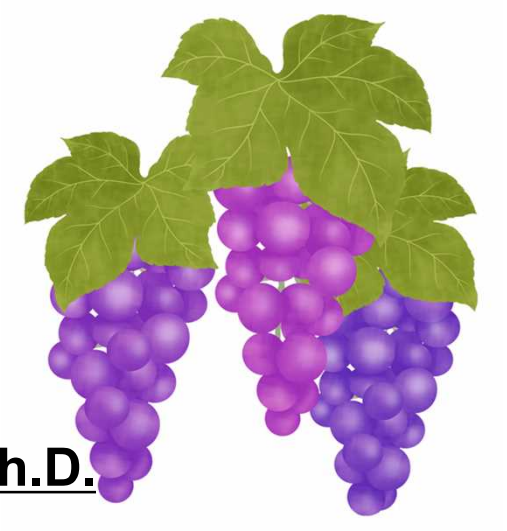
41st IRCMS Seminar



Date: **September 25, 2018 (Tue)**

Time: 17:00-18:00

Venue: 1F Meeting Lounge
International Research Center
for Medical Sciences(IRCMS)

Speaker: **Hideki Makishima M.D., Ph.D.**
Associate Professor
Department of Pathology and Tumor Biology, Kyoto University

Title: **Sequential acquisition of mutations in myelodysplastic syndromes**

Abstract: Recent progress in next-generation sequencing technologies allows us to discover frequent mutations throughout the coding regions of myelodysplastic syndromes (MDS), potentially providing us with virtually a complete spectrum of driver mutations in this disease. As shown by many study groups these days, such driver mutations are acquired in a gene-specific fashion. For instance, *DDX41* mutations are observed in germline cells long before MDS presentation. In blood samples from healthy elderly individuals, somatic *DNMT3A* and *TET2* mutations are detected as age-related clonal hematopoiesis and are believed to be a risk factor for hematological neoplasms. In MDS, mutations of genes such as *NRAS* and *FLT3*, designated as Type-1 genes, may be significantly associated with leukemic evolution. Another type (Type-2) of genes, including *RUNX1* and *GATA2*, are related to progression from low-risk to high-risk MDS. Overall, various driver mutations are sequentially acquired in MDS, at a specific time, in either germline cells, normal hematopoietic cells, or clonal MDS cells.

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