

DYSTONIA EUROPE

NEWS

AUTUMN 2016

Dystonia Europe in the EU Parliament: Living with a Brain Disorder – Taking Control of your Life



Dystonia Europe Executive Director Monika Benson and Dr. Jean-Pierre Lin, Paediatric Neurologist, Evelina London Children's Hospital.
Read more on page 4.

Connecting people for dystonia

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Dystonia Europe
connecting people to
spread information
raise awareness &
promote research

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From Dystonia Europe to all of you,
a Happy Holiday Season and all the
best for the New Year!



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www.dystonia-europe.org

President's Message



Dear all,

We are happy to present you with another newsletter with information on activities and happenings around Europe and beyond.

It is soon the end of 2016 and we look forward to 2017 to continue our work to improve the quality of lives for people living with Dystonia.

We are now in the process of planning the next D-DAYS conference, which will be held in Rome 13 & 14 May 2017. More information on venue and programme will be available on our website beginning of 2017.

As Dystonia Europe is a member of EFNA (European Federation for Neurological Association) and now also a member of EPF (European Patient Forum), we continue to participate in workshops and communicate with other patient organisations that also represent brain disorders. Dystonia Europe participated in a workshop in Dublin this autumn. There are always new things to learn on how to move forward, to do the best possible job for those living with dystonia or have a loved one who is affected. You can read more about it in this newsletter.

MyDystonia is an APP owned by Dystonia Europe. It is a valuable tool for many dystonia patients and can facilitate the communication between you and your healthcare specialist. The APP is available online at www.mydystonia.com and for smartphones on both Android and IOS. For more information read about the APP in this newsletter. I also want to take this opportunity and thank the MyDystonia Ambassadors for engaging and taking on the task to support MyDystonia users. As there are more users every day it will be very helpful for them to get support in their national language.

If you have access to internet you can check out our website www.dystonia-europe.org and our different Facebook pages Dystonia Europe, Jump for Dystonia and MyDystonia. By liking our pages you get regular updates on our activities and the latest news. Facebook offers an excellent platform to spread awareness of dystonia through initiatives such as the EFNA "Undertheumbrella" campaign which everybody is invited to take part in. We still have a long way to go to spread information about dystonia to healthcare professionals and the public. There are often misunderstandings about movement disorders and if more people know about dystonia there would be less stigma.

Finally, I want to express our sincere appreciation and to thank all of you who help people affected by dystonia. Whether you help in a patient organisation, work in healthcare, as a researcher or in any other way assist with the work to improve the lives of dystonia patients, your work is very valuable and appreciated.

Wishing you all a Happy Holiday Season and all the best for the New Year!

Merete Avery
President

Anyone for tennis? Living with a neurological brain dis-

As a political lobbyist working in Brussels I have attended many meetings in the European Parliament. But I have never seen 50 people seated with tennis balls clasped between their jawbone and their shoulder. This was the way Monika Benson invited the audience to 'walk in her shoes.' It was a striking way to introduce dystonia, the little known brain disorder, to the Health working group in the European Parliament.

Monika went on to describe the symptoms, the numbers of persons affected in Europe and explained there was no known cure for dystonia although treatments are available. She had been invited to speak as new technologies such as mobile health apps are high on the agenda of the European Union. She demonstrated the MyDystonia app with the help of a short animation www.mydystonia.com. By providing an accurate picture of a patients' symptoms and their impact on daily life MyDystonia assists patient/physician communication and helps make efficient use of the limited consultation times. Monika described how it can also help improve therapy outcome, well-being and help overcome other symptoms such as depression. MyDystonia has the potential to develop into a central hub for dystonia and provide a possible data base as it is only when we understand the prevalence that health systems can provide the resources to treat dystonia.



Consultant Paediatric Neurologist Dr Jean-Pierre Lin gave a moving description of generalised dystonia that affects children for a lifetime, he believes that mobile health apps can assist a child transition into adulthood and help put the patient in control.

Other speakers included Frédéric Destrebecq, from the European Brain Council who gave an overview of brain disorders. He said the human brain is the most complex living structure known in the universe, but is also the origin of many chronic and disabling diseases that have a huge impact on the lives of people affected by them, as well as the healthcare systems that support them. According to the World Health Organisation brain disorders account for 35% of the burden of all diseases in Europe and are predicted to become the major medical need of the 21st century.



Latest News

Nicola Bedlington, from the European Patients' Forum echoed Monika's call for more patient involvement in new technologies such as mobile health apps and said we must ensure that mhealth helps to reduce health inequities and contributes to patient empowerment.

Dr Terje Peetso, from the European Commission and several members of the European Parliament (MEPs) were on the platform.

Linda McAvan MEP spoke from personal experience of the huge impact of brain disorders as her

family has been impacted by three different neurodegenerative diseases.

She paid tribute to patient organisations who help give practical and moral support to families so they do not feel so alone.

It was a great privilege to work with Dystonia Europe developing this meeting. The presentations will soon be available on the EU website.

Heather Clarke, EU Affairs Consultant



Heather Clarke



EFNA Neurology Advocacy Awards 2016

These awards recognise the contribution of an individual or group to the development and promotion of advocacy for people with neurological disorders in Europe. This year's winners were presented with their awards at a Gala Dinner in Dublin on October 13th.



All winners, from left: Audrey Craven, Alistair Newton, Marian Harkin and Paola Giunti

The winning European Policy-Maker for 2016 was **Marian Harkin MEP**.

She was nominated for her award, jointly, by the International Bureau for Epilepsy and Pain Alliance Europe. Marian is currently a co-chair of the MEP Interest Group on Brain, Mind and Pain – but has been long-term champion of health issues in the European Parliament.

She was presented with her award by last year's winner – Gay Mitchell, former MEP.

The second award of the evening was for European Health Professional, and went to **Professor Paola Giunti**.

An ataxia specialist, Prof. Giunti was nominated for her work in the field – including the establishment of a specialist ataxia centre – a bespoke health service made in partnership with patients. Speaking on the evening, Cathalijne van Doorne of Euro-Ataxia also commended Prof. Giunti for her voluntary work with patient organisations and the wider patient community.

She was presented with her prize on the evening by Prof. David Vodusek, member of the judging panel – representing the European Academy of Neurology.

The final award of the evening was presented to **Alistair Newton** – the winning European Patient Advocate.

Not only was Alistair involved in the foundation of Dystonia Europe but also in organisations such as EFNA and the European Brain Council.

Explaining why he was put forward for the award, Monika Benson of Dystonia Europe said: *"We thank Alistair for his dedicated work and all his efforts over the last 30 years, a true patient advocate whose mission has stimulated fruitful collaboration and created many successful partnerships to benefit patients within neurology, especially dystonia."*



Dystonia Europe: Monika Benson, Maja Relja, Alistair Newton and Merete Avery

Audrey Craven, Past President of the European Federation of Neurological Associations [EFNA] was presented with a Lifetime Achievement Award; acknowledging her long-time commitment to the cause. This was presented by Eveline Sipido of the European Academy of Neurology last year's winner, who said:

"In all these years and in spite of the condition she lives with, Audrey never lost the good spirits nor gave up so I think that with Oscar Wilde's help I can perfectly summarise what she did: The world is divided into two classes, those who believe the incredible, and those who do the improbable."

DYSTONIA
EUROPE

Ipsen committed
to **patient** care
over the long term
supporting

The David Marsden Award 2017

10 000€

to stimulate research on dystonia

The award, introduced by Dystonia Europe in 2003 is presented every two years to stimulate developing knowledge of and interest in dystonia through publications on aetiology, pathogenesis, diagnosis or therapies in dystonia or on the psycho social effects.

The deadline for submissions is 31 January 2017

All information regarding the 2017 David Marsden Award are available at www.dystonia-europe.org/activities/awards/

www.dystonia-europe.org

European Network on the Study of Dystonia Syndromes-COST action BM1101

The European Network on the Study of Dystonia Syndromes (RN) is a coordinated effort of European Scientists specializing in movement disorders, in particular dystonias, funded, in part, under the COST action BM1101. This scientific network was created with the support of a four-year grant awarded in 2011 by COST (Co-operation in Science and Technology in Europe – www.cost.eu), a European intergovernmental organization which pre-dates the European Union. The network has functioned as a ‘**COST Action**’ during the period of COST grant (November 2011-November 2015). COST grant does not provide funds for research projects – only support to established greater international collaboration in research. Thus, the project was intended to create better networking among dystonia specialists around Europe, and to raise awareness of dystonia, covering the expenses of management and collaborative research meetings, training courses and Short-term Scientific Missions (STSMs). In addition, the goal of this effort was to better understand the causes of dystonia, to improve diagnosis and treatment as well as care for patients.

