

DEPARTMENT OF COMPUTER SCIENCE

SEMINAR

2023 SERIES

Repeat-aware Insertion Calling and its Clinical Applications

DATE & TIME

10 OCT 2023 (TUE) 2:00 PM – 3:00 PM

VENUE

Mr. and Mrs. Lee Siu Lun Lecture Theatre (WLB 205),
The Wing Lung Bank Building for Business Studies, Shaw Campus



PROF. WING-KIN SUNG

Global Stem Professor
Department of Chemical Pathology
The Chinese University of Hong Kong

ABSTRACT

Insertions are one of the major types of structural variations and are defined as the addition of 50 nucleotides or more into a DNA sequence. Several methods exist to detect insertions from next-generation sequencing short read data, but they generally have low sensitivity. Our contribution is two-fold. First, we introduce INSuVeyor (Nature communication, to appear), a fast, sensitive and precise method that detects insertions from next-generation sequencing paired-end data. Using publicly available benchmark datasets (both human and non-human), we show that INSuVeyor is not only more sensitive than any individual caller we tested, but also more sensitive than all of them combined. Furthermore, for most types of insertions, INSuVeyor is almost as sensitive as long reads callers. Furthermore, we will highlight the practical application of our improved method.



SPEAKER'S
BIOGRAPHY



REGISTER NOW