

## Epigenetic modifications in breast cancer as biomarkers

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Biomarkers are substances that are present in a considerable amount in the body that can be measured when a change in body or development of disease occurs. Any molecule, genetic material, proteinaceous compounds and epigenetic alterations can serve as a biomarker. Breast cancer is a complex disease and mammography which is the screening tool used for detection of breast cancer is not highly sensitive and specific. Epigenetic modifications in breast cancer like DNA hypermethylation can serve as biomarkers which in turn help in the early detection of cancer. The promoter region is methylated in tumour suppressor genes. This methylation pattern has been observed in the bloodstream of breast cancer patients. BRCA1 (breast cancer gene 1) gene is known to be a tumour suppressor gene. Inactivation of this gene leads to the progression of breast cancer and tumourigenesis in sporadic type breast cancer. Loss of BRCA1 will result in the prevention of repair of DNA breaks which is the mode of action of platinum-based chemotherapy. Therefore, BRCA1 hypermethylation is sensitive to platinum-based chemotherapy. It has been observed that these DNA methylation patterns occur in the bloodstream of a patient since way before breast cancer is detected. Hence, studying methylation patterns can help in diagnosing breast cancer diagnosis at a very early stage. Other substances which can serve as biomarkers have also been found. For example, hypermethylation of hyaluronoglucosaminidase can be detected in the early stages of breast cancer. RASSF1A (ras association domain-containing protein 1) and RAR $\beta$ 2 (retinoic acid receptor beta 2) also undergo DNA hypermethylation and act as biomarkers in the early stages of breast cancer. Since there are several types of breast cancer, one kind of biomarker might work for the detection of a particular cancer type and not for others. Hence, a misdiagnosis is possible. All biomarkers for all breast cancer types have yet not been found. Other epigenetic modifications like histone modifications can also be studied to serve as biomarkers. Hence, it is important to study epigenetic alterations as they can act as biomarkers in the early detection of breast cancer.

*Keywords: Biomarker, DNA methylation, Chemotherapy, Breast cancer, BRCA1*

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