# ASHBi SEMINAR

## Genome analysis platforms in the era of massive publicly available sequence data and pangenomes


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Division of Genome Analysis Platform Development
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**Date** Monday, 17 June 2024

**Time** 15:00 – 16:00 [JST]

**Venue** Conference Room
(B1F, Faculty of Medicine Bldg. B)

*Register via the right QR code

## Abstract


As various types of omics data continue to accumulate, the development of algorithms and infrastructure for analyzing them is becoming increasingly important. In this lecture, I would like to introduce two topics.

### 1. Autonomous knowledge acquisition platform from large-scale public transcriptome sequencing data

One of our primary research goals is to develop a system that autonomously derives "knowledge" from large-scale data, such as the Sequence Read Archive, to drive innovation in science and medicine. I would like to introduce our efforts in systematically developing registries of genomic variants causing splicing changes using massive publicly available transcriptome data (Shiraishi et al., Nature Communications, 2022; Iida et al., bioRxiv, 2024), and discuss how these can be applied to future medicines and therapeutics.

### 2. Centromere analysis using long-read sequencing and pangenome

There are still large portions of dark matter regions in human genomes, including centromere sequences, that have remained largely unexplored by short-read technologies. I will present our analysis of unbalanced translocation der(1;7)(q10;p10) by reconstructing centromere sequences using extensive long-read sequencing data. Additionally, we will introduce our attempts to maximize the utility of short-read sequencing data for centromere analysis by effectively utilizing pangenome reference sequences.


Organizer : Graduate School of Medicine Institute for the Advanced Study of Human Biology (WPI-ASHBi)


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