The Identification of a Potential Novel Breast Cancer Biomarker Through Hypomethylation Analysis at Transcription Ending Site (TES)

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Breast cancer (BC) is the most frequently diagnosed disease among women, with 2.3 million new cases and 669,000 deaths identified in 2022 alone. Most studies on epigenetic diagnosis biomarkers for BC focus on hyper/hypomethylation patterns of genes at the transcription starting sites, near the promoter region. The aim of this work is to explore the role of epigenetic patterns at the transcription ending sites (TES). Since transposable elements can act as alternative polyadenylation signals, it was hypothesized that hypomethylation at transcription ending sites (TES) may serve as a biomarker for breast cancer because increased activity in transposon regions can induce mutations. The methylation levels of bisulfite-treated reads from publicly-accessible healthy and cancerous human breast cell lines data were compared by extracting methylation metrics at each cytosine in the dataset. Upon producing a visualization plot of the average methylation levels across the gene set, a hypomethylation downward spike pattern was observed at TES. Subsequently, calling upon differentially methylated regions (DMRs) reported statistical significance ($q < 0.05$) in the observed pattern, suggesting that hypomethylation at TES is a significant candidate for a BC biomarker. The identified hypomethylated TES pattern provides a new potential BC risk diagnosis pathway that is more cost-efficient than imaging techniques and less invasive than biopsy-based diagnostic methods, as the hypomethylation pattern could be identified through circulating tumor DNA in blood.

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