Formal **Management Committee** - 40 dystonia specialist, (clinicians and researchers) represented the **24 countries** (23 across Europe, plus Israel) whose national governments are affiliated to COST. RN **Chair** (prof A. Albanese, Italy), **Vice Chair** (prof M. Vidailhet, France) and members of its **Steering Committee** (K Bhatia, J Ferreira, T. Gasser, R. Goodchild, A Pisani, M Relja, M de Koning-Tijssen, Y Yebenes) were all elected at a first meeting of the RN’s Management Committee on 14th November 2011 in Brussels.

At the same meeting Dystonia Europe (DE) was elected Grant Holder of the Network (Alistair Newton). The members of the Steering Committee were appointed to the DE medical and scientific advisory board (MSAB) (six of the scientists were already members of MSAB).

To facilitate work and functioning of RN, 4 Workgroups (WG) were formed, all chaired by members of the Steering Committee.

WG1. *Genetic studies of DS* (K. Bhatia, T. Gasser) ; **WG2.** *Animal models of DS* (R. Goodchild, A. Pisani) **WG3.** *Standardisation and harmonization of clinical practice* (M. de Koning-Tijssen, J. Ferreira); **WG4.** *E-infrastructure for European cooperation on DS* (M. Relja, Y. Yebenes)



ORIGINAL ARTICLE

Management of dystonia in Europe: a survey of the European network for the study of the dystonia syndromes

A. Valadas^{1,2,*}, M-F. Contarino^{3,4,*}, A. Albanese⁵, K. P. Bhatia⁶, C. Falup-Pecurariu⁷, L. Forsgren⁸, A. Friedman⁹, N. Giladi¹⁰, M. Hutchinson^{11,12}, V. S. Kostic¹³, J. K. Krauss¹⁴, A. Lokkegaard¹⁵, M. J. Marti¹⁶, I. Milanov¹⁷, Z. Pirtosek¹⁸, M. Relja¹⁹, M. Skorvanek^{20,21}, M. Stamelou^{22,23}, A. Stepiens²⁴, G. Tamás²⁵, A. Taravari²⁶, C. Tzoulis^{27,28}, W. Vandenberghe²⁹, M. Vidailhet^{30,31}, J. J. Ferreira³² and M. A. Tijssen³³

Results are presented in *Table 1*, showing the expertise in movement disorder and dystonia available in participating EU countries. In conclusion of this work it is evident that *'Internationally, collaboration in training, advanced diagnosis, treatment and research of DS and, locally, in each country the creation of multidisciplinary teams for the management of dystonia patients could provide basis for improving all aspects of dystonia management across Europe'*.

Table 1 Expertise in movement disorders and dystonia per country

	BE	BG	DE	DK	EL	ES	FR	HR	HU	IE	IL	IT	LV	MK	NL	NO	PL	PT	RO	RS	SE	SI	SK	UK
Movement disorders																								
Society	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Experts	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Stages for residents in neurology	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Teaching courses for residents/GNs	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Teaching courses for GPs	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Specific training for nurses	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Specific training for speech therapists	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Specific training for physiotherapists	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Clinical trials	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Dystonia																								
Networking group	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Experts	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Tertiary centers	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Teaching courses for residents/GNs	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Teaching courses for GPs	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Specific training for nurses	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Specific training for speech therapists	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Specific training for physiotherapists	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Clinical trials	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
National patients association	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+

BE, Belgium; BG, Bulgaria; DE, Germany; DK, Denmark; EL, Greece; ES, Spain; FR, France; HR, Croatia; HU, Hungary; IE, Ireland; IL, Israel; IT, Italy; LV, Latvia; MK, Republic of Macedonia; NL, Netherlands; NO, Norway; PL, Poland; PT, Portugal; RO, Romania; RS, Serbia; SE, Sweden; SI, Slovenia; SK, Slovakia; UK, United Kingdom.

GPs, general practitioners; GNs, general neurologists; +, yes; -, no.



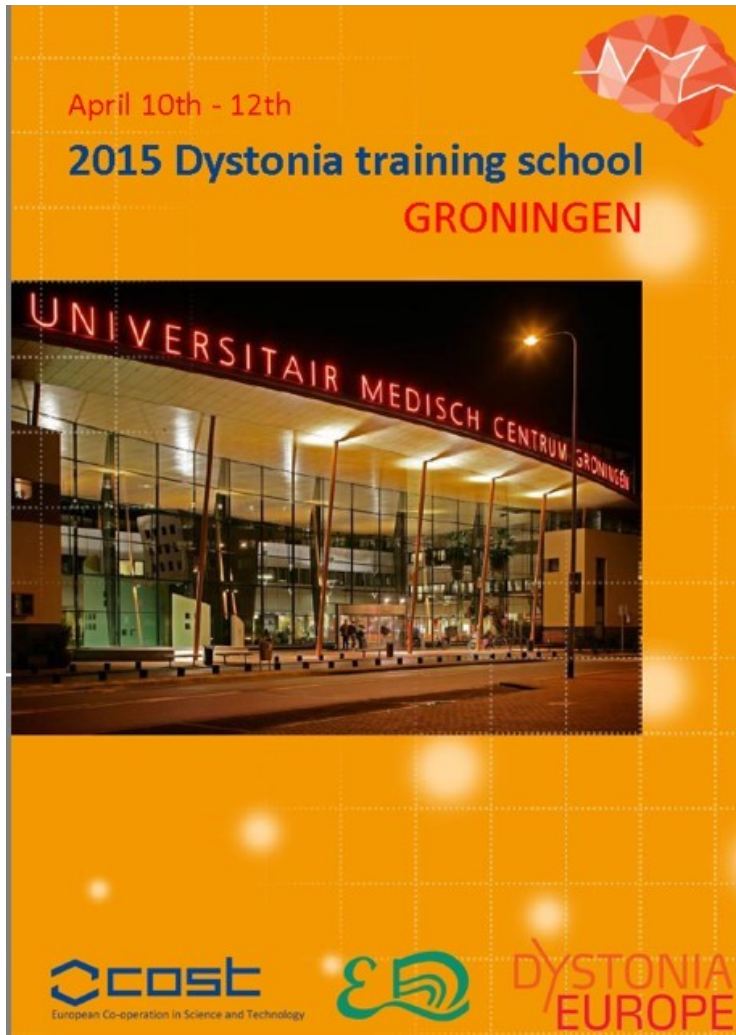
*Prof. Maja Relja,
Prof. Kailash Bhatia,
and
Prof. Marina de
Koning-Tijssen*

RN has organized **3 Training Schools**. First TS was organized by prof Maja Relja in Croatia (Bol, 20-23 September 2012). The 2nd TS was organized by prof Kailash Bhatia in UK (London 22-24 November 2013), and the 3rd TS was organized by prof Marina de Koning-Tijssen in NE (Groningen 10-12 April 2015) A total of 120 young trainees under the age of 35 attended the three training schools.

In addition **23 STSMs** were granted to young scientists from Italy, Germany, Croatia, Romania, Hungary, Israel, Czech Republic, Poland and UK, who spent 1-2 months in the centers where they carried out their projects (The Netherlands, France, Belgium, Italy, Portugal, UK and Spain).

Closing conference was held in Rozzano near Milan on 16-17 October 2015 organized by prof Alberto Albanese, Chair of the European Network on the Study of Dystonia Syndromes.

As a joint effort of all RN members a survey was conducted in European countries via the management committee to document managing strategies, facilities and expertise available in various EU countries in order to identify which measures should be implemented to improve the management of DS in EU. Original article was published in *European Journal Neurology* in January 2016.



Second review article was *Submitted* for publication in *Frontiers in Neurology*, section Movement Disorders, *Oct 2016*.

Title: **Clinical practice: evidence-based recommendations for the treatment of cervical dystonia with botulinum toxin**

Authors are: Maria Fiorella Contarino, Joost Van Den Dool, Yacov Balash, Kailash Bhatia, Nir Giladi, Johannes Koelman, Annemette Lokkegaard, Maria Jose Marti, Miranda Postma, Maja Relja, Matej Skorvanek, Johannes Speelman, Evelien Zoons, Joaquim Ferreira, Marie Vidailhat, Alberto Albanese, Marina de Koning-Tijssen.

In this review article evidence-based recommendation for the treatment of the most frequent type of focal dystonia: cervical dystonia was analysed by RN's dystonia experts in order to improve treatment of DS. Final publication is expected as summary of RN closing conference in Milan.

