

DBMR Research Conference

Seminar room EG050
Murtenstrasse 24, 3008 Bern

Date: Thursday, October 13, 2022, 17:00 – 18:00
Title: Evolution of minor introns and their role in development, disease, and therapeutics
Speaker: Ass. Prof. Rahul N Kanadia, Ph.D.
Department of Physiology and Neurobiology
University of Connecticut (USA)

Bio: Dr. Rahul Kanadia did his Ph.D. in Molecular Genetics at the University of Florida, Medical School, in the laboratory of Dr. Maurice Swanson. His graduate work focused on testing the hypothesis that Myotonic Dystrophy, which is caused by (CTG)_n repeats in the 3' untranslated region of DMPK is caused by toxic gain of function such that the mutant DMPK-(CUG)_n mRNA sequesters an alternative splicing factor called Muscleblind. In essence, the disease is caused by the indirect loss of function of Muscleblind. He generated a Muscleblind knockout mouse model that recapitulated the cardinal symptoms of myotonic dystrophy. This seminal discovery has led to new therapeutic strategies that are focused on rescuing Muscleblind function in patients. He then continued to his postdoctoral training in Dr. Constance Cepko's laboratory at the Howard Hughes Medical Institute, Department of Genetics, at the Harvard Medical school. There he explored the role of alternative splicing in retinal development. As Associate Professor in the Physiology and Neurobiology at the University of Connecticut, he explores the role of minor intron splicing in evolution, development, disease and therapeutics. His is one of the few labs that has explored the role of minor intron splicing with the production of mouse models, bioinformatics pipelines and various biochemical and molecular approaches to better understand minor intron splicing.

Abstract: The importance of the relatively under explored minor intron splicing has recently gained attention with its links to a growing number of developmental diseases. The moniker "minor introns" refers to their relatively low number (<0.5%) in the human genome. In fact, minor introns are often found inserted as a single intron in ~700 genes that mostly consist of the canonical or the major introns. Since major and minor introns are spliced by their respective major and minor spliceosomes, the expression of minor intron-containing genes (MIGs) is inherently more complex. Therefore, it is unclear why these genes acquired minor introns and that they are maintained across evolution. In fact, these minor introns are highly conserved in their splice site sequence elements and their position. This talk will focus on our efforts to understand the evolution of minor introns and their role in regulating proliferation of stem cells during brain development and the effects of perturbed embryonic development on postnatal life and behavior. The specific focus here will be on postnatal growth, feeding behavior such as hyperphagia that results in obesity.

Ass. Prof. Rahul N Kanadia has been invited by Prof. Mark A. Rubin, MD, Director DBMR and Dr. Anke Augspach, Cancer Therapy Resistance (CTR), Department for BioMedical Research, University of Bern.

Next DBMR Research Conference

Monday, 31 October 2022, 17:00 – 18:00
Prof. Dr. Ron. M. A. Heeren, The Maastricht MultiModal, Molecular Imaging Institute, Maastricht University, The Netherlands
Title: "Translational molecular imaging; Using a mass spectrometer as a microscope."

The DBMR Research Conference will be followed by an apéro.



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