

# 2022 Annual NHGRI Centers of Excellence in Genomic Sciences (CEGS) Meeting: AGENDA

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**October 18 – 20, 2022**

**Duke University**

**Durham, NC**

**Monday, October 17, 2022**

**PM**

**6:00**

**Welcome Meetup**

Join us as we kickoff this year's meeting and celebrate 20 years of CEGS.

**Bull McCabe's Irish Pub**

427 W Main St, Durham, NC 27701 | (919) 682-3061

**Tuesday, October 18, 2022**

**AM**

**7:00**

**Shuttle**

Shuttle service available from Washington Duke Inn and 21C Durham to Trent Semans Center.

***Group ground transportation from other hotels not provided. Please coordinate transportation to meeting independently.***

**7:00-8:00**

**In-person Check-in and Breakfast**

**TSC – Great Hall**

Mary Duke Biddle Trent Semans Center for Health Education (TSC)

Duke University

8 Searle Center Drive

Durham, NC 27710

**8:00-8:30**

**Welcome and Opening Remarks**

Dean Mary Klotman, MD

Dean, Duke University School of Medicine

Vice Chancellor for Health Affairs at Duke University

Adam Felsenfeld, PhD

Program Director, NHGRI

Tim Reddy, PhD

Duke CEGS / CCGR

**8:30-9:30**

**Center for Live Cell Genomics**

**Introduction (5 mins)**

### **Live Cell Genomics**

Sofie Salama, UC Santa Cruz (15 mins)

Holger Schmidt, UC Santa Cruz (15 mins)

Mircea Teodorescu, UC Santa Cruz (15 mins)

### **Q&A**

Moderated by Laavanya Sankaranarayanan, Duke UPGG PhD student

## **9:30-10:30 Center for Dynamic RNA Epitranscriptomes**

### **Sequencing RNA Modification at Base Resolution**

Chuan He, University of Chicago (30 mins)

### **Transcriptome-wide Profiling and Quantification of $N^6$ -Methyladenosine by Enzyme-assisted Adenosine Deamination**

Weixin Tang, University of Chicago (10 mins)

### **RBrowser: A Multimodal Data Exploration Platform for RNA and its Modifications**

Mengjie Chen, University of Chicago (10 mins)

### **Synthetic Biology Approaches to Harness the Epitranscriptome**

Bryan Dickinson, University of Chicago (10 mins)

## **10:30-10:45 Break**

### **Walk to Duke Gardens (10:30 – 11:30)**

A few groups will walk from TSC to Duke Gardens for a moment to stretch and brief view of what's blooming. *We recommend you join us in leaving at the beginning of the break.*

[TSC - Walking Map](#)

## **10:45-11:30 Trainee Career Panel**

This Q&A session will highlight various post training career pathways, explore the current state of academic careers and biotech collaborations as well as the current state of the genomic workforce and related challenges.

### **Panel**

Lisa Chadwick, PhD; Program Director, NHGRI

Shane Liddelow, PhD; Assistant Professor of Neuroscience and Ophthalmology, NYU Langone

Bill Majoros, PhD; Assistant Professor of Biostatistics & Bioinformatics, Duke

Sudarshan Pinglay, PhD; Postdoctoral Fellow, NYU Langone

### **Moderators**

Makenzie Beaman, Duke MSTP MD/PhD student

Apoorva Iyengar, Duke UPGG PhD student

Schuyler Melore, Duke UPGG PhD student

## **11:30-12:30 Center for Personal Dynamic Regulomes**

### **Overview**

Howard Chang, Stanford University (12 mins)

**Oncogenes Outside Chromosomes: Form and Function**

King Hung, Stanford University (12 mins)

**Scaling Single Cell Analysis to Millions of Cells**

Ben Parks, Stanford University (12 mins)

**Machine Learning Identifies Genetic Basis of Amyotrophic Lateral Sclerosis**

Sia Zhang, Stanford University (12 mins)

**Q&A**

Moderated by Laavanya Sankaranarayanan, Duke UPGG PhD student

**PM**

**12:30-1:30**

**Lunch**

Pick up boxed lunch in TSC lobby and enjoy outdoors and nearby tables.

