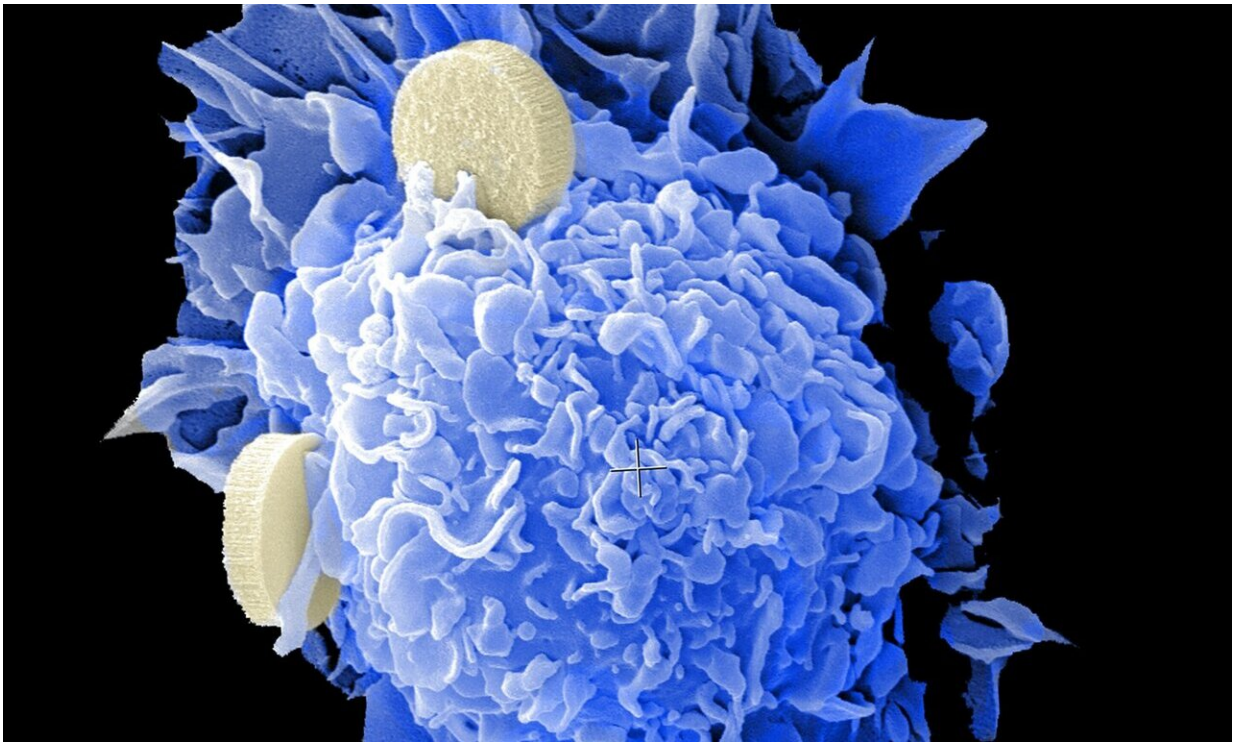
What you need to know about the PALB2 breast cancer gene

October 12 2021



Credit: Unsplash/CC0 Public Domain

Most of us have heard about the *BRCA1* and *BRCA2* genes. Men and women with a personal and/or family history of breast cancer have been offered testing for mutations in these two genes for a number of years to determine if they are at greater risk for breast and other cancers. But what about the *PALB2* gene, which stands for Partner And Locator of

BRCA2? A recent study in the *New England Journal of Medicine* showed women who carry a mutation in the *PALB2* gene were 35% more likely to be diagnosed with breast cancer by age 70, compared with women who don't carry the mutation.

"*PALB2* is a gene that encodes a *BRCA2*-interacting protein. The *BRCA2-PALB2* interaction is necessary for DNA damage repair that normally happens in our cells," said Mariya Rozenblit, MD, Instructor of Medicine (Medical Oncology) at Yale Cancer Center. "When this protein is not functioning normally, DNA damage is not fixed, and cells become abnormal, leading to cancer."

Rozenblit said patients are becoming more aware of genetic testing and its importance in determining their breast cancer risk. If they have concerns, she recommends women discuss testing with their primary care doctor and see a genetic counselor if needed. Patients counseled by the Smilow Cancer Genetics & Prevention Program are referred based on their personal and/or family history of cancer. A genetic counselor will take a thorough family history and recommend the most appropriate genetic test(s), which are often performed through a blood test. Genetic tests to evaluate for hereditary breast cancer risk now often consist of a panel of genes, including *BRCA1, BRCA2, PALB2*, and others.

"Results of genetic testing typically take two to three weeks and the genetic counselor will review the results of the genetic testing and their implications with the patient," said Claire Healy, MS, CGC, Co-Manager Genetic Counselor for the Smilow Cancer Genetics and Prevention Program. "If a mutation is identified the genetic counselor can also help connect the patient to providers for high-risk cancer screening and medical management, if needed."

Healy added that clinical testing of *PALB2* has not been available for as long as genetic testing of the *BRCA1* and *BRCA2* genes, so patients who

completed testing prior to 2013 may not have had an analysis including the *PALB2* gene. These patients should consider discussing with their providers whether additional genetic testing is indicated.

"One of the BEST things patients can do if they are worried about their risk to develop cancer is ensure that they have collected detailed information about their family history," said Healy. "Ideally, patients should be aware of who in their family has had cancer, the type of cancer their relative was diagnosed with, i.e. in which organ the cancer started, and the age at which they were diagnosed."

More information: Antonis C. Antoniou et al, Breast-Cancer Risk in Families with Mutations in PALB2, *New England Journal of Medicine* (2014). DOI: 10.1056/NEJMoa1400382

Provided by Yale University