Huda Y. Zoghbi, MD

2023 Eminent Scholar Lecturer
Hagler Institute for Advanced Study

Investigator, Howard Hughes Medical Institute; Distinguished Service Professor of Pediatrics, Molecular & Human Genetics, Neurology, and Neuroscience at Baylor College of Medicine

Eminent Scholar Lecture

Molecular Neurobiological Studies in Rett Syndrome and *MECP2* Disorders

Rett syndrome is a delayed-onset childhood disorder, typically found in girls, that causes a broad range of severe neurological disabilities, including loss of the ability to speak and socialize, and the development of tremors, ataxia, seizures, autonomic dysfunction, and stereotypic hand-wringing movements. We discovered that loss-of-function mutations in the *MECP2* gene cause Rett syndrome, and before long it became clear that milder mutations in *MECP2* can also cause other neuropsychiatric phenotypes ranging from autism to bipolar disorder. Using genetically-engineered mice, we 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. Nonetheless, normalizing MeCP2 levels reverses symptoms in a humanized mouse model of *MECP2* duplication syndrome, a disorder that results from excess MeCP2 and typically affects boys. Most recently we have found an approach that could delay onset of Rett symptoms, suggesting that earlier diagnosis through screening may be worthwhile for these disorders.

Please RSVP to hias@tamu.edu

HAGLER INSTITUTE FOR ADVANCED STUDY
at Texas A&M University

April 24, 2023 / 4:00 PM
MSC Gates Ballroom, Rm 2400
Reception immediately to follow

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