セミナーのお知らせ

"Using Drosophila to discover and study new human disease causing genes"

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Date: Jan 24th Friday
Time: 4pm to 5 pm
Location: Biken Hall, 1st Floor, Main building, Research Institute for Microbial Diseases (微研ホール　微生物病研究所 本館 1F)

Many rare and undiagnosed diseases are caused by mutations in the patient's genomic DNA. Identification of mutations responsible for these disorders can facilitate the understanding of biological functions of these genes in human biology. In the past decade, we have been using both forward and reverse genetics approaches in Drosophila melanogaster (fruit flies) in combination with state-of-the-art human genomics techniques to discover new human disease causing genes. Through sophisticated genetics tools in flies, we have been able to further study and dissect the underlying molecular mechanisms of many rare neurological diseases, which often provides new insights into more common diseases including Alzheimer's disease and Zika virus mediated microcephaly. Opportunities for basic scientists to directly contribute to clinical and translational research are increasing globally due to establishment of collaborative consortiums such as Undiagnosed Diseases Network (UDN, USA), Rare Disease Models and Mechanisms Network (RDMM, Canada) and Initiatives for Rare and Undiagnosed Diseases (IRUD, Japan), and model organism researchers are playing a critical role in this world-wide endeavor.

参考文献

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2. Splinter et al, 2018 N Eng J Med "Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease." PMID: 30304647
3. Wangler et al, 2014 Genetics "Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research." PMID: 28874452
4. Bellen et al., 2019 Hum Mol Genet "The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases." PMID: 31227826

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