Update on Breast Cancer and Genetic Risk

High-risk genes

- The vast majority of breast cancer in the UK is sporadic and the inheritance of high-risk BRCA1/2 gene mutations only account for 1.6% of all breast cancer cases.
- However, the results of the POSH study showed that more than 10% of young women (under 40) with breast cancer have BRCA 1/2 gene mutations regardless of their family history.
- The BRCA genes are inherited in an autosomal dominant fashion and pass down the male and female lines so children of mutation carriers have a 50% chance of carrying the mutation too.
- The latest estimates indicate that a woman with a BRCA 1 gene mutation has a 40-87% risk and a woman with a BRCA 2 gene mutation has a 27-80% risk of developing breast cancer up to the age of 70. The wide variation in probability suggests that lifestyle factors such as obesity can modify the inherited risk.

Intermediate and Low-risk genes

- There are a dozen or so intermediate risk genes for breast cancer that include ATM, CHEK2 and PALB2.
- Low risk genes are single nucleotide polymorphisms (SNPs), which are much more common than BRCA1/2 mutations. Around 100 breast cancer SNPs have been identified and although each one carries a minimal risk, the risk can add up if a woman is found to carry multiple breast cancer SNPs.
- The PROCAS study looked at classical risk factors (obesity, menopausal status etc) and breast density on mammograms together with 18 SNPs and found that the information from the SNPs added important information for risk assessment.
- In the future, the results of the PROCAS study may be used as the basis to stratify breast cancer screening according to risk.

Routine testing for breast cancer SNPs and intermediate risk genes are not currently available on the NHS.

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