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Screening for Cancers in Males with Family History of Breast Cancer

Department  Nursing
Degree  Master of Science

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Signature  __Amanda L Schmieg, FNP-S__

Date  __March 1, 2018__
Abstract

**Background:** Men with family members who have breast and ovarian cancer have increased risks of both male breast and prostate cancer, and likely increases their risks of other genetically linked cancer. Routine identification of their genetic status and follow-up screenings related to their prognosis are often missed, spurring a need to further educate the public and providers about what screening schedules are recommended for these high-risk men.

**Case Description:** A 31-year-old female presented to the clinic to establish care with a family history significant for multiple 1st and 2nd degree relatives with known breast or ovarian cancer at an early age. Recommendations were made to have genetic screening. Concern was raised about how this screening may also impact others, such as her brother.

**Literature Review:** A review of literature was made through the University of North Dakota medical school database, including databases of CINAHL, PubMed, JAMAevidence, EBSCO and ERIC. Keywords utilized included male, breast cancer, genetic, BRCA, and family history. This search resulted in 22 articles related to the background as above, with a final 10 being utilized to synthesize recommendation for this paper. Another single article was brought forward by a colleague with knowledge of the background.

**Conclusion:** Although guidelines exist leading to screening recommendations in females with a family history of breast and ovarian cancer, guidelines regarding males strongly focus on genetic counseling. This option, although sometimes sought, is not always followed through on or affordable, leaving primary care providers to provide the initiative for early or non-routine screening schedules.
Screening for Cancers in Males with Family History of Breast and Ovarian Cancer

Women with family members who have been diagnosed with neoplasm of either the breast or ovary are closely followed, monitoring for any signs or symptoms of cancer, along with genetic testing, to determine if they carry the genetics that will put them at higher risk for these same cancers and more. Men are rarely assessed for family history of breast or ovarian cancer, much less routinely screened for related cancers as it is deemed unnecessary since they have neither breasts nor ovaries (Skop, Lorentz, Jassi, Vesprini, & Eisenstein, 2018). Unfortunately, genetics play into cancer diagnoses in men as much as women, and routine screening and genetic testing may be advisable for male individuals whose relatives have been diagnosed with breast or ovarian cancer. Is the lack of screening and testing due to lack in protocol surrounding these diseases in male patients or lack of willingness to further test by the male patients themselves? Further research needs to be done to better guide providers and educate the public about the increased risks males also face when a family has a history of breast and ovarian cancer.

**Background**

Triggering the investigation is an encounter with a young woman presenting to establish care. This woman has no significant personal history but does have a concerning family history, including multiple women in her family with breast or ovarian cancer. Recommendations made for this patient include early and frequent breast screening along with pelvic ultrasounds to monitor for developing neoplasm. Along with personal screenings, recommendations were made to follow up on any genetic testing that had been performed or could still be performed on living family members with known neoplasm. Although genetic concerns about neoplasm is most concerning for our patient, she has a sibling, a brother, who may also benefit from testing and early screenings.
When thinking about breast and ovarian cancer and genetics, most health care providers express concerns about female patients, almost completely ignoring the impact these same genetic markers may have on males also. Risk of male breast cancer in BRCA 1/2 carriers is increased and further research into other genetic mutations linked with neoplasms is in progress (Skop et al, 2018). BRCA 1/2 also increases a man’s risk of prostate cancer significantly (Kajula, Kaarianen, Moilanen, & Kyngas, 2016). Guidelines for providers are unclear on how to better screen and monitor these patients, so it is often left out of patient visits (Skop et al, 2018). As males make up ~50% of the population, and prostate cancer is the most common form of cancer in males, closer follow up and care of these patients is warranted.

Breast and ovarian cancers are not just viewed by providers as a woman only problem, the general public also thinks of these cancers in this way. This causes a stigma that raises concerns about the willingness and readiness of male patients to follow through with screenings should a genetic link be made to their female relative’s neoplasms. Even the word “breast” in referral to a male with said cancer has been conceived as negative and demasculating (Skop et al, 2018). This public idea of breast cancer as only a female problem also leads men to not complain about their symptoms and harbors noncompliance with recommended screening if diagnosis is made (Moynihan et al, 2017). Providing knowledge of the risks of cancer for both men and women related to family history of breast and ovarian cancer is essential to releasing these stigmas.

