The Impact of CRISPR on Imprinting Disorders

An educational event for researchers, practitioners, and students in health care, genetics and genomics

Saturday, June 10, 2017

Rome Commons Ballroom
626 Gilbert Road, Ext
University of Connecticut
Storrs, Connecticut

Sponsored by

The Institute for Systems Genomics
http://isg.uconn.edu/
&
The Health Care Genetics Professional Science Masters Degree Program
http://healthcaregenetics.uconn.edu/
A Note from Our Conference Organizer

Welcome! This is the 3rd continuing education conference I have hosted on the Storrs campus and am looking forward to an exciting day of science with exceptional educators, scientists and professionals. The purpose of this conference is to provide you with information from our experts about the mechanisms of imprinting, examples of human imprinting disorders, as well as the hows and whys of CRISPR modifications. Examples of CRISPR modified cell lines which mimic imprinting disorders and the Chromosome Core testing pipeline will convince you of the excitement and future clinical relevance of CRISPR technology; but in conclusion –what is our ethical standpoint about bio-editing the human genome??

A special thank you to our speakers for sharing a Saturday and to Stephanie Holden for her assistance!

Best,

[Signature]
Schedule

9:00-9:25  Registration and Continental Breakfast

9:30-10:00  Judy Brown, Ph.D. Welcome: How are we keeping the CRISP fresh excitement of learning for genetic and genomic students at UConn?

10:00-10:45  Joseph Tucker, M.D. Clinical aspects of Angelman syndrome and Prader-Willi syndrome

10:45-11:00  Break

11:00-11:30  Michael O’Neill, Ph.D. Locus-Specific Imprinting on the Mouse X chromosome

11:30-12:00  Christopher Stoddard CRISPRs: The adjustable wrench in the toolbox for geneticists

12:00-1:00  Lunch
Meet the scientists; students will have an opportunity to lunch and talk with speakers and other scientists in attendance

1:00-1:30  Stormy Chamberlain, Ph.D. Using CRISPR technology to dissect mechanisms of imprinting in Angelman syndrome

1:30-2:00  Marc Lalande, Ph.D. Knockout of zinc finger protein 274 re-activates imprinted maternal RNA expression at the Prader-Willi syndrome locus

2:00-2:15  Break

2:15-2:45  Rachel O’Neill, Ph.D. Chromosome and Genome Analyses: technology for the science and safety of stem cell research

2:45-3:15  Robert Bird, J.D. Ethical, Legal, and Social Implications of CRISPR Technology

3:15-3:30  Open Discussion

3:30  Closing remarks/workshop adjourns
**BIOGRAPHIES**


**Judy Brown**, an Associate Professor in Residence in the Dept of Allied Health Sciences is the director of the Diagnostic Genetic Sciences undergrad and post-baccalaureate training programs, and the Health Care Genetics PSM Degree Program at UConn. Originally trained in the clinical lab, Judy has dual certifications from ASCP in cytogenetics and molecular biology with a doctorate in genetics and genomics. Dr. Brown has provided training and expertise using cytogenomics tools to assess the impact of CNVs and repeats on chromosome structure and function in both model and non-model systems for diverse collaborative projects to expand grant-funded efforts in the growing field of study: chromosome biology. As Director of the Chromosome Core, Dr. Brown receives Stem Cell Research Grant Funding to examine the genome integrity of human induced pluripotent stem cells created to mimic diseases such as autism, breast cancer, Prader-Willi, Angelman syndrome and psychiatric disorders. It is Judy’s goal to lead a genomics health education initiative and she is continuously looking for opportunities to expand the quality and quantity of opportunities for UConn students and the services for CT workforce partners and patients.
**Stormy J. Chamberlain** is the Raymond and Beverly Sackler Assistant Professor of Genetics and Genome Sciences at the University of Connecticut Health Center. Stormy obtained a B.A. degree in Molecular Biology from Princeton University and a Ph.D. in Genetics from the University of Florida. She performed post-doctoral research at the University of North Carolina-Chapel Hill and the University of Connecticut Health Center prior to joining the faculty at the University of Connecticut Health Center. Her lab uses induced pluripotent stem cell models of Angelman syndrome and 15q duplication syndrome to understand genetic and epigenetic regulation at the imprinted chromosome 15q11-q13 locus.

