2024 Annual NHGRI Centers of Excellence in Genomic Sciences (CEGS) Meeting AGENDA

October 28-30, 2024

Scripps' Seaside Forum
La Jolla, California

All times PT

Monday, October 28

8:00-8:45 am	**In-person Check-in** Scripps' Seaside Forum 8610 Kennel Way, La Jolla, California 92037 Coffee
8:45 - 9:00 am	**Welcome and Opening Remarks**
9:00-10:00 am	**Center for Genome Imaging – Harvard Medical School** (Virtual Presentations) • Introduction – Ting Wu • Towards a 3D structure of chromosomal ends: Developing the methods – Fei Zhao • Towards 1-kb in 3D: Integrative modeling using human genome sequences, Hi-C, and super-resolution imaging – Marc Marti-Renom • Spacewalk: A 3D visualization web application for super resolution imaging – Douglass Turner • Bringing the 3D genome to K-12: A partnership with the Pinhead Institute – Nicola Neretti
10:00-11:00 am	**Center for Genome Editing and Recording – Whitehead Institute for Biomedical Research** • Introduction – David Liu (Virtual Presentation) • A platform for multimodal in vivo pooled genetic screens reveals regulators of liver function – Reuben Sanders • TBD – Smriti Pandey • CGER Outreach – Maxine Wang
11:00-11:20 am	Coffee Break

11:20-11:45 pm **Flash Talks Session 1**

Each CEGS Center will nominate two speakers from their center to present flashtalks. Flashtalks will be two minutes each with no transition time.

Center for Genome Imaging – Harvard Medical School (Virtual Presentations)

1. Imaging chromosome territories in 3D using Oligopaints – Mikko Sokka
2. Lord of the Rings – Guy Nir

Center for Genome Editing and Recording – Whitehead Institute for Biomedical Research

1. Bacterial single-cell RNA-sequencing identifies host immune responses to phage infection – Bruce Wang
2. High-resolution footprinting of transcription factors involved in 3D chromatin interactions – Corriene Sept

Center for Genomic Information Encoded by RNA Nucleotide Modification – Weill Cornell Med.

1. Molecular heterogeneity of ribosomal RNA modifications and their biological relevance – Adrianna Dabrowska
2. New tools for studying mRNA methylation – Kate Meyer

Center for Dynamic RNA Epitranscriptomes – University of Chicago

1. Building better pipelines for nanopore sequencing of the epitranscriptome – Tao Pan
2. Engineering CIRTS to rebalance protein expression from mRNA in haploinsufficiencies – Riley Sinnot

Center for Synthetic Regulatory Genomics – New York University School of Medicine

1. Refactoring the Mouse IFN-I Locus for Functional Studies – Skyler Uhl

Genetic & Social Determinants of Health: Center for Admixture Science and Technology – Yale University

1. Admixture Mapping in the All of Us Research Program – Wilfredo Gonzalez-Rivera

11:45-12:30 pm **Trainee Career Session**

Students will gather at round tables with faculty members for 4, 10-minute rotations. Each table and faculty member will have a default conversation topic such as early career, grant writing, finding post doc positions, etc.

12:30-1:30 pm Lunch

1:30-2:30 pm **Center for Genomic Information Encoded by RNA Nucleotide Modification – Weill Cornell Med.**

- Introduction to the center – Samie Jaffrey
- Control of multilineage hematopoiesis by 5' cap-proximal modification – Andrew Levine
- 5’ end mononucleotide expansion diversifies mRNA in eukaryotic transcriptome – Jianheng Liu

- Mass Spectrometry-Based De Novo Direct Sequencing of tRNAs – Shenglong Zhang
- Parallelized smFRET reveals differential drug sensitivity of ribosomes bearing endogenously encoded rRNA sequence variation – Ryan Brady
- Outreach Presentation – Kate Meyer

2:30-3:30 pm **Poster Session 1**

3:30-3:50 pm Coffee Break

3:50-4:50 pm **Center for Dynamic RNA Epitranscriptomes – University of Chicago**

- RNA m5C oxidation in leukemogenesis – Chuan He
- CAM-seq to quantitatively map RNA m6A at base resolution – Michelle Zhao
- Novel chemical approach for Pseudouridine sequencing across RNA types and multiple species – Chang Ye
- Non-canonical mode of action and function of snoRNA targeting – Bei Liu
- Website & Outreach – Mengjie Chen & Tao Pan

5:00-5:30 pm **Bus to Stone Brewing**

5:30-8:30 pm **Dinner – Stone Brewing**
2816 Historic Decatur Rd, Unit 116
San Diego, CA

5:00-5:30 pm **Bus to La Jolla Shores Hotel**

Tuesday, October 29

7:30-8:00 am Coffee

8:00-9:00 am **Center for Synthetic Regulatory Genomics – New York University School of Medicine**

- Introduction – Matthew Maurano
- Lighting up the genome in living cells – Timothee Lionnet
- A Novel Mouse Model of X-Linked Dystonia Parkinsonism Implicates Oligodendrocytes as Drivers of Disease Pathology – Priya Prakash
- Dissecting a complex genetic association at the *CACNA1C* pan-psychiatric locus – Raquel Moya
- Tissue-specific gene expression in humanized mouse models – Tiffany Tsou
- Assessing Type 2 Diabetes Risk using Genome Engineering Techniques in Human iPSCs – Noor Chalhoub
- Exploring Genomic and Phenotypic Evolution Through Synthetic Biology Approaches – Francisca Real (Virtual Presentation)