As a result of the work of the Research Network during the 4-year period we could say that closer collaboration in dystonia research and management in EU countries was achieved. Very successful Training Schools and STSMs have encouraged many younger scientists to focus on dystonia. Very good platform for further improvement of DS management in EU is achieved.

Prof. Maja Relja, Vice President Dystonia Europe

Young scientists in dystonia research

By Lisa Rauschenberger and Susanne Knorr

Department of Neurology, University Hospital of Würzburg, Germany



Our first contact with Dystonia Europe was at this year's **International Congress on Treatment of Dystonia** in Hannover and at the International Congress of Parkinson's Disease and Movement Disorders in Berlin. Besides meeting Merete Avery and Monika Benson, we participated at the project "Jump for Dystonia" in order to raise awareness for the disease. This was not the first time we were confronted with the fact that dystonia encompasses a rare and heterogeneous group of disorders not many people are aware of. Even doctors and biologists often have limited knowledge about this disease. At the same

time, cause and mechanisms behind the development of dystonia are still not entirely understood. It is being assumed that the nature of the disease is a dysfunction involving multiple brain networks. In most cases, the disorder leads to a strong disability with severe diminishment of the quality of life.

Despite constant medical advances, therapeutic possibilities for dystonia patients are still limited. So far botulinum toxin and deep brain stimulation are the treatment options with most benefit. However, considering the wide variety of disorders encompassed by the term dystonia there is still a need for novel therapeutic options as well as improvement of the existing methods. The dystonia research community is rather small in comparison to other neurological research fields such as Parkinson's disease and Alzheimer's disease. As the working group for experimental research in movement disorders, with PD Dr. Ip as head of laboratory, in the neurological department of Professor Volkmann in Würzburg, we work on two different animal models for dystonia. Our focus lies on animal models for dystonia 1 and dystonia 12, both leading to generalized dystonic symptoms with however different genetic mutations as the underlying cause. Dystonia 1 is the most common form of hereditary dystonia with onset in early childhood. Dystonia 12 is characterized by an abrupt onset of symptoms in young adulthood, triggered in most cases by a stressful event. Under the assumption that multiple areas of the brain are involved, our aim is to analyze possible changes taking place on a variety of levels. We perform different forms of gait analyses; search for metabolic differences on protein and RNA level as well as structural changes in the brain. For this purpose, we have established a multitude of analytical methods in order to examine the disorder from all sides.

Research

As a biologist and a medical doctor, we have different strengths and are able to assist each other. With the help of these animal models, we hope to get a better understanding of the nature of the disease. With a better knowledge of the mechanisms behind the disorder we aim to develop new therapeutic approaches.

The development of animal models and basic research is a time-consuming process, demanding perseverance and commitment. Very often, one can merely expect to make small steps forward. However, thanks to these projects, our interest in this rare group of diseases has been awakened and the limited knowledge of the mechanisms

behind dystonia has given us additional motivation to advance this area of research. Especially in experimental research it is important not to lose sight of those affected by the disease. Through Dystonia Europe, we got the possibility to talk to patients whose personal experiences showed us once more the necessity for basic research, but also the importance of raising awareness. Just like Dystonia Europe, we hope to be able to spread knowledge about the disease among friends, family and colleagues and to motivate them to “Jump for Dystonia”.



*Lisa Rauschenberger
and
Susanne Knorr,
Jumping for
Dystonia*

A Non-Profit Approach to Accelerate Therapy Development for the 95% of Rare Diseases With No Therapy



*Barbara Handelin,
CEO BioPontis*

The facts and figures about so-called rare diseases illuminate how common and globally devastating this group of disorders is. When viewed collectively, these 7,000+ rare diseases affect some 350 million globally- that is more than all the AIDS and cancer patients combined. Yet today, less than 5% have any treatment available. Since 80% of rare diseases are of genetic origin, most occur in children and represent a major reason for admission to children's hospitals.

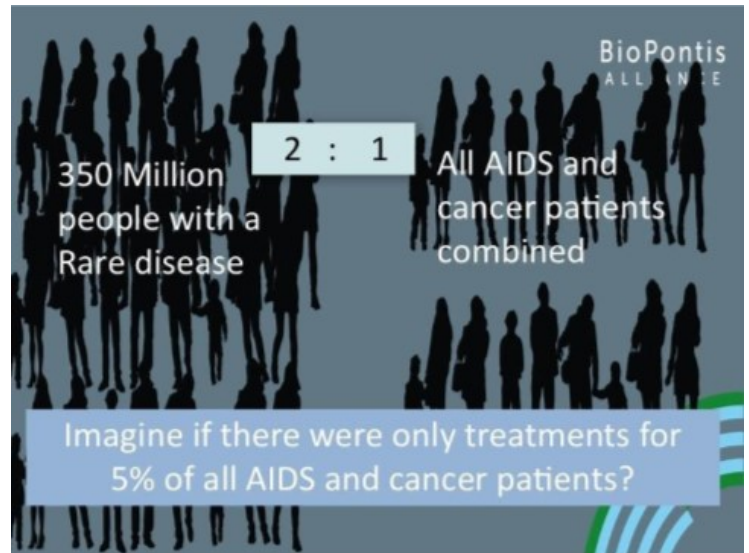
In the US, with the Orphan Drug Product Act's regulatory incentives, 556 programs are in the clinic or under agency review by the FDA as of 2016 (Medicines in Development Report (PhRMA), the majority of these being for rare cancers and genetic diseases. These statistics are startling considering that publicly funded biomedical research has identified the genetic cause of 4,000 of those 7,000 rare diseases) and the drug development technology to potentially devise effective treatment is widely available. (NIH Rare Disease Day speech, Francis Collins, 2016).

But academia, where fundamental discoveries are made, is not where product development expertise lives. The rare disease patient organizations that may fund basic research also lack expertise and/or sufficient funding to carry forward into medicines development. The exceptional successes seen in Cystic Fibrosis and Muscular Dystrophy communities are laudable, but cannot be replicated across 7,000 diseases without duplicating expertise and the massive investments made for those two diseases.

Developing any drug is extremely difficult, expensive and rife with early stage failures. The pharmaceutical industry's model requires them to allocate their research dollars to later stages with lower risks (e.g., at clinical trial stage).

A new solution must open the flow of treatments to patients with many more rare diseases. BioPontis Alliance for Rare Diseases has been established to deliver drug product candidates that have been developed to industry standards and that promise to halt or reverse the target disease, not just treat symptoms.

The BioPontis Alliance innovative model overcomes the limitations of patient organizations, academic researchers and biopharmaceutical companies to deliver on this solution alone. Our strategy and collaborative model directly addresses the fundamental problems preventing basic research from being converted into new therapies:



Professionally led and executed drug discovery

programs: We identify promising rare disease discovery science programs using strict selection criteria, and utilize philanthropic and public funds to generate lead compounds and develop these into therapy candidates up to the point they are ready for first-in-human clinical trials. Drug discovery and pre-clinical development is planned for and managed by BioPontis scientists, together with the academic group behind the science and selected contract research companies.

Patients integral to therapeutic objectives: Unlike other pharmaceutical patient-oriented practices, often limited to help in study recruitment and clinical protocol design, we bring patient input in the very beginning, at the drug discovery phase. Information on disease progression, symptoms, quality of life and other parameters are fed directly into the vision for the new therapy to be developed.

In this way, therapeutic treatment objectives are aligned between drug developers and patients and clinical endpoint selection and interpretation issues are avoided at the later clinical stages. We have conducted two patient integration workshops to produce draft methods to involve patients in BioPontis development pathways – with participation from 17 patients' organizations from the USA and Europe.

Using the right funding for each stage of de-

risking product development: We apply philanthropic funding at the earliest, highest risk steps (high-throughput screening for drug candidates, or lead candidate optimization) and will continue to commit such funds all the way through preclinical development, if warranted by technical success. We also have the option to invite private sector investment in later, less risky stages, through IND filing.

Research

- a) the general lack of pharmaceutical and regulatory expertise in academia and patient organizations;
- b) the risk tolerance limits on profit-based commercial developers;
- c) the need to create a professional-product development team from 'scratch' by a patient organization; and
- d) the lack of patient integration into the design/development of their own therapies.



Finally, we offer patient organizations and academic institutions equitable ownership and economic returns back from all product licensing that BioPontis negotiates with each company that acquires one of our therapeutic product candidates. This economic model is designed to allow BioPontis to become self-sustaining and to expand to work on many rare diseases over the next 50 years.

