ASHBi SEMINAR

Multitudes of genomes: pan-genomic methods and applications

Lecturer: Cristian Groza

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Date Monday, 29 May 2023

Time 11:00 – noon [JST]

＼Hybrid*／

Venue Seminar Room / Zoom Online

1F, Faculty of Medicine Bldg. B

*Register via the right QR code

Abstract

Reference genome sequences facilitate many aspects of daily research, from designing primers for PCR assays in molecular biology experiments to analyzing massive amounts of sequencing data with highly parallel bioinformatic methods. However, reference genomes are just one example of a possible genome for any given organism and can only approximate the actual genomes that are encountered in the lab. Therefore, genomic methods are slightly biased by these differences and may fail due to "reference bias".

Fortunately, long read sequencing technologies and assembly algorithms are now producing many reference quality genomes of humans and model organisms. To take advantage of this new wealth of data, pan-genomic methods have been developed to create genome graphs, a data structure that represents a population of genomes by collapsing conserved sequences and highlighting polymorphic sequences. Thus, the genetic diversity of a species is not ignored, which provides multiple benefits for downstream analysis and paves the way for other applications.

For example, the sensitivity and specificity of common tasks such as genotyping is improved. In particular, pangenomic methods detect 104% more structural variants compared to methods that use a single reference genome. Moreover, it is now possible to interrogate the expression and epigenomic status of structural variation using RNA-seq, long read methylation or even short read epigenomic data. In this seminar presentation, I will review other ways in which pangenomic methods may augment biological research.

Organizer：Graduate School of Medicine

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