Afternoon moderated by Tim Reddy, Duke CEGS / CCGR PI

**1:30-2:30**

**Center for Synthetic Regulatory Genomics**

**Introduction to SyRGe, The Center for Regulatory Genomics**

Jef Boeke, NYU Langone (5 mins)

**Architecture of Mammalian Gene Regulation**

Matt Maurano, NYU Langone (8 mins)

**Default Chromatin States in Yeast and Mammalian Cells**

Brendan Camellato, NYU Langone (5 mins)

**Genomically Rewritten and Tailored Genetically Engineered Mouse Models (GREAT-GEMMs)**

Weimin Zhang, NYU Langone (7 mins)

**Pooled Big DNA Deliveries and Phenotyping**

Raquel Ordoñez Ciriza, NYU Langone (4 mins)

**CRISPR Engineering of Episomes in Yeast (CREEPY)**

Yu Zhao, NYU Langone (4 mins)

**Engineering Brain Genes**

Shane Liddelow, NYU Langone (7 mins)

**Q&A**

Moderated by Laavanya Sankaranarayanan, Duke UPGG PhD student

**2:30-3:30**

**Center for Combinatorial Gene Regulation**

**Overview of Center for Combinatorial Gene Regulation (CCGR)**

Tim Reddy, Duke (5 mins)

**Multiplex Epigenome Editing with CRISPR/Cas12**

Schuyler Melore, Duke (5 mins)

**Epigenome Editing to Modulate Gene Expression in the Prader-Willi Syndrome Locus**

Dahlia Rohm, Duke (5 mins)

**Improving Analyses of Genome-wide CRISPR Screens**

Maria ter Weele, Duke (5 mins)

**Modeling cooperative transcription factor binding**

Kyle Pinheiro de Oliveira, Duke (5 mins)

**Identifying Rare Disease Populations with Undiscovered Genetic Etiology**

Makenzie Beaman, Duke (5 mins)

**Identifying and Reversing a New Cause of GSD IX**

Apoorva Iyengar, Duke (5 mins)

**A Portal for Results of High-Throughput Functional Genomic Studies**

Warren Kibbe, Duke (5 mins)

**Q&A**

Moderated by Laavanya Sankaranarayanan, Duke UPGG PhD student

**3:30-3:40 Break**

**3:40-4:10 Flash talks**

Each CEGS Site to select two individuals to present a flash talk (3 min each).

<b>Alexander Urban</b>	Center for Personal Dynamic Regulomes	Advanced Methods to Resolve the Sequence of Inaccessible Regions of the Human Genome
<b>Naomi Pacalin</b>	Center for Personal Dynamic Regulomes	Bidirectional Epigenetic Editing with Single-cell Profiling
<b>Matt Elliott</b>	Center for Live Cell Genomics	WetAI: An IoT Integrated Device Environment to Automate Scalable Cerebral Organoid Experiments
<b>Mohammed A. Mostajo-Radji</b>	Center for Live Cell Genomics	Cloud Controlled Microscopy Enables Project-Based Learning in Underserved Communities
<b>Tiffany Tsou</b>	Center for Synthetic Regulatory Genomics	Hox Genes and a Generic Landing Pad Line
<b>Antonio Vela-Gartner</b>	Center for Synthetic Regulatory Genomics	Deploying Machine Learning to Optimize CRISPR Cutting in Yeast
<b>Zhongyu Zou</b>	Center for Dynamic RNA Epitranscriptomes	FMRP Phosphorylation Modulates Neuronal Translation through YTHDF1
<b>Feng Zhang</b>	Center for Dynamic RNA Epitranscriptomes	Epitranscriptomic regulation of cortical neural stem cell maintenance via Mettl8-dependent mitochondrial tRNA m3C modification
<b>Keith Siklenka</b>	Center for Combinatorial Gene Regulation	Characterizing the role of gene regulatory elements in the differentiation and function of mouse CD4+ T cell subsets
<b>Sean McCutcheon</b>	Center for Combinatorial Gene Regulation	Epigenome Editing Screens to Map Transcriptional Regulators of Human CD8 T Cell State

**4:30 Shuttle to Carolina Theatre for Downtown Dinner Tour**

Shuttle service available from Trent Semans Center to Carolina Theatre for dinner event (pre-registration for dinner event is required). **Individuals will need to coordinate transportation following dinner.**

**4:30 Shuttle to Washington Duke Inn and 21C Durham**

Shuttle service available from Trent Semans Center to Washington Duke Inn and 21C Durham.

