Hundreds more breast cancer patients should be tested for BRCA1 gene

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(Medical Xpress) -- Leading breast cancer experts are calling for women under 50 who are diagnosed with triple-negative (TN) breast cancer to be offered testing for faults in the BRCA1 gene, according to a report published in the British Journal of Cancer.

The researchers – funded by Cancer Research UK and led by scientists at The Institute of Cancer Research – looked at more than 300 women with TN breast cancer and found BRCA1 mutations in nearly one in five women diagnosed under 50.

But due to the cost of testing for the BRCA1 mutation, NICE guidance recommends that BRCA1 should be offered if the likelihood of detecting a mutation is greater than 20 per cent, although many testing centres offer it if the likelihood is between 10-20 per cent.

Centres use a range of criteria and methods to determine if a patient is eligible for testing. But this requires specialist knowledge and software and potentially misses hundreds of women a year.

Based on their findings the researchers estimate more than one in three women with TN breast cancer caused by BRCA1 mutations would not have been tested using the current criteria.

Lead author Professor Nazneen Rahman, a Cancer Research UK funded researcher at the Institute of Cancer Research and the Royal Marsden Hospital, said: “Our findings show that women diagnosed with triple-

negative breast cancer under 50 should be offered BRCA1 testing. Using a simple age criteria for testing will provide a clear and understandable guide for doctors and women to follow, and should result in many more women benefitting from the optimised care that genetic information makes possible."

Changing the current testing guidelines could mean an extra 1,200 tests a year, which the researchers acknowledge will place an extra burden on current genetic testing services.

Professor Rahman added: "There are hurdles to overcome so that NHS testing services can cope with more BRCA1 testing, but we're moving towards an era of faster and cheaper genetic testing, so it will soon be possible."

Women carrying the BRCA1 mutation have up to a 65 per cent chance of developing breast cancer by the time they are 70. But only around one in 900 women in the general population carry a BRCA1 mutation.

Breast cancers with BRCA1 mutations can respond well to treatment with platinum-based drugs, such as carboplatin and cisplatin, and also to radiotherapy. But they can develop resistance to the treatment and start to grow again.

Rowena Kincaid, 36, from Cardiff, was diagnosed with breast cancer in July 2009 after finding a lump. It was confirmed as triple negative and she underwent a lumpectomy followed by four months of chemotherapy and radiotherapy. She is now doing well, is back at work and took part in Race for Life in 2011.

She said: "I am interested to know about the genetic testing, as I know there has been cancer in the family. To be offered the chance to find out if I carry the BRCA1 mutation would not only give me insight to my

own disease but also allow me and my family to discuss with our doctors if we carry the mutation what the next steps would be."

Professor Peter Johnson, chief clinician at Cancer Research UK, said: "It's important that we identify women and their families who carry BRCA1 mutations. They're more likely to develop breast and ovarian cancer, so armed with this knowledge doctors can offer targeted screening and tailored treatments to these women.

"The NHS needs to adapt so that tests for BRCA1 can be offered to women who are likely to carry the mutation. This approach will be cost-effective for the NHS in the long-term, leading to a substantial reduction in the number of breast and ovarian cancers by offering preventative treatments for those women and their families who are at greatest risk."

More information: L. Robertson et al BRCA1 testing should be offered to individuals with triple-negative breast cancer diagnosed below 50 years *British Journal of Cancer* (2012). doi:10.1038/bjc.2012.31

Provided by Cancer Research UK