In 2016 we initiated our first preclinical development program. The program is aimed at developing a treatment for one type of Charcot Marie Tooth disease (CMT), a rare, progressive and crippling neuropathy. There are no disease-modifying therapies available for CMT patients today, although it affects 1/2,500 worldwide. The project will advance the work of Dr. Albena Jordanova at the University of Antwerp and in collaboration with VIB. The partnership blends the basic research and early-stage translation resources at VIB with the professional therapeutics development expertise in the rare disease field at BioPontis Alliance. By joining forces, both partners can step in earlier than most for-profit developers could justify.

BioPontis Alliance for Rare Diseases is a 501(c)(3) Public Charity in North Carolina (USA) and a Foundation of Public Utility in Brussels (Belgium). Managed jointly by an international Board and executive team, BioPontis partners with patients' organizations and academic researchers to bridge the gap from promising science to medicines, ready for uptake by the biopharmaceutical industry. BioPontis Alliance for Rare Diseases is reaching out to all stakeholders in the biopharmaceutical industry, to institutions, charitable funds and foundations, and to individuals who want to directly impact the healthcare outcomes for rare disease patients who live with no hope of treatment today. Contributions to fund and support our programs and mission can be made at <http://biopontisalliance.org>.

Richard Basile, Barbara Handelin and David Spencer

BioPontis Alliance for Rare Diseases

The following articles, page 17–20, make reference to certain companies and their products and any opinions expressed should not be taken to be the views of Dystonia Europe or its members.

The articles are written by respected authors and are provided for the information of Dystonia Europe News readers.

Medtronic Receives CE Mark for SURETUNE2™ Software for Deep Brain Stimulation Therapy

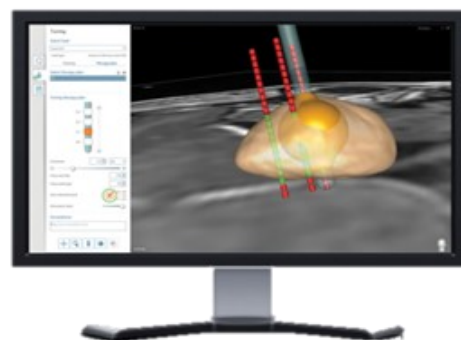
New Visual-Based Platform Helps Physicians Make Informed Programming Decisions by Providing an Intuitive Visualization of Patient-Specific Images and Data

Medtronic announced it has received CE Mark for SureTune2™ software, **which provides patient specific visualization to help physicians make decisions on how to program – or tune – their patient’s deep brain stimulation (DBS) therapy.**

DBS therapy applies mild electrical stimulation to precise targets in the brain in order to modulate specific symptom control. The brain targets are stimulated through lead(s) inserted into the brain and connected to an implantable neurostimulator through extensions running under the skin. A medical professional uses an external programmer to set and adjust stimulation settings.

SureTune2 software is designed for Medtronic DBS therapy and other DBS therapy delivery systems **to help physicians more efficiently select the optimal stimulation settings** on their programmer by visualizing patient-specific information in one comprehensive view including anatomy, physiology, and calculated stimulation field. Users can segment structures using a greyscale threshold within a region of interest, or by outlining shapes of interest from a patient image.

"SureTune will have an important impact on the care of patients with deep brain stimulation because it allows me for the first time to visualize activation patterns of DBS within the individual segmented patient anatomy," said Professor Jens Volkmann, MD, PhD, FEAN, chairman and professor of neurology in the University Clinic of Würzburg. "It simplifies the trial and error process associated with DBS programming by helping me identify the best contacts, which saves me time."



"Medtronic is committed to providing advanced technology to the multidisciplinary teams who are helping DBS patients, and I'm convinced that SureTune will provide them with easy-to-use tools to aid in optimizing therapy outcomes," said Lothar Krinke, PhD, vice president and general manager of the Brain Modulation business in Medtronic's Restorative Therapies Group. "SureTune is a key aspect of Medtronic's commitment to providing integrated solutions **for improving accuracy and confidence from surgery to post-operative DBS patient management.**" Medtronic helped pioneer DBS, and more than 140,000 patients worldwide have received Medtronic DBS Therapy TODAY.

Any forward-looking statements are subject to risks and uncertainties such as those described in Medtronic's periodic reports on file with the Securities and Exchange Commission. Actual results may differ materially from anticipated results.

Medtronic DBS Therapy is not for everyone. Not everyone will receive the same results. Please consult your doctor for further information.

Brief Statement

See the device manual for detailed information regarding the implant procedure, indications, contraindications, warnings, precautions, and potential adverse events.

Paroxysmal Dyskinesias May be Sign of Glut1 Deficiency Syndrome

Genetic Testing May Detect Rare but Underdiagnosed Genetic Disorder

Investigators have discovered that certain dystonia-causing genes are associated with other diseases. In these cases, dystonia may be one possible manifestation of a more complex syndrome.

Paroxysmal dyskinesias, which often include episodes of dystonia, provide one example. Several genes for paroxysmal dyskinesias have been identified: DYT8, DYT9, DYT10, and DYT18. As it turns out, investigators ultimately determined that DYT9 and DYT18 are the same. Commonly referred to as SLC2A1/Glut1, this gene is associated with Glucose Transporter Type 1 deficiency syndrome (Glut1 DS), a rare genetic disorder that can present with a spectrum of signs and symptoms.

Glut1 DS may include paroxysmal dyskinesias (DYT9/18), seizures, and additional findings such as intermittent ataxia, choreoathetosis, dystonia, and alternating hemiplegia.

Melanie Brandabur, MD is a movement disorders specialist at University of California San Francisco and Medical Director at Ultragenyx Pharmaceutical Inc., a company focused on the development of therapies for rare diseases. “There are a lot of different movement disorders that can be symptoms of Glut1 deficiency syndrome,” she explains. “The dystonia that is associated with Glut1 DS presents a bit differently than some of the other paroxysmal dyskinesias. Symptoms are often evoked by exercise or stress and are generally not responsive to the drugs that other paroxysmal dyskinesias are. Some patients will have other types of movement disorders during an episode. They may have dystonic movements, choreic movements, tremor, or even movements that look like parkinsonism.”

Glut1 DS is caused by a genetic defect in how sugar reaches the brain. Sugar molecules provide the primary source of energy for the brain and Glut1 DS deprives the brain of the fuel it requires to function properly. There are currently no treatments approved by the Food & Drug Administration (FDA) specifically to treat Glut1 DS. Special diets, such as the ketogenic diet (high fat, low carbohydrate and protein), may be prescribed for some patients. Studies suggest 3,000 to 7,000 people are affected by Glut1 DS in the United States.

A genetic test for Glut1 DS may be part of the work-up for children with intractable epilepsy, so these children may get a timely diagnosis. “It’s more complicated when the patient presents at an older age just with movement disorders, and they don’t have a history of seizures,” explains Dr. Brandabur. “These are the patients I worry about being underdiagnosed because they didn’t have the typical epilepsy that would have brought them to the attention of a clinician experienced with diagnosing Glut1 DS.”

Glut1 DS is also sometimes diagnosed in the parent of a child being evaluated for intractable seizures or other symptoms of Glut1 DS. “Sometimes the symptoms are mild enough that it doesn’t interfere with life too much, but for other people the symptoms may be really profound. Glut1 DS is quite a spectrum and I don’t think we have a complete understanding yet of the older Glut1 DS population,” Dr. Brandabur continues. Genetic testing for Glut1 DS may help identify patients. “People who have movement symptoms that come and go, and that are provoked by things like fasting, stress, exercise, fever, fatigue, changes in activity level—these are the people who should talk to their doctor about getting tested,” says Dr. Brandabur.

