

# Department Colloquium Department of Mathematics



Friday, December 4, 2:00 pm PST

Zoom Meeting Room Number 981 5791 3939

## Our Guest Speaker



**Dr. Liliana Florea**, Associate Professor of Medicine in the McKusick-Nathans Department of Genetic Medicine and the Department of Computer Science at [the Johns Hopkins University](#), develops computational methods and tools for analyzing large-scale sequencing data to help characterize molecular mechanisms of diseases. Before joining the Johns Hopkins faculty in 2011, Dr. Florea served on the faculty at the University of Maryland and the George Washington University. Previously, she was a member of the team of scientists at Celera Genomics that assembled the first sequence of the human genome. Her work has been recognized with a Sloan Research Fellowship in Computational and Evolutionary Molecular Biology and has been funded by several NSF and NIH awards. Dr. Florea holds a Ph.D. degree from the Pennsylvania State University (2000).

## Our Event

### Computational approaches to characterize gene alternative splicing variations from short read sequencing data

Eukaryotic genes are interrupted, appearing on the genome as informative segments (exons) interspersed with 'spacer' DNA (introns). During the process of splicing, introns are removed and exons are joined together to form the gene sequence (mRNA). Further, through alternative splicing the gene can select different combinations of exons or exon segments, leading to different gene products called splice isoforms or mRNA transcripts, and ultimately contributing to functional diversity in an organism and through evolution. We first describe computational methods to reconstruct the repertoire of genes and splice isoforms and to quantify their abundance from abundant short read sequencing data, a process known as transcript assembly. Our suite of tools CLASS, CLASS2 and PsiCLASS represent increasingly complex algorithms centered on the splice graph data structure. Secondly, we will describe statistical methods for identifying divergent splicing patterns between different cellular conditions, such as between tissue samples from patients versus healthy controls. Our tool MntJULiP uses a zero-inflated negative binomial distribution and a Dirichlet multinomial distribution to model two aspects of the problem, namely differential splicing ratio versus differential isoform abundance. We discuss the problems, heuristics, performance evaluations, and applications of the methods on real data.

The Department Colloquium will be preceded by the [Problem Solving Seminar](#), starting in the same Zoom room at 1:00 pm. The speaker on December 4 is Dr. Adam Glesser, who will present

## Niels Abel's Darkest Secrets