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## Mutation pattern in *BRCA1/2* genes in Indian hereditary breast/ovarian cancers

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Hereditary breast/ovarian cancers are responsible for 5-10% of breast cancers and 7-10% of ovarian cancers. Breast cancer is one of the most common cancers in Indian women. Women carrying *BRCA1* or *BRCA2* mutations have a lifetime risk of between 60% and 85% for development of breast cancer, and 26% and 54% for development of ovarian cancer for *BRCA1*, and between 10% and 23% for *BRCA2*. In this study, 61 breast and/or ovarian cancer patients with a positive family history of breast and/or ovarian cancer were screened for *BRCA1/2* mutations. For mutation screening, conformation sensitive gel electrophoresis was used, followed by DNA sequencing, were heteroduplexes were detected. In the *BRCA1* gene, 15 mutations were identified; (mutation frequency, 24.6%) and in the *BRCA2* gene, two mutations were detected (mutation frequency, 3.28%). Of the *BRCA1* mutations identified, 3 were novel mutations and 3 more were previously reported mutations. The mutation, 185delAG was found in 10 patients at a very high frequency of 16.4%. This mutation is detected in high proportion in Ashkenazi Jewish population (18% in breast/ovarian cancers and 1% in general population). A large number of polymorphisms were also detected in *BRCA2* gene, which were normal population variants. The mutation spectrum of *BRCA1/2* in other Indian studies also indicate a higher incidence of 185delAG mutation, and the important studies shall be reviewed. Haplotype analysis was carried out, and it was found to be different from Ashkenazi Jewish population. The possibility of founder mutation status need to be considered for *BRCA1* 185delAG mutation.

### Biography

Kannan Vaidyanathan, MBBS, MD (Biochemistry) did his Postdoctoral fellowship from Indian Institute of Science, Bangalore, India. He is Professor & Head, Department of Biochemistry and Deputy Medical Superintendent, Pushpagiri Medical College Hospital, Tiruvalla, Kerala. He has 75 publications, 25 indexed in PubMed, 3 textbooks and 2 textbook chapters. His textbook on Biochemistry sells ~25000 copies worldwide every year and has been translated to other languages, including Slovakian and Spanish. Awards won by him include APFCB (Asia Pacific Federation of Clinical Biochemists) Silver Jubilee scholarship (2007) and KP Sinha – PS Krishnan award for best original research article in *Indian Journal of Clinical Biochemistry* (2011).

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