

Genetic epidemiology of breast cancer

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Cancer causation is multi-factorial, and, in most individuals, cancer risk is a consequence of genetics and epidemiological factors. Identification of the underlying variation in genes that promote breast cancer and their interactions with epidemiological factors will allow us to better understand the causes of this disease. This knowledge will allow researchers to develop more accurate models to assess breast cancer risks, to target those women at highest risk of developing the disease for prevention strategies, and to provide new paradigms for treating the disease. A family history of breast cancer has been identified as a major risk factor for the development of breast cancer, with estimates of a two to ten-fold increased risk to first-degree relatives of a breast cancer case. Mutations in *BRCA1* and *BRCA2* only explain a proportion of the familial risk, suggesting that there are other genetic effects, probably of low to moderate penetrance, explaining the remainder. *ATM*, *CHEK2*, *TP53*, and *PTEN* have also been shown to play modest roles in accounting for breast cancer risk. Much research is currently being devoted to identifying genes of low to moderate penetrance that increase breast cancer risk in the general population. This talk will focus on describing the approaches that are being used to identify these genes and the genes and pathways that are being studied.

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