



# Update

## EUROPEAN DYSTONIA FEDERATION (DYSTONIA EUROPE)

MEMBERS' NEWSLETTER  
WINTER 2011

### BARCELONA – FOCUS OF THE DYSTONIA WORLD IN 2011

Three articles on different aspects of  
this historic event

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## 5TH INTERNATIONAL DYSTONIA SYMPOSIUM - EDF AND ITS PARTNERS ACHIEVE A GREAT SUCCESS

*After 3 years of planning and a huge amount of hard work, in October this year EDF proudly welcomed more than 550 dystonia specialists from 39 countries to the 5<sup>th</sup> International Dystonia Symposium in Barcelona.*

The venue – an excellent modern auditorium in central Barcelona, was chosen in summer 2009 and potential sponsors were approached for support. An experienced local conference management company was selected, and caterers, hotels and other suppliers were identified. The EDF Medical Advisory Board approved a draft scientific programme, and the process of inviting speakers and publicizing the event was ready to begin by early summer 2010.

During that summer, the Dystonia Coalition became our partner for the conference. The Coalition is an important network of scientists based in USA but with contributing scientists from Europe and other continents, supported by a large grant from the US National Institutes of Health and by all the US dystonia patient organisations. So - the original conference title of “Dystonia-Europe 2011” became “5<sup>th</sup> International Dystonia Symposium”.

The Coalition scientists, including several from Europe, created a new, longer and even more ‘scientific’ programme with 53 speakers and chairs with sessions over 3 full days, while EDF concentrated on the organisation and financing. An enlarged Organising Committee led by EDF, now included the Dystonia Medical Research Foundation, National Spasmodic Dysphonia Association, and a representative from the Coalition.

In Barcelona from 20 to 22 October, the venue was excellent, the programme and speakers were of

very high quality, and the atmosphere during the whole event was one of excitement and satisfaction that the arrangements ran so smoothly. The social events were very much enjoyed, and during all breaks/lunchtimes/evenings a huge amount of networking was going on. Many of the specialists said it was the best conference they had ever attended!

Most large-scale medical conferences in neurology cover sessions on several different illnesses, which brings complication and clashes in the programme. Our meeting in Barcelona avoided these difficulties because it focused on dystonia alone. The success of our Symposium shows clearly that the specialists were very happy to concentrate on dystonia, without the distractions of sessions on other conditions.

This was the biggest international scientific event solely on dystonia ever to have been held anywhere in the world. Its success is already stimulating even greater interest in dystonia among scientists in many countries, and has brought the dystonia ‘community’ even closer together. All of this can only push forward research on dystonia and the possibilities for faster results in improving treatments and finding a cure.

We are extremely proud of our achievement and we are very grateful to the Coalition, DMRF and NSDA who became our partners and provided such strong support during the important final year of the process.

The Symposium sponsorship from Allergan, Medtronic, Ipsen and Merz was much appreciated as always. But the grants from DMRF, Movement Disorder Society, Bachmann-Strauss Foundation, the Dystonia Society and, above all, the strong early support of the Foundation for Dystonia Research, allowed us to finalise the arrangements which made the event such a success. We are grateful to them all.

*Executive Director EDF, Alistair Newton*

## PRESIDENT'S MESSAGE



*Monika Benson*

### **Dear Friends and Colleagues,**

We are now Dystonia Europe! A much broader platform to improve the focus for all European dystonia 'stakeholders'.

At the 2011 General Assembly, in Barcelona, the EDF delegates agreed on the new name and several changes to the statutes. Now there is a lot of work being done in order to launch the "new look" around February 1st 2012.

**DYSTONIA  
EUROPE**

*February 1st – launch of new name and new profile.*

EDF was founded in 1993 and since then the world of dystonia has changed. Patient 'self-help groups' have become 'patient advocacy organisations'. Today, they have a much stronger voice and more influence. Research efforts and treatment options have increased,

and there is a lot more interest in dystonia today, from clinicians, researchers and the public. And by broadening our membership and strengthening our Board, we hope to see Dystonia Europe move forward strongly into this new 'world' and continue to play an important role in improving the lives of people living with dystonia.

