BRCA変異と対側乳がんのリスク（Abstract # S4-2）

BRCA変異を有する乳がん既往者において対側の新たな乳がんを発症するリスクが高い

Risk for developing contralateral new cancer increased for survivors with BRCA mutation

BRCA1またはBRCA2遺伝子変異を有する乳がん既往者は対側の乳がん（CBC）を発症するリスクが高く、この群の女性の一部は診断時年齢および最初の腫瘍の状態によってさらにリスクが高いとのデータが2011年CTRC-AACRサンアントニオ乳がんシンポジウムで発表された。研究者らは、オランダの10の病院で片側の浸潤性がんと診断された女性5,061人を調査した。211人（4.2%）がBRCA1またはBRCA2のキャリアであった。フォローアップ期間中央値8.4ヵ月の時点で、全体の8.6%がCBCを発症した。10年間のCBC総発症率はノンキャリアで6.0%であり、一方キャリアのリスクは17.9%であった。最初の乳がんを40歳未満で診断されたキャリアの10年間のCBCリスクは26.0%に跳ね上がった。40～50歳に最初の乳がんを診断されたキャリアのリスクは11.6%であった。さらに、最初の腫瘍がトリプルネガティブであった変異キャリアの10年間累積CBCリスクは18.9%であり、それと比較し最初の腫瘍がトリプルネガティブでなかったキャリアにおいては11.2%であった。

Full Text

Breast cancer survivors who carry the BRCA1 or BRCA2 genetic mutation are at high risk for developing contralateral breast cancer, and certain women within this group of carriers are at an even greater risk based on age at diagnosis and first tumor status, according to data presented at the 2011 CTRC-AACR San Antonio Breast Cancer Symposium.

"Our studies show that certain subgroups of women [with this mutation] who have already had cancers are also at risk for developing a second new cancer in their other breast, much more so than survivors who do not carry the mutation," said Alexandra J. van den Broek, M.Sc., a doctoral candidate at the Netherlands Cancer Institute. "Our study is, as far as we know, the first study showing that within certain carriers of BRCA mutations, subgroups with an increased or decreased risk for contralateral breast cancer (CBC) can be made."

Researchers surveyed 5,061 women diagnosed with unilateral, invasive breast cancer at 10 hospitals in the Netherlands. Two hundred eleven women (4.2 percent) were carriers of the BRCA1 or BRCA2 mutation. Overall, at a median of 8.4 years of follow-up, 8.6 percent of participants developed CBC.

Van den Broek and colleagues found that the overall 10-year risk for developing CBC in noncarriers was 6.0 percent, while risk for carriers was 17.9 percent.

For carriers diagnosed with their first breast cancer when aged younger than 40 years, the 10-year risk for CBC jumped to 26.0 percent. For carriers between the ages of 40 and 50 years at first diagnosis, the risk was 11.6 percent. In addition, mutation carriers with a triple-negative first tumor had a 10-year cumulative CBC risk of 18.9 percent compared with 11.2 percent among carriers with a non-triple-negative first tumor.

Although these numbers can be overwhelming to carriers who have already survived breast cancer, van den Broek said it is crucial to know who is most at risk and by how much.

"Guidelines for prophylactic measures and screening in the follow-up of patients with breast cancer carrying the BRCA1 or BRCA2 mutation are important to provide patients with the best information and counseling," she said. "If these results are confirmed, [it will be] possible to personalize the guidelines for these specific subgroups."

The next step will be to confirm the results in larger studies and to look at other factors that define subgroups of patients with an increased or decreased risk for CBC.

TOPICS