that their children will have FAP. This is similar to other heritable diseases such as Huntington's disease.¹¹ Despite the authors' qualitative observations of psychosocial impact, they astutely noted the current lack of evidence-based recommendations for addressing the psychosocial needs of such individuals.¹⁰

At-risk AYAs may also face challenges unique to their "cancer-free" medical status. Complexities of medical decision-making are common among these AYAs (e.g., prophylactic mastectomy/oophorectomy for BRCA; prophylactic colectomy for FAP; frequent invasive screenings).^{10,12} At-risk AYAs often receive care within designated cancer centers, where

they may feel out of place because they are not themselves cancer survivors. At our institution, some at-risk individuals have reported feeling “guilty” for utilizing cancer center psychosocial services, as they are cancer-free and thus “undeserving.” Furthermore, because they do not have a cancer diagnosis, this group often lacks access to many psychosocial resources available to their cancer survivor peers (e.g., financial aid from nonprofits such as the American Cancer Society or CancerCare). While nonprofit organizations such as Facing our Risk of Cancer Empowered (FORCE) and Bright Pink strongly represent persons at risk for women’s cancers, AYAs at risk for other heritable forms of cancer (e.g., colorectal cancers arising from FAP) have fewer resources.

Case Example

The following case example illustrates some of the challenges at-risk AYAs may face and how multidisciplinary psychosocial care can be useful to this group. Because of the complexities of decision-making within this population, the choices that individuals make regarding genetic counseling, genetic testing, and treatment options when mutations are detected can vary widely from person to person. This is a summary of one individual’s experience and not necessarily representative of every at-risk AYA. The supportive care she received is not meant to represent an optimal model of care, as there are likely other effective means of providing care to this population (e.g., community-based referrals).

In the summer of 2011, “Mrs. Brown,” a married 37-year-old woman, was referred by her internist to the cancer genetics program at our comprehensive cancer center due to her extensive family history of breast and ovarian cancers diagnosed at young ages (four family members diagnosed in their 30s), as well as prostate and thyroid cancers. She chose to pursue this referral and received genetic counseling based on her personal risk. After pre-test counseling, genetic testing for hereditary breast and ovarian cancer syndrome was offered, and she chose to complete testing. Results indicated a deleterious BRCA mutation and thus a high risk for developing cancer in the future. Subsequently, she received further genetic counseling to discuss this result.

Even with the counseling she received, Mrs. Brown reported that when looking back on this period of her care, she did not initially realize the full scope of decisions that would result from genetic testing. She felt tremendous pressure to make life-changing decisions in a short timeframe. At the age of 37, she faced difficult decisions regarding invasive prophylactic treatments, including bilateral mastectomy and bilateral salpingo-oophorectomy. Also discussed were decisions about additional surgery for breast reconstruction and treatments for premature menopause if she chose the prophylactic surgeries. The complexity of the above decisions was aggravated by her difficulty in weighing the possible heightened risk of breast cancer associated with hormone replacement therapy against the potential risk of developing conditions such as osteoporosis or heart disease without it.

While postponing or forgoing prophylactic treatments was discussed, based on her understanding of personal risk and her personal values, Mrs. Brown chose to undergo bilateral mastectomy and salpingo-oophorectomy followed by bilateral breast reconstruction. Understandably, these decisions and treatment experiences produced emotional distress.

In addition to fears associated with developing cancer and acute fears associated with undergoing and recovering from multiple surgeries, Mrs. Brown had concerns about changes in her body image and sexual functioning that could result from the surgeries and premature menopause. She was apprehensive about how these physical changes and medical procedures might impact her young marriage, her career, and her longevity. Mrs. Brown also expressed communication challenges when discussing her treatments with others, including feeling as if she sometimes had to “justify” her decisions for what others perceived to be “drastic” measures. To address these concerns, Mrs. Brown was able to engage with the cancer center’s multidisciplinary supportive oncology team via the breast center’s nurse navigator. She was successfully connected with a cancer center clinical health psychologist for psychotherapy with emphasis on emotional adjustment to genetic risk and prophylactic treatments. She reported benefit from participating in psychotherapy over the extensive course of prophylactic and reconstructive surgeries.

Further, Mrs. Brown did not have children and was planning to have a family in the near future with her husband. She was suddenly confronted with the fear that she may never have a child and, as such, found herself making decisions about fertility-preserving procedures. In addition to psychotherapy, Mrs. Brown was also connected with the fertility navigator for counseling regarding fertility preservation options prior to her surgeries. Mrs. Brown and her husband chose to pursue embryo cryopreservation. She successfully underwent in-vitro fertilization (IVF) and delivered their first child, a baby girl, in the fall of 2012.

Overall, Mrs. Brown reported improved quality of life and reduced health-related anxiety as a result of the care she received. Mrs. Brown expressed that she would not have been able to navigate through her many decisions without the help of the supportive oncology team. However, we fear that Mrs. Brown is the exception to the rule and that many AYAs with genetic risk for cancer do not have access to the support and resources they need. Too little is known to either confirm or dismiss these fears. Anecdotally, despite the positive outcomes observed and reported by Mrs. Brown, financial and insurance barriers continue to inhibit optimal care. For example, at one point, Mrs. Brown chose to discontinue psychotherapy because of the financial burdens associated with IVF and suboptimal insurance coverage for psychotherapy services.

Conclusion

AYAs with known genetic risk for cancer may face challenges in common with their cancer survivor peers in addition to their own unique challenges. However, a scientific foundation has not been laid that clearly identifies the needs of this group. Prospective longitudinal studies examining the health-related quality of life and distinct age-specific psychosocial consequences for at-risk AYAs are warranted. In turn, such studies will provide evidence-based recommendations for addressing the psychosocial needs of these individuals. This is a call to action for AYA practitioners and researchers to engage in advocacy, research, and clinical initiatives that will help these young men and women access valuable support and more clearly mark their place within the AYA oncology spectrum.

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