The 2011 General Assembly followed the greatest success in EDF's history – the 5th International Dystonia Symposium. This was, by far, the biggest and most successful meeting which has ever taken place in the world of dystonia. We are very proud of this great achievement, and grateful for the support of our friends and colleagues in the Dystonia Coalition and the Dystonia Medical Research Foundation.

Since EDF's previous international medical conference in Hamburg 2008, the number of participants more than doubled from 250 to 560 specialists, who came from 39 countries over the world. Fifty three dystonia experts contributed to a programme of very high quality and to the realisation of the largest conference ever held solely on dystonia. In the first coffee-break an excited neurologist exclaimed "This is more like a cocktail-party!" The buzzing sound of networking filled the large room outside the auditorium and the atmosphere was full of excitement and anticipation. Our strategy of reaching out to the professionals for an specialised event, without other movement disorders, has succeeded and, from now on, it is very obvious that "dystonia deserves its own symposium"!

I am very grateful to all the people in the Scientific Committee, the Organising Committee and the Local Organising Committee, all of whom contributed to making our Barcelona event such a success. I would especially like to thank our own

Executive Director Alistair Newton for his dedicated work on the conference for the last three years!

Without his passion and commitment the symposium would not have been so successful.



*From left: Monika Benson, the members of the local Conference Managers staff and Alistair Newton*

Another very exciting development for Dystonia Europe is the new scientific initiative led by Professor Alberto Albanese – “European Network for the study of Dystonia Syndromes”. This is a 4-year project that is intended to create a large network of dystonia researchers from all over Europe. Dystonia Europe participated at the first meeting of around 25 scientists in November and was appointed as the Grantholder of the project. As your President, I have been appointed to the project Managing Committee, and Dystonia Europe will provide the administrative arm of this important new scientific organisation.

Göran Bylund has decided to retire from the EDF Board, and I take this opportunity to thank him for his great efforts and contribution to our work over the last two

years. Göran will be very much missed and we wish him all the best for the future. New board members are Prof. Maja Relja from Croatia and Prof. Marina De Koning-Tijssen from the Netherlands. I welcome them both to the board and I look very much forward to our first meeting with the “new team” in January 2012.

As you will see later in this Update, we have also appointed 4 more specialists to our Medical and Scientific Advisory Board and the new Polish Dystonia Society has become a member of Dystonia Europe.

We welcome all of them and look forward to their contribution to our work. Looking back at the year of 2011, I think we can all feel proud of what the old EDF achieved in its final year with that name – we have been part of organising the largest ever scientific conference on dystonia; there are new and exciting projects developing in dystonia across Europe; the European dystonia family was enlarged by the recent membership of the Polish national group, and we have now started to make contacts for a new group in the Czech Republic. I hope this is just the beginning of our important work of reaching out to countries where there are still no dystonia patient organisations.

Finally I would like to thank YOU for all the work you do for people with dystonia in your country. Whether it is producing the national newsletter, organising an awareness activity, or support a newly diagnosed it is just as important. ALL of those activities, together, create our ‘Dystonia Europe’.

I wish you a wonderful Christmas and all the best for 2012.

*Monika Benson*

## EDF GENERAL ASSEMBLY 2011

### A new name and a new approach after 19 years!

*After the excitement and the hard work of the scientific symposium in Barcelona last October, the week's activities ended with a busy General Assembly day for EDF.*