**Group ground transportation from other hotels not provided. Please coordinate transportation to independently.**

**4:45**      **Downtown Dinner Tour with Carolina Taste (registration required)**

This optional event will be a great opportunity to network with other meeting attendees and explore the Downtown Durham food scene. For more information, <http://bit.ly/CEGSDINNER>.

Meet at the **Carolina Theatre**  
309 W Morgan St, Durham, NC 27701

**Wednesday, October 19, 2022**

**AM**

**7:00**      **Shuttle**

Shuttle service available from Washington Duke Inn and 21C Durham to Trent Semans Center.

**Group ground transportation from other hotels not provided. Please coordinate transportation to meeting independently.**

**7:30-8:30**      **Breakfast**

**TSC – Great Hall**  
Mary Duke Biddle Trent Semans Center for Health Education (TSC)  
Duke University  
8 Searle Center Drive  
Durham, NC 27710

Morning moderated by Greg Wray, Duke CEGS / CCGR PI

**8:30-9:30**      **Center for Admixed Science and Technology**

**Welcome and Summary**

Lucila Ohno-Machado, UCSD (5 mins)

**Exploring the Contribution of Complex Variants to Polygenic Traits Across Diverse Population**

Melissa Gymrek, UCSD (15 mins)

**Enabling Collaborative Studies Across Data Silos Using Secure and Federated Algorithms**

Hoon Cho, Broad Institute of MIT and Harvard (20 mins)

**A Pilot Study of Standardizing Social Determinants of Health Data**

Hua Xu, University of Texas Health Science Center (20 mins)

**9:30-9:45**      **Break**

If you are presenting a poster during AM or PM sessions, please hang now on the 6<sup>th</sup> floor.

**9:45-10:45**      **Poster Session**

This AM poster session will include the ODD numbered posters. Join upstairs!

**TSC – 6th Floor**

Mary Duke Biddle Trent Semans Center for Health Education (TSC)

**11:00-12:00** **Center for Integrated Cellular Analysis**

**CICA Overview**

Rahul Satija, New York Genome Center (6 mins)

**CICA Session A: Epigenomics**

**Integrated Single-cell Genotyping and Chromatin Accessibility Charts JAK2-V617F Human Hematopoietic Differentiation**

Franco Izzo, Cornell University (9 mins)

**Nanobody-tethered Transposition Allows for Multifactorial Chromatin Profiling at Single-cell Resolution**

Ivan Raimondi, Cornell University (9 mins)

**CICA Session B: Perturbation**

**Efficient Combinatorial Targeting of RNA Transcripts in Single Cells with Cas13 RN Perturb-seq**

Harm Wessels, New York Genome Center (9 mins)

**Deciphering the Regulation of Cleavage of Polyadenylation at Single-cell Resolution with CPA-Perturb-seq**

Maddie Kowalski, New York Genome Center (9 mins)

**CICA Session C: Integration**

**Dictionary Learning for Integrative, Multimodal, and Scalable Single-cell Analysis**

Yuhan Hao, New York Genome Center (9 mins)

**Cross Species Comparisons Utilizing a Cell Type Atlas of the Spanish Ribbed Newt Brain**

Jamie Woych, Columbia University (9 mins)

**PM**

**12:00-1:00** **ExCITE Challenge Lunch**

Join us as we work in small groups to overcome some of genomics biggest challenges. Pickup your lunch in the lobby then return to Great Hall and find a table. Bonus if you do not already know everyone at the table!

Afternoon moderated by Greg Crawford, Duke CEGS / CCGR PI

**1:00-2:00** **Center for Genome Editing and Recording**

**The Center for Genome Editing and Recording**

Jonathan Weissman, Whitehead Institute/MIT (10 mins)

**Lineage Tracing Reveals the Phylodynamics, Plasticity, and Paths of Tumor Evolution**

Dian Yang, Whitehead Institute (10 mins)

**Engineered Virus-like Particles for Efficient *in vivo* Delivery of Therapeutic Proteins**

Samagya Banskota, Broad Institute (10 mins)

**Deciphering Cell States and Genealogies of Human Hematopoiesis with Single-cell Multi-omics**

*Remote Presentation*

Chen Weng, Whitehead Institute (10 mins)

**Mapping the DNA Repair Process that Enable Genome Editing**

Britt Adamson, Princeton University (10 mins)

**Q&A**

Moderated by Makenzie Beaman, Duke MSTP MD/PhD student

**2:00-2:10 Break**

**2:10-3:10 Poster Session**

This PM poster session will include the EVEN numbered posters. Join upstairs!

