



## Office for People With Developmental Disabilities

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### **Institute for Basic Research in Developmental Disabilities Awarded \$1.95 Million NIH Grant to Study Rare Diseases**

The New York State Office for People With Developmental Disabilities' (OPWDD) Institute for Basic Research in Developmental Disabilities (IBR) has received a \$1.95 million grant, for a five-year period, from the National Institute of Health's National Institute of General Medical Sciences (NIGMS) to support IBR physician-scientist Gholson Lyon, MD, PhD, in his research on rare diseases. The grant is part of the NIGMS's Maximizing Investigators' Research Award for Early Stage Investigators initiative, which provides science investigators who have demonstrated ability to make major contributions to medical science the freedom to embark on ambitious, creative, and/or longer-term research projects.

Dr. Lyon plans to expand his studies related to the discovery and genetic characterization of rare human diseases. Among these diseases are ones involving severe developmental delay and physical malformations, such as Ogden syndrome. Dr. Lyon discovered and named this disease, which is characterized by craniofacial abnormalities, hypotonia, global developmental delays, cryptorchidism, cardiac anomalies, and cardiac arrhythmias. Ogden syndrome is estimated to occur in 1 of 1,000,000 births.

The rare diseases Dr. Lyon studies are associated with a modification of proteins called N-terminal acetylation (NTA), which is crucial for the regulation and function of different proteins and is believed to play an important role in cells' ability to proliferate. Despite the importance of NTA, its function is not well understood. Dr. Lyon's research focuses on understanding the function of NTA and its role in the origin and development of Ogden syndrome and other diseases involving mutations in the same gene, *NAA10*, as found in Ogden syndrome, or with mutations in a related gene, *NAA15*.

Dr. Lyon has dual roles at IBR: he is both a psychiatrist at IBR's George A. Jervis Clinic and a researcher, as head of the Genomic Medicine Laboratory in the Department of Human Genetics. With this grant, he will conduct clinical studies for families with *NAA10*- and *NAA15*-related syndromes to better understand the genetic basis of the syndromes and undertake basic research studies at the molecular level. He plans to reveal the biological pathways associated with these diseases.

"These studies will be a critical step toward revealing the role of NTA in human health and disease, including neurodevelopmental diseases," said OPWDD Commissioner Theodore Kastner, MD, MS.

IBR Acting Director Joseph J. Maturi said, "Dr. Lyon's extensive medical and scientific training and experience will help him successfully undertake these ambitious and important studies."

"These diseases have a profound impact on families," said Dr. Lyon. "I am grateful for this support from OPWDD and NIGMS."

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**About OPWDD and IBR:**

*The Institute for Basic Research in Developmental Disabilities (IBR) is the research arm of the New York State Office for People With Developmental Disabilities (OPWDD). OPWDD is responsible for coordinating services for nearly 140,000 New Yorkers with developmental disabilities, including intellectual disabilities, cerebral palsy, Down syndrome, autism spectrum disorders, Prader-Willi syndrome and other neurological impairments. It provides services directly and through a network of nonprofit service-providing agencies*