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NIH network addresses research needs in rare disorders

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Clinical Research Network for Dystonia: The Dystonia Coalition

By Ami Rosen, MS, CGC and H. A. Jinnah, MD, PhD

Dystonia is a neurological disorder characterized by involuntary muscle contractions that lead to twisting or repetitive movements and abnormal postures. Focal dystonias involve an isolated region of the body. Examples include cervical dystonia, blepharospasm, spasmodic dysphonia, and limb dystonias. Segmental dystonias involve two contiguous body regions, and there is broader involvement in the generalized dystonias. Despite widely varying clinical manifestations, many studies have suggested the dystonias share certain mechanisms of pathogenesis. Studies of one type of dystonia, therefore, provide valuable lessons for others.

Dystonia research historically has faced two big challenges. The first challenge is dystonia is relatively uncommon, making it difficult for any single center to collect enough patients for meaningful studies rapidly. The second challenge is that different forms of dystonia often are seen by different subspecialists. For example patients with cervical dystonia tend to see neurologists, patients with blepharospasm go to ophthalmologists, and patients with spasmodic dysphonia see otolaryngologists or speech pathologists.

The *Dystonia Coalition* hopes to address these challenges with a 5-year grant recently awarded by the NIH Office of Rare Disease Research and the National Institute of Neurological Disorders & Stroke, by encouraging collaboration among institutions and subspecialty domains. Clinical and translational research relevant to the primary dystonias will be the primary focus, with an operational plan that is different from other NIH-sponsored consortia.

Specifically, the *Dystonia Coalition* has adopted a pharmaceutical model that permits investigators to participate in different studies at different times. Currently, there are 34 participating sites in 5 different countries. Emory University in Atlanta serves as the organizational hub with the Dystonia Medical Research Foundation and other patient advocacy groups integrally involved in providing logistical support for planned activities.

The Dystonia Coalition is planning three multi-center studies. Project 1 is a prospective natural history study for all primary dystonias, with a linked biorepository to store and distribute samples of DNA and other materials for research. This project is being led by Dr. Joel Perlmutter at Washington University in St. Louis. It was conceived after several retrospective studies showed the focal dystonias evolve over many years, a finding that questioned prevailing beliefs that they were relatively static disorders. A linked biorepository will provide a vital resource for gene and biomarker discovery, and a universal data and sample collection plan will address the limitations inherent in sharing data and samples across smaller pre-existing local collections.

Project 2 is devoted to developing more comprehensive assessment tools for cervical dystonia, and is being led by Dr. Cynthia Comella at Rush University, with added support from the Dystonia Study Group and Allergan, Inc. This project emerged as a result of prior meetings where experts recognized limitations of existing scales, along with a growing appreciation of psychiatric comorbidities and their potential impact on quality of life. A new scale will encompass all of these multifaceted aspects as a more accurate reflection of the whole condition to aid in monitoring outcomes of trials involving new therapeutic interventions. A teaching video also will be prepared so that others may learn the scale quickly and efficiently.

Project 3 aims at establishing a new diagnostic and rating tool for spasmodic dysphonia that can be used both in the clinic and for research studies. This project is being organized at James Madison University by Dr. Christy Ludlow, and was conceived following a consensus conference designed to delineate research priorities for spasmodic dysphonia sponsored by the National Spasmodic Dysphonia Association and the NIH. This new diagnostic tool will facilitate early recognition of the disorder, so patients can be directed more quickly to appropriate therapies. The tool also will be critically valuable for monitoring disease severity in response to potential new treatments.

In addition to these projects, the *Dystonia Coalition* will provide resources to foster the development of new projects and investigators interested in clinical and translational research in the dystonias. Proposals for the Pilot Projects Program will be considered twice yearly with decisions for funding based on relevance to primary dystonia, impact in the clinical arena, potential as a seed funding, and value for promoting collaboration. Proposals for the Career Development Program will be considered annually, with decisions for funding based on the candidate's track record and plan, local resources and mentorship, and impact on career development.

This new grant provides an unprecedented platform to bring the research community together in a common mission to address the most important issues in clinical and translational research for dystonia. Our next meeting is planned for 21-22 October 2010 in Atlanta, and we welcome feedback on current and future plans. For more information, please contact Ami Rosen at arosen3@emory.edu.

Ami Rosen, MS, CGC is a Project Coordinator and Genetic Counselor in Emory University's Department of Neurology. She received her BA in Psychology from the University of Pennsylvania and her MS in Genetic Counseling from Mount Sinai School of Medicine. She has coordinated and provided genetic counseling for Emory's presymptomatic genetic testing program for Huntington's Disease for the past nine years. Over that time period, she has also coordinated many research studies in the Department of Neurology focused on Alzheimer's Disease, Frontotemporal Dementia, Parkinson's Disease, Stroke, and Dystonia. She has provided educational lectures to lay

and professional groups and published papers as first author in both Pediatrics and Neurogenetics. She is currently the Program Coordinator for the ORDR/NINDS-sponsored *Dystonia Coalition*.



H. A. (Buz) Jinnah, MD, PhD is Professor in the Departments of Neurology, Human Genetics, and Pediatrics at Emory University School of Medicine in Atlanta, Georgia. He got his MD and PhD degrees with a focus in Neurosciences at the University of California in San Diego, then completed a Neurology Residency & Fellowship at Johns Hopkins University. He has a longstanding research interest in the dystonias, both primary and secondary forms. His studies have focused on basic mechanisms of pathogenesis, as well as clinical and translational aspects. His work has been funded by multiple grants from the NIH and patient advocacy groups, with more than 100 peer-reviewed publications, chapters, and reviews. Currently, he is the Program Director for the ORDR/NINDS-sponsored *Dystonia Coalition*. To contact Dr. Jinnah, e-mail hjinnah@emory.edu

