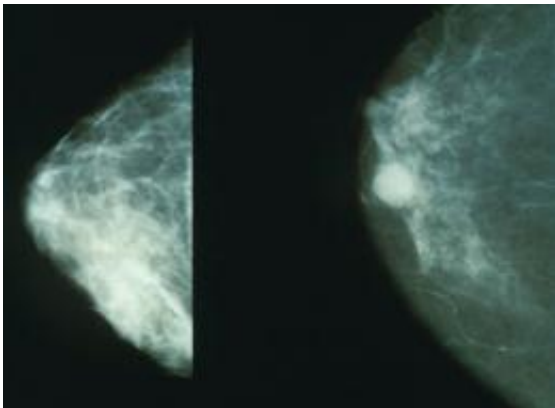


Gene increases risk of breast cancer to one in three by age 70

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Mammograms showing a normal breast (left) and a cancerous breast (right).
Credit: Wikipedia.

Breast cancer risks for one of potentially the most important genes associated with breast cancer after the BRCA1/2 genes are today reported in the *New England Journal of Medicine*. Women with mutations in the PALB2 gene have on average a one in three chance of developing breast cancer by the age of seventy.

In a study run through the international PALB2 Interest Group a team of researchers from 17 centres in eight countries led by the University of Cambridge analysed data from 154 families without BRCA1 or BRCA2 mutations, which included 362 [family members](#) with PALB2 gene mutations. The effort was funded by the European Research Council,

Cancer Research UK and multiple other international sources.

Women who carried [rare mutations](#) in PALB2 were found to have on average a 35% chance of developing breast cancer by the age of seventy. However, the risks were highly dependent on family history of breast cancer where carriers with more relatives affected by breast cancer, were at higher risk. Only a very small proportion of [women](#) worldwide carry such mutations and the researchers point out that additional studies are required to obtain precise estimates of mutation carrier frequency in the population.

PALB2 is known to interact with both the BRCA1 and BRCA2 and was first linked with breast cancer in 2007. As is the case for women who carry mutations in BRCA1 or BRCA2, women with PALB2 mutations who were born more recently tended to be at a higher risk of developing breast cancer than those born earlier. The reason why is unclear, but the researchers speculate that it may be related to factors such as later age at first childbirth, smaller families and better surveillance leading to earlier age of diagnosis.

Dr Marc Tischkowitz from the Department of Medical Genetics at the University of Cambridge, who led the study, says: "Since the BRCA1 and BRCA2 mutations were discovered in the mid-90s, no other genes of similar importance have been found and the consensus in the scientific community if more exist we would have found them by now. PALB2 is a potential candidate to be 'BRCA3'. As [mutations](#) in this gene are uncommon, obtaining accurate risk figures is only possible through large international collaborations like this.

"Now that we have identified this gene, we are in a position to provide genetic counselling and advice. If a woman is found to carry this mutation, we would recommend additional surveillance, such as MRI breast screening."

The researchers at Addenbrooke's Hospital, part of Cambridge University NHS Hospitals Trust, have developed a clinical test for PALB2, which will become part of their NHS service. Clinical testing for PALB2 will be available also in certain other diagnostic laboratories worldwide.

There is evidence that cells carrying the PALB2 mutation are sensitive to a new class of drugs known as PARP inhibitors that are currently being trialled in BRCA1/2-related breast cancers. It is possible that these drugs would also work in PALB2-related breast cancer.

Dr Antonis Antoniou from the Centre for Cancer Genetic Epidemiology at the University of Cambridge adds: "Knowing the key genes that significantly increase cancer risk and having precise cancer risk estimates ultimately could help assess the breast [cancer risk](#) for each woman and allow better targeting or surveillance."

Professor Peter Johnson, Cancer Research UK's chief clinician, said: "We're learning all the time about the different factors that may influence a woman's chances of developing [breast cancer](#). This particular mutation doesn't make people certain to develop cancer, but it's another piece of information to help women make proper informed choices about how they may help to minimise their own risk."

More information: Antoniou, A. et al. Inherited loss-of-function mutations in PALB2 and breast cancer risk. *NEJM*; 6 Aug 2014.

Provided by University of Cambridge

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