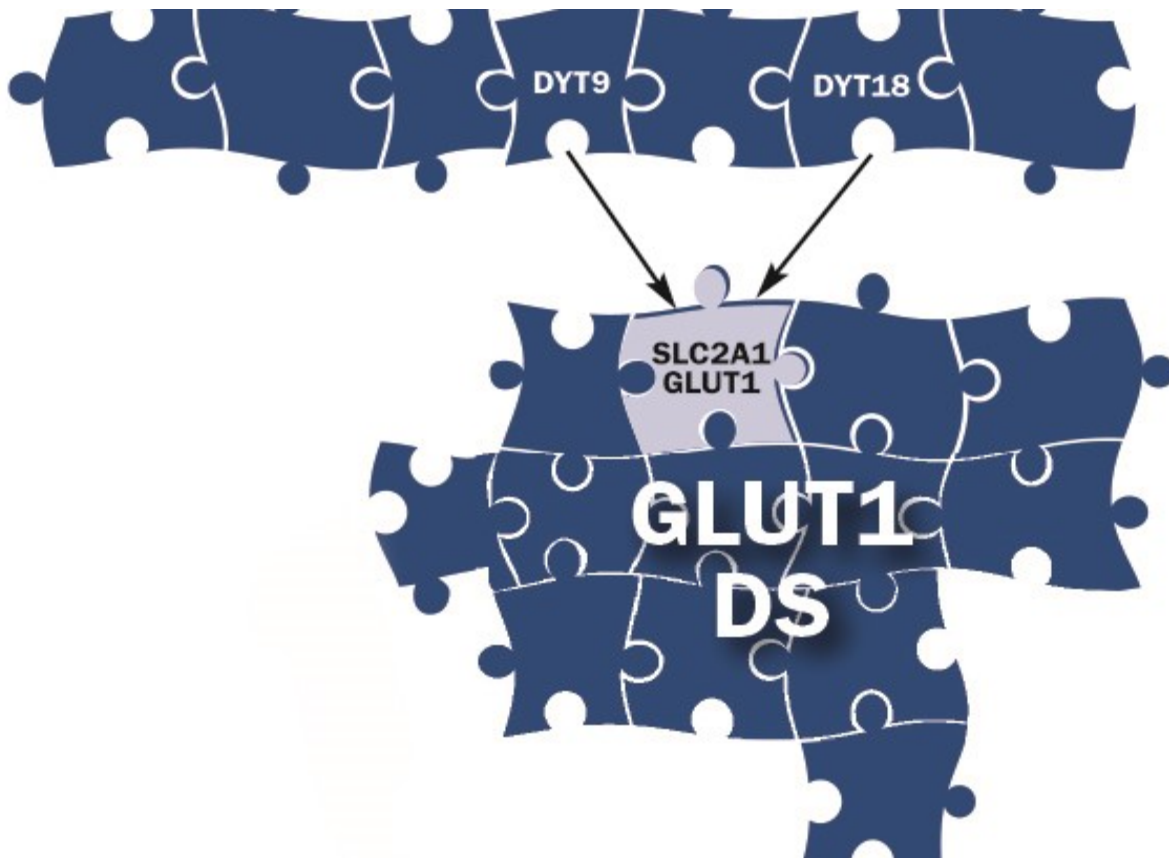
Research

Ultragenyx is developing an experimental treatment for Glut1 DS. A randomized double-blind placebo controlled study testing UX007 (triheptanoin) in Glut1 DS patients with movement disorder symptoms will begin recruiting in the near future. For more information, visit Glut1study.com or contact Kim Mooney, Associate Director of Patient Advocacy, at 415-483-8872 or kmooney@ultragenyx.com.

PAROXYSMAL DYSKINESIAS are a group of movement disorders characterized by episodes of involuntary movements.

The movement disorder symptoms disappear in between the episodes.

Acknowledgement: This article came from the Dialogue/Dystonia Medical Research Foundation, US.



PUTTING THE PIECES TOGETHER

Researchers discovered that the *DYT9* and *DYT18* genes responsible for paroxysmal dyskinesias are the same gene. Known as *SLC2A1/GLUT1*, this gene is associated with Glut1 DS, a rare genetic disorder.

Deep Brain Stimulation – Industry Perspective



Three questions for
Vincent Sourdain
*Director Europe,
Neuromodulation
Division at Boston
Scientific*

Tell us a little about your work in dystonia.

As a company, we are still young in the field of neuromodulation. However, we are moving fast and are innovating quickly. In 2012, we launched our first Vercise™ Deep Brain Stimulation (DBS) System. It was followed by seven new devices and solutions – in only three years. For me, it is very exciting to work in neuromodulation, since it is a field of constant change and we're only beginning to understand the human brain. In dystonia, we are particularly proud of our partnerships with trusted physicians and patients. They help us take the next step in developing better technologies for improving the lives of people living with dystonia. Seeing the results of our work and what we can achieve together every day is incredible – I couldn't think of a better job.

What are you currently working on?

We are continuously evolving our devices, technologies and services in Deep Brain Stimulation. Using DBS systems in dystonia is showing impressive results for patients, bringing the symptoms down to a level where many people can lead a normal life again.

Challenging the status quo is in our DNA and at the core of what we do. We recently introduced a directional system which allows very targeted and personalized therapy for each person, thereby offering very fine control of the stimulation of select brain areas and reducing potential side effects as much as possible. This is a significant improvement in the way we are able to help people living with dystonia.

What's in the future for treating dystonia and chronic motor diseases?

With DBS, we've already come a long way to help improve the lives of people living with dystonia, but there is so much more to do. We will continue to take risks, try out new things, investigate in further areas and indications with the best possible



outcomes for the patients in mind. To us, the future is happening now, and that's why we continue to chart new paths, closely working with the patient and physician community to help us understand what's worth exploring.

Cycling across the Alps with Parkinson's disease

Read how Tony Seidl from Germany set things in motion and became a role model for many of us

On average about 48 kilometers – that is the distance that Tony Seidl cycled every day during his eleven day tour from Altötting, Germany to Padua, Italy. For most people this would be a real challenge. But the 49-year old patient from Altötting, Upper Bavaria, traded the challenge for an adventure: it marked the beginning of a second life after his Deep Brain Stimulation (DBS) surgery.

His motivation: exchanging experiences with other patients

Why did Tony cycle 517 kilometers? Because of a promise he made to himself: If his Deep Brain Stimulation (DBS) surgery went well, Tony swore that he would visit the grave of his namesake Saint Anthony of Padua by bike so he can meet other patients along the way. “When I recall the time prior to my surgery, I really missed learning from other patients who had already undergone DBS and could have shared their experience with me. Unfortunately, networking among DBS patients is still at an early stage.” Just in time for his 50th birthday and exactly two years after the life changing surgery, Tony kicked off his tour in Altötting using a recumbent bike which allows him to maintain better balance.

Red hair as sign for courage

As a sign of his new courage to face life Tony dyed his hair red. “The red rose stands for love and the red tulip is also a sign of the Parkinson's disease movement. Every smile that I receive for my red hair gives me strength and I need friendly faces in my daily life”, Tony said.

“Tony is such an inspiration to all of us, and his story is the reason why we work in Deep Brain Stimulation”, said Vincent Sourdain, Director Neuromodulation Europe at Boston Scientific. “What sounds like a fairytale – from being in a wheelchair to cycling over the Alps – has actually become a reality for Tony. Our teams couldn't be happier and more motivated to carry on inventing, innovating and shaping the future of what is possible with DBS.”

During his tour, Tony met with people with Parkinson's and their associations and exchanged personal experiences and challenges with regard to the disease and its therapy. Being welcomed and having his cause supported by so many people motivated Tony every day to carry on with his tour to Padua.



Preparing and training for the tour, and also finally being on the road, gave Tony not just physical, but also a lot of emotional strength. Still overwhelmed by his feelings when thinking back to the tour earlier this year, Tony feels there is little he cannot achieve. And he smiles when he tells us that he is already planning his next adventure. Stay tuned!



Tony Seidl on his bicycle

Dystonia and Parkinsons Patients in the Limelight at MDS Congress in Berlin

The 20th International Congress of Parkinson's Disease and Movement Disorders took place in Berlin last June where over 5000 specialists had come to attend the 4-day meeting. Both Dystonia Europe and EPDA (European Parkinson's Disease Association) had centrally located booths in the exhibition hall which was made possible by a donation from Medtronic. By having a booth in the centre Dystonia Europe:

- was better branded and more visible.
- had a welcoming look and feel (chairs and tables) and many visitors took time to sit down and discuss dystonia patient association development, research opportunities, and other projects such as MyDystonia - the digital patient diary, and our awareness campaign "Jump for Dystonia".
- had a tremendous increase in visitors from both old and new connections.



Dystonia Europe's booth at the MDS

But the focus on patients' needs was not just limited to the exhibition hall. In the innovative Medtronic symposium "Who wants to be a DBS-Master?" two DBS specialists had to compete and answer questions on dystonia with short educational sessions on research and development updates in-between. The final prize of Euro 10 000 was awarded to Dystonia Europe and EPDA. The Medtronic patient centric view was clearly demonstrated by these initiatives and we are very grateful for all the support and efforts to make sure that Dystonia Europe and our work for dystonia patients was centrally exposed at the MDS congress. We appreciate very much the positive partnership between our organisations as well as our joint work for better quality of life for dystonia patients.



Dystonia Europe and EPDA receive the prize from the symposium «Who wants to be a DBS-Master?»



