

Research-Dystonia Coalition

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I was privileged to represent the BEBRF at the first meeting of the Dystonia Coalition on November 5, 2009 in Washington D.C. This coalition should not be confused with the Dystonia Advocacy Coalition, the first cooperative venture of some of the dystonia groups. The Dystonia Coalition is one of 19 coalitions funded through the National Institutes of Health's National Institute of Neurological Diseases and Strokes and their Office of Rare Diseases Research. (ORDR). The emphasis of these coalitions is on collaboration between researchers from North America and Europe and patient advocacy groups. Representatives from both, as well as representatives of industry attended the meeting and there was great excitement in the room.

The initial focus of the Dystonia Coalition is to advance clinical research on primary focal dystonias and to find better therapies. It will have a five year \$6 Million award to move us closer to our goals. The multicenter Coalition will be led by H.A. Jinnah, MD, PhD, professor of neurology and human genetics at Emory University School of Medicine and Joel Perlmutter, MD, professor of neurology and radiology at Washington University School of Medicine. Dr. Perlmutter is an old friend of the BEBRF who led Scientific Day at our 1998 International Conference and has received research grants from the BEBRF in the past. According to Dr. Jinnah, "The misconception that adequate therapies are available for dystonia is impeding the development of better ones. This funding will allow our investigators to address several unmet needs and deepen our understanding of these rare and devastating diseases."

The meeting helped all the attendees to learn more about the dystonia organizations, about the research programs and the application process, how the research will be conducted, and data handled. Pilot Projects will provide seed money to foster promising ideas and promising research projects. Some of the patient groups have already agreed to collaborate on funding some projects. The BEBRF will co-fund one of the initial Pilot Projects--"Development and Validation of Clinical Diagnostic Guidelines and a Novel Severity Rating Scale for Primary Blepharospasm." (See Dr. Hallett's article). Other approved projects are related to Spasmodic Dysphonia (their first research program) and Cervical Dystonia.

Other aspects of the Coalition--

- In an attempt to increase the number of researchers interested in dystonia, the Coalition will contain a Career Development Program to help junior

faculty staff in neurology to start and develop careers in research.

- Researchers need patient samples to conduct research and Dr. Perlmutter's project will be: 1) creating a biorepository for focal dystonias to store the samples (DNA, serum and cells) that will serve the whole research community and 2) collecting data on their natural history.
- The Rare Diseases Clinical Research Network (RDCRN) will coordinate research on more than 95 rare diseases. Their goal is "to contribute to the research and treatment of rare diseases by working together to identify biomarkers for disease risk, disease severity and activity, and clinical outcome, while also encouraging development of new approaches to diagnosis, prevention, and treatment." Their website includes a Patient Registry where Coalition patients can go to register to be notified when clinical trials are available on their diseases.
- Because patient advocacy organizations are a very important part of this venture, another important coalition exists--the Coalition of Patient Advocacy Groups (CPAG) which represents the perspective and interests of all patient advocacy organizations associated with the various coalitions.

Through the years patients have wondered why dystonia groups didn't work together more to find a cure for dystonia. The leaders of the dystonia organizations acknowledged that this is a milestone that has come about as a result of cooperation and perseverance. Answers will come slowly and we have to be patient but this is our moment and it is an exciting time.