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*Relating enhancer genetic variation across mammals to
complex phenotypes using machine learning*

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Abstract: Abstract: Many mammalian characteristics have evolved multiple times throughout history. For example, humans and dolphins have larger brains relative to body size than their close relatives, chimpanzees and killer whales. We want to identify the parts of the genome associated with these characteristics by comparing the genomes of hundreds of mammals. Rather than focusing on the small proportion of the genome that encodes genes, which cannot fully explain many of characteristics' evolution, we present a new approach that uses machine learning to find conserved patterns of sequences at candidate enhancer regions, which control the levels of genes expressed in specific tissues. We established a new set of evaluation criteria for these machine learning models and used these criteria to compare our models to previous methods for this task. When applying our approach to the brain, we identified dozens of new enhancers associated with the evolution of brain size relative to body size and vocal learning.

Bio: Irene Kaplow received her B.S. in Mathematics with a minor in Biology from the Massachusetts Institute of Technology in 2010. There, she began her career as a computational biologist while doing research with Bonnie Berger. She then went to graduate school at Stanford University, where she received her Ph.D. in Computer Science in 2017. At Stanford, she worked in the Hunter Fraser and Anshul Kundaje's labs to develop methods to analyze novel high-throughput sequencing datasets to better understand the roles of DNA methylation and Cys2-His2 zinc finger transcription factor binding in gene expression regulation. Irene is now a Lane Postdoctoral Fellow in Andreas Pfenning's lab in the Computational Biology Department at Carnegie Mellon University, where she is developing methods to identify enhancers involved in the evolution of neurological characteristics that have evolved through gene expression.

Friday, October 21, 2022, 1:00 pm
Mathematics and Science Center: MSC W201

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