D5 Medical & Life Science Seminar (Elective 2 credits)

Academic Year 2018 "International Biomedical Research Seminars"

Title: # Runx tales: a small transcription factor family and human diseases

Speaker: **Motomi Osato, M.D., Ph.D.**

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Date: **May 30, 2018 (Wednesday)**

Time: 17:30 –

Venue: IRCMS 1F Meeting Lounge

Abstract

In 2016, WHO revised its classification of myeloid neoplasm and acute leukemia, incorporating several new clinical entities associated with specific genetic changes, one of which is a point mutation in the *RUNX1* gene. Notably, this genetic alteration was world-first detected by us in Kumamoto in the late '90s. In addition, *RUNX1* and its heterodimeric partner *CBFB* are also frequently affected in chromosomal abnormalities such as t(8;21), t(12;21) and inv(16). Germline *RUNX1* mutations cause an inherited hematological disease, called familial platelet disorder with predisposition to acute myeloid leukemia (FPD/AML). Gene targeting studies in mice demonstrated that *Runx1* and *Cbfb* are both essential for the generation of hematopoietic stem cells. Altogether, *RUNX1* is now widely believed to be the master regulator for hematopoiesis. Similarly, another family gene, *RUNX2*, is the master regulator for bone formation. Its knockout mice completely lack the ossified bone and its haploinsufficiency in human results in craniofacial anomaly, named cleidocranial dysplasia (CCD). In the era of ever-accelerating population aging, osteoporosis is one of the most common geriatric health concerns. Pharmacological RUNX2 activation is a promising therapeutic avenue for osteoporosis. The third family gene, *RUNX3*, is recently found to cause congenital progressive neurological disease in human. RUNX3 is also a key molecule in immunology and associated with cancers. Such widespread involvements of RUNX in a plethora of human diseases are good indicative of its fundamental role throughout multiple organs. Efforts made by us and others in attempt to unveil multifaceted RUNX functions will be described in the seminar, with emphasis on historical notes and episodes behind the seminal discoveries.

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