

DiSCourse Seminar

The Digital Science Center would like to invite you to the following presentation by our guest speaker:

Catie Welsh
Rhodes College

Detection of Mitochondrial DNA Variants from Next-generation Sequencing Data
Recent sequencing studies have extensively explored the tumor-specific variants present in the nuclear genomes of cancers. Although mitochondria are known to control energy metabolism and apoptosis, the origins and impact of cancer-associated mutations in mtDNA are unclear. In this talk, I will describe a pipeline for locating and categorizing mitochondrial mutations in whole-genome sequencing data. This pipeline was applied to a large cohort of pediatric cancer samples to determine the frequency of particular variants within the population and within subtypes of cancer. We looked at the data through a number of lenses to better understand the functional significance of these mtDNA mutations, including validating our results using RNA-sequencing and single-cell sequencing. Our study found that tumor-specific mtDNA variants are enriched for variants that are rare in the general population and those that are predicted to be deleterious.

About the speaker

[Catie Welsh](#) is an Associate Professor of Computer Science at Rhodes College in Memphis, USA. Her research areas are bioinformatics and computational genetics. As a Fulbright Fellow, she is currently completing a four-month research visit at the University of Innsbruck.

Date and Time: Friday, 16 April 2021, 12:00 (CEST)

Please note: This presentation will be given **online** in [Big Blue Button](#). Participants do not need to register.