# ALS CONSORTIUM MEETING

New York Genome Center (NYGC)  
June 24-26, 2018  
Sponsored by The ALS Association Greater New York Chapter

**Sunday, June 24, 2018**  
**Keynote Address and Reception**

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| 5:00pm-5:30pm | Optional: NYGC Facility Tour  
(1st Floor Lobby)                                                          |
| 5:30pm - 6:45pm | Welcome, Opening Remarks and Keynote Address  
(1st Floor Auditorium)  
Lucie Bruijn, PhD, MBA, Chief Scientist, ALS Association – Research  
Tom Maniatis, PhD, Scientific Director & CEO, New York Genome Center  
Speaker: Cori Bargmann, PhD, Head of Science at the Chan Zuckerberg Initiative, Head of the Lulu and Anthony Wang Laboratory of Neural Circuits and Behavior and the Torsten N. Wiesel Professor at the Rockefeller University |
| 6:45pm-8:00pm | Reception  
(1st Floor Cafeteria) |

**Monday, June 25, 2018**

All sessions will be held in the 1st floor Auditorium unless noted otherwise.

<table>
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<th>Time</th>
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<td>8:15am-9:00am</td>
<td>Breakfast</td>
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| 9:00am-9:10am | Welcome and Opening Remarks  
Hemali Phatnani, PhD, Director, Center for Genomics of Neurodegenerative Disease at NYGC  
Lucie Bruijn, PhD, MBA, Chief Scientist, ALS Association - Research |
| 9:10am-10:30am | NYGC ALS Consortium Updates  
**Moderator: Samantha Fennessey, MS**, Associate Scientific Project Manager at NYGC  
- Outline of Agenda and Overview of Progress  
  *Speaker: Hemali Phatnani, PhD*, Director, Center for Genomics of Neurodegenerative Disease at NYGC  
- Workflow Updates and Transition to NovaSeq  
  **Speakers: Michael Zody, PhD**, Senior Director of Computational Biology at NYGC (WGS)  
  **Nicolas Robine, PhD**, Assistant Director of Computational Biology at NYGC (RNA-seq) |

Last Updated: June 26, 2018
• MetroNome
  **Speaker:** Christian Stolte, Data Visualization Designer at NYGC

• Discussion

10:30am-10:45am  Coffee Break
  *(1st Floor Cafeteria)*

10:45am-12:15pm  NYGC ALS Consortium Updates – continued
  **Moderator:** Michael Zody, PhD, Senior Director of Computational Biology at NYGC
  • Genetic Modifiers
    **Speaker:** Terry Heiman-Patterson, MD, Professor of Neurology at Temple University
  • Familial ALS gene discovery
    **Speaker:** John Landers, PhD, Professor of Neurology at the University of Massachusetts Medical School
  • Repeat expansion and structural variants
    **Speakers:** Robert Sebra, PhD, Associate Professor of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai
    Giuseppe Narzisi, PhD, Lead Bioinformatics Scientist at NYGC
  • Discussion
    o What should we incorporate into our production pipeline?
    o What do we need in order to fill in the gaps?
    o How should we move forward?

12:15pm-1:15pm  Lunch
  *(1st Floor Cafeteria)*

1:15pm-2:15pm  Updates from Other Consortia and Partnerships
  **Moderator:** Lucie Bruijn, PhD, MBA, Chief Scientist, ALS Association - Research
  • Project MinE updates and repeat expansions
    **Speaker:** Jan Veldink, PhD, Professor of Neurology and Neurosurgery at the University Medical Center Utrecht
  • GTAC
    **Speaker:** Matthew Harms, MD, Associate Professor of Neurology at Columbia University College of Physicians and Surgeons
  • CReATe
    **Speaker:** Gang Wu, PhD, Group Lead of Genomics/Genetics Analysis, Department of Computational Biology at St. Jude Children’s Research Hospital

2:15pm-3:30pm  Updates from Other Consortia and Partnerships - continued
  • Answer ALS
    **Speakers:** Jeffrey Rothstein, MD, PhD, Professor of Neurology and Neurosciences at Johns Hopkins University School of Medicine
    Leandro Lima, PhD, Bioinformatics Software Engineer II at Gladstone Institute of Neurological Disease
  • DIALS
    **Speaker:** Katharine Nicholson, MD, Department of Neurology at Harvard Medical School
• Discussion
  o Areas of synergy
  o How can we streamline data flow?

