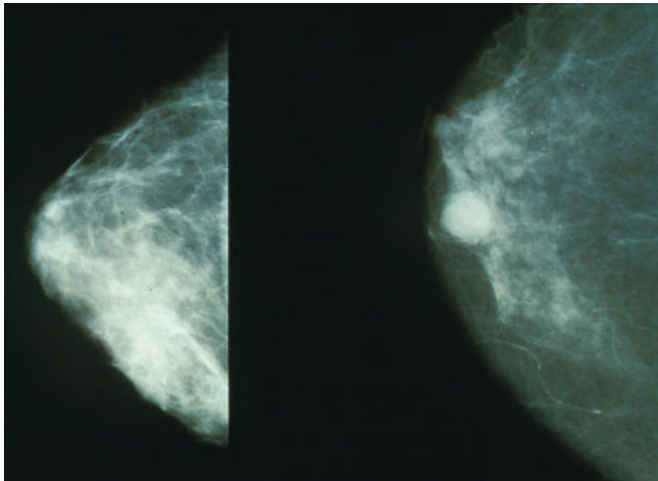


# Genetic link confirmed between DCIS and development of invasive breast cancer

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Mammograms showing a normal breast (left) and a breast with cancer (right). Credit: Public Domain

Scientists funded by Breast Cancer Now have confirmed inherited genetic links between non-invasive cancerous changes found in the milk ducts – known as ductal carcinoma in situ (DCIS) – and the development of invasive breast cancer, meaning that a family history of DCIS could be as important to assessing a woman's risk as a history of invasive breast cancer.

DCIS is a non-invasive form of early breast cancer, where [cancerous cells](#) are entirely contained within the milk ducts of the breast. 5,000 women are diagnosed with DCIS each year in the UK, with two thirds of these cases being diagnosed through routine breast screening, rather than symptomatic detection.

But while DCIS accounts for 1 in 10 breast cancers in the UK, very little has been understood about its [genetic risk factors](#) and [genetic](#) relationship with [invasive breast cancer](#), until now.

In new research published in the journal *Breast*

*Cancer Research*, scientists at King's College London analysed over 200,000 genetic markers in DCIS patients, making it the largest study of inherited genetic predisposition to DCIS to date.

The team, led by Dr Elinor Sawyer and Professor Montserrat Garcia-Closas, compared DNA extracted from the blood samples of over 5,000 women with DCIS and 24,000 with invasive ductal cancer (the most common form of invasive breast cancer), finding that most of the genetic changes linked to invasive ductal cancer were also linked to DCIS, and there were no unique changes linked only to DCIS.

These findings provide the strongest evidence yet of a shared genetic susceptibility between DCIS and invasive ductal cancer, giving us important insight into the biology of DCIS.

This conclusion also confirms the need for a [family history](#) of DCIS to be considered when assessing familial risk of breast cancer.

It is currently estimated that only 50% of DCIS cases will develop into invasive breast cancer if left untreated. However, at present it is not possible to predict which DCIS cases will progress to invasive breast cancer and therefore all women are offered treatment after diagnosis.

This research is therefore just the first step. No genetic change unique to DCIS (and not invasive breast cancer) was discovered in this study, leaving researchers still without a genetic marker to find cases of DCIS with a very low risk of progressing to life-threatening breast cancer. Being able to identify these patients could potentially allow them to be spared unnecessary treatment.

It is clear that preventing overtreatment of DCIS requires further research along a number of different avenues. In the short term, it is hoped that a biomarker test could be developed to predict the

risk of progression for DCIS patients, with a number of promising molecules identified in recent years.

Dr Elinor Sawyer, Clinical Reader in Oncology at King's College London and Consultant Oncologist at Guys and St Thomas' NHS Foundation Trust, said:

"This study provides the strongest evidence to date of a shared genetic susceptibility between DCIS and invasive ductal cancer, suggesting that both develop through a common pathway.

"This is a great leap in our understanding of DCIS, but it is also just the first step. We now hope to carry out a larger study assessing more genetic changes to discover whether there are any that predispose solely to DCIS, something that could help us predict whose non-invasive cancer is unlikely to progress."

Katie Goates, Senior Research Communications Officer at Breast Cancer Now, said:

"This largest-of-its-kind study is invaluable as we have previously known so little about the inherited genetics of DCIS, something that affects around 5,000 women each year.

"The results confirm how similar DCIS is on a genetic level to invasive [breast cancer](#), greatly increasing our understanding of the small genetic changes linked to this very early condition.

"However, we still have not cracked the tough nut of identifying which women's DCIS won't progress to invasive cancer, and further research will now be crucial to finally preventing cases of overtreatment in the future."

**More information:** Christos Petridis et al. Genetic predisposition to ductal carcinoma in situ of the breast, *Breast Cancer Research* (2016). [DOI: 10.1186/s13058-016-0675-7](#)

Provided by Breast Cancer Now

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