医学セミナー・第 17回分子遺伝疫学セミナー
"Seminars in Medical Sciences" Lecture

ゲノム医科学リサーチユニット　難治性免疫疾患・アレルギーリサーチユニット

Approaches to identify disease causing or susceptible functional variants out of millions of variations detected by human whole-genome sequencing

Speaker: Dr. Yuki Hitomi (Assistant Professor, Department of Human Genetics, The University of Tokyo)（人見祐基博士、東京大学大学院医学系研究科人類遺伝学分野助教）

Date:　October 3, 2018 (Wed)

Time:　17:00-18:15

Venue: Clinical Lecture Room B（臨床講義室 B）

This seminar is one of the seminars for the subject "Seminar in Medical Sciences" in Doctoral Programs in Biomedical Sciences and Clinical Sciences. The seminar will be given in English, but questions in Japanese are also welcome.

This seminar will NOT be recorded; therefore, be sure to attend if are interested.

Abstract:

Any of the genetic variations in human genome may cause differences in disease risk. So far, whole genome sequencing (WGS) using short-read type Next-Generation Sequencing (NGS) and Genome-Wide Association Study (GWAS) have become indispensable methods for comprehensive screening of "causal variants in rare disorders" and "susceptibility gene loci in common disorders", respectively. However, molecular genetic approach for the identification and evaluation of disease-causal/susceptibility variants from millions of genetic variations in the human genome should be included in follow-up studies.

By performing systematic searches for disease causing or susceptible variants including their functions, we will be able not only to understand the molecular mechanisms of pathogenesis, but also to establish novel drugs based on the information obtained from the functional effects of the variants.

In this seminar, I would like to introduce the molecular genetic approaches for identification and evaluation of functional disease-causal/susceptible variants from comprehensive whole-genome analysis (WGS and GWAS) in rare neurological disorders and common immune-related disorders.

References:

1. Hitomi Y and Tokunaga K. Significance of the functional disease-causal/susceptible variants identified by whole-genome analyses for the understanding of human diseases. ***Proc Jpn Acad Ser B Phys Biol Sci*** 93: 657-676, 2017.
2. Hitomi Y, *et al.* Identification of the functional variant driving *ORMDL3* and *GSDMB* expression in human chromosome 17q12-21 in primary biliary cholangitis. ***Sci Rep*** 7: 2904, 2017.
3. Epi4K Consortium, *et al. De novo* mutations in epileptic encephalopathies. ***Nature.*** 12; 501: 217-221, 2013.

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