The programme, on Sunday 23 October, first gave the EDF delegates a review of the Barcelona Symposium from Prof Buz Jinnah, Chair of the Dystonia Coalition's Steering Committee. This was followed by information on the new European Network for the Study of Dystonia Syndromes from its new Chair, Prof Alberto Albanese. We then heard a presentation by Amanda Benson, a consultant in corporate social responsibility and marketing who has been advising the EDF Board on the use of modern communication media such as Facebook and other social networks. If Dystonia Europe is to succeed in its aim to broaden its appeal, we must make ourselves more accessible to a greater number of people, using the communication channels which are now so universal, especially to younger sections of society. Amanda's presentation was very well-received and delegates were encouraged to re-examine the modes of communication of their national organisations with their members and others. Then, the 2011 winner of the EDF David Marsden Award, Dr David Bradley, presented his paper on "Temporal discrimination threshold as an endophenotype in adult-onset primary torsion dystonia." Many of the scientists, who had already heard his presentation during the symposium, were very complimentary about the quality of the research involved.

Then the formal part of the Annual Assembly took place, with several important features this year, which will launch our organisation

into a new era as "Dystonia Europe".

First, the meeting agreed that membership fees should remain at the present level and be reviewed in 2012. They then considered the Board's proposals for modernising the name and the approach of the Federation. These are intended to broaden our membership, strengthen our Managing Board and establish our organisation even more firmly as a pan-European platform for all dystonia 'stakeholders' across the continent. These proposals were agreed by the meeting and Professor Maja Relja representing the Croatian Dystonia Society and Professor Marina de Koning-Tijssen of the Netherlands Society, were unanimously elected to the Managing Board.

Dr Göran Bylund, from the Finnish Dystonia Society, had decided to retire from the Managing Board at this meeting and the President, Monika Benson, thanked him for his work on the Board over the previous two years. His positive approach and his thoughtful and logical advice will be much missed. After some discussion regarding the work of EDF and the future activities of Dystonia Europe (DE), the formal meeting closed and the delegates continued their discussions over lunch.

The logo for Dystonia Europe, featuring the word "DYSTONIA" in a large, orange, sans-serif font above the word "EUROPE" in a smaller, red, sans-serif font. The letters are slightly offset to the right.

**The new profile of Dystonia Europe** is being created by a public relations consultant in Stockholm, who is giving time and expertise for free in support of our organisation and our work to help dystonia patients. We are very grateful for this generous contribution. The new profile, website and other changes are expected to be ready to launch by 1st February 2012.

*Alistair Newton*



### **Highlights from**

## **5<sup>TH</sup> INTERNATIONAL DYSTONIA SYMPOSIUM**

**OCTOBER 20-22, 2011 •  
BARCELONA, SPAIN**

### **Introduction**

"This is a momentous year," remarked Stanley Fahn, MD on day 1 of the 5<sup>th</sup> 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 5<sup>th</sup> 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 5<sup>th</sup> 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

