

# ASHBi SEMINAR

**Mutation and selection on tandem repeats promote genetic and phenotypic diversity across species.**

**Lecturer: Dr. Maria Anisimova**

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**Date** Tuesday, 10 October 2023

**Time** 16:00 – 17:00 [JST]

**Venue** Conference Room

**B1F, Faculty of Medicine Bldg. B**

*\*Register via the right QR code*



## Abstract

Tandem repeats (TRs) are frequent in genomes across the tree of life. TRs contribute to fundamental biological functions and can be associated with virulence, resistance, and human diseases. TR diversity offers a perfect playground for natural selection. However, TR studies are typically non-trivial due to their high mutability. In this talk I will discuss some statistical methods for analyzing TRs and present several applications. For example, we analyzed TRs with long repeat units (> 10-15 residues) across eukaryotic proteomes. We found that most TRs are ancient, with TR regions preserved since distant speciation events. In contrast, short TRs (STRs, with units of 1–6 bp) are orders of magnitude more mutable than other well-studied genotypic variants; STRs therefore make major contribution to genetic and phenotypic divergence. Our current work shows that STR mutations might regulate gene expression in colorectal cancer (CRC). We identify putative expression STRs (eSTRs) for which the STR copy number is associated with gene expression in CRC tumors. Linear models describing eSTR-gene expression relationships allow for predictions of gene expression changes in response to eSTR mutations. Furthermore, we found an increased mutability of eSTRs in MSI tumors. This may be an early indication that eSTR mutations confer a selective advantage for some CRC tumors. Our evidence of gene regulatory roles for eSTRs in CRC highlights a largely unexplored way through which tumors may modulate their phenotype.

Organizer : Graduate School of Medicine

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