IWSG2017 サテライトセミナー

日時：2017年 6月 26日（月）18:15–19:15
場所：最先端医療イノベーションセンター棟
　　　1F マルチメディアホール

演題：

Mapping cellular quantitative trait loci in human IPS-based cell model systems

講師： Dr. Daniel Gaffney
　　　 (Wellcome Trust Sanger Institute)

Induced pluripotent stem cells (IPSCs) are a powerful model system for studying the cellular basis of human health and disease. The Human IPSCs Initiative is a collaborative project generating IPSC lines with genotype and multi-omic phenotype information from >300 healthy individuals, which will be made available to the research community and industry. I will discuss describe the initial analysis of the genomes, epigenomes, transcriptomes, proteomes and cell behaviours of >700 IPS lines from healthy individuals. Through genome-wide profiling we find that 5-46% of the variation in different iPSC phenotypes arises from differences between individuals. We assess the phenotypic consequences of rare, genomic copy number mutations that are repeatedly observed in iPSC reprogramming and present a comprehensive map of common regulatory variants affecting the transcriptome of human pluripotent cells. Finally, I will discuss our work using the HIPSCI resource to examine how common genetic variants alter cellular phenotypes in immune response.

1. Kilpinen, H. *et al*. Common genetic variation drives molecular heterogeneity in human iPSCs. *Nature* 2017.
2. Kumasaka, N. *et al*. Fine-mapping cellular QTLs with RASQUAL and ATAC-seq. *Nature Genetics* 2016.
3. Rouhani, F. *et al*. Genetic background drives transcriptional variation in human induced pluripotent stem cells. *PLoS Genetics* 2014.