H. A. Jinnah, MD, PhD, Emory University School of Medicine - USA

Ryuji Kaji, MD, PhD, Tokushima University Hospital - Japan

Christine Klein, MD, University of Lübeck - Germany

Anthony Lang, MD, FRCPC, Toronto Western Research Institute - Canada

Alistair Newton, European Dystonia Federation - France

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**

Janet Hieshetter, Dystonia Medical Research Foundation – USA

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

Rodger Elble, MD, PhD, Southern Illinois University School of Medicine - USA

Steven Frucht, MD, Mount Sinai School of Medicine - USA

Victor Fung, MBBS, PhD, FRACP, The University of Sydney - Australia

Thomas Gasser, MD, University of Tübingen - Germany

Robert Gross, MD, PhD, Emory University School of Medicine - USA

Kathrin Grundmann, MD, University of Tuebingen - Germany

Mark Hallett, MD, National Institute of Neurological Disorders and Stroke - USA

Ellen Hess, PhD, Emory University School of Medicine - USA

H. A. Jinnah, MD, PhD, Emory University School of Medicine - USA

Ryuji Kaji, MD, PhD, Tokushima University Hospital - Japan

Christine Klein, MD, University of Lübeck - Germany

Joachim Krauss, MD, PhD, Medical University of Hannover - Germany

Mark LeDoux, MD, PhD, University of Tennessee Health Science Center - USA

Stephane Lehericy, MD, PhD, Hôpital Pitié-Salpêtrière - France

Jean-Pierre Lin, MB, ChB, MRCP, PhD, Evelina Children's Hospital - UK

Katja Lohmann, PhD, University of Lübeck - Germany

Andres Lozano, BSc, MD, BMedSci, FRCSC, PhD, Toronto Western Hospital - Canada

Jonathan Mink, MD, PhD, University of Rochester Medical Center - USA

Angelo Quartarone, MD, University of Messina Medical Center - Italy

John Rothwell, PhD, University College London - UK

Young Ho Sohn, MD, PhD, Yonsei University Medical Center - South Korea

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*

*November 30, 2011*

## **Dystonia Europe sets the COMPASS for the future**

Early in October, the EDF board received the good news from Medtronic Foundation that the grant application for a development project, COMPASS, had been approved.

This is a one-year project intended to reach out for human and economic resources for Dystonia Europe in order to secure the stability of the organisation for the future. Monika Benson, President of Dystonia Europe, will be the project manager and report directly to the Dystonia Europe managing board.

The activities of the project will focus on the following main areas:

### **Developing partnerships with sponsors**

By reviewing existing partnerships and searching for new possibilities, including the European Union grant programmes and other sources external to the pharma and medical device industries.

### **Developing membership involvement and recruiting board members**

Including closer contact with existing member organisations and recruitment of new members. Finding potential new Board members to match the needs of Dystonia Europe.

### **Broadening the partnership with the Medical/Scientific professions**

To create and realise more opportunities to work closely with the clinicians and researchers with projects such as: The David Marsden Award Academy; the new European Network for the Study of Dystonia Syndromes; the expected "European Year of the Brain" in 2014, driven by the European Brain Council, in

which Dystonia Europe is centrally involved.

### **Communication**

Develop and modernise the overall communication policy of Dystonia Europe, and manage the introduction of the use of social media such as Facebook

The DE board believes that the strategy of setting the COMPASS in the above areas will strengthen and develop the impact and role of Dystonia Europe for the future.

*Monika Benson*

## **ANNUAL GENERAL ASSEMBLY 2012 Bol, Island of Brač, Croatia 21/23 September 2012**

The 2012 General Assembly will take place on 21/23 September on the Island of Brač and we ask you to put the dates in your agenda now. It's an exciting location and we will have a stimulating programme. Travel is normally via the airport of Split, and more information will be sent to you soon.



*Bol is an attractive tourist destination with good conference facilities on the south coast of the island of Brač*

## Personal summary of the 5<sup>th</sup> Dystonia Symposium 20. – 22. October 2011 in Barcelona, Spain

*Merete Avery, Norway*

As we travelled to Barcelona, for the 5<sup>th</sup> International Dystonia Symposium, we had high expectations that this would be an event where we would get new insights into this movement disorder and discover how much research is being carried out on the illness. We were not disappointed. We were delighted to return from Barcelona with information that there is a great interest in dystonia and an increasing amount of research. This was the biggest and most successful scientific event ever to have taken place for dystonia, and many of the scientists were loud in their praise of the organisation.

We arrived in Barcelona on the 19<sup>th</sup> October and stayed at a hotel only minutes from the location of the Symposium. On the morning of the 20<sup>th</sup> we walked to AXA Auditorium, which is a very comfortable venue for an audience of up to 620 people. We had 560 people registered to attend over the three days of the conference.



*Monika Benson*

The President of European Dystonia Federation (now Dystonia Europe), Monika Benson, opened the Symposium with a welcome speech, followed by a short presentation from Prof Buz Jinnah, principal scientist of the Dystonia Coalition. Dystonia Europe has done an outstanding job initiating and putting this event together over the past three years since their successful medical conference in Hamburg 2008, with the partnership of the Dystonia Coalition and organizational support from DMRF (Dystonia Medical Research Foundation) since summer 2010.

