Study shows benefit of universal genetic testing after breast cancer diagnosis

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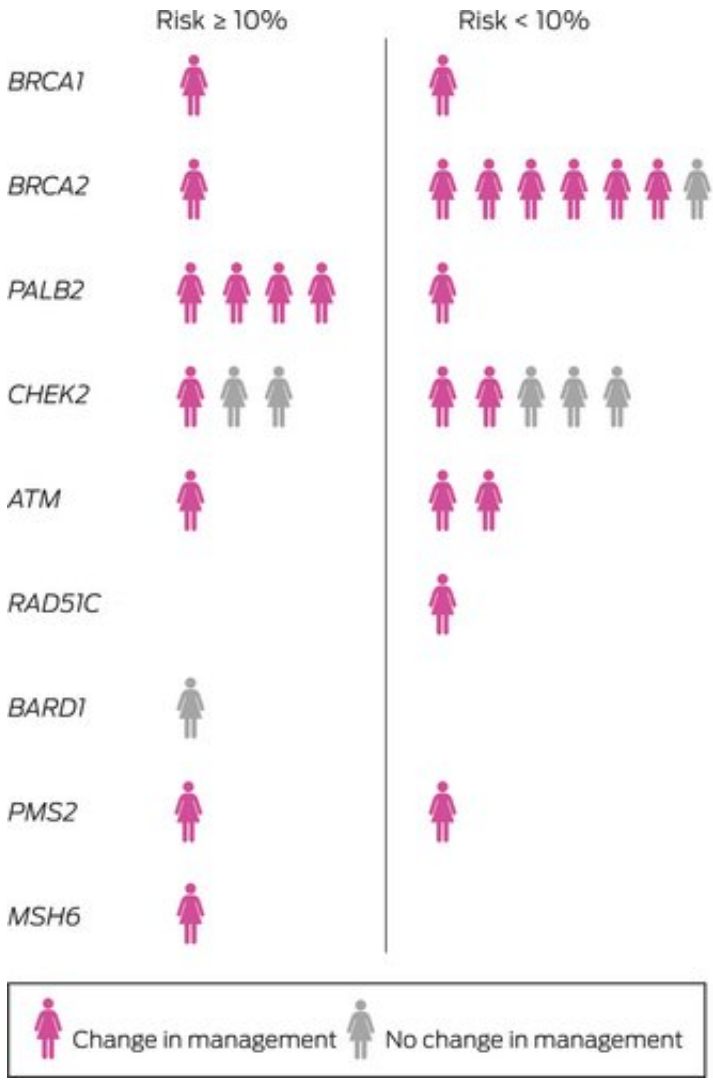

Classification of 31 participants found to carry pathogenic gene variants, by gene, estimated risk of having a pathogenic gene variant,* and effect on treatment recommendations. Pathologic gene variants estimated with the CanRisk or Manchester algorithms. Credit: *Medical Journal of Australia* (2023).

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According to Australian guidelines, women with newly diagnosed breast cancer should be offered genetic testing when they meet certain strict criteria—such as being diagnosed at a very young age or if many family members have had breast cancer—amounting to a 10% or more risk of having a rare, hereditary gene mutation.

The Melbourne-based study provided genetic testing to 474 women with newly diagnosed breast cancers irrespective of their risk of having a gene abnormality, such as BRCA1 or BRCA2 mutations.

Faulty cancer-causing genes were identified in 31 of the women and more than half (18) of these patients would not have been eligible for genetic testing under the current guidelines.

The study is a collaboration between the Parkville Breast Service (Peter MacCallum Cancer Center, Royal Melbourne Hospital and Royal Women's Hospital), the Parkville Familial Cancer Center and WEHI, and results are published in the *Medical Journal of Australia*.

Universal screening provides missing vital link for many

Peter Mac's Professor Ian Campbell said the study showed that current Medicare-funded testing guidelines mean many patients miss out on vital information that could change the course of their treatment.

"Our study expanded this genetic testing to all women with newly diagnosed breast cancer and found 58% of patients identified as having a

faulty gene had a risk factor of less than 10%, meaning these patients would not have been picked up under current screening guidelines," said Prof Campbell, who is head of the Cancer Genetics Laboratory at Peter Mac.

"This demonstrates that more than half of breast cancer patients are missing out on their treatment being properly managed based on additional genetic information currently unavailable to them and their treating team."

"The adoption of universal genetic testing for women with newly diagnosed breast cancer has the potential to improve outcomes by identifying hereditary mutations in patients and their families, leading to better treatment options and outcomes," he said.

Genetic risk factors impact treatment decisions

Approximately 5% of breast cancers occur in people who have inherited a gene abnormality from a parent.

WEHI's Professor Geoff Lindeman said identifying these cases was vital to providing the best medical advice and intervention to patients and their wider families, and it often impacted surgery decisions.

"For example, a patient with a BRCA1 or BRCA2 gene mutation may choose to have a double mastectomy, rather than radiation therapy," said Prof. Lindeman, who is also a medical oncologist at Peter Mac and the Royal Melbourne Hospital.

"Similarly, a patient with a high risk of developing ovarian cancer might elect to have their ovaries removed as a preventative measure."

"When a faulty gene is found, it also means family members can be

tested and referred to a familial cancer clinic to manage their risk factor for developing breast or ovarian cancer."

Professor Bruce Mann, Head of the Parkville Breast Service, said for most women in the study identified as having a gene abnormality, their recommended treatment changed as a result.

"With this additional information, the multidisciplinary team modified treatment recommendations for 77% of the women with hereditary mutations, including those not currently eligible for Medicare-funded testing," Prof. Mann said.

"We were able to incorporate this genetic testing into our existing multidisciplinary model of care, expediting treatment for patients."

"Having their doctors initiate genetic counseling and testing streamlined the process and improved access to testing."

No regrets about being tested

Associate Professor Lesley Stafford, a clinical psychologist at the Royal Melbourne Hospital and the Parkville Familial Cancer Center, said the research also found strong acceptance of genetic testing.

"Of the women who participated in our study, 87% agreed or strongly agreed that all women with breast cancer should be offered genetic testing," she said.

"There was no decision regret among the participants in our study about having the genetic testing."

More information: Dilanka L De Silva et al, Universal genetic testing for women with newly diagnosed breast cancer in the context of

multidisciplinary team care, *Medical Journal of Australia* (2023). DOI: 10.5694/mja2.51906