



**Dr. Neville Sanjana Receives Young Investigator Award from  
the Brain & Behavior Research Foundation for Autism Functional Genomics**

*Combining CRISPR and Human Neurons to Dissect Autism Genetics*

NEW YORK, NY (August 29, 2018) -- Neville Sanjana, PhD, Core Faculty Member at the New York Genome Center, Assistant Professor of Biology, New York University, and Assistant Professor of Neuroscience and Physiology at NYU School of Medicine, is the recipient of the Brain & Behavior Research Foundation's Young Investigator Award. This grant will support the Sanjana Lab's research on single-cell functional genomics of mutations implicated in severe forms of autism spectrum disorder (ASD) and intellectual disability (ID).

In 2016, the Centers for Disease Control estimated that 1 in 68 children were diagnosed with ASD, a disorder of the nervous system whose causes and treatment remain elusive in many cases, despite its prevalence. In order to better understand the mechanisms underlying the mutations found in autism, Dr. Sanjana and his team will develop novel gene editing methods for high-throughput, genetically-based subtype identification of *de novo* ASD variants. *De novo* mutations are alterations in the DNA of a child that are not found in the parents. They can arise from mutations in the germ line cells (sperm or egg) of the parents. Emerging evidence from whole-exome and whole-genome sequencing studies in individuals with ASD and ID highlights a prominent role of *de novo* mutations. One goal of Dr. Sanjana's research is to rapidly deduce which mutations alter cortical neuron function and whether shared mechanisms exist between different mutations implicated in ASD and ID.

"We are pleased to support Neville Sanjana's promising autism research," said Dr. Jeffrey Borenstein, President & CEO of the Brain & Behavior Research Foundation. "The Young Investigator grants are part of the continued investment BBRF makes in brilliant young scientists conducting neurobiological research that is most likely to lead to breakthroughs in understanding and treating mental illness."

"We are very grateful for this support from the Brain & Behavior Research Foundation and are excited to develop CRISPR-based screens to speed progress in understanding the genetics of ASD," said Dr. Sanjana. His research utilizes high-throughput methods to identify changes in gene expression after ASD-associated *de novo* mutations are introduced into human cells. After designing and introducing a pooled CRISPR library into human pluripotent stem cells, Dr. Sanjana then differentiates human cortical neurons and analyzes changes in neuronal gene expression for each mutation.

Progress in ASD research has been slow, as mouse models of ASD mutations have been met with limited success. An important difference in Dr. Sanjana's approach is the use of human

cortical neurons – in effect, ‘human neurons in a dish.’ Dr. Sanjana explained, “Neurons are something that obviously we can’t get directly from human patients. However, gene editing in stem cells and differentiating the stem cells into cortical neurons gives us a powerful platform for understanding what is going wrong in neurons with autism-associated mutations.”

As the cost of sequencing a human genome continues to fall, physicians are better able to diagnose severe forms of autism and intellectual disability at early stages but treatment options are often limited. The Sanjana Lab’s research is focused on addressing this unmet need: to understand the genetic mechanisms underlying ASD in order to rationally design effective therapies.

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### **About the New York Genome Center**

The New York Genome Center is an independent, nonprofit academic research institution at the forefront of transforming biomedical research with the mission of advancing clinical care. A collaboration of premier academic, medical and industry leaders across the globe, the New York Genome Center has as its goal translating genomic research into the development of new treatments, therapies and therapeutics against human disease. NYGC member organizations and partners are united in this unprecedented collaboration of technology, science and medicine, designed to harness the power of innovation and discoveries to advance genomic services. Their shared objective is the acceleration of medical genomics and precision medicine to benefit patients around the world. For more information, visit our website at <http://www.nygenome.org>.

### **About the Brain & Behavior Research Foundation**

The Brain & Behavior Research Foundation awards research grants to develop improved treatments, cures, and methods of prevention for mental illness. These illnesses include addiction, ADHD, anxiety, autism, bipolar disorder, borderline personality disorder, depression, eating disorders, OCD, PTSD, and schizophrenia. Since 1987, the Foundation has awarded more than \$394 million to fund more than 4,700 leading scientists around the world, which has led to over \$3.9 billion in additional funding. 100% of every dollar donated for research is invested in our research grants. Our operating expenses are covered by separate foundation grants. For more information visit our website [bbrfoundation.org](http://bbrfoundation.org).

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