EDF was successful in obtaining sponsorship from Allergan, Foundation for Dystonia Research, Ipsen, Medtronic, Merz, and the Dystonia Society. With the later partnership of the Coalition, which made the Symposium a much longer and even more 'scientific' event, more funding was needed. The strong support of DMRF brought sponsorship from The Movement Disorder Society and The Bachmann-Strauss Foundation, with a very generous grant from DMRF itself.

The large majority of the 560 participants were neurologists and other health care specialists, with many basic research scientists from 39 countries around the world. A number of dystonia patient groups were represented, from Europe and USA.

During the 3-day Symposium in Barcelona, The David Marsden Award 2011 was presented by the EDF President, Monika Benson, to Dr. David Bradley, of St Vincent's University Hospital, Dublin, Ireland, for his research paper "*Temporal Discrimination Threshold: VBM evidence for an endophenotype*". Dr Bradley then presented his paper as part of the Symposium programme, and later in the week, again presented his research results to EDF's General Assembly.

There were 53 scientific speakers and chairpersons involved in the Symposium, and all topics focused clearly on dystonia. The main themes were: “Clinical issues”; “Functional Anatomy of Dystonia”; “Physiology and Pathophysiology”; “Genetics and Molecular Biology”; “Emerging Issues in Therapy”.

Two satellite sessions were organized by Merz and Medtronic, and over 100 posters describing different research studies were accepted for display. As is normal at scientific conferences, the authors of these posters all were available in the poster sessions, to answer questions on their research.

In addition, senior scientists led groups of less-experienced researchers and clinicians on ‘Poster Tours’, where specific topics were chosen to stimulate scientific discussion, led by the senior people. There was also a video session, led by more experienced clinicians, when many different and unusual dystonia symptoms were discussed.

This was a tremendous opportunity for specialists to meet, network and learn in so many ways and increase their knowledge on one subject – DYSTONIA. In Barcelona, this was possible without the usual distractions of other conferences where more illnesses, such

as Parkinson’s or other neurological conditions are involved, and attract more attention.

The 5<sup>th</sup> International Dystonia Symposium was a great event for the worldwide dystonia community, where researchers, clinicians and other health care professionals shared their knowledge and brought attention to new areas of interest for further research. All this is of great benefit for the many people in all countries of the world who already live with dystonia, and for those whose diagnosis is yet to come.

I believe the great success of Barcelona has been a huge step in the progress towards new treatments and, one day, a cure for dystonia...



*15<sup>th</sup> December, 2011  
Merete H. Avery,  
Norwegian Dystonia Association*



*The Symposium lunch network, AXA Auditorium*

## COST - (European Cooperation in Science and Technology)

COST is one of the longest-running European instruments supporting cooperation among scientists and researchers across Europe. It is an intergovernmental framework of 36 countries for European cooperation in these fields, allowing the coordination of nationally-funded research on a European level. It is not an EU organisation, but has close contacts with the European Commission.



*Prof Alberto Albanese*

Earlier this year (2011), Prof Alberto Albanese – with the support of a large number of dystonia specialist in centres across Europe, and from EDF - submitted a proposal to COST applying for a grant to create a European network for the study of dystonia syndromes. These applications are made in competition, with scientists from other specialties reviewing and commenting on the proposals. The competition from other applicants was very strong but the quality of the dystonia proposal was enough to win one of only three grants in 2011. Following on the great success of our Barcelona symposium, and as more scientists are made aware of this initiative, an increasing number of institutions are making requests to join the network.

The project to create the network will run for four years and the first meeting of the project Management Committee took place in November. Prof Albanese was appointed Chairman, with Prof Marie Vidailhet as Vice Chair. EDF was appointed Grantholder of the project, with our President Monika Benson as the Legal Representative and our Executive Director Alistair Newton as Financial Representative. This means that EDF is centrally involved and will deal with the finances and the administration of the network. Monika is also a member of the project Management Committee.

The aims of the project are described in the following two paragraphs taken from the COST website.

