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Huda Zoghbi

Neuroscientist Huda Y. Zoghbi, MD, Receives \$500,000 Gruber Neuroscience Prize for Pioneering Work in Revealing the Genetic and Molecular Underpinnings of Neurological Disorders

June 15, 2011, New York, NY – Huda Y. Zoghbi, MD, will receive the 2011 Neuroscience Prize of The Peter and Patricia Gruber Foundation for her pioneering work in unlocking the genetic and molecular mysteries behind a number of devastating neurological disorders, including Rett syndrome, spinocerebellar ataxia type 1, and brain tumors called medulloblastomas. Her contributions to these discoveries have greatly advanced our scientific understanding not only of these disorders, but also of more common ones, including autism, Parkinson’s disease, and Alzheimer’s disease.

Zoghbi, 56, is a professor of pediatrics, molecular and human genetics, neurology, and neuroscience at Baylor College of Medicine in Houston, Texas. She is also director of the Jan and Dan Duncan Neurological Research Institute at Texas Children’s Hospital and a Howard Hughes Medical Institute investigator. Her work has inspired many researchers in the broad field of neurological disorders, and serves as an exemplar of how complex brain disorders can be better understood by basic genetics and molecular neuroscience.

She will receive the award November 13 in Washington D.C. at the Annual Meeting of the Society for Neuroscience and will deliver a lecture titled “Rett syndrome: Linking Epigenetics and Neuronal Plasticity.”

“There are numerous examples of basic science that has inspired translation into clinical treatment,” says Carol Barnes, Regents’ Professor in the Departments of Psychology and Neurology, Director of the Evelyn F. McKnight Brain Institute and Research Scientist in the ARL Division of Neural Systems, Memory & Aging at the University of Arizona, and chair of the Selection Advisory Board to the Neuroscience Prize. “What stands out about Dr. Zoghbi’s discoveries is that the original inspiration for her science was her clinical observations – and her determination to ‘go to the bench’ to solve the mystery of the disorder. Her work has revealed probable underlying mechanisms of a number of postnatal neurologic disorders, for which the path to clinical treatment can now be realistically followed – this trajectory to discovery and potential treatment is simply inspirational.”

Zoghbi began her career as a clinical pediatric neurologist, and much of her research has evolved from her early experiences with patients with rare neurological disorders. Her first major research breakthrough occurred in 1993 when she co-discovered, with long-time collaborator Dr. Harry Orr, a mutation in the gene *ATXN1* that is responsible for spinocerebellar ataxia type 1, a deadly neurodegenerative disorder characterized by a progressive loss of movement. That mutant gene, as Zoghbi and collaborators also discovered, produces a sticky protein called ataxin-1, which accumulates in brain cells and causes neuronal dysfunction through abnormal protein interactions. Scientists have subsequently found that something similar occurs in other brain disorders,



such as Huntington's, Alzheimer's, and Parkinson's diseases. The search is now on for drugs that might slow or stop that process.

In 1999, Zoghbi's lab identified the mutation in the gene *MECP2* that causes Rett syndrome, an autism spectrum disorder that leads seemingly healthy female toddlers to avoid eye contact, cease talking, engage in obsessive behaviors (such as constant hand-wringing), and develop other devastating symptoms. Before Zoghbi tracked down *MECP2*, scientists were not sure that this brain disorder was genetic. Further research by Zoghbi has uncovered a host of other information about *MECP2* mutations, including the finding that doubling the levels of *MECP2* can cause a host of neuropsychiatric features. These discoveries have opened exciting new approaches to the study of autism and other psychiatric disorders.

Another major finding from Zoghbi's lab has been the identification of *Math1*, a gene central to the formation of hair cells in the inner ear and of specialized neurons in the cerebellum involved in balance and proprioception (the sense of one's position in space). The finding that *Math1* can generate inner ear hair cells may lead to more effective treatments for certain types of deafness – especially age-related deafness and vestibular problems. Zoghbi and her team have also discovered that continuous activation of *Math1* can cause granule cells in the hindbrain in mice to grow nonstop. These findings are relevant for a common childhood brain tumor called medulloblastoma, and open up a new pathway for therapy in cancer.

"*Math1* turned out to be a medically relevant gene," says Zoghbi. "It's a beautiful example of how doing science for science's sake opens up new pathways that benefit mankind."

Additional Information

In addition to the cash award, the recipient will receive a gold medal and a citation that reads:

The Peter and Patricia Gruber Foundation proudly presents the 2011 Neuroscience Prize to Huda Zoghbi for her pioneering work on revealing the genetic underpinnings of neurological disorders.

Huda Zoghbi discovered that mutations in the MECP2 gene cause Rett syndrome, an autism spectrum disorder. Her work on MECP2 offers a new understanding of gene regulation in brain disorders and opens a new approach to studies of autism and other psychiatric disorders. She also illuminated the genetic basis of several spinocerebellar ataxias and provided insights on the role of protein aggregation in neurodegeneration.

Huda Zoghbi's work has inspired many other researchers in the broad field of neurological disorders, and serves as an exemplar of how complex brain disorders can be better understood by basic genetics and molecular neuroscience.

Laureates of the Gruber Neuroscience Prize:

- **2010: Robert H. Wurtz**, for pioneering work concerning the neural bases of visual processing in primates.
- **2009: Jeffrey C. Hall, Michael Rosbash, and Michael Young**, for revealing the gene-driven mechanism that controls rhythm in the nervous system
- **2008: John O'Keefe**, for discovering place cells, which led to important findings in cognitive neuroscience



- **2007: Shigetada Nakanishi**, for pioneering research into communication between nerve cells in the brain
- **2006: Masao Ito** and **Roger Nicoll**, for work on the molecular and cellular bases of memory and learning
- **2005: Masakazu Konishi** and **Eric Knudsen**, for work on the neural basis of sound localization
- **2004: Seymour Benzer**, for applying the tools of molecular biology and genetics to the fruit fly, *Drosophila*, and linking individual genes to their behavioral phenotypes

The Prize recipients are chosen by the Neuroscience Selection Advisory Board. Its members are:

Carol A. Barnes, University of Arizona, **Stephen Heinemann**, Salk Institute, **Masao Ito**, RIKEN Brain Institute, **David Lewis**, University of Pittsburgh, **Erwin Neher**, Max-Planck Institute, **Li-Huei Tsai**, Massachusetts Institute of Technology.

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The Gruber International Prize Program honors contemporary individuals in the fields of Cosmology, Genetics, Neuroscience, Justice and Women's Rights, whose groundbreaking work provides new models that inspire and enable fundamental shifts in knowledge and culture. The Selection Advisory Boards choose individuals whose contributions in their respective fields advance our knowledge, potentially have a profound impact on our lives, and, in the case of the Justice and Women's Rights Prizes, demonstrate courage and commitment in the face of significant obstacles.

The Neuroscience Prize honors scientists for major discoveries that have advanced the understanding of the nervous system.

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The Peter and Patricia Gruber Foundation honors and encourages educational excellence, social justice and scientific achievements that better the human condition. For more information about Foundation guidelines and priorities, please visit www.gruberprizes.org.

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Media materials and additional background information on the Gruber Prizes can be found at our online newsroom: www.gruberprizes.org/Press.php

By agreement made in the spring of 2011 the Gruber Foundation has now been established at Yale University.