SOUTHWESTERN NEWS

Media Contact: Amy Shields
214-648-3404
amy.shields@utsouthwestern.edu

UT SOUTHWESTERN SCIENTIST RECEIVES GRANT TO FURTHER RESEARCH INTO MECHANISMS INVOLVED IN MENTAL RETARDATION

DALLAS – April 16, 2002 – A UT Southwestern Medical Center neuroscientist has been awarded a nationally competitive three-year, $300,000 grant from the McKnight Endowment Fund for Neuroscience to study the underlying mechanisms of the most common inherited form of mental retardation.

Dr. Kimberly Huber, an assistant professor in the Center for Basic Neuroscience, will study Fragile X syndrome, which is characterized by attention deficit disorder, high anxiety and symptoms of autism. It affects one in 1,500 men and one in 2,500 women in the United States.

The syndrome is caused by a mutation of the *fragile X mental retardation 1 gene (Fmr1)*, which results in the loss of the fragile X mental retardation protein, or FMRP. There is no cure for fragile X syndrome.

Huber and her colleagues will focus on how synapses – connections between brain cells responsible for transferring information – change during brain development and during adulthood. The researchers will study these changes in a mouse model of fragile X syndrome. This "fragile X mouse" is a genetically altered mouse that lacks the FMRP. Through this evaluation, they hope to understand how two vital components of synaptic function are changed in fragile X syndrome.

"There is little known about the neural mechanisms that underlie mental retardation, and there are no gross abnormalities of the nervous system thought to give rise to fragile X syndrome," said Huber.

"Instead, there is evidence that the structure of synaptic connections between neurons is abnormal. The fact that this disorder is caused by the loss of one protein provides an extraordinary opportunity to discover the neural mechanisms of mental retardation and devise therapeutic strategies."

Specifically, Huber and other UT Southwestern scientists will study a form of synaptic

(MORE)

weakening, known as long-term depression, in the mice to understand how synaptic function is altered in fragile X syndrome. The researchers aim to define the function of the missing protein in neuronal communication.

"By understanding the developmental functions of this protein we will be able to determine if FMRP is really essential during early neuronal development or if we can reintroduce the protein into the neuron and re-establish its function after the neuron is developed," Huber said. "Results of our research will provide the framework for future clinical trials and facilitate progress towards a treatment for fragile X syndrome."

The McKnight Endowment Fund for Neuroscience is an independent organization funded solely by the McKnight Foundation of Minneapolis, Minn., and led by a board of prominent neuroscientists from around the country. The foundation has supported neuroscience research since 1977. Huber was one of six awarded the grant in competition with 121 other applicants.

###