**Case Report**

As stated above, the initial encounter was with a 31 year old female presenting with no concerns to establish care with a new provider. Her visit was overly uncomplicated and non-concerning with the exception of her family history, which presented a concern about the
possibility of a genetic link to her family’s breast and ovarian cancer. This also presented a concern about others in her family in need of further genetic screening, including a brother, who would need screening also, even as a male. Full visit note complete with ICD-10 codes can be found in the appendix of this paper.

**Literature Review**

As research into the genetic components of cancer, specifically breast and ovarian cancer, continues to grow, there is still very little in the way of research about how positive genetic concerns for breast and ovarian cancer affect males and their need for screening. Current risks for men with a family history of breast and ovarian cancer have been identified with the well-known BRCA1/2 genes, along with lesser known sites such as the PALB2, CHEK2, and NBN genes (Daly et al, 2017). While each gene presents a different risk, each of these genes increases a males risk for either male breast or prostate cancer. Overall recommendations based on Daly et al (2017) is genetic risk evaluation for anyone with a family history of a known gene mutation in immediate family, breast cancer under the age of 50 in the family, triple negative breast cancer under age 60 in the family, two family members with known breast cancer, personal history of breast cancer with a close blood family member with either breast or ovarian cancer, or family history of male breast cancer. These recommendations are nonspecific to males but are recommendations to all patients overall. While genetic risk evaluation is beneficial, it precludes the screenings that could be presented by primary care providers in situations where either a patient or family member declines genetic screening, such as in the case of lack of affordability.

Genetic screening is not affordable, and not always covered by health insurance. Even those covered by health insurance can find the cost of their high co-pay extremely debilitating. White et al (2018) found that 38% of patients, both male and female, who would qualify and
desired testing for genetic testing based on family history declined due to financial concerns. Nearly 25% of patients who would qualify also declined screening for personal reasons (White et al, 2018). This lack of affordability may leave further clinical screening for breast and prostate cancers in males up to the primary practitioner, requiring more guidance and information be given to these providers in regard to males with family history of breast and ovarian cancers.

Even when taking genetic screening out of the equation, the numbers point to a need for further primary care screening in patients with known family history of breast and ovarian cancer. In patients with known male breast cancer, 20% have a family history of breast cancer in females in their families (Silvestri et al, 2015). Even more striking, about 20% of males with breast cancer will go on to develop a second primary cancer in their life time, most frequently prostate or colon cancer (Silvestri et al, 2015). Of the above mentioned genetic mutations, BRCA1/2, CHEK2, and PALB2, only 10-15% of those with male breast cancer are positive for these mutations (Silvestri et al, 2015). This points to further need for genetic research to ascertain other areas of genetic risk in males with significant family history of cancer. In fact, research is now pointing to familial testing utilizing multiple genetic mutation variables rather than focusing on just one, such as BRCA, as there has been a swift increase in the number of associations made with cancers and minute genetic mutations (Desmond et al, 2015).

More striking than the numbers directly related to genetic mutation is the increased risk associated with these genetically associated cancers, where males are often diagnosed later than women (Nielsen, Petersen, Therkildsen, Skytte, & Nilbert, 2016). Along with late diagnosis, male breast cancer related to genetic mutation also tends to be more likely to involve ductal and lymph node involvement (Nielsen et al, 2015). When looking at prostate cancer in BRCA1/2 mutations, they have higher Gleason scores, again with more lymph node involvement and
higher staging, all associated with a poor prognosis (Nielsen et al, 2015). In fact, both types of cancers, when associated with genetic mutations have lower survival rates and more aggressive progression (Nielsen et al, 2015). These associations require a need to push for earlier and better diagnostic screenings for patient by their primary care providers and if possible, a genetic counselor. So why then do males with a known family history or positive genetic screening not participate in more routine screenings?

