

Genetic Science Spotlight

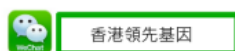
Peter MacCallum Cancer Centre Revealed that Women with BRCA1 Mutations Linked to Infertility



According to the National Cancer Institute, mutations in *BRCA1* are associated with a 60% lifetime risk of breast cancer in women and a 40% lifetime risk of ovarian cancer. Mutations in *BRCA2* are associated with a 49% lifetime risk of breast cancer and an 18% lifetime risk of ovarian cancer. Although all women who have the mutations are usually advised to have children at an earlier age, there is little evidence regarding the effects of the *BRCA1* and *BRCA2* mutations on female fertility. Researchers from Peter MacCallum Cancer Centre investigated the anti-Mullerian hormone (AMH) levels of 693 women and the results showed an average concentration of AMH that was 25% lower in women who carried the *BRCA1* mutation than in non-carriers. There was no evidence of an association between AMH concentration and *BRCA2* mutation status. The mechanism behind could be explained by the role of *BRCA1* in repairing DNA double stranded breaks and the lack of functional *BRCA1* will lead to inefficient DNA repair which can then lead to accelerated aging in a woman's eggs, hence female infertility. On the basis of these findings, it is highly advisable that women with the *BRCA1* mutation to either consider having children earlier or consider having their eggs frozen in order to keep open the possibility of having children in the future.

<https://academic.oup.com/humrep/article/31/5/1126/1750205/Anti-Mullerian-hormone-serum-concentrations-of>

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