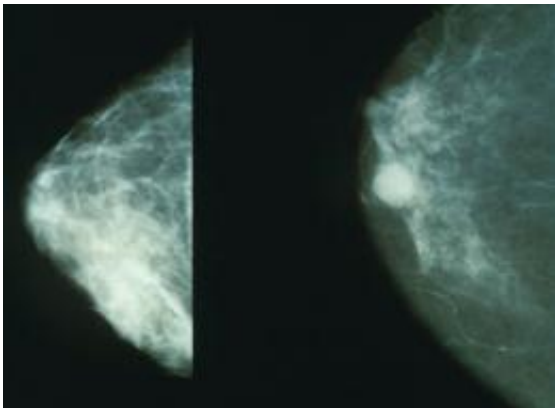


Genetic testing could improve breast cancer prevention

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

Scientists used mathematical models to show that analysing genetic data, alongside a range of other risk factors, could substantially improve the ability to flag up women at highest risk of developing breast cancer.

Their study showed that prevention strategies could be improved by testing not only as currently for major cancer predisposition genes such as BRCA1 and BRCA2 - which identify a small percentage of women at very high risk - but also by factoring in data on multiple gene variants that individually have only a small effect on risk, but are more common in the population.

The research was carried out by researchers at The Institute of Cancer Research, London, and the National Cancer Institute in Bethesda, US - and is published today (Thursday) in the *Journal of the National Cancer Institute*.

The study received funding from The Institute of Cancer Research (ICR), Breakthrough Breast Cancer and the National Cancer Institute.

Researchers stressed that their study was a computer modelling analysis and would need to be confirmed by further research aimed at validating the models they used and assessing real-life prevention approaches.

But they said identifying women at highest risk using genetic and other factors could allow preventative treatments and tailored advice to be offered more effectively, reducing the number of women who develop [breast cancer](#). And [genetic testing](#) could be done using currently available technology.

In the study, researchers modelled the potential risk stratification of eight hypothetical scenarios. Depending on the risk factors measured under each scenario, they would use a mixture of a GP or self-administered questionnaire, genetic profiling and mammography scans to measure breast cancer density to calculate risk.

The risk factors were:

- genetic profile
- family history of breast cancer
- age at menarche
- number of births and age at first live birth
- oral contraceptive use
- combined menopausal therapy (MHT) use
- body mass index [BMI]

- alcohol consumption
- smoking status
- personal history of benign breast disease [BBD]
- [breast tissue density](#).

The most effective of the eight models at predicting [breast cancer risk](#) combined analysis of all the risk factors. For instance when applied to 50 year-old women, such a model could identify the most at-risk 10.2 per cent of women - who account for 32.2 per cent of all breast cancer cases.

That means doctors could tell an individual 50-year old woman if she was in the most at-risk 10 per cent of the population - and potentially identify lifestyle changes or preventative treatments to reduce her risk, or provide better advice on the potential benefits and harms of taking hormone replacement therapy to reduce menopausal symptoms.

In the UK, more detailed risk information is only routinely collected for women with a family history of cancer, whose care can be altered if they are thought to have a high chance of carrying a very high-risk genetic mutation, such as one of the BRCA genes.

NICE guidelines recently recommended considering the use of drugs to prevent breast cancer in apparently healthy women found to be at moderate to high risk of developing breast cancer, based on their family history or carrying a very high-risk genetic mutation.

But the great majority of breast cancers occur in women without a known family history of the disease. The new study shows additional genetic testing and risk factor assessment could help more accurately identify [women](#), with and without [family history](#), who are at elevated risk and could benefit most from preventive strategies.

Study leader Professor Monserrat Garcia-Closas, Professor of

Epidemiology at The Institute of Cancer Research, London, said:

"Our study shows that genetic testing has the potential to improve strategies for preventing breast cancer, and suggests that multiple, small-effect genetic [risk factors](#) could be included alongside the major [breast cancer genes](#) to assess risk. We hope it helps to start a wider discussion about how to add more accurate risk prediction tools to future prevention programmes.

"Our models are theoretical and rely on a number of assumptions, and they are only the starting point in this discussion - much more detailed and practical research studies are needed to say exactly how combined risk factor analysis

Provided by Institute of Cancer Research

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