Current way of detecting gene mutations misses people at high risk of cancer

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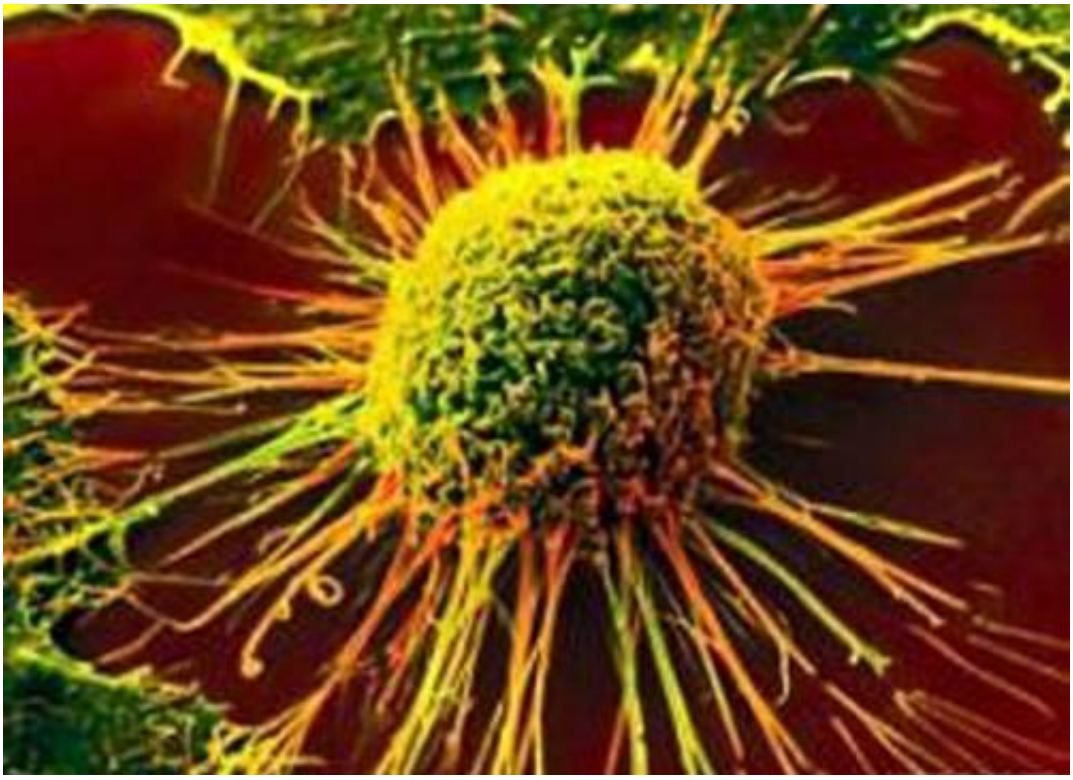

(Medical Xpress)—Research led by a University of Manchester academic on the BRCA gene mutation in the Jewish population shows that the current process of identifying people which relies on assessing someone's family history, misses half the people who have the mutation and are at risk of developing cancer.

Women carrying a BRCA 1 or 2 gene mutation have approximately a 15%-45% chance of getting ovarian cancer and a 45-65% chance of getting breast cancer. Women who know they are carriers can better manage their risk of developing cancer by enhanced screening or risk-reducing surgery.

Researchers running the Genetic Cancer Prediction through Population Screening (GCaPPS) study were funded by The Eve Appeal – the UK's only dedicated gynaecological research cancer charity.

They assessed the difference between family history-based testing and population screening in the Ashkenazi Jewish community, which is known to have a significantly higher number of people carrying the BRCA gene mutations than the non-Jewish population.

The researchers also carried out a cost-effectiveness analysis, and a study of the impact of testing on psychological health and quality of life. The results from both papers are published in the *Journal of the National Cancer Institute*.

In the study, 56% of people carrying a BRCA mutation were not identified by the existing family history criteria but were identified by population testing. Other key findings included:

- No significant difference in the short-term psychological impact or quality of life between those tested through population screening than those through family history testing.
- Screening all Ashkenazi Jewish women aged 30 and over, compared with family history testing, would cut the number of ovarian and breast cancers that occur and could potentially save the NHS £3.7million.

Athena Lamnisos, CEO, The Eve Appeal, said: "Women at increased

risk of cancer deserve far more than today's genetic screening process gives them. This study shows that broadening genetic testing beyond just family history saves more lives and more money."

Professor Ian Jacobs, Chief Investigator, GCaPPS and Vice-President and Dean of the Faculty of Medical and Human Sciences, University of Manchester, said: "Our findings have important implications. For the Ashkenazi Jewish community specifically, they suggest that population testing for BRCA1/2 mutations could save lives, is felt to be acceptable, and would be cost-effective.

"More broadly, they raise the possibility of new approaches to testing populations for cancer predisposing inherited genetic changes.

"As knowledge of the genetic basis of cancer increases, the acceptability to society of this type of testing rises and the cost of testing falls, we are likely to see rapid change in the way we deliver cancer genetic testing in health care."

Ranjit Manchanda, Trial Co-ordinator, GCaPPS and Clinical Lecturer in Gynaecological Oncology and Fellow in the Familial Cancer Clinic, UCH London, said: "This approach does not cause short-term psychological harm or impact quality of life on a population basis compared to the traditional approach of family history based testing. This is reassuring and indicates the change we may see in future screening."

Caroline Presho, 41, underwent a preventative double mastectomy when she was 36 years old after testing positive for a BRCA gene mutation three years earlier. Caroline's family is of Ashkenazi Jewish descent but despite losing both her aunt and father to cancer she didn't meet the family history criteria to have genetic testing.

She said: "The current NICE guidelines meant initially I wasn't eligible for BRCA screening, even with my Ashkenazi Jewish ancestry. Subsequent access to BRCA testing empowered me to make informed decisions about my personal health. This type of proactive healthcare not only saves lives, but will also save the NHS considerable amounts of money."

Provided by University of Manchester