*Dystonia syndromes (DS) are the third most common movement disorder, yet they are poorly understood and are underdiagnosed. The causes of these disabling neurological conditions are poorly known and the mechanisms that give rise to the uncontrollable involuntary movements have not been elucidated. Particularly the role played by genetic and environmental factors is unclear. Scientific initiatives are fragmented across Europe and hampered by insufficient dimension.*

*This project will foster the creation of an interdisciplinary network of experts to promote genetic studies of DS, develop experimental animal models, standardise and harmonise patient care and educate the public and professionals about the disorder. It will create a unified platform for sharing expertise and exchanging procedures among centres in COST countries. Short-term scientific missions, training schools and workshops will be organised to train early-stage scientists with specific skills in the field of DS. Best practice guidelines will be set and national initiatives will be unified into a large-scale*

*effort to improve knowledge of DS and increase standards of care for patients.*

So, our new format of “Dystonia Europe” – intended to offer a broader platform for **all** dystonia ‘stakeholders’ in Europe – has already established our central place in this important initiative to help dystonia patients.

As the project develops, we will report on progress.

*COST website:*

[http://www.cost.esf.org/domains\\_actions/bmbs/Actions/BM1101](http://www.cost.esf.org/domains_actions/bmbs/Actions/BM1101)

*Alistair Newton*

## David Marsden Award 2011- European Dystonia Federation

*"This year's David Marsden Award was generously sponsored by Ipsen Pharma and won by Dr David Bradley of St Vincent's University Hospital, Dublin. The presentation of the Award was made by EDF President, Monika Benson during the 5th International Dystonia Symposium in Barcelona, at which Dr Bradley also presented his paper. The following article on Dr Bradley's paper has been provided by Prof Michael Hutchinson, Dublin"*

### **Temporal discrimination threshold as an endophenotype in adult-onset primary torsion dystonia. Brain 2009;132:2327-35**

*David Bradley , Robert Whelan, Richard Walsh, Richard B Reilly, Siobhan Hutchinson, Fiona Molloy, Michael Hutchinson.*

*St Vincent's University Hospital, Beaumont Hospital, University College Dublin and Trinity College Dublin.*



*David Bradley, the winner of the David Marsden Award 2011. From left: Monika Benson, David Bradley and Sandrine Guillebaud of Ipsen Pharma*

## Introduction

Adult onset primary torsion dystonia is a movement disorder with a number of different clinical manifestations and is classified by the part of the body affected. The disorder includes cervical dystonia (spasmodic torticollis) (neck), blepharospasm (muscles of eye-closure), oro-mandibular dystonia (mouth, tongue and jaw), spasmodic dysphonia (vocal cords) and focal hand dystonia (writer's cramp) (hand and forearm muscles), which tends to begin after 30 years of age. The term “primary” indicates that there is no other disease causing the dystonia and all investigations, including MRI of the brain, are normal.

Although most individuals with adult onset primary torsion dystonia have no other family member affected (and are thus termed sporadic), there are familial forms with two or more members of a family affected. Most often, when two or more relatives are affected, the members of the one family have the same type of adult onset primary torsion dystonia (for example both have cervical dystonia). However a significant proportion of affected familial individuals have different clinical manifestations (for example, an aunt with cervical dystonia and a nephew with focal hand dystonia).

## **Adult onset primary torsion dystonia as a genetic disorder**

There is growing evidence that most, if not all, adult onset primary torsion dystonia patients have a genetic disorder. However, the genetic effect is very poorly penetrant; the overwhelming majority of individuals who inherit the genetic tendency never develop the disorder. It appears from a number of studies that the gene or genes is/are inherited in an autosomal dominant manner: that is, both men and women can have the gene and that they can pass it on to one half of their offspring in a random manner [the chance of inheriting the gene is the same chance as flipping a coin (50%)].

