Underutilization of Genetic Testing and Breast Imaging Leads to Delays in Diagnosis

In the United States, the number of women diagnosed with breast cancer under the age of 40 is increasing. However, these young women are typically diagnosed at later stages and have poorer prognoses than older women. Risk factors for early age-of-diagnosis breast cancer include a family history of breast and/or ovarian cancer and mutations in genes such as \textit{BRCA1} or \textit{BRCA2}. Genetic testing can help to identify young women who are at high risk for breast cancer and for whom heightened surveillance and/or prevention strategies may be employed. If utilized appropriately, advances in breast imaging technologies, such as digital mammography and breast MRI, may also improve early detection of breast cancer in young women. A recent study examined whether advances in imaging technologies and genetic testing have translated into earlier detection of breast cancer for young women.


Purpose:
To examine the contributions of digital mammography, breast MRI and genetic testing, both actual and potential, to early detection of breast cancer in women diagnosed at age 40 or younger.

Design and Methods:
The authors conducted a retrospective study of 628 women diagnosed with breast cancer at age 40 or younger. Patient and tumor characteristics, means of diagnosis, imaging results, and use of genetic testing were reviewed.

Results:
- 50% of women in this study had a family history of breast and/or ovarian cancer and in 2/3 of these women, family history would have been missed by concentrating solely on first-degree relatives
- The mammogram performed at or after diagnosis was the initial mammogram for 81% of women in this study
- Breast cancers detected in these women were larger (median 2.0 cm) than previous studies have been able to visualize (1.2 cm and 1.1 cm)\textsuperscript{1,2} making it reasonable to conclude that evaluation of family history, combined with screening would have been able to detect cancer in these women earlier
- Only 40% of women had genetic testing (nearly all post-diagnosis) and 25% of these women were found to carry a \textit{BRCA1} or \textit{BRCA2} mutation

"Young women have not yet achieved the full benefit possible from application of current imaging technologies or from the use of genetic testing."

Bottom Line:
This study emphasizes the importance of taking a family history that extends beyond first-degree relatives. "These data confirm the need for earlier identification of potential risk gene mutation carriers for genetic testing so that appropriate high-risk screening and prevention options could be used."