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Responses to BRCA gene mutation & related medical issues

In view of the keen discussions on BRCA gene mutation, its screening, as well as the preventive measures of breast cancer among the general public recently, the Hong Kong Academy of Medicine (HKAM), being the statutory body and authority in the medical training and accreditation in Hong Kong, would like to take this chance to contribute our professional knowledge in raising the general awareness on the above stated topics.

1. *What is BRCA gene mutation? Is it common in Hong Kong?*

BRCA gene mutation is one of the known genetic factors that would raise the risks of developing certain cancers. It can be further subdivided into BRCA 1 and BRCA 2 mutations. According to the latest local data, there are only 8.5% of 914 selected high-risk families being detected to carry the BRCA mutations. If we consider in a broader sense, BRCA gene mutations just account for way less than 1% of the general population. Therefore, BRCA gene mutations are not common in this locality. This is consistent with findings which have been published in Western literature.

Not all carriers of BRCA mutations will have cancer. Medical evidences suggested that, the lifetime risk of developing breast cancer and ovarian cancer for BRCA1 carrier are 51-75% and 22-51% respectively, while those for BRCA 2 carrier are 33-54% and 4-18% respectively. Individual's risks also rely on other factors apart from the gene mutations, such as age, and presence of genetic modifiers

2. *Who need to be screened for BRCA gene mutations?*

As BRCA gene mutations are not common among the general public, HKAM only recommends those who are at high risk to consider taking the genetic testing after receiving detailed genetic counseling by specially trained medical personnel. People who are at high risk include:

- Female breast cancer patients, with:
 - at least 1 first degree relative having breast cancer or ovarian cancer, OR
 - bilateral breast cancer, OR
 - triple negative breast cancer [i.e. estrogen receptor (ER) -ve, progesterone receptor (PR) -ve and HER2 -ve]
- Male breast cancer patients
- Pre-menopausal ovarian cancer patients

3. *Where can BRCA gene mutations screening be done?*

For those who want to know more about their own breast cancer risk, or who are considering taking the BRCA gene mutation screening, they should first consult their family physician who would be in the best position to have the initial history taking and examinations done. Only under a doctor's referral should one receive the screening services in a certified center with well validated testing modality.



If someone seeks genetic testing under no supervision, no counseling and inaccurate data interpretation, not only will he/she be subjected to extra medical risks, but he/she may also have a chance of getting non-validated testing result, leading to unnecessary worries or a false sense of security because of the lack of professional assessment.

4. *What procedures are included in the screening process apart from the genetic testing?*

Before the laboratory works, a well-trained doctor, nurse or genetic counselor will first conduct a detailed genetic counseling with the individuals who are considering taking the test. Apart from the testing methodology and associated risks, the counseling session will also cover the clinical meaning of various test results and the potential effects on the test subject and his/her family members (including the emotional burdens, family planning, further screenings, treatments, etc.) Decision to take the test or not should only be made after a well-informed counseling session.

5. *After confirmation of carrying the BRCA mutated genes, is prophylactic mastectomy the only option for preventing breast cancer?*

After an individual has been confirmed of carrying the BRCA mutated genes, prophylactic mastectomy is one of the preventive measures, but not the only one, that can be taken. One should be also reminded that prophylactic mastectomy cannot guarantee a 100% reduction in risk of developing breast cancer. Other preventive measures include salpingo-oophorectomy, which is usually advised at forties or above when the family is completed, the use of specific hormonal drugs (including tamoxifen and raloxifene approved by FDA, and exemestane, supported by clinical research data) and regular examinations (mammography, ultrasonography or MRI scan) to detect early cancer and, therefore, receive treatment earlier.

6. *Besides screening BRCA gene mutation, what other measure should be taken to help detect breast cancer?*

For any women, with or without BRCA mutations, HKAM suggests them to observe for symptoms of breast cancers which include

- Change in size or shape of the breast
- Change in skin texture of the breast
- Rashes around nipple
- Bloody nipple discharge
- Presence of new lump or thickening in the breast or axilla

Whenever any of the above symptoms appear, one should seek medical attention as soon as possible for early detection and treatment.

Information provided by:

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Media members are welcome to enquire for more information of the above health issues through our hotline:

Media Enquiry Hotline of Hong Kong Academy of Medicine: 2505-5795

Note: The Hong Kong Hereditary Breast Cancer Family Registry provides free advice by genetically trained professionals