Genetic Research Leads to New Discovery in Understanding Rare Neurological Disorder

February 1, 2009 - A study led by Laurie Ozelius, PhD at Mount Sinai School of Medicine has identified a gene associated with the development of primary torsion dystonia, also known as DYT6 dystonia. With funding provided by the Dystonia Medical Research Foundation (DMRF), Dr. Ozelius and her colleagues have found that mutations in the THAP1 gene cause DYT6 dystonia in Amish-Mennonite families, as well as in other ethnic groups.

Dystonia is a neurological movement disorder characterized by sustained, uncontrolled muscle contractions and spasms. There are multiple forms affecting men, women, and children of all ages and backgrounds. More than 300,000 people in Canada and the United States are estimated to have some form of primary dystonia. In terms of secondary dystonias, which can be caused by trauma, other disorders, conditions or diseases, the numbers can be propelled into the millions. There is no cure.

Since the pathogenic mechanisms of primary torsion dystonias are very poorly understood any new information about genetic causes of these diseases is desperately needed to facilitate new studies toward the understanding of those mechanisms. The newly discovered DYT6 gene and protein can now be used as drug discovery targets.

"Primary torsion dystonia are rare and devastating diseases," explains the Foundation's Science Officer, Dr. Jan Teller. "This is the second gene identified for this type of dystonia. Its discovery will greatly contribute to our knowledge about molecular mechanism of all the dystonias."

"We are greatly excited about Dr. Ozelius's findings, as they will help us to better understand the many different factors responsible for this puzzling and disabling disorder," says Dr. Mahlon DeLong, Scientific Director of the Foundation. "Dr. Ozelius has been a pioneer in genetic research on dystonia and we are grateful for her unwavering commitment to this and the broader dystonia community."

"I am grateful to the Foundation for their support of this research," adds Dr. Ozelius. "The DMRF has supported gene identification studies throughout its history and has been a leader in recognizing that these studies represent an important first step leading to molecular insights into the disease."

The Dystonia Medical Research Foundation is dedicated to advancing research for more treatments and ultimately a cure, promoting awareness and education, and supporting the needs and well being of affected individuals and families. DMRF Canada works in partnership with the Dystonia Medical Research Foundation in the United States to ensure funding of the best and most relevant dystonia medical research worldwide. To learn more about dystonia, contact Dystonia Medical Research Foundation Canada at 1-800-361-8061 or www.dystoniacanada.org

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