**TSC – 6th Floor**

Mary Duke Biddle Trent Semans Center for Health Education (TSC)

**3:20-4:20 Center for SubCellular Genomics**

**Towards Organelle Genomics**

Junhyong Kim, University of Pennsylvania (15 mins)

**Measuring the Small Molecules in Individual Organelles with Mass Spectrometry**

Jonathan Sweedler, University of Illinois (15 mins)

**Connecting Subcellular Morphology with Molecular States Using Computer Vision**

*Remote Presentation*

James Zou, Stanford University (15 mins)

**Q&A**

Moderated by Makenzie Beaman, Duke MSTP MD/PhD student

**4:20-4:50 Flash talks**

Each CEGS Site to select two individuals to present a flash talk (3 min each).

<b>Yasemin Atiyas</b> <i>Remote Presentation</i>	Center for SubCellular Genomics	Towards the Isolation and Molecular Analysis of Single Lysosomes from Single Cells
<b>David Froelicher</b> <i>Remote Presentation</i>	Center for Admixture Science and Technology	Secure and Federated Genome-Wide Association Studies
<b>Matteo D'Antonio</b>	Center for Admixture Science and Technology	HLA Types are Associated with Hundreds of Complex Traits and Diseases in an Ancestry-dependent Manner
<b>Aidan Daly</b>	Center for Integrated Cellular Analysis	Spatiotemporal Pathology of Colon Aging: An Integrative Spatial Transcriptomics and Single-nuclear Modeling Approach
<b>Ignacio Vazquez-Garcia</b>	Center for Integrated Cellular Analysis	Genomic Instability as a Determinant of Immune Escape in Ovarian Cancer
<b>Aaron Lin</b>	Center for Genome Editing and Recording	Building a Molecular Recorder for Viral Infection
<b>Vikram Pattanayak</b> <i>Remote Presentation</i>	Center for Genome Editing and Recording	Population-scale Off-target Assessment of Cas9 and Other Gene Editing Enzymes

**5:00 Shuttle to Washington Duke Inn and 21C Durham**

Shuttle service available from Trent Semans Center to Washington Duke Inn and 21C Durham.

***Group ground transportation to other hotels not provided. Please coordinate transportation to independently.***

***Group ground transportation to and from dinner not provided. Please coordinate transportation independently.***

**6:00** **Bull City Run Club meet up (optional)**  
Join the [Bull City Run Club](#) for a 3 mi, 4 mi, 6.4 mi route

**6:30** **Dinner (included for meeting registrants)**  
This dinner and meeting reception will include local and regional favorites along with options for vegetarian, vegan, and gluten-free diners. Starters offered at 6:30 pm with dinner planned for 7 pm. Additional registration not required.

**The Pit**  
321 W. Geer Street, Durham, NC 27701 | (919) 282-3748

## **Thursday, October 20, 2022**

**AM**  
**7:00** **Shuttle**  
Shuttle service available from Washington Duke Inn and 21C Durham to Trent Semans Center.

***Group ground transportation from other hotels not provided. Please coordinate transportation to meeting independently.***

**7:30-8:30** **Breakfast**  
**TSC – Great Hall**  
Mary Duke Biddle Trent Semans Center for Health Education (TSC)  
Duke University  
8 Searle Center Drive  
Durham, NC 27710

Morning moderated by Warren Kibbe, Duke CEGS / CCGR PI

**8:30-9:30** **Center for the Multiplexed Assessment of Phenotype**  
**Developing New Methods to Assess Genetic Variation at Scale**  
Douglas Fowler, University of Washington (10 mins)  
**Expanding Cellular Context for Mutation Scans of Clinically Relevant Genes**  
Lea Starita, University of Washington (10 mins)  
**Doubling Rare Variant Burden-based Gene-trait Associations via Computational Predictor Choice**  
Fritz Roth, University of Toronto (10 mins)  
**Deep Molecular, Cellular, and Temporal Phenotyping of Developmental Perturbations at Whole Organism Scale**  
Cole Trapnell, University of Washington (10 mins)  
**Multiplexed Variants – Multiple Contexts**  
Christine Queitsch, University of Washington (10 mins)



