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Singapore launches open-access tool to decode DNA change patterns in breast cancer

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New study specifically examined breast cancer genomes from The Cancer Genome Atlas (TCGA) and METABRIC



A study led by Dr Jason Pitt, Principal Investigator at the Cancer Science Institute of Singapore (CSI Singapore), has identified eight new "signatures" of DNA patterns (gains and/or losses) in breast cancer.

By analysing nearly 2,800 genomes, the team systematically profiled changes in the number of DNA copies in breast cancer, with the goal of better understanding the underlying mechanisms of tumour development and evaluating how these structural genomic changes relate to clinical outcomes.

The identified signatures could help refine future diagnostic tools, as well as to better match breast cancer patients with targeted therapies.

While genomic instability is a hallmark of cancer, previous research often relied on broad patterns that apply across many different types of the disease. This new study specifically examined breast cancer genomes from The Cancer Genome Atlas (TCGA) and METABRIC, both of which are open-access databases.

The research team was able to break down known, broad genetic signatures into more detailed, disease-specific categories, revealing a complex interaction between genome instability and the tumour's immune microenvironment.

To ensure the findings benefit the wider scientific community, the researchers launched the CNA Visualizer. This open-access web tool allows scientists worldwide to interact with and visually explore the massive dataset of various cancer genomes.

The next phase of this research will focus on validating these genetic signatures in clinical settings to assess their reliability in predicting patient responses to targeted therapy. Additionally, Dr Pitt and his team plan to further explore how the interplay between genome instability and the tumour microenvironment influences long-term clinical outcomes.