### **Temporal discrimination**

If two lights flash at a very close interval between them, say 20 milliseconds, then the lights are perceived as being one single flash. As one increases the time interval between the flashing lights, then at about 30-40 milliseconds between the flashes, they are perceived as being two distinct light flashes. This change, from seeing one to two flashing lights, is called the temporal discrimination threshold. Most people can distinguish two flashes of light as being two (and not one), when there are slightly more than 30 milliseconds between the flashes. The same is true for tactile (touch) stimuli. Bradley and his co-workers have noted in people with adult onset primary torsion dystonia that there is an abnormality in the ability to discriminate between two stimuli (visual or tactile) happening in close succession in time. The temporal discrimination threshold is prolonged in most people with this form of dystonia. In people with adult onset primary torsion dystonia, the temporal discrimination threshold may be increased up to about 100

or 150 milliseconds. This has no practical significance in everyday life.

However if one tests unaffected first-degree relatives (siblings, offspring, parents) of people with adult onset primary torsion dystonia, then one finds that about 50% (one half) of these adults have an abnormal temporal discrimination threshold and the other half have a normal temporal discrimination threshold.

Bradley and his colleagues examined unaffected first-degree relatives of people who had evidence of familial adult onset primary torsion dystonia (three or more family members affected). They found that about half of the unaffected relatives had abnormal temporal discrimination thresholds. They postulate that an abnormal temporal discrimination threshold represents an endophenotype, that is evidence of gene carriage without any other clinical manifestation. In support of this they also found that unaffected first-degree relatives with abnormal temporal discrimination thresholds also had enlargement of the putamen, a finding also reported in people with adult onset primary torsion dystonia.

Subsequently the same research group has examined relatives of people who have adult onset primary torsion dystonia with no family history, which is the most common, (called sporadic) form of dystonia. They found also that 50% of their unaffected first-degree relatives had abnormal temporal discrimination thresholds.

## Implications of this research

The authors of this study hypothesise that by using the temporal discrimination threshold test one can identify individuals who have inherited the gene or genes for adult onset primary torsion dystonia. These individuals have an extremely low risk for developing the disorder clinically- probably less than 10% risk. There is no immediate practical benefit from this research. However, the usefulness of this work is that it will aid researchers looking for the genetic cause(s) of adult onset primary torsion dystonia. It may also be of use in identifying environmental risk factors for adult onset primary torsion dystonia. Why does only one individual in a family where half the individuals have the abnormal gene, develop the disorder? It is probable and likely that additional environmental factors are needed to develop the disorder. Lastly the identification of a disorder in temporal discrimination as a marker (endophenotype) and the associated enlargement of the putamen, suggests a disordered mechanism involving a nerve pathway through the

putamen as a cause of adult onset primary torsion dystonia in its many manifestations.

*The authors are grateful to Maria Hickey of Dystonia Ireland and all the people with dystonia and their relatives who have generously supported this research both financially and by giving of their time in taking part in these studies. This research was funded by Dystonia Ireland, a non-profit patient support charity, ([www.dystonia.ie](http://www.dystonia.ie)), and by the Health Research Board of Ireland, a state funding agency ([www.hrb.ie](http://www.hrb.ie)) (HRB Grant number H01341).*

