Highlights from
5TH INTERNATIONAL DYSTONIA SYMPOSIUM
OCTOBER 20-22, 2011 • BARCELONA, SPAIN

Introduction

"This is a momentous year," remarked Stanley Fahn, MD on day 1 of the 5th International Dystonia Symposium held on October 20-22, 2011 in Barcelona, Spain. "The first definition of dystonia was published exactly 100 years ago in 1911." So it was all the more fitting that this year was marked by the most extensive, comprehensive scientific symposium on dystonia to date. Dr. Fahn went on to praise the debate and discussion that was taking place at the symposium, commenting that this is what is needed to move the field forward.

The 5th International Dystonia Symposium (IDS5) represents the latest chapter in a definitive series of truly international dystonia meetings organized by the Dystonia Medical Research Foundation (DMRF) since 1975, the last of which was held in 2002 in Atlanta with 150 attendees. This year, the symposium attracted 560 dystonia clinicians and researchers from 38 countries. The audience was an encouraging mix of established dystonia experts and up-and-coming movement disorder specialists.

The symposium was a joint organizational effort of the European Dystonia Federation, the Dystonia Coalition, and the Dystonia Medical Research Foundation (DMRF). Individual dystonia patient groups funded travel scholarships for young investigators and industry sponsors provided additional support.

IDS5 brought together renowned researchers and clinicians for an extensive, three-day program of dystonia topics including clinical issues, functional anatomy, pathophysiology, genetics and molecular biology, treatments, and the very latest developments in treatment approaches. The Symposium was the most thorough, comprehensive meeting on dystonia to date.

The purpose of the meeting was not simply to rehash what is known about dystonia but to challenge current thinking, explore controversial topics, and push forward into a deeper understanding of the disorder and how to treat those who are affected. Sessions and topics were designed to spark discussion between speakers and the audience, and at least one especially provocative topic was served up for debate every afternoon. In addition to oral presentations, poster sessions presenting preliminary findings of ongoing research were scheduled each day: clinical, anatomy and physiology, genetics and treatment. The full spectrum of dystonia, from primary and secondary forms and beyond, were addressed including examinations of what fundamental lessons specific forms can teach us about all dystonias. Discussions consistently circled back to how the topic at hand was relevant to benefitting patients. Every section of the program concluded with a panel discussion of issues raised during the presentation and important considerations for the future.

It was not uncommon to hear attendees remark that this was the best dystonia symposium they had ever attended. The symposium demonstrated how much is known about dystonia, challenged how to best apply what we know, and began to chart a course through what has yet to be learned, guided consistently by a compass that points to better treatment for affected individuals.

Highlights & Headlines: About IDS5

The 5th International Dystonia Symposium (IDS5) was a collaborative effort of the European Dystonia Federation, the Dystonia Medical Research Foundation (DMRF), and the Dystonia Coalition.

The symposium was a truly international meeting dedicated to dystonia. A record 560 attendees from 38 countries attended.

The participants included a mix of established dystonia experts and up-and-coming researchers and clinicians. Several dystonia patient organizations funded travel scholarships to allow young investigators to attend.

The agenda included three full days of presentations with updates on key areas and discussion of controversial and emerging areas of research.

The program consisted of succinct, 20-minute presentations which were followed by open discussion. The meeting was designed to be interactive, stimulate debate, and identify areas for future collaboration.

In addition to oral presentations, over 100 posters represented some of the very latest dystonia studies.

Proceedings from every International Dystonia Symposium have been published. *Dystonia 5* will be published shortly to summarize the IDS5 presentations.

The ideas shared at the symposium will lead to further studies, new collaborations, and scientific workshops. The next International Dystonia Symposium is tentatively scheduled for 2015.

Highlights & Headlines: About Dystonia

As researchers learn more about dystonia, the definition of dystonia is changing. A Dystonia Classification Committee met just before the symposium to revisit the currently used descriptions and classifications and to propose a new scheme. Because dystonia occurs in diverse forms and with a spectrum of clinical presentations and symptoms, creating an accurate system to classify dystonia is essential to understanding the disorder better. Properly classifying dystonia has a direct impact on proper diagnoses and treatment.

It has become clearer than ever that dystonia is more than a “movement disorder.” Research is demonstrating that individuals with dystonia display differences in how their brains process sensory information compared to unaffected individuals. There may be additional non-motor components to some forms of the disorder that may be overlooked.

Areas of the brain implicated in dystonia are associated with not only movement but also behavior, cognition, and emotions. There may be psychological components, such as depression, in some forms of the disorder that are sometimes overlooked in the treatment process. As a result, quality of life may be compromised. Researchers are investigating what it truly means to treat the “whole person.”

Rating scales used to assess dystonia in patients are being expanded and revised to more accurately measure motor symptoms as well as additional factors such as impact on daily living, pain, emotional health, and overall quality of life.

Dystonia is not just a disorder of the basal ganglia, as once thought. Symptoms in some forms of dystonia appear to arise as well from the complicated interaction between the basal ganglia and the cerebellum. Other brain areas including the motor cortex, thalamus, cerebellum, and corpus callosum are implicated in dystonia.

The basal ganglia remain an important point of focus in dystonia research. The concept of "reduced inhibition" in controlling movement and abnormal neural plasticity as significant contributors to dystonia are linked to basal ganglia dysfunction.

New imaging studies are revealing that an area of the basal ganglia called the putamen is enlarged in individuals with several forms of dystonia.

