Inherited gene fault influences breast cancer survival

December 11 2012

Researchers have shown that an inherited gene fault influences the chances of some women surviving breast cancer. It also increases the risk of women developing a second breast cancer. The research is published in this week's *Journal of Clinical Oncology*.

The scientists, funded by Cancer Research UK, found that women with the fault, and who had the oestrogen-receptor-positive form of breast cancer, were more likely to die from the disease - 62 out of 100 women who carry the fault will be alive ten years after being diagnosed compared with 73 of 100 who do not carry the fault.

The research also found that women with the fault were more likely to develop a second cancer. In the study, 24 per cent of women with the fault developed a second breast cancer compared to seven per cent

without the fault.

The gene fault, called CHEK2*1100delC, creates a faulty protein that interrupts a cell's ability to repair damaged DNA, so increasing the number of DNA mistakes that can lead to cancer.

The fault is carried by about one per cent of all women and it is already known that women with it are more likely to develop breast cancer.

Dr Paul Pharoah, lead researcher based at the University of Cambridge, said: "This is the first time we've shown how CHEK2 faults influence the long-term prognosis of breast cancer. We need further studies to see if we can find other similar faults that affect how the disease develops so that one day we can test and predict how each individual woman's breast cancer will behave."

The researchers also predict that women with this genetic mistake are more susceptible to other forms of cancer, contributing to the increased risk of early death after a breast cancer diagnosis.

Dr Julie Sharp, senior science information manager at Cancer Research UK, said: "We've made huge progress improving the diagnosis and treatment of breast cancer - in the 1970s around five out of 10 women with breast cancer survived beyond five years but now it's more than eight out of 10. But, we still need better ways to predict how the cancer will behave to help doctors treat the disease more effectively.

"These results suggest that more widespread testing for the CHEK2 fault could help identify women with oestrogen-positive breast cancers who are at a greater risk of developing a second breast cancer. Further research needs to be done to see if these women would benefit from long-term treatment with anti-oestrogen drugs, such as tamoxifen, to try and reduce this risk."

More information: Weischer, M et al CHEK2*1100delC heterozygosity in women with breast cancer associated with early death, breast cancer specific death, and increased risk of a second breast cancer *Journal of Clinical Oncology* (2012). jco.ascopubs.org/content/30/35/4308.full

Provided by Cancer Research UK