Third Annual Gale and Ira Drukier Lecture in Children's Health Research

November 10, 2017

Rett Syndrome: From the Clinic to Genomes, Epigenomes, and Neural Circuits featuring Huda Y. Zoghbi, MD Ralph D. Feigin Professor of Pediatrics, Neuroscience, and Molecular and Human Genetics at Baylor College of Medicine Investigator, Howard Hughes Medical Institute Director, Jan and Dan Duncan Neurological Research Institute at Texas Children’s Hospital Reception to follow at 5:00 PM. Rett syndrome is a delayed-onset childhood disorder, typically found in girls, that causes a broad range of severe neuropsychiatric disabilities, including loss of the ability to speak and socialize, and the development of tremors, ataxia, seizures, and stereotypic hand-wringing movements. The Zoghbi lab discovered that mutations in the gene MECP2 cause Rett syndrome, and before long it became clear that mutations in MECP2 can also cause autism and other neuropsychiatric phenotypes. Using genetically-engineered mice, the Zoghbi lab learned that the brain is acutely sensitive to MeCP2 levels; both decreases and increases in the amount of MeCP2 protein can lead to neurological problems that are also observed in humans. The research team showed that normalizing MeCP2 levels can reverse disease-like features in a mouse model of the human MECP2 duplication syndrome, a disorder that is usually found in boys and results from excess MeCP2. Zoghbi and collaborators have been gradually pinpointing the neurons and circuit abnormalities that mediate various symptoms. Building on this understanding of the relationship between neural circuits and the features of Rett syndrome, they showed that deep brain stimulation of a specific neural network improved learning and memory in a Rett syndrome mouse model.