ASHBi SEMINAR

Toward a cell-type-specific understanding of complex diseases

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Time 13:30 – 14:40 [JST]

Venue Conference Room Onsite Only*
B1F, Faculty of Medicine Bldg. B

Register here

Abstract

High-throughput genotyping and sequencing have uncovered thousands of disease-associated variants, most of which reside in non-coding regions, making their functional mechanisms unclear. Investigating genetic effects in the appropriate cell types is crucial for understanding complex diseases. We highlight the use of cell-type-specific assays in three diseases. For coronary artery disease (CAD), which is 40-60% genetically driven, we identified five risk genes, including TCF21 and SMAD3, by integrating eQTL and GWAS data, with validation in mouse models. In age-related macular degeneration (AMD), we discovered that a retinal pigment epithelium (RPE) eQTL variant regulates splicing in RDH5, leading to RNA decay, establishing RDH5 as a key risk gene. Finally, using ~1M PBMC single cells from the Asian Immune Diversity Atlas (AIDA), we demonstrated the utility of single-cell sQTLs in dissecting autoimmune disease mechanisms.

Organizer : Graduate School of Medicine
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