CDB SEMINAR

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Monday, May 15, 2017

16:00~17:00 Seminar Room A7F

## *Drosophila* genetics for human biology and medicine

Many rare undiagnosed diseases are caused by mutations found in the patients' genomic DNA. Identification of mutations responsible for these disorders can facilitate the understanding of biological functions of these genes in human biology. However for many neurological and most psychiatric diseases, the molecular causes are yet to be identified. By performing genetic screens in *Drosophila*, characterizing the *in vivo* function of novel genes in development, function, and maintenance in the fly nervous system, and further identifying patients with rare deleterious variants in their human homologs, we have been successful in identifying a number of new disease causing genes in human (ANKLE2 in microcephaly: Yamamoto et al., 2014 *Cell*, NRD1 & OGDHL in neurodegeneration: Yoon et al., 2017 *Neuron*). More recently, we developed a novel method to "humanize" a *Drosophila* gene (Bellen and Yamamoto, 2015, *Cell*) to study the functions of newly identified human disease-liked genes and variants in vivo in flies (TM2D3 in Alzheimer's disease: Jakobsdottir et al., 2016, *PLoS Genetics*, EBF3 in a novel neurodevelopmental disorder: Chao et al., 2017, *AJHG*).

In this seminar, I will discuss how both forward and reverse genetic strategies can aid in the function annotation of novel disease-linked human genes and variants, and will emphasize the importance and value of team science in biomedical research.

References:

Yamamoto S et al. A *drosophila* genetic resource of mutants to study mechanisms underlying human genetic diseases. *Cell*. 2014 Sep 25;159(1):200-14. PMID: 25259927

Yoon WH et al., Loss of Nardilysin, a Mitochondrial Co-chaperone for α-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. *Neuron*. 2017 Jan 4;93(1):115-131. PMID: 28017472

Bellen HJ, Yamamoto S. Morgan's legacy: fruit flies and the functional annotation of conserved genes. *Cell*. 2015 Sep 24;163(1):12-4. PMID: 26406362

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