Dystonia Europe Summer Think Tank 2016

The very busy congress season ended with our second Think Tank (TT), held on 6 and 7 July in Lund, Sweden. The Dystonia Europe Board and representatives from our Platinum sponsors had come for the meeting. The goal of this TT was to discuss strategy and if our activities are driven by our objectives. We came to the following conclusion:

The Dystonia Europe **vision** is to **improve the quality of lives of people living with Dystonia in Europe, while supporting the search for a cure.**

We will achieve our vision through the following core activities:

- **Raise awareness** and be the leading voice in Europe on the impact of Dystonia (Jump for Dystonia, Under the umbrella, congresses).
- **Share the knowledge** – provide the latest information and resources for our members (DDAYS) and also be the source of information for the other stakeholders (data collection, Dystonia newsletter).
- **Connect people** with Dystonia, member organizations, healthcare professionals, European policymakers and the treatment industry to close the gaps of today's dystonia treatments (MyDystonia app).
- **Foster training and education** and educate multi disciplinary teams and stakeholders along the dystonia journey to optimize care and improve quality of life (training and education initiatives).



Monika Benson, Nathalie Courtine, Maja Relja, Adam Pinter, Jennifer Fluck, Sorin Ionescu, Erhard Mätzener and Alvaro Gonzales, jumping for dystonia.

Dystonia Europe at the Evelina Children's Hospital London

Dr. Jean-Pierre Lin and his specialist team welcomed DE Executive Director Monika Benson and a team from the medical device company, Medtronic, at the Evelina Children's Hospital in London last October, to discuss DBS barriers, MyDystonia for Kids and rehabilitation for children with dystonia. Next year OT (occupational therapist) Hortensia Gimeno, is planning to host an event in London for children with dystonia and their families. More information will soon be on our website.



*Monika Benson with specialist DBS team:
Sarah Perides (Specialist Paediatric DBS nurse)
Hortensia Gimeno (Occupational therapist) and
Esra Serdaroglu (EPNS Fellow from Turkey)*

Connecting with Dystonia Advocates in Berlin

During the MDS Congress in Berlin there was also the opportunity to connect with some passionate dystonia advocates, former board member of Dystonia Europe, Heike Wolf and our special dystonia photographer Stephan Röhl, who both live in Berlin.

Stephan has been our photographer at our annual meetings in Bol 2012, Edinburgh 2013, Paris 2014 and this year in Oslo, Norway. He also worked with us at the MDS Congress in Berlin. If anybody is interested in his services, we can highly recommend him and make the connection.



Heike Wolf and Stephan Röhl

Heike was on the DE board until the last GA in Oslo this year. We would like to take this opportunity and thank Heike for all the valuable work she did during her time on the Dystonia Europe Board.

We also appreciate very much that both Stephan and Heike continue to support Dystonia Europe in our activities.

Training Initiatives for Neurology Advocates – Launch

In 2016, the European Federation of Neurological Associations [EFNA] launched its Training Initiatives for Neurology Advocates [TINA].

For many years EFNA has run workshops on Health Technology Assessment and, more recently, Pharmaceutical Policy – Pricing, Access and Reimbursement at the London School of Economics.

However, EFNA feels it is increasingly important to ensure that its training activities are targeted to the neurology sector and the specific obstacles faced therein.

Working with other stakeholders in the field e.g. neurologists, neuroscientists, industry, regulators and payors is also part of the envisaged way forward.

This approach has led to the launch of TINA in autumn-winter 2016, with three events across Europe tackling the issues mentioned above, from both a national and pan-European perspective.

The overarching theme is:
PUTTING NEUROLOGY PATIENTS AT THE
HEART OF RESEARCH AND BEYOND

The Dystonia Europe President, Vice-President and Executive Director attended the first pan-European event in Dublin on October 13th and 14th, 2016.

The DE President Merete Avery said: “It was a valuable and informative workshop with many lectures of high quality providing important knowledge within the areas of research and patient reported outcomes, social media and communication, principles for patient partnerships with other stakeholders, which will all be useful in the work of our own association.”

National workshops, focussing on country-specific challenges, also form part of TINA – with workshops having taken place in Bucharest and Utrecht on October 6th and November 5th, respectively.

Once again, Dystonia Europe was represented. Attending the Romanian event, Sorin Ionescu said: “It is important with this kind of patient training on national level to increase patient advocacy and involvement in order to influence decision-makers, especially within the field of health.”

Together under the umbrella – Focus on invisible disabilities

Invisible disability is a term that captures a whole spectrum of hidden symptoms or challenges that are primarily neurological in nature.

Many brain and brain-related disorders cause symptoms that are not obvious to an onlooker. These symptoms can include pain, fatigue, sleeping problems, dizziness, cognitive dysfunction, brain injuries, learning difficulties and mental health disorders, as well as hearing and vision impairments.

While these symptoms may not be visible, they can limit the person’s ability to perform daily activities and severely impact quality of life. Even in brain disorders that have immediately apparent symptoms, the secondary invisible symptoms can be just as, or even more, disabling – and worsened by the stigma that such unseen disabilities can create.

That’s why, the European Federation of Neurological Associations [EFNA] got involved in supporting the annual Invisible Disabilities Week.

Using it's *Together Under the Umbrella* campaign, it asked supporters to take a photo hidden behind an umbrella and to post this to Twitter; listing their disease area and the associated invisible symptoms.

Many groups got involved, including DE and its members.

The photo below shows Adam Kalinowski of Dystonia Poland, behind his umbrella.



The wider *Together Under the Umbrella* campaign – supported by DE – runs until April 2017. To get involved or to learn more, see: www.undertheumbrella.eu or check out #UnderTheUmbrella on social media.

#MakeWorkWork with EFNA!

People with neurological and chronic pain conditions may find it difficult to get or keep a job. Unemployment and under-employment can worsen the situation for patients by impacting self-esteem, mental health and increasing isolation.

Rose living with Dystonia said: “When I went to see them I sat there and my head was totally twisted to the left. The health insurance officer gave me a cold look and said to me, ‘If you turn your left side of your body towards the desk you can write on the computer’. My eyes filled with tears, I remember feeling so hurt and misunderstood. She doesn’t know anything about what it feels like not being able to control your movements the way you want, I thought. There is nothing else I would like than being able to hold my head straight and go to work.”

To help Rose and the millions in Europe living with Dystonia and other neurological disorders, the MEP Interest Group on Brain, Mind and Pain is working on this topic of equitable opportunities at work.

On October 25th, a *Written Declaration on Access to Employment for those affected by Neurological Disorders and Chronic Pain Conditions* was launched in the European Parliament.

A Written Declaration is statement of policy which is placed before the Parliament for signature. Any MEP can sign if they support the Written Declaration and would like this to become the official position of the European Parliament but 376 MEP signatures are required for this to happen.

You can read the full Declaration at: www.brainmindpain.eu

EU Activities

If you'd like to get involved, then you may consider contacting your regional MEPs and requesting their signature. The involvement of national organisations and individuals is absolutely vital in ensuring that the goal is reached.

You can find a list of MEPs by country and template letters for both organisations and individuals to help you on the website listed above.

You can also contact Heather Clarke, EFNA's EU Policy Advisor for further assistance:

euaffairs@efna.net

You can help us to #MakeWorkWork (check out this hashtag on Twitter too!)



MEP Heinz Becker

EFNA is an umbrella of pan-European disease-specific neurology patient organisations. Its slogan is 'Empowering Neurology Patient Groups' – which encapsulates its work under the four pillars of: advocacy, awareness, empowerment and engagement. In 2016, it signed a Memorandum of Understanding with EAN as its patient partner.



My Dystonia

The “MyDystonia” app has been available since 2015. This digital diary for dystonia patients was developed by Dystonia Europe with the support of Merz Pharmaceuticals. And now another dimension has been added with the MyDystonia Ambassador

They come from Germany, Norway, Sweden, Spain, the UK, Finland, Denmark, and Poland, and they all have a mission: to be ambassadors for MyDystonia. At a kick-off workshop in Oberursel near Frankfurt, they found out exactly what that means. The “MyDystonia Ambassador” program was launched to support the app’s introduction in individual European countries and is based on the simple idea that the best people to promote an app developed by patients for patients are also patients themselves.



Using the free app, patients can keep an electronic diary of their neurological condition. Normally, people who suffer from dystonia see their doctor every three months, and so physicians and those affected by the condition have not, until now, had any practical means of tracking the course of their illness between appointments. Now, the users of the MyDystonia app will be able to document the pattern of their illness each day in a process that only takes two minutes.

The MyDystonia Ambassadors are now launching their mission to make the benefits of the app accessible to as many people as possible. They are active users of the app themselves and will be running local activities to spread the word to other patients. The aim is clearly defined: to increase awareness of MyDystonia and boost user numbers.

