

***BRCA1* and *BRCA2* germline mutations in Asian and European populations**

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ABSTRACT

Women who carry a pathogenic mutation in the breast cancer susceptibility genes *BRCA1* or *BRCA2* (*BRCA*) have markedly increased risks of developing breast and ovarian cancers during their lifetime. It has been estimated that their breast and ovarian cancer risks are in the range of 46-87% and 15-68%, respectively. Therefore, it is of utmost clinical importance to identify *BRCA* mutation carriers in order to target unaffected women for prevention and/or close surveillance and to help affected women choose the best chemotherapy regimen.

Genetic testing for *BRCA* germline mutations is expanding in clinical oncology centers worldwide. Given the high costs of complete *BRCA* gene screens, a lot of effort has been expended on deciding upon whom to test. Relevant issues involved in decision making include the prior probability of a woman having a *BRCA* mutation, which is a function of her age and her disease status, her ethnic group, and her family history of breast or ovarian cancer.

The frequency and spectrum of mutations in these genes show considerable variation by ethnic groups and by geographic regions. Most studies have been conducted in European and North American populations, while studies in Asian, Hispanic, and African populations are fewer. In most populations, many *BRCA* mutations were identified, which were distributed all over the genes. However, in some populations, a relatively small number of specific *BRCA* mutations are recurrent and account for the majority of all mutations in that population. Many of the recurrent mutations are founder mutations, which were derived from a common ancestor. Founder mutations are present in Ashkenazi Jewish, European, and Islander (Faroe, Easter, and Pitcairn) populations. Such mutations have also been identified in patients from several Asian, South American, and African countries. Population-specific genetic risk assessment and genetic mutation screening have been facilitated at low costs. Given that mutations in the *BRCA* genes are distributed in populations throughout the world, it is important that the benefits of genetic testing and of targeted therapies be made available not only to women from developed countries in Europe and North America, but also to those from less developed countries in Asia, Africa and South America.

Keywords: *BRCA1*, *BRCA2*, founder mutations, hereditary breast cancer

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