Open-access genetic screening for hereditary breast cancer is feasible and effective

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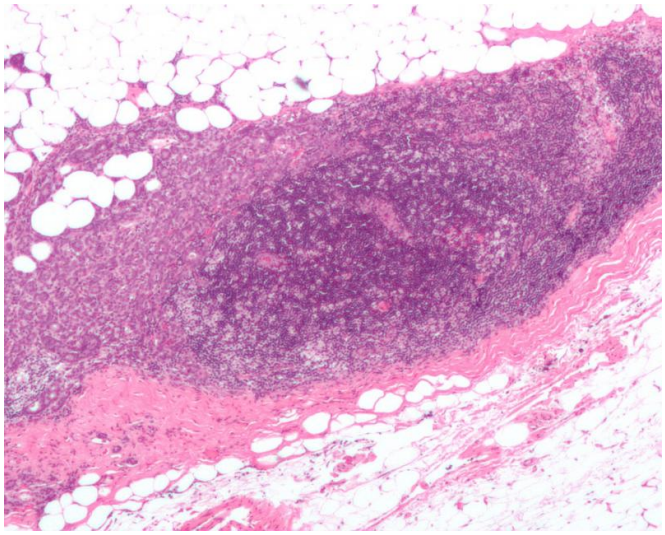

Micrograph showing a lymph node invaded by ductal breast carcinoma, with extension of the tumour beyond the lymph node. Credit: Nephron/Wikipedia

Ashkenazi Jewish women are known to have a predisposition to the inherited breast cancers BRCA1 and BRCA2, but currently genetic testing in this group is limited to women affected by breast and ovarian cancers and those who are unaffected but have a family history of the disease.

Ms Sari Lieberman, a genetic counsellor at the Shaare Zedek Medical Centre, Jerusalem, Israel, will tell the annual conference of the European Society of Human Genetics tomorrow (Sunday) that offering open-access BRCA testing to Ashkenazi women unaffected by cancer, regardless of their family history, enables the identification of carriers who would otherwise have been missed. Carrying one of the mutations for the BRCA genes means that women affected have a 50-80% risk of developing breast cancer and a 20-50% risk for ovarian cancer.

"We knew that half of these carriers have no family history of cancer, and therefore would not have been identified had the test been offered on the current personal and family history criteria," she says. "As a genetic counsellor, it is frustrating and saddening to see the results of this policy, where patients are often only identified as BRCA carriers once they have been diagnosed with cancer."

The researchers streamlined the pre-test process so that traditional genetic counselling, which can be time-consuming and difficult, was excluded. Instead they provided written information about the BRCA genes, the genetic test, and about the implications of being a carrier.

"Current strategies for testing focus on women who are 50 and older, which is not the optimal age for effective prevention. In order to address this, we would like to continue this study and look for other approaches that could include younger women," says Ms Lieberman.participants in the study either referred themselves or were recruited by health professionals. Two-year follow up of the 1771 women tested included looking at psychosocial outcomes and health behaviours. Both groups reported a high level of satisfaction (94%) and low stress. Those who had referred themselves tended to be more knowledgeable about breast cancer issues than those who were recruited.

"Among the 25 women carriers we identified, 94% expressed satisfaction and 92% endorsed the idea of population screening. Their stress was understandably higher, but it declined over time, and their knowledge was greater than in non-carriers. All of them had breast surveillance, and three underwent risk-reducing bilateral mastectomy. Of those aged over 40, fifteen out of a

total of 16 had their ovaries and Fallopian tubes removed in order to reduce risk," Ms Lieberman reports.

The researchers say that their study provides convincing evidence that open access genetic testing overcomes major barriers; not just lack of family history, but also referral and bureaucratic barriers, and that it is acceptable to those likely to be affected and their families.

"We were concerned that 'low risk' participants, with no family history, might not be able to cope with being offered BRCA testing and particularly with positive test results. We also worried that being found not to be a carrier might provide false reassurance and cause women to think they had no cancer risk and therefore avoid standard surveillance. We were pleasantly surprised on both counts," Ms Lieberman will say. In fact, mammography screening rates did not decline post-test in non-carriers, and even increased in some.

Falling prices for genetic sequencing and new techniques to avoid evaluating irrelevant gene variants will most likely make mutation screening available to wider populations in the near future. "We believe that our results are useful and highly relevant for other populations. On a personal note, I hope that this new approach means that one day I will not have to counsel someone with no family history and therefore no awareness of increased risk who says to me that she only wished she had known before," Ms Lieberman will conclude.

Chair of the ESHG conference, Professor Joris Veltman, Director of the Institute of Genetic Medicine at Newcastle University, Newcastle, United Kingdom, said: "This important study highlights the importance of population-wide genetic screening to identify women at risk of developing breast and ovarian cancer because of a genetic predisposition. The study also showed that most people cope very well with this genetic information; carriers of these mutations undertake breast cancer surveillance, whereas non-carriers are aware they can still develop breast cancer."

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