My Dystonia

And so, on two days in fall, the MyDystonia Ambassadors came together to devise activities for the mission. As MyDystonia Ambassador Gill explained: “I came here without any preconceptions about what being an ambassador means. Now I have a very clear idea of the many things I can do.”

So what are the ambassadors' exact duties? They are local contacts for the app and can explain it to people who are interested. They contact other MyDystonia users, help them, and offer them support if they have questions or problems. One way in which they do this is by the targeted distribution and use of information material about MyDystonia. The ambassadors pass on user feedback from their MyDystonia network to MyDystonia's developers.

In order to facilitate the ambassador's mission, a platform is going to be developed as a working tool to enable them to communicate better. During the workshop, the different functionalities of this platform were jointly determined by all of them. But the exchange is not only taking place through this platform, but also in a founded Facebook group or regular calls. And of course also at the regular meetings of Dystonia Europe, like next May in Rome, where there will be even



The dashboard as a resource for ambassadors.

a special session for the MyDystonia ambassadors.

The ambassadors' wholehearted commitment to the app and enthusiasm about performing their role became clear during the workshop. They are all convinced of the app's benefits. Equipped with the knowledge acquired during the workshop, their mission can now start in earnest. By their efforts, they are actively helping to improve communication between people with dystonia and physicians, optimize treatment success, create greater quality of life for dystonia patients, and strengthen dystonia research in the future by gathering anonymous data.



*MyDystonia
Ambassadors*

Jump for Dystonia

Jump for Dystonia Skydive in Sweden

I have always been terribly afraid of heights. I get a dizzy feeling and it's almost like the floor is moving and I am pulled towards the edge. I have tried to get over it. Once I went on the "Free fall ride" at the Gröna Lund theme park in Stockholm, and I only say – never again! My fear of heights got even worse.

That day I wake up by the alarm as usual. When remembering that this is the day my daughter Michelle is going to do a skydive I get up immediately. Last year I signed Michelle up for the JUMP for Dystonia contest organised by Dystonia Europe. Michelle had told me once that she wished to try skydiving, so this must be fate I thought. I took a photo of Michelle when we were visiting Milan in Italy, where she was jumping for our disorder in front of the famous cathedral, Il Duomo. And guess how surprised I was when she was one of the four winners of a Jump for Dystonia Skydive.

When we head towards Gryttjom airport, close to Tierp, north of Stockholm, Sweden, the weather is gorgeous. The sun is shining and there is a nice and warm breeze. Arriving at the destination we meet with Bengt-Erik Calles, the President of the Swedish Dystonia Association, his brother Olle and some of Michelle's friends. We take some photos and we have a coffee and a snack. Michelle is unusually calm and I feel a bit stupid not feeling the same way. She has told me afterwards that she was not the least nervous, only looking forward to the activity. Her co-jumper, Alexander, introduces himself and then they put on the security equipment. He tells her how to do the jump and informs her about all safety routines. Michelle shares with him why she is going to Jump and that it's for a good cause: for all of us who have dystonia. She explains what dystonia is. Alexander tells me that I should jump as well, but I don't think it is such a good idea.

When I hear the plane take off with my daughter to reach a height of 4000 metres so she can throw herself out the door, I catch myself thinking: "at least we are lucky with the weather". We all try to follow the plane higher and higher in the sky and I take as many photos as I can. The plane is at a long distance

and it becomes more and more difficult to see. At the end I lay flat on the ground looking up into the sky. It is neither fun nor very easy having cervical dystonia and to be standing still and trying to follow a small plane in the sky. Good it's the first and only time. I lose of course sight of the plane. After a while Bengt-Erik tells me that he sees parachutes in the sky. We know that Michelle has an orange parachute since we asked for it. But there are three other skydivers who also have orange parachutes. Therefore we don't know which one of the four is Michelle. We follow the skydivers when they glide towards the ground and I take many photos of all parachutes that look a bit orange. I don't want to miss this occasion. When Michelle is finally sitting on the ground again I sigh of relief. She comes walking towards us with a big smile. The other skydivers are greeted by their friends and families.



Michelle Rydow

A lot of people cheer and scream of excitement. "So cool!" Michelle with the happiest smile on her face says: "I think I want to jump again."

I still cannot understand the excitement and why people want to fall free for 50 seconds, like my daughter did. I would never do the same thing. But if you have no fear of heights I'm sure it must be an incredible experience. For Michelle this is a memory for life. She is so happy that her dream came true. She wants to go skydiving again and she also wants to do a bungyjump.

Jump for Dystonia

Many thanks to Dystonia Europe and the Jump for Dystonia campaign which made this possible. Michelle is so happy and I am very proud of her for doing both the skydive and the winner Jump in Milan - two important JUMPs for all of us with dystonia.

Johnny Rydow

Vice President, Swedish Dystonia Association



Michelle Rydow ready to Jump for Dystonia from the sky.



Jump for Dystonia Skydive in Finland

(Translated from Newsletter 3/2016 of the Finnish Dystonia Association)

The Finnish Dystonia Association was very happy and proud when they were one of the four winners of a Skydive in the the Jump for Dystonia contest. The skydive took place beginning of July at the Malmi Airport outside of Helsingfors. The association had donated the skydive to Finnish MEP (Member of EU Parliament) Merja Kyllönen. She had already jumped for dystonia when Dystonia Europe organized a dystonia awareness day in the European Parliament in Strasbourg in 2014. Now she made a much bigger jump and also bigger for

dystonia awareness. Merja Kyllönen said: "It was a great feeling, a feeling of being weightless. All people living with a disorder should be able to experience that kind of feeling of being free from your body." She had enjoyed the beautiful scenery when floating in the air. "Thanks a lot for giving me this opportunity, it was a great honor for me to receive this gift and to JUMP once again for Dystonia." The Finnish Dystonia Association had gathered a group of happy members at the airport. All dressed in orange JUMP t-shirts, just like Merja, they cheered and welcomed Merja after her JUMP.

We especially thank Susanne Olenius for all her work in promoting the campaign in Finland.



The Finnish Jump for Dystonia supporters



*Finnish MEP (Member of EU Parliament)
Merja Kyllönen*

www.dystonia-europe.org

Jump for Dystonia

Jump for Dystonia Skydive in Romania

Romania was one of the four winners in the Jump for Dystonia Skydive Contest. We were actually the very first winner in this campaign that was promoted in our country by the two associations Dystonia Asociatia and Children's Joy. The Romanian winning JUMP came from Ramona Pop and her photo received all together a total of 600 votes!



Dystonia Asociatia

Due to personal issues she could not take part in the Skydive and she therefore decided to donate her prize to Dystonia Association President Sorin G. Ionescu. We thank Ramona for her participation in our contest and for her support in raising awareness of dystonia.

On May 29 this year Sorin headed for the airfield in Lunca, about 60 km from the city of Cluj-Napoca in Romania.

Here "Transylvania Skydive" welcomed him and informed him about the day and how to JUMP. The jump took place at a height of 3000 meters, together with a skydive instructor. This was all filmed to help raise awareness of dystonia.

The video can be found on the Jump for Dystonia Facebook page:
www.facebook.com/jumpfordystonia

Just like in many other countries around the world the Jump for Dystonia Campaign was a great success in Romania. There were many Jump for Dystonia events around the country, many photos submitted to the contest and we thank everybody who voted for our photos.

The campaign was featured in various Romanian media on local and national level. On behalf of the Romanian Dystonia Association we congratulate and thank all the people who were involved in the campaign. It was a great initiative from Dystonia Europe and it has been a pleasure working on this project together with dystonia advocates from all over the world!

Jump for dystonia – together for dystonia!



*Sorin G. Ionescu
President, Asociatia
Dystonia and
Board Member
Dystonia Europe*

Meet Board Member: Sorin Ionesco

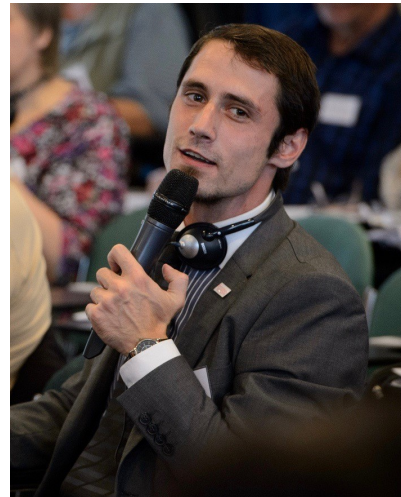
Sorin Ionesco, the chairman of the Romanian Dystonia Association – Asociatia Distonia - was elected to the DE Board at the General Assembly 2015 in Rotterdam. He was dubbed as “The man who turned disability into success” by author Cristiana Cicau in a major article in the newspaper Transylvania Reporter in September 2013 about Sorin’s incredible life story. This article is an excerpt from that text:

Sorin had many odds against him when he was born with umbilical cord strangulation after 24 hours spent without amniotic fluid. The lack of oxygen caused progressive neurological problems visible as involuntary movements and tremor.