9:00-10:00 am **Genetic & Social Determinants of Health: center for Admixture Science and Technology – Yale University**

- Introduction to CAST, Center for Admixture Science and Technology – Melissa Gymrek
- PIPSORT: Multi-ancestry fine-mapping – Tara Mirmira
- Secure discovery of genetic relatives across distributed datasets – Matthew Hong
- Extracting Social Determinants of Health Information from Clinical Documents using Large Language Models – Hua Xu
- Outreach Overview – Wilfredo Gonzalez-Rivera

10:00-10:15 am Coffee Break

10:15 – 11:15 am **Duke Center for Combinatorial Gene Regulation – Duke University**

- TBD – Tim Reddy
- TBD – Schuyler Melore
- TBD – Grayson Rice
- TBD – Christian McRoberts Amador
- TBD – Apoorva Iyengar
- Outreach – Shannon Clark

11:15–11:30 am **Flash Talk Session 2**

Duke Center for Combinatorial Gene Regulation – Duke University

1. TBD – Tania Guerrero-Altamirano
2. TBD – Micah Dailey

Center for Integrated Cellular Analysis – New York Genome Center

1. Single-cell joint analysis of DNA damage and transcriptome reveals selective genome vulnerability – Dongsheng Bai
2. A Panoramic View of Cell Population Dynamics in Mammalian Aging – Zehao Zhang

A Phenomics-First Resource for Interpretation of Variants – University of North Carolina, Chapel Hill

1. The Ontology of Biological Attributes and the GWAS Catalog use cases – Ibrahim Arwa
2. Combining Computational Phenotyping and Skin Organoid Disease Modeling of Hypermobile Ehlers-Danlos Syndrome to Reveal Underlying Mechanisms – Megan Kraus

Center for Live Cell Genomics – UC Santa Cruz

1. Title TBD – Sofie Salama
2. Title TBD - Kateryna Voitiuk

Center for Multiplexed Assessment of Phenotype - University of Washington

1. Principles of single-nucleotide plasmid chromatinization within mammalian cells – Ben Mallory
2. Uncovering the functional impact of missense mutations proteome-wide using mistranslation and mass spectrometry – Matt Berg

11:35-12:15 pm **Blue Sky Session 1**

5 minute - moderator introduce session
Breakout groups to discuss each bold prediction (4 predictions/2-3 groups per prediction), put major talking point into google slide deck (40 minutes)

12:15-1:15 pm Lunch

1:15-2:15 pm **Center for Integrated Cellular Analysis – New York Genome Center**

- Center for Integrated Cellular Analysis – Rahul Satija
- Optics-free Spatial Genomics for Mapping Mouse Brain Aging – Abdulraouf Adbulraouf
- Mapping transcriptional responses to cellular perturbation dictionaries with RNA fingerprinting – Isabella Grabski
- Tissue and cellular spatiotemporal dynamics in colon aging – Aidan Daily
- Single-cell genotype-phenotype mapping identifies therapeutic vulnerabilities in VEXAS syndrome – Saravanan Ganesan

2:15-3:15 pm **Poster Session 2**

3:15-3:30 pm Coffee Break

3:30-4:30 pm **A Phenomics-First Resource for Interpretation of Variants – University of North Carolina, Chapel Hill**

- Phenomics First: an integrated resource for interpretation of variants – Chris Mungall
- PhEval and the Phenopacket-Store: Benchmarking Variant and Gene Prioritisation Algorithms for Rare Disease Diagnostics – Yasmin Bridges
- Why we need all the organisms: evaluating contributions of different organisms to mechanistic disease understanding – Katherina Cortes
- Bridging Knowledge Across Species: Unifying Trait and Phenotype Ontologies – Ray Stefancsik
- Sustaining Mondo Disease Ontology: Harnessing Community Power for Long-Term Success – Sabrina Toro
- Engaging and serving the community – Chris Mungall

Wednesday, October 30

7:30-8:00 am Coffee

8:00-9:00 am **Center for Live Cell Genomics – UC Santa Cruz**

- Introduction – David Haussler
- TBD – Viktor Yurevych
- TBD – Ravipa Losakul
- TBD – Abir (S M Saiduzzaman)
- Education Outreach – Samira Vera-Choqqueccota

9:00-10:00 am **Center for Multiplexed Assessment of Phenotype - University of Washington**

- Introduction **–** Douglas Fowler
- Optical Pooled Phenotyping at Nucleotide Resolution – Sriram Pendyla
- Deep learning-based prediction of E3 ubiquitin ligase substrates – Chase Suiter
- Landscapes of human alpha-1 antitrypsin missense-variant effects reveal pathogenic variation and genetic interaction – Warren van Loggerenberg
- Building a Variant Effects Atlas: Technology Sharing and Global Community Engagement – Lara Muffley

10:00-10:15 am Coffee Break

10:15-11:00 am **Blue Sky Session 2**
Presentations from discussions in Session 1

11:00-11:10 am **Closing Remarks**