REVIEW

Hereditary Breast Cancer: High Risk Genes, Genetic Testing and Clinical Implications

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SUMMARY

About one in eight to ten women living in Western countries will develop breast cancer during her lifetime and between 5-10% of these cases result from an inherited susceptibility to the disease. Within the past few years, a number of genes associated with a high risk of breast cancer have been identified, including BRCA1, BRCA2, TP53, PTEN, MLH1, MSH2, and STK11. The identification of these genes, together with the rapid advances in molecular genetic analyses, should improve the diagnosis and therapy of breast cancer. This article reviews the genetic basis of hereditary breast cancer, in particular the contribution of BRCA1 and BRCA2 and discusses the clinical application of this new molecular knowledge with regard to molecular testing, surveillance and prevention in women with a hereditary predisposition to breast cancer. (Clin. Lab. 2000;46:447-461)