*Michael Hutchinson*

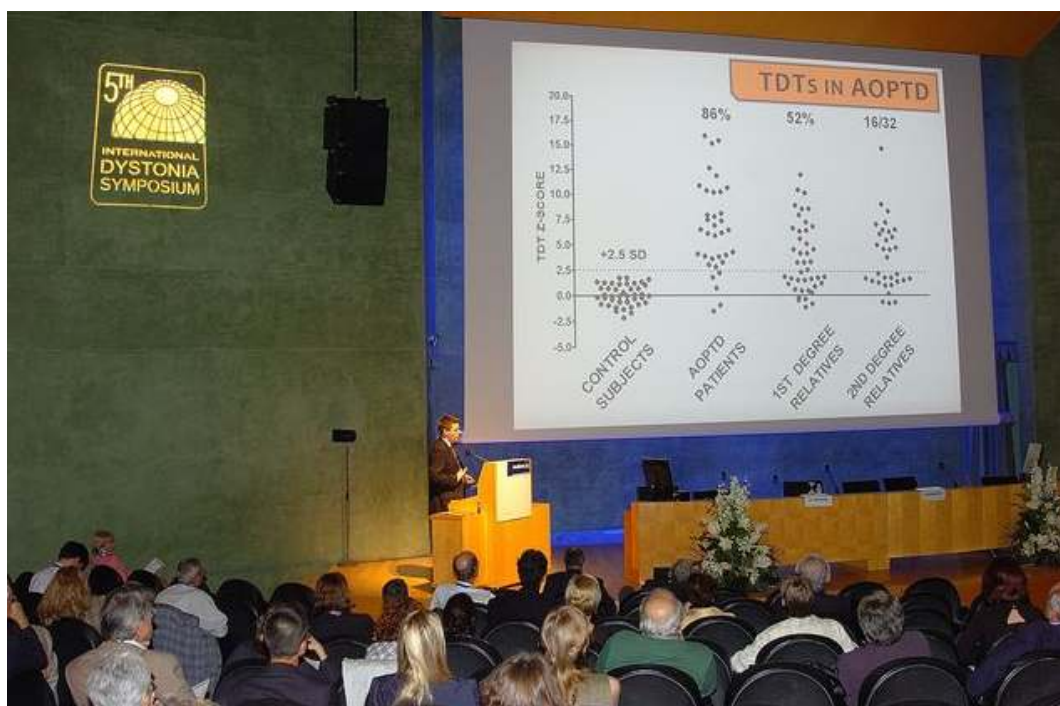
*Consultant Neurologist*

*Newman Clinical Research Professor*

*St Vincent's University Hospital*

*University College Dublin*

*December 2011.*



*David Bradley, winner of the David Marsden Award 2011*

## **NEW APPOINTMENTS TO OUR MEDICAL AND SCIENTIFIC ADVISORY BOARD**

EDF is very pleased to announce that the following distinguished dystonia specialists have accepted an invitation to join our Medical and Scientific Advisory Board:



*Prof Marie Vidailhet, Paris, France*



*Prof Marina de Koning-Tijssen, Groningen, The Netherlands*



*Dr Jean-Pierre Lin, London, UK*

We are grateful to these specialists for their willingness to support our work and we welcome them with great pleasure.

### **Medical and Scientific Advisory Board**

Prof Alberto Albanese  
Prof Alfredo Berardelli  
Prof Kailash Bhatia  
Prof Andreas Ceballos-Baumann  
Prof Marina de Koning-Tijssen  
Prof Joachim Krauss  
Dr Jean-Pierre Lin  
Prof Maja Relja  
Prof Eduardo Tolosa  
Prof Tom Warner  
Prof Marie Vidailhet

## **NEW MEMBER ORGANISATION POLISH DYSTONIA SOCIETY JOINS EDF**

At the Barcelona General Assembly, 2011, EDF was delighted to welcome the recently-formed Polish Dystonia Society into membership. Their representative, Dr Stanislaw Ochudlo attended the meeting and was asked to take back the warm greetings of the other EDF members to his colleagues in Poland.

We look forward to working with this new society and with other prospective EDF members in Eastern Europe.

**EDF is very grateful to the following organisations which have supported our core activities in 2011:**

### **Allergan**



Marlow, United Kingdom

### **Ipsen Pharmaceutical**



Paris, France

### **Medtronic Europe**



Tolochenaz, Switzerland

### **Medtronic Foundation**



Minneapolis, USA

### **Medborgarskolan**



Malmö, Sweden



## **EUROPEAN DYSTONIA FEDERATION**

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Prof Marina de Koning-Tijssen

Prof Maja Relja

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### **Editorial Committee**

**Editors:** Monika Benson and Alistair Newton

**Layout and graphic design:** Sølvi Engeland

### **Disclaimer**

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