Taming batch effects in cell-free DNA data

Prof. WONG Lim Soon
KITHCT Chair Professor
School of Computing
National University of Singapore
Singapore

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Abstract

Cell-free DNA (cfDNA) is a noninvasive window into human health, with applications from early cancer detection to prenatal testing. Yet cfDNA analysis is challenging: GC-related biases and batch effects can strongly confound results, especially in low-pass sequencing, limiting the sensitivity of applications such as liquid biopsy and noninvasive prenatal testing. I will present GCfix, a fast, single-sample method for correcting GC bias and batch effects in cfDNA. GCfix requires no training or adaptation and works directly on individual samples. Built on a detailed understanding of cfDNA-specific bias at both the region and fragment-length level, it consistently outperforms existing tools, particularly in low-pass sequencing scenarios. This work is based on research by my student Rafeed Rahman.

About the Speaker

Prof. Wong Limsoon is the Kwan-Im-Thong-Hood-Cho-Temple Professor in the School of Computing at the National University of Singapore (NUS) and an Honorary Professor of Pathology at NUS Medicine. He was elected Fellow of the ACM in 2013 and Fellow of the Singapore National Academy of Science in 2024, in recognition of his contributions to database theory and computational biology. His honours include the FEER Asian Innovation Gold Award (2003) for advances in childhood leukemia treatment optimization, and the ICDT Test of Time Award (2014) for his work on naturally embedded query languages. He co-founded Molecular Connection (Molcon) in India and, beyond the company's 400-fold growth over the past two decades, is particularly proud that Molcon has become one of the best companies in India for women to work in.