



Cancer Genomics: Chapter 13. Breast Cancer Genomics

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Breast cancer is the most common cancer in women worldwide and the second leading cause of cancer deaths. Although early diagnosis, outcome prediction and treatment options are the ultimate objectives when assessing breast cancer patients, the methodology behind this clinical assessment varies and has gradually evolved from using standard clinical criteria into incorporating high-throughput genome-wide analysis. Early methods involved evaluating tumor size and spread as well as histological assessment (tumor grade). Later, the expression of hormone/growth receptors (ER, PR, and HER2) was added to the standard stratification of breast cancer patients. More recently, molecular approaches, which are based on the expression of a well-defined set of genes, have subdivided patients into five clinically relevant subtypes which not only predict prognosis and dictate treatment choice but also complement standard assessment. The advent of genome-wide analysis has produced the most robust classification system of breast cancers by coupling specific genetic aberrations (single nucleotide mutations and gene copy number variations) with gene expression profiles. Although these genome-wide approaches offer a promising future for breast cancer prognosis and treatment options, they are still not clinically feasible for standard population-based screening. Nonetheless, these approaches are becoming faster and more reliable in understanding the molecular architecture of breast cancer and are slowly paving the way towards personalized treatments which are tailored to individual patients. In the light of a rapidly evolving field of breast cancer genomics, this chapter highlights key standard and upcoming approaches for diagnosis, prognosis and treatment and discusses the feasibility of genome-oriented personalized treatments.

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