Genomics and Human Genetics: The New York Symposium

January 26, 2024
8:30 AM - 5:00 PM
Schwartz Hall E
NYU Langone Health

SPEAKERS

Wendy Bickmore, PhD
MRC Human Genetics Unit,
University of Edinburgh

Jef Boeke, PhD
Institute for Systems Genetics,
NYU Langone Health

Karen Miga, PhD
UC Santa Cruz Genomics Institute,
University of California Santa Cruz

Arnold Munnich, MD, PhD
Imagine Institute for Genetic Disease & Hospital Necker, Paris

Teresa Davoli, PhD
Institute for Systems Genetics,
NYU Langone Health

Pilar Ossorio, PhD
Center for Law and Bioethics,
University of Wisconsin

Gilad Evrony, MD, PhD
Center for Human Genetics & Genomics, NYU Langone Health

Bing Ren, PhD
Center for Epigenomics,
University of California San Diego

Julie Makani, MD, PhD
Sickle Cell Program, Dar-es-Salaam & Imperial College, London

Sue Slaugenhaupt, PhD
Center for Genomic Medicine,
Mass General Research Institute

Organizer:
Aravinda Chakravarti, PhD
Center for Human Genetics & Genomics,
NYU Grossman School of Medicine

Organizer:
Eric Green, MD, PhD
National Human Genome Research Institute, NIH

Sponsored by:
<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Speaker(s)</th>
</tr>
</thead>
<tbody>
<tr>
<td>8:30am - 8:45am</td>
<td>Opening Remarks</td>
<td>Aravinda Chakravarti, PhD</td>
</tr>
<tr>
<td>8:45am - 9:15am</td>
<td>&quot;Expanding studies of global genomic diversity with complete, telomere-to-telomere (T2T) assemblies&quot;</td>
<td>Karen Miga, PhD</td>
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<tr>
<td>9:15am - 9:45am</td>
<td>&quot;Unlocking the Genome’s Regulatory Code&quot;</td>
<td>Bing Ren, PhD</td>
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<tr>
<td>9:45am - 10:15am</td>
<td>“Throwing light on gene regulation from the non-coding genome”</td>
<td>Wendy Bickmore, PhD</td>
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<tr>
<td>10:15am - 10:45am</td>
<td>Coffee Break</td>
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<tr>
<td>10:45am - 11:15am</td>
<td>&quot;High-fidelity DNA sequencing of the single-strand origins of mutations&quot;</td>
<td>Gilad Evrony, PhD, MD</td>
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<tr>
<td>11:15am - 11:45am</td>
<td>“The Dark Matter Project”</td>
<td>Jef Boeke, PhD</td>
</tr>
<tr>
<td>11:45am - 12:15pm</td>
<td>&quot;Friedreich ataxia: for disease mechanisms to clinical trials in patients&quot;</td>
<td>Arnold Munnich, PhD, MD</td>
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<tr>
<td>12:15pm - 2:15pm</td>
<td>Lunch</td>
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<td>2:15pm - 2:45pm</td>
<td>Poster session with coffee</td>
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<td>2:45pm - 3:15pm</td>
<td>&quot;The long road from gene to drug for an ultra-rare disease.&quot;</td>
<td>Sue Slaugenhaupt, PhD</td>
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<tr>
<td>3:15pm - 3:45pm</td>
<td>&quot;Engineering specific aneuploidies to study their role in cancer and congenital disorders.&quot;</td>
<td>Teresa Davoli, PhD</td>
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<td>3:45pm - 4:15pm</td>
<td>&quot;Gene Therapy for Sickle Cell Disease: Advances in Science and Health in Africa.&quot;</td>
<td>Julie Makani, PhD, MD</td>
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<tr>
<td>4:15pm - 4:45pm</td>
<td>Conclusions</td>
<td>Eric Green, PhD, MD</td>
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<tr>
<td>5:00pm - 6:00pm</td>
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About the Presenters

Wendy Bickmore, PhD
"Our lab investigates how individual genes are organized and packaged in the nucleus and how they move in the cell cycle and during development. Current research focuses on how the spatial organization of the nucleus influences genome function in development and disease."

Jef Boeke, PhD
"The Boeke lab has long been known for foundational work on mechanistic and genomic aspects of retrotransposition in both yeast and mammalian systems. Our lab is heavily involved in the development of novel technologies in genetics, genomics and synthetic biology."

Aravinda Chakravarti, PhD
"Our laboratory focuses on the development and application of genetic, genomic, and computational technologies for discovery of genes and mechanisms in a variety of complex (non-Mendelian) human diseases: Hirschsprung disease and autism, two neurological developmental disorders, Hypertension and sudden cardiac death, and age at onset cardiovascular diseases."

Teresa Davoli, PhD
"The Davoli lab utilizes a variety of experimental and computational approaches, from large-scale genetic screens in human cells to prediction of survival in cancer patients."

Gilad Evrony, MD, PhD
"Our lab’s mission is to develop foundational new technologies to advance neuroscience and human genetics and to help solve diagnostic mysteries in neurology and psychiatry. We also aim to accelerate the implementation of genomics in clinical medicine, especially for children with rare undiagnosed diseases."

Eric Green, MD, PhD
"His work included significant, start-to-finish involvement in the Human Genome Project. His laboratory also pursued human genetics studies and identified and characterized several human disease genes, including those implicated in certain forms of hereditary deafness, vascular disease and inherited peripheral neuropathy."

Julie Makani, MD, PhD
"Her work aims to use sickle cell disease as a model to establish scientific and healthcare solutions in Africa, that are locally relevant and globally significant. The approach integrates public health interventions with genomics including gene therapy to improve health and cure sickle cell disease."

Karen Miga, PhD
"The Miga lab combines innovative computational and experimental approaches to produce the high-resolution sequence maps of human centromeric and pericentromeric DNAs. We aim to uncover a new source of genetic and epigenetic variation in the human population, which is useful to investigate novel associations between genotype and phenotype of inherited traits and disease."

Arnold Munnich, MD, PhD
"Arnold Munnich devoted his research to the identification of genes responsible for the neurological, metabolic and malformative handicaps of the child."

Pilar Ossorio, PhD
"Dr. Ossorio’s research interests revolve around research ethics and the protection of research participants, including: governance of large bioscience projects; data sharing in scientific research; the use of race in biomedical and social science research; ethical and regulatory issues in human subjects’ research; and the regulation and ethics of online research."

Bing Ren, PhD
"Our laboratory is interested in understanding how the complex gene regulatory networks in mammalian cells control cellular proliferation and differentiation. Our research areas are: 1) the development of genomics and bioinformatics tools that allow genome wide identification of regulatory targets for transcription factors and 2) the application of these tools to the study of transcription factors that play critical roles in cancer."

Sue Slaugenhaupt, PhD
"My research focuses on two neurological disorders, familial dysautonomia (FD) and mucolipidosis type IV (MLIV), as well as the common cardiac disorder mitral valve prolapse (MVP). Our work is focused on gene discovery and therapeutic development, specifically targeting RNA splicing. My work in the Mass General Research Institute is focused on supporting investigators and promoting research at the hospital."