

Screening for a Young, Asymptomatic Woman with a Family History of Breast Cancer

Pujara MP

A Story from the Front Lines

A previously healthy 26-year-old woman presented to the clinic for follow-up on an ED visit for a hyperextension injury of her right thumb after a bicycle accident. In review of her family history, it was discovered that her mother had passed away from breast cancer after a diagnosis at age 32. Her mother's genotype was unknown to the patient and neither she nor her mother underwent genetic testing for hereditary cancer syndromes. Her paternal grandmother had also passed of breast cancer, though her age at diagnosis was unknown. She was unaware of any family history of ovarian, tubal, or other cancer. She requested information on screening for breast cancer given her mother's early diagnosis and expressed interest in genetic testing. At the time of this visit she was asymptomatic without breast masses, skin changes, or systemic symptoms of metastasis. She was referred Genetic Medicine and an order for screening mammography was placed after considering her vague but concerning family history of early breast cancer and the patient's personal concern over her risk of breast cancer.

Her screening mammography was classified as BI-RADS 0 with a "1.6 cm right axillary lymph node" and no evidence of malignancy of the left breast. She subsequently underwent right axillary ultrasound that elucidated "multiple lymph nodes with preserved fatty hilum but lobulated cortices" with "the thickest measured portion...up to 0.33 cm (upper limits of normal 0.3 cm)". Based on these findings her BI-RADS category was upgraded from 0 to 4 and the joint decision was made with the patient to pursue ultrasound-guided biopsy. The procedure was completed successfully and pathology showed a "benign lymph node". However, later that day she was seen in the ED for nausea, vomiting, right axillary swelling, and 8/10 pain. Three days later she continued to have 6/10 pain and worsening swelling, and was found to have a chest wall hematoma on ultrasound without evidence of infection or hemodynamic instability. With conservative management her symptoms have since resolved.

A Teachable Moment

Breast cancer screening is a topic that has developed increasing complexity over the past decade in part due to new evidence and in part related to many recommendations and guidelines created by multiple professional organizations. However, it is important to note that these screening guidelines are meant for the general population who are not already at high risk for developing breast cancer. Specifically, the USPSTF recommendations on breast cancer screening pertain to women age >40 without prior diagnosis of breast cancer or high-risk breast lesion who do not have a known underlying genetic mutation or history of chest radiation at a young age^[1].

Therefore, the important branch points in determining whether our patient is at higher risk and warrants concern for early-onset breast cancer are exposure to radiation at a young age (which was not the case) and whether or not she has a BRCA mutation. The USPSTF recommends that women with at least one family member with breast, ovarian, or other BRCA-related cancer should undergo risk stratification to determine the need for referral for in-depth genetic counseling using either the Ontario Family History Assessment Tool, the Manchester Scoring System, the Referral Screening Tool, the Pedigree Assessment Tool, or the FHS-7 – all of which have sensitivity estimates of >85%^[2,3]. Of these five screening tools, only the FHS-7 recommended further Genetic Medicine workup, with the MSS suggesting a <10% chance of BRCA mutation and the B-RST suggesting a <5% chance.

In reviewing these recommendations and re-evaluating this case, it is clear that our initial order for mammography did more harm than good and was likely unwarranted. Simply a family history of early breast cancer does not warrant imaging, but rather an evaluation of a patient's risk for having a BRCA mutation. Based on the five genetic counseling referral screening tools, this patient may not have even warranted further workup for a familial cancer syndrome. However, this would likely still have been a joint decision with the patient after discussing the risks and benefits. Ultimately, this patient experienced one of the more common adverse events of a false positive mammogram and was informed of this risk prior to the procedure, after which she recovered appropriately with minimal intervention. However, it was the fact that this imaging study was unnecessary that makes this a clear case of medical harm.

References

1. *Final Update Summary: Breast Cancer: Screening. U.S. Preventive Services Task Force.* September 2016. [https://www.uspreventiveservicestaskforce.org/Page/Document/UpdateSummaryFinal/breast-cancer-screening1?ds=1&s=breast cancer](https://www.uspreventiveservicestaskforce.org/Page/Document/UpdateSummaryFinal/breast-cancer-screening1?ds=1&s=breast%20cancer)
2. *Final Recommendation Statement: BRCA-Related Cancer: Risk Assessment, Genetic Counseling, and Genetic Testing.* U.S. Preventive Services Task Force. December 2013. <https://www.uspreventiveservicestaskforce.org/Page/Document/RecommendationStatementFinal/brcarelatedcancer-riskassessmentgeneticcounselingandgenetic-testing>
3. Nelson HD, Fu R, Goddard K, Mitchell Priest J, Okinaka-Hu L, Pappas M, Zakher B. *Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer: Systematic Review to Update the U.S. Preventive Services Task Force Recommendation.* Evidence Synthesis No. 101. AHRQ Publication No. 12-05164-EF-1. Rockville, MD: Agency for Healthcare Research and Quality; April 2013.