Researchers are uncovering multiple genes associated with certain forms of dystonia. 21 genes and gene markers are known to date. Various relationships and associations among different dystonias previously thought to be separate forms are being uncovered.

Molecular genetic studies point to a growing number of proteins believed to contribute to the disorder at the cellular level. A new understanding of how these proteins interact with each other is taking shape.

The role of the torsinA in neurons is becoming clearer. TorsinA is now known to interact with many proteins including other dystonia-related proteins such as THAP1 and SGCE.

Researchers continue to study the role of a protective factor, an additional mutation occurring in the DYT1 gene that appears to reduce the risk of a person with the DYT1 dystonia-causing mutation from developing symptoms.

Although deep brain stimulation (DBS) for dystonia has proven to be highly effective, there is a renewed consideration of ablative brain surgeries such as the pallidotomy to treat dystonia. DBS is not the only viable surgical option. Additional therapeutic stimulation methods, such as cerebellar and premotor cortical stimulation, are under investigation.

Physical therapy may be an underrated and underused therapeutic tool. Larger, more complete studies are needed.

Increased brain plasticity appears to play a major role in some forms of dystonia. This might point toward possible biomarkers to assist in diagnosis and identify individuals at risk of developing dystonia. Increased plasticity appears to be a quality that distinguishes dystonia from other movement disorders.

Contrary to previous ideas about cell loss in dystonia, preliminary studies suggest that there may be a subtle loss of Purkinje cells in adult onset primary focal dystonia. Purkinje cells, large neurons located in the cerebellum, are lost according to a recent study.

Several speakers stressed the role of the physicians' training in treatment success with botulinum neurotoxin and deep brain stimulation (DBS) highlighting the importance of patients consulting qualified medical teams.

IDS5 Scientific and Program Committee

Alberto Albanese, MD, Istituto Nazionale Neurologico Carlo Besta – Italy
Mahlon DeLong, MD, Emory University School of Medicine - USA
Wendy Galpern, MD, PhD, National Institute of Neurological Disorders and Stroke – USA
Mark Hallett, MD, National Institute of Neurological Disorders and Stroke - USA
Janet Hieshetter, Dystonia Medical Research Foundation – USA
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Ryuji Kaji, MD, PhD, Tokushima University Hospital - Japan
Christine Klein, MD, University of Lübeck - Germany
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Jan Teller, MA, PhD, Dystonia Medical Research Foundation – USA
Marie Vidailhet, MD, PhD, Hôpital de la Salpêtrière - France
Tom Warner, PhD, FRCP, University College London - UK

IDS5 Organizing Committee

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Kimberly Kuman, National Spasmodic Dysphonia Association – USA
Alistair Newton, European Dystonia Federation - France
Jody Roosevelt, Dystonia Medical Research Foundation – USA
Ami Rosen, Emory University School of Medicine - USA

IDS5 Session Chairs

Mahlon DeLong, MD, Emory University School of Medicine - USA
Stanley Fahn, MD, Columbia University Medical Center - USA
Joseph Jankovic, MD, Baylor College of Medicine - USA
Sabine Meunier, MD, PhD, Institut National de la Santé et de la Recherche Médicale – France
Nardo Nardocci, MD, Istituto Nazionale Neurologico Carlo Besta - Italy
Laurie Ozelius, PhD, Mount Sinai School of Medicine - USA
Joel Perlmutter, MD, Washington University School of Medicine - USA
Antonio Pisani, MD, University of Rome Tor Vergata - Italy
Hartwig Siebner, MD, Danish Research Center for Magnetic Resonance - Denmark
Eduardo Tolosa, MD, University of Barcelona - Spain
Marie Vidailhet, MD, PhD, Hôpital de la Salpêtrière - France
Giovanni Defazio, MD, PhD, University of Bari – Italy

IDS5 Speakers

Alberto Albanese, MD, Istituto Nazionale Neurologico “Carlo Besta” - Italy
Eckart Altenmuller, MD, University for Music, Drama and Media - Germany
Kailash Bhatia, MD, DM, FRCP, University College London - UK
Xandra Breakefield, PhD, Massachusetts General Hospital - USA
Robert Chen, MA, BChir, MB, MSc, Toronto Western Hospital - Canada
Cynthia Comella, MD, Rush University Medical Center - USA
Marina de Koning-Tijssen, MD, PhD, University of Amsterdam - The Netherlands
Guenther Deuschl, MD, Christian-Albrechts-Universität zu Kiel - Germany
Bogdan Draganski, MD, Centre Hospitalier Universitaire Vaudois - Switzerland
Dirk Dressler, MD, Hannover Medical School - Germany
David Eidelberg, MD, The Feinstein Institute for Medical Research - USA

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Angelo Quartarone, MD, University of Messina Medical Center - Italy
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Caroline Tanner, MD, PhD, The Parkinsons Institute and Clinical Center - USA
Michele Tinazzi, MD, Università di Verona - Italy
Tom Warner, PhD, FRCP, University College London - UK
Kirsten Zeuner, MD, University of Schleswig-Holstein, Campus Kiel - Germany
Mateusz Zurowski, MD, Toronto Western Hospital – Canada

Extra-curricular Activities

A number of additional meetings took place during and surrounding IDS5, including:

- Travel Stipend Recipients Dinner (October 18)
- Dystonia Coalition Steering Committee Meeting (October 19)
- Classification Meeting (October 20)
- Genetics Meeting (October 22)
- Dinner Meeting of International Dystonia Societies (October 22)
- European Dystonia Federation General Assembly (October 23)

Written by the DMRF
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