3:30pm-4:15pm Coffee Break (1st Floor Cafeteria)

4:15pm-6:00pm Facilitating Open and Collaborative Access to Data and Resources
Moderator: Lyle Ostrow, MD, PhD, Assistant Professor of Neurology at Johns Hopkins University School of Medicine
  • Presentations from Target ALS Data Users
  • Discussion

6:00pm-8:00pm Dinner

Tuesday, June 26, 2018

All sessions will be held in the 1st floor Auditorium unless noted otherwise.

8:15am-9:00am Breakfast

9:00am-10:15am NYGC ALS Consortium Updates
Moderator: Delphine Fagegaltier, PhD, Staff Scientist, Center for Genomics of Neurodegenerative Disease at NYGC
  • Analysis of post-mortem samples
    o pH/RIN
      Speaker: Lyle Ostrow, MD, PhD, Assistant Professor of Neurology and Pathology at Johns Hopkins University School of Medicine
    o Splicing
      Speakers: Delphine Fagegaltier, PhD, Staff Scientist, Center for Genomics of Neurodegenerative Disease at NYGC
      Phaedra Agius, PhD, Senior Bioinformatics Scientist at NYGC
    o ALS Biomarkers in cell-free DNA
      Speaker: Barbara Celona, PhD, Molecular and Cell Biology Specialist at UCSF

10:15am-10:30am Coffee Break
(1st Floor Cafeteria)

10:30am-12:00pm NYGC ALS Consortium Updates – continued
  • Retroelements
    Speakers: Molly Hammell, PhD, Associate Professor of Genomics at Cold Spring Harbor Laboratory
    Avindra Nath, MD, Clinical Director of the National Institute of Neurological Disorders and Stroke (NINDS) at the NIH
  • Spatial Transcriptomics
    Speaker: Silas Maniatis, PhD, Staff Scientist, Center for Genomics of Neurodegenerative Disease at NYGC
  • Spatial Transcriptomics in Human Post-Mortem Tissue
    Speaker: Jenna Gregory, PhD, ECAT SCREDS Clinical Lecturer and Honorary StR in Histopathology, The University of Edinburgh

12:00pm-1:00pm Lunch

Last Updated: June 26, 2018
1:00pm-1:30pm  Measuring Progression in ALS  
**Speaker:** Ammar Al-Chalabi, MD, PhD, Professor of Neurology and Complex Disease Genetics, Kings College London; Director, King's MND Care and Research Centre

1:30pm-3:00pm  Presentations from industry: Analytics  
**Moderator:** Lucie Bruijn, PhD, MBA, Chief Scientist, ALS Association - Research  
- GNS Healthcare  
  **Speaker:** Diane Wuest, PhD, Director of Precision Medicine Initiatives at GNS Healthcare, Inc.  
- Origent Data Sciences  
  **Speaker:** Dave Ennist, PhD, MBA, Chief Science Officer at Origent Data Sciences  
- BenevolentAI  
  **Speaker:** Mark Rackham, PhD, Drug Discovery Scientist at BenevolentAI  
- Sage Bionetworks  
  **Speaker:** Ben Logsdon, PhD, Director of Neurodegenerative Research at Sage Bionetworks  
- Discussion

3:00pm  Closing Remarks  
**Lucie Bruijn, PhD, MBA, Chief Scientist, ALS Association – Research**

4:00pm-6:00pm  Greater New York Area ALSA Clinics Discussion (for ALSA Clinics only)  
**Moderator:** Kristen Cocoman, President & CEO, The ALS Association Greater New York Chapter  
- How to encourage participation in the ALS Consortium  
- Multicenter IRB  
- Clinical Research Coordinator/Research Nurse Support

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Guest Wireless

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Password: welcome101