## Q&A

Moderated by Apoorva Iyengar, Duke UPGG PhD student

### 9:30-10:30 **A Phenomics First Resource for Interpretation of Variants**

#### **Interoperable Phenotype Definitions to Improve Disease Diagnosis and Discovery**

Melissa Haendel, University of Colorado Anschutz Medical Campus (30 mins)

#### **Phenopackets, Tools for Sharing Individual-level Phenotypic Information**

*Remote Presentation*

Peter Robinson, The Jackson Laboratory for Genomic Medicine (15 mins)

## Q&A

Moderated by Apoorva Iyengar, Duke UPGG PhD student

### 10:30-10:40 **Break**

Please remove your poster from 6<sup>th</sup> floor.

### 10:40-11:40 **Center for Genomic Information Encoded by RNA Nucleotide Modifications**

#### **Introduction to the Center**

Sammie Jaffrey, Cornell University (5 mins)

#### **Uncovering the Diversity and Function of rRNA and Protein Modifications in the Human Ribosome Using Single-molecule FRET and cryoEM**

*Remote Presentation*

Scott Blanchard (27.5 mins)

#### **Next-generation Mass Spectrometry-based de novo Methods for Direct Sequencing and Quantitative Mapping of RNA Modifications**

Shenglong Zhang, New York Institute of Technology (27.5 mins)

### 11:40 **Lunch and ExCITE Report Back**

Pickup your lunch in the lobby and return as we hear from group on ExCITE discussions.

Afternoon moderated by Charlie Gersbach, Duke CEGS / CCGR PI

## PM

### 12:40-1:40 **Center for Genome Imaging**

#### **Imaging the Genome at Super-resolution: Easy to Say, but....**

*Remote Presentation*

Ting Wu, Harvard University (7 mins)

#### **How to Transform Whole Genome Imaging Challenges into Opportunities**

*Remote Presentation*

Antonios Lioutas, Harvard University (7 mins)

#### **Quality Control and Reproducibility of Genomic Super-resolution Imaging**

*Remote Presentation*

Sara Aufmkolk, Harvard University (7 mins)

#### **Ultraconserved Elements: Solving a Mysterious Genome-wide**

## Phenomenon

*Remote Presentation*

Tae Ryu, Harvard University (7 mins)

## A Story About Spooning Homologs

*Remote Presentation*

Jumana Alhaj Abed, Harvard University (7 mins)

## Oligopaint Probe Design to Paint and Image Genomic Repetitive Regions *in situ*

*Remote Presentation*

Nuno Martins, Harvard University (7 mins)

## Q&A

Moderated by Apoorva Iyengar, Duke UPGG PhD student

1:40-2:05

## Flash talks

Each CEGS Site to select two individuals to present a flash talk (3 min each).

<b>Nicholas Popp</b>	Center for Multiplexed Assessment of Phenotype	Comprehensive Characterization of Missense Variation in Coagulation Factor IX Reveals Features Necessary for Secretion and Gamma-Carboxylation
<b>Matthew Berg</b>	Center for Multiplexed Assessment of Phenotype	Proteome-wide Alanine Scanning Using Mistranslating tRNA Variants
<b>Monica Munoz-Torres</b>	A Phenomics First Resource for Interpretation of Variants	Ontologies for Phenotyping and Disease
<b>Katerina Cortes</b>	A Phenomics First Resource for Interpretation of Variants	Revealing Potential Mechanisms for Ehlers Danlos Syndrome using Knowledge Graphs
<b>Fei Zhao</b> <i>Remote Presentation</i>	Center for Genome Imaging	Genome-wide Super-resolution Fluorescence Imaging of Subsubtelomeric Chromosome Regions
<b>Dimos Gkoutaroulis</b> <i>Remote Presentation</i>	Center for Genome Imaging	Leveraging Video Compression Codecs to Effectively Reduce Microscopy Data Size

2:05

## Closing Remarks

**Tim Reddy, PhD**

Duke CEGS / CCGR PI