**Marc Lalande** received a Ph.D. in Medical Biophysics from the University of Toronto in 1981. His postdoctoral training was with Dr. Samuel A. Latt in the Department of Pediatrics, Harvard Medical School and Children's Hospital, Boston, MA. At Harvard Medical School, he was later an Associate Professor of Pediatrics and an Assistant Investigator of the Howard Hughes Medical Institute. In 1998, he moved to the UCONN School of Medicine. He is Director of the Stem Cell Institute and of the newly created Institute for Systems Genomics, a group with representation from 10 different UConn Schools and Colleges, the Jackson Laboratory and Connecticut Children’s. His research laboratory focuses on using induced pluripotent stem cell (iPSC) technology to study Prader-Willi syndrome, a chromosome 15q11-q13 imprinting disorder.
Michael O’Neill is Assistant Director of the Institute for Systems Genomics and Associate Professor in Genetics and Genome Sciences, Department of Molecular and Cell Biology at the University of Connecticut. He is also an investigator in the Connecticut Institute for the Brain and Cognitive Sciences. Dr. O’Neill obtained his BA and PhD from the University of Texas at Austin with Dorothea Bennett and Karen Artzt studying mouse t-haplotypes. He received post-doctoral training at the University of Melbourne with Dr. Andrew Sinclair studying the genetics of sex determination and at Princeton University with Dr. Shirley Tilghman studying genomic imprinting. Dr. O’Neill’s work has focused on the evolution of genomic imprinting in vertebrates and on the role of X-linked imprinted genes in reproductive- and neuro- development.

Rachel O’Neill received her PhD in Genetics and Human Variation from La Trobe University. Currently a Professor in the Dept of Molecular and Cell Biology, Director of the Center for Genome Innovation within the Institute for Systems Genomics and Co-Director of the Chromosome Core, Dr. O’Neill has built her career on understanding how genomes maintain stability over time. Her research group uses molecular, cytogenetic, and computational approaches to study genomic conflict involved in retroelement transcription and centromere function and the role of novel small RNAs in chromosomal and genome stability. Her lab uses a comparative genomics approach encompassing both traditional and non-traditional model systems, including human, non-human primates, rodents, marsupials, and several marine species of relevance to environmental genomics. Over the past 20 years, her lab has established a suite of genome-scale techniques for their research, including ISH, cytogenetics, molecular genetics, genetic engineering, artificial chromosomes, bioinformatics, cell assays, and next-generation sequencing methods (10X genomics, Oxford Nanopore, Bionano Irys, Illumina, pyrosequencing, sequencing by ligation, Ion Torrent).
Christopher Stoddard was born and raised in Connecticut. He left to go to college at Saint Michaels College in Colchester, Vermont majoring in Biology and minoring in Chemistry. Upon completion Chris returned to Connecticut and began working at the UConn Health Center in the Gene Targeting and Transgenic Facility. There he worked on creating mouse models to be used in research through microinjection of zygotes and targeting mouse embryonic stem cells using custom made vectors. In the past few years, since the discovery of powerful gene editing tools that makes engineering possible in human cell lines, he shifted to gene editing in human cell lines. This includes many cancer cell lines, human embryonic stem cells, and patient derived induced pluripotent stem cells. Currently he operates the human genome editing core at UConn Health. In the core we develop cell line models used by researchers to aid in their research projects.

Joseph W. Tucker, an Assistant Professor of Genetics and Genome Sciences and Pediatrics, is a clinical medical geneticist at UConn Health and Connecticut Children’s Medical Center. He obtained a B.S.E. degree in Electrical Engineering from the University of Pennsylvania and M.D. from State University of New York – Downstate. He completed both residency in pediatrics and residency in clinical medical genetics at UConn Health and Connecticut Children’s Medical Center. His clinical practice includes care of individuals with rare disorders from infancy throughout adulthood.
SPECIAL THANKS TO OUR SPONSORS

Agilent Technologies

Biological Industries
Culture of Excellence

Dharmacon™
part of GE Healthcare

Rainbow Scientific, Inc.
Serving Laboratories throughout North America

Thermo Fisher Scientific
The world leader in serving science