Think Tank Debates Genetic Screening and Clinical Practice

"Genetics and Biology of Dystonia" was the focus of our eighth annual Think Tank on Dystonia this past November as scientists, clinicians, and geneticists came to New York from around the world to discuss the latest research and exchange ideas.

This preeminent, international group reviewed a wide spectrum of promising new data, but it was the discussion of whether genetic findings are shaping clinical practice that sparked the broadest range of opinion.

While anecdotal evidence and early research suggest that the different subtypes of dystonia may respond to different treatments, the discussion indicated broad variance in how doctors currently use genetic testing in their practices, e.g., if they do send dystonia patients for genetic screening, whether the findings are used to make treatment decisions or, instead, to inform a diagnosis.

Future directions

The discovery of genes that reveal the biological pathways involved in dystonia and the development of targeted treatment continue to be central to much of the new work underway. While discovering the differences between the subtypes of dystonia may be key to making a diagnosis, clinicians are interested in the commonalities that can be used to find therapies that treat as many patients as possible.

"Sharing patient and genetic material will be needed during the next stage of genetic analysis," said Ted Dawson, MD, PhD of Johns Hopkins University School of Medicine and Chair of our Scientific Advisory Board. Laurie Ozelius, PhD of Mount Sinai School of Medicine added that "Worldwide collaborations in genetic research will be paramount, particularly for the more common focal dystonias."

Read more. An executive summary of our recent Think Tank on Dystonia is on www.dystonia-parkinson.org.

Dystonia Coalition Forms

A $6 million commitment over the next five years by the National Institutes of Health (NIH) has led to the establishment of a new Dystonia Coalition, a collaboration of scientists, institutions, patient advocacy organizations and the National Institutes of Health – all united to advance clinical research for dystonia.

The NIH commitment will support a Dystonia Coalition of 34 centers in the United States, Canada and Europe. This is the largest sum ever provided for a single project devoted to clinical and translational research in the dystonias. The Coalition was established as part of a $117 million expansion of the NIH’s Rare Diseases Clinical Research Network, which will coordinate research on more than 95 rare diseases.

H.A. Jinnah, MD, PhD, professor of neurology and human genetics at Emory University School of Medicine, will direct the Dystonia Coalition; Joel Perlmutter, MD, professor of neurology and radiology at Washington University School of Medicine, is co-director.

"I am very pleased that the NIH has recognized our program for clinical and translational research in dystonia with this big award. Lots of folks worked really hard to make it happen," said Dr. Jinnah. "We hope to be able to catalyze the development and testing of new treatments. It’s an exciting time for clinical research, and I’m happy to be a part of it."

Our Foundation, which has previously provided funding to Dr. Jinnah’s work, is a member of the Coalition.