“Guys don’t have breasts” (Skop et al, 2017). The stigma that males have a chest, and overall association of the word “breast” with females is a driving force in resistance to further screenings, such as mammograms (Skop et al, 2017). Along with this, males do not think about their breasts in terms of risk of cancer, as they may for prostates, but more in terms of physical fitness, meaning that routine breast exams are not routinely discussed or taught during male physicals (Skop et al, 2017). Although the primary symptoms seen in male breast cancer is gynecomastia, gynecomastia itself does not confer male breast cancer, again making it difficult to determine need for further screenings such as mammogram (Ouzounakis, Tsiligiri, & Kourkouta, 2014). In terms of screenings offered, in a cohort of 15 males with known BRCA1/2 mutation, only 8 received mammogram screening, 6 were not offered mammograms and 1 declined mammogram screening (Skop et al, 2017). Of the males undergoing mammograms, many felt awkward and excluded in the mammogram waiting room as the focus of the room was on women’s health and prevention (Skop et al, 2017). They also felt that the mammogram technicians were uncomfortable with screening males (Skop et al, 2017). Along with inconsistencies in mammogram screenings, males also found inconsistencies in their genetic counseling regarding the BRCA1/2 mutation (Kajula, Kaarianen, Moilanen, & Kyngas, 2016).
While genetic counseling is advised for males with a family history of breast and ovarian cancer, concerns about the benefit and quality of this counseling can preclude actual benefit. Of males receiving counseling, Kajula et al (2016) found that less than half of them felt their counseling included enough social support. Many also felt that psychosocial support needs to be added (Kajula et al, 2016). Most were satisfied with the genetic counseling offered overall, but many felt that it decreased their quality of life as it increased their concerns about possible cancer and passing it on to their offspring or potential offspring (Kajula et al, 2016).

Even as they are concerned about what having a high-risk mutation may mean for their children, they struggle to inform their family (Suttman, Pilarski, Agnese, & Senter, 2018). Further, they struggle to inform male children what being positive for a genetic mutation commonly associated to breast and ovarian cancer such as BRCA1/2 entails and why it is important to their health (Moynihan et al, 2017). Even more concerning, these patients do not follow through with informing their primary care providers of their genetic risks, leaving their primary care providers in the dark (Suttman et al, 2018). The screening for breast and prostate cancer ends up in the hands of genetic counselors, who are well advised about these screenings but may not see the patients as often to monitor as closely as primary care providers.

Although guidelines regarding male screenings in family history of breast and ovarian cancer are rare, the NCCN makes minor recommendations for those with known family or personal BRCA1/2 history. Daly et al (2017), recommends education in self-exam starting at age 35 with a clinical breast exam at least every year after this age. The recommendations regarding prostate cancer are less specific, with only a recommendation for prostate cancer screening in BRCA2 carriers over age 45, and consideration given to those with BRCA1 mutations (Daly et al, 2017). These guidelines also mention a lack of recommendations about pancreatic cancer and
melanomas, both increased in occurrence in BRCA1/2 carriers, further advising this should be followed on an individual basis (Daly et al, 2017). No guidelines require routine mammograms and prostate screening is nonspecific, neither mentioning prostate exams or PSAs. Overall, the recommendations both for when genetic screening should occur and when it should be advised are murky and unclear, leading both primary care providers and genetic counselors to often skip over the risks to males with family history of breast and ovarian cancer, instead focusing on the females.

**Learning Points**

- Primary care providers are on the front lines when approaching male patients about their family history. This requires them to closely examine family history in order to make referrals to genetic counseling in those at risk. They also need to closely follow the increased screening recommendations for these patients, especially when these patients decline genetic screening, whether due to personal reasons or financial burden.

- Males with family history of breast and ovarian cancer have a higher risk than the general male population for both breast and prostate cancer. Their cancers are often found later. Along with a higher risk, their overall prognosis is poorer, with more lymph node involvement and higher staging being common.

