Genetic tests for patients with a high risk of breast and ovarian cancer are now covered by Medicare, and can be ordered by non-genetic specialists who may not have the training and experience to correctly interpret the results, warn the authors of a Perspective published in the Medical Journal of Australia.

Associate Professor Judy Kirk, Head of the Familial Cancer Service at Westmead Hospital in Sydney, and colleagues wrote that new Medicare item numbers for genetic testing available since 1 November 2017 meant that any specialist or consultant physician (other than general practitioners) could now order genetic testing for breast and ovarian cancer under certain criteria, and those tests were now covered by the Medicare Benefits Schedule.

"Clinical genetic testing for heritable, germline mutations in two major genes … (BRCA1 and BRCA2) that are associated with a high risk of breast and ovarian cancer came into Australian practice in the mid-1990s, and were offered free of charge (but not under Medicare) to appropriate patients in public clinics," Kirk and colleagues wrote.

"Until now, testing, which has proven clinical utility, has mostly been offered through a network of family cancer clinics and genetics services that provide expert genetic counselling and testing of these genes in the context of familial breast and ovarian cancer.

"These tests are now being mainstreamed and they can be ordered for selected patients, with a new Medicare benefit, by non-genetic specialists in either public or private practice.

"It is … essential that any clinician who orders breast cancer genetic testing understands the complexities and implications generated."

The problems arise when a negative result — that is, when no mutation is found — is returned.

"When a mutation is not found using the initial mutation search in an affected family member, this negative result is not truly negative, but rather is uninformative," Kirk and colleagues wrote.

"No predictive test can be done for family members if that is the case.

"There are many families with a strong family history for which there must be some genetic cause, and yet it cannot be found. Limitations in knowledge and technology need to be understood, and individuals from these families remain at potentially high risk.

"Such uninformative results require careful counselling so that families (and their clinicians) are not falsely reassured by the ‘no mutation found’ result."

The authors concluded that non-genetic specialists ordering the tests "receive appropriate education and support to enable safe, evidence-based practice in keeping pace with the genomics revolution".

"This will be optimised if ordering clinicians develop a close relationship with a local family cancer clinic or clinical genetics service, which are ideally placed, and willing, to educate and support non-genetic practitioners in the transition to mainstream genetic testing."

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