David A. Knowles, PhD, Joins the New York Genome Center Core Faculty

He will present his application of machine learning and use of novel genomic technologies to determine the mechanisms underpinning neurological disease at a NYGC Five Points Lecture on February 20, 2019

New York, NY (January 24, 2019) – The New York Genome Center (NYGC) announces the appointment of Dr. David Knowles, PhD, to the NYGC Core Faculty, and jointly to Columbia University as a tenure-track assistant professor. Dr. Knowles' primary appointment at Columbia is in the Department of Computer Science with a secondary appointment in Columbia's Department of Systems Biology. "We are very excited to welcome Dr. Knowles to the core faculty at the Center," said Tom Maniatis, PhD, Scientific Director and Chief Executive Officer of the NYGC. "David has excelled at every stage of his academic career, and most recently has made important contributions to the application of machine learning to the genomics of RNA splicing. His laboratory will significantly advance our mission of developing and applying new computational methods and genomic tools to both basic and translational genomic research."

Dr. Knowles joins the NYGC from Stanford University, where he was a postdoctoral researcher in genetics and pathology, under the mentorship of Drs. Sylvia Plevritis (Center for Computational Systems Biology/Radiology), Jonathan Pritchard (Genetics), and Daphne Koller (Computer Science). During this postdoctoral training, Dr. Knowles applied his statistical machine learning expertise to understanding the interacting effects of genetic and environmental factors on gene expression and RNA splicing. He developed the novel method "LeafCutter," which identifies, quantifies, and tests variable RNA splicing events, and the computational tool EAGLE, which leverages allele-specific expression as a test of the influence of environment factors on distinct genetic backgrounds. "I look forward to working with the NYGC team as well as its affiliate members and founding institutions to strengthen the Center's genomic research and foster collaborative scientific discoveries, particularly in neurological diseases," Dr. Knowles said.

Dr. Knowles earned a PhD in the Machine Learning Group of the Cambridge University Engineering Department under the mentorship of Dr. Zoubin Ghahramani. While at Cambridge, Dr. Knowles developed Bayesian nonparametric approaches to factor analysis, network modeling, and hierarchical clustering, and also extended variational inference methods, computationally-attractive alternatives to Markov chain Monte Carlo to a broader class of probabilistic models.

Dr. Knowles' research accomplishments have been recognized by numerous awards, including the outstanding Poster Award at the 2017 Stanford Cancer Systems Biology Symposium, the International Society for Bayesian Analysis's New Researcher Travel Award for best invited Bayesian paper, and the Society's Dennis V. Lindley Prize for innovative research in Bayesian statistics. He was also a Roger Needham Scholar at Cambridge's Wolfson College, funded by Microsoft Research.

At the NYGC, Dr. Knowles and his lab members will focus on the development of novel machine learning methods and their application to data analysis challenges in genomics. A key aim is to understand the role of transcriptomic dysregulation across the spectrum from rare to common genetic diseases, which involves characterization of the genetic and environmental factors contributing to messenger RNA expression and splicing variation. The Knowles Lab will collaborate with research groups at the NYGC, and with the wider New York academic community, with an emphasis on collecting and analyzing large-scale genomics datasets, particularly in the context of neurological diseases. Dr. Knowles' aim is to leverage cutting-edge machine learning to make best use of novel genomic technologies, including single cell methods, forward genetic screens, and long-read transcriptomics, to better understand the mechanisms underlying genetic diseases.

Dr. Knowles will deliver a Five Points Lecture to discuss his research on Wednesday, February 20, at the NYGC. The lecture, titled *"Probabilistic Models of Transcriptome Dysregulation in Genetic Disease,"* will be moderated by Dr. Maniatis. For more information and to register for the event, please visit Five Points: David A. Knowles, PhD.

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About the New York Genome Center

The New York Genome Center (NYGC) is an independent, nonprofit academic research institution focused on furthering genomic research that leads to scientific advances and new insights and therapies for patients with neurodegenerative disease, neuropsychiatric disease, and cancer, leveraging our strengths in whole genome sequencing, genomic analysis, and development of new genomic tools.

The NYGC serves as a nexus for collaboration in genomic research for the New York community and beyond, building on the combined strengths of our faculty, member institutions, scientific working groups, affiliate members, and industry partners. Essential to this commitment is an outstanding faculty who are leading independent research labs based at the NYGC and one of our member institutions. They also support the NYGC scientific mission and engage in research programs that support the NYGC and the wider scientific community.

Member institutions include: Albert Einstein College of Medicine, American Museum of Natural History, Cold Spring Harbor Laboratory, Columbia University, Hospital for Special Surgery, Icahn School of Medicine at Mount Sinai, The Jackson Laboratory, Memorial Sloan Kettering Cancer Center, New York-Presbyterian Hospital, The New York Stem Cell Foundation, New York University, Northwell Health, Princeton University, The Rockefeller University, Roswell Park Cancer Institute, Stony Brook University, and Weill Cornell Medicine. For more information on the NYGC, please visit: http://www.nygenome.org.

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