- Males who have undergone genetic screening and are positive for a genetic mutation are unlikely to tell their primary care provider. They may also struggle to tell family and friends due to the stigma surrounding their diagnosis. Counseling is essential to overcoming some of these obstacles.

- Stigma can play a big role in screenings for patients with genetic risks, including discomfort with mammogram screening and lack of knowledge about males' ability to get breast cancer.
Public education is needed to overcome this obstacle and enlighten the public about the existence of male breast cancer and the increased risk associated with both breast and prostate cancer in those testing positive for genetic mutations.

- Further education is also needed for primary care providers in order to monitor and properly screen high risk patients. No cohesive guideline exists that focuses primarily on males whose family has a history of breast or ovarian cancer.
References


Appendix

Case Study Progress Note

Patient is a 31 year old Caucasian female.

S: Chief Complaint: Establish Care

HPI: Patient presents to the clinic today to establish care in our facility. She is healthy with no concerns noted today. She is on OCP with no concerns about this. LMP:1/28/18 – normal.

PMH/SH: Appendicitis with appendectomy

Social History: Art teacher at the local high school for 5 years. In a monogamous relationship with boyfriend for 2 years.

Allergies: Penicillin

Family History: Father – Unknown; Mother – Breast Cancer – deceased at age 40; Brother – healthy – living; MGM – Ovarian cancer – deceased in 40s; MAunt – Breast cancer – alive, in 50s; Maternal Cousin – Breast Cancer – Alive, in 40s

Medications: Oral contraceptive- unsure of which one; Multivitamins occasionally

ROS: General: Denies for headaches, weakness, dizziness, nausea

HEENT/Neck: Denies for sore throat, vision changes, hearing loss or congestion

Cardio: Denies chest pain, palpitations, discomfort

Resp: Denies chronic or acute cough; no shortness of breath noted

GI: Denies diarrhea, constipation, abdominal pain, no blood in stool

GU: Denies difficulty voiding, changes in voiding pattern, no blood in urine

Musculo: Denies any weakness, pain, tremors, or swelling.

Neuro: Denies dizziness, syncope, weakness or fatigue, denies changes in vision

Psych: Denies previous history of any mental illness or depression
**O:** Vitals: P: 82, regular; R- 14; B/P – 110/66  Left arm Height: 67” Weight: 155# BMI: 24.27

General: Patient appears healthy, with no noticeable ailments noted

HEENT/Neck: Nontender with normal coloration; PERRLA, no nystagmus, red reflex intact, Tympanic membranes pearly with good light reflection; no nasal/oral discharge or redness

Cardio: Regular rhythm with no murmurs, clicks or gallops noted

Resp: Lungs clear to auscultation, no crackles or wheezes

Peripheral vascular: Pink, warm and dry. Equal and regular pulses in all extremities with no edema and normal cap refill.

Abdomen: Bowel sounds in all quadrants, no pain, tenderness or firmness noted

Breast: Normal, no lumps or fibrocystic changes noted, nipples with no discharge or tenderness

GU: Vagina and cervix normal, no adnexal masses noted. Uterus normal size, no cervical motion tenderness.

Musculoskeletal: Regular gait with no limp, grips equal

Neuro: AOx3, no speech impairment noted, pleasant, and appropriate

Labs & Diagnostics:

Mammogram – scheduled

**A:**

Establish care – Z01.89

Family history of Breast Cancer – Z80.3

**P**

1. Encouraged to continue current healthy lifestyle.

2. Recommend start yearly mammograms d/t family history, educated about importance of self breast exam and continued yearly exams.
3. Discussed genetic testing, she will think about this and discuss with living aunt/cousin if they have had performed. She does not think mother or grandmother had genetic testing. Recommended genetic counseling at this time to further discuss options and screenings, such as MRI.

4. Discussed contraceptive issues, recommend no estrogen d/t increased risk of breast cancer. She is not comfortable with the IUD or Nexplanon. Progestin only OCP discussed, she is comfortable with this. Educated to start next cycle, s/e discussed, recommend giving 3 months to regulate cycles, will call if concerns arise.

5. Follow up annually or sooner if concerns arise.

CPT: 99203