“It was often hard because there were times when I had to hide from other children. It was easy probably because I grew up in a small community where acceptance for my condition was easier to digest”, says Sorin.

Despite these barriers, Sorin played in the school soccer team and participated in competitions, always among the best, determined not to let his disability compromise his life. Sorin confessed that he had unconditional support from family to a degree where he sometimes even felt overburdened with protection.

When he moved from country to city for higher education he slowly managed to take the reins of his life into his own hands. In the beginning he was still financially dependent on his family, but during his second year in college he worked at a playground, go-cart track administration, as a marketing agent in private pension and even in interior design, as far as his medical condition allowed him, in order to pay his way through college, studying Sociology. After his graduation he found a job in the social field but continued to carry out voluntary work and social projects.



Sorin Ionesco

After dozens of different diagnoses and even more treatments without results he was finally diagnosed with generalized dystonia at the age of 25, and a high degree of disability was finally recognized. Sorin feels that the biggest obstacle he encountered was not the disability itself but the mentality of society about disability which forms an invisible barrier which is, in his words, not invincible.

Today Sorin is working as an operator in a market research company. He founded and leads the Romanian Dystonia Association in order to help patients and to raise awareness among society and in the medical field. Sorin is also running a website on “Dystonia in Romania” and he wrote a book about “(dis)abilities in experience”.

Sorin says, everything he does is through small, steps. His involvement in community and life achievements have not gone unheard. He was even called to give a lecture at the faculty he attended. But Sorin feels most rewarded by the gratitude of the patients he has been able to support and help, by “a smile, a tear, a sincere thank you”. He feels that his way leads to the fullness of life. What a wonderful message that is to the estimated number of 2000 dystonia sufferers in Romania!

Around Europe & Beyond

Norway

Final Workshop of 3-Year Project on Physiotherapy & Dystonia

Last week of August the final workshop of the Norwegian project to train physiotherapists on dystonia throughout the country, took place in Tromsø, in Northern Norway. Fifteen participants had come to the 2-day workshop.

Chairwoman of the Norwegian Dystonia Association, NDF, Anniken Hansen welcomed everybody and then gave a short update on background and development of the project. It is very clear that this project is a great success in Norway. "Now I know about specific movements and exercises to use with dystonia patients which I had never thought of before" one participant said. "We now also have this network of physiotherapists throughout Norway who are skilled in dystonia and to whom I can refer patients."

The project has also attracted international interest with a visit from an Austrian physiotherapist some time ago.

The project is funded by Ekstrastiftelsen, a Norwegian Foundation, which has supported the project with about NOK 1 200 000 over the three years.

The programme has consisted of 2 Workshops – step 1 and step 2. These have been organized in various cities throughout Norway: Bergen, Oslo, Arendal, Molde and Tromsø in order to give physiotherapists from all over the country the opportunity to attend.

The first workshop gave an introduction to dystonia and treatment possibilities such as with botulinum toxin and physiotherapy. Dystonia patients took part in the workshop for hands-on training. Before the second workshop the participants got homework to prepare for the meeting. Here they could report by video or bring their patient to the workshop.



From left:

*Monika Benson,
Anniken Hansen,
Anette Holmelid Elvik,
and Johanna Blom*

Around Europe & Beyond

It was a great pleasure for me to attend this workshop on behalf of Dystonia Europe since I also took part in the very first workshop in May 2014 in Bergen. It was exciting to see how the level of the workshop and the activities have been taken forward. The high quality of trainers and presentations contributed to an excellent workshop and it's my hope that this programme can continue to develop. The participants were interested and engaged in the discussions and they had all prepared their homework very well. Day 1 they presented a dystonia patient, in person or on video, and made an anamnesis and described how they thought about the dystonic symptoms. Day 2 they all demonstrated how they thought about exercises and training for their patients. Physiotherapist Johanna Blom from Malmö University Hospital in Sweden, guided and added value to these patient demonstrations. Anniken Hansen who is a physiotherapist herself and also has dystonia, and physiotherapist Anette Holmelid Elvik both also contributed largely .

Neurologist Grit Richter from the University Hospital in Tromsø participated in the second part of the workshop and gave presentations on the plasticity of the brain, how to optimize medical treatment and what scoring tools are available.



Physiotherapist Anette Holmelid Elvik gave a presentation on "Coping with Dystonia" and what an important role the physiotherapist also has in coaching the dystonia patient.

Trainers who have been involved in the project are: Jean-Pierre Bleton from France, Johanna Blom from Sweden and Anette Holmelid Elvik from Norway.

A great congratulation to Anniken Hansen and NDF for a very well performed project and we wish you much good luck and success in your continued work to improve skills and knowledge of dystonia among Norwegian physiotherapists.

Monika Benson



*Physiotherapist
Johanna Blom
demonstrating
exercises for
dystonia*

Germany

Dystonie Förderverein Deutschland e.V. (DFVD e.V.) - Who we are and what we do!

The DFVD e.V. was founded in 2014 and has been a member of Dystonia Europe (DE) since the DE General Assembly last April this year.

Martina Kühn is the chairwoman and Elke Schilling is the vice chairwoman of the association.

One of our tasks is to support Deutsche Dystonie Gesellschaft e.V. (DDG e.V.). Furthermore, the association can support individuals if they are not able to pay for treatment themselves.

The first project is the translation into English of the booklet "Dystonia - when the brain plays a trick on the muscles" so it can be used internationally. We hope we can finish this project within a few weeks.

This year, Dr. Aloysius Domingo received the Openheim Award from DDG e.V. for his work on "x-linked dystonia parkinsonism" (XDP). This form of dystonia has so far only been found on Panay Island, an area in the Philippines. The particularity of this disease is the overlap of dystonia and Parkinson's disease.

Dr. Domingo described in his work a new gene (TAF-1), which is responsible for the formation of XDP.

Those affected are predominantly men. One of these patients is Alain. Alain was a happy family father before the onset of the illness. His passion was photography. Because of his illness he lost his family and his profession.

One possibility for him is surgery, either DBS - deep brain stimulation, or a pallidotomy. The surgical treatments are as expensive in the Philippines as in Europe. Therefore only Pallidotomy is possible for Alain, because this is only about 10% of the cost of DBS. But Alain doesn't have that kind of money either. That's why we want to help him, and if possible, many others with Dystonia Parkinsonism in the Philippines. We have started a fund raising campaign which will hopefully bring us enough funding in order to help this very severely affected dystonia sufferer.



Martina Kühn

In the Uni-Klinik in Lübeck, where Dr. Domingo worked as a exchange-doctor until the summer of 2016, twelve Philippine patients were treated with DBS as part of a project.

Prof. Christine Klein, who is very involved in this project, said in an interview with the Lübeck news in 2014 that these patients in their homeland were regarded as demons. They would be hidden away so neighbors wouldn't see them because of their terrifying appearances, and sometimes they would even have to live in cages behind the house. There they would often die from malnutrition or infections.

In the Dystonie-Förderverein Deutschland e.V. we would like to help these patients. We are in contact with Dr. Aloysius Domingo, Dr. Cid Diesta, neurologist from Manila and Prof. Christine Klein from Lübeck, who are all involved in the project.

We need help so we can help. Everybody can help with something. We are looking forward to any kind of support.

*Martina Kühn
President
DFVD e.V.*



*Oppenheim-Preis 2016
Dr. Aloysius Domingo, Prof. Christine Klein, Ute Kuhn.*

Around Europe & Beyond

Czech Republic

Dystonia Awareness in Prague

The Czech dystonia patient organization was established in 2013. Our organization has 25 members with dystonia. About 15 members are involved in various activities such as: sewing toys and pillows; spreading information about dystonia; providing a video about the disease; gathering information on the effect of treatment and rehabilitation; and participation in discussions on social networks.

Two members, Jana Vičarová and Věra Chybová, are leading the project "Awareness campaign to support patients with dystonia" funded by the Ministry of Health of Czech Republic.

We organize meetings between patients and doctors in cities such as: Chomutov, Prague, Olomouc, Brno. One of these "meet the experts"-meetings took place at the Neurology Clinic of the Medical Faculty of Charles University in Prague on 19 September. On the same day we held a Jump for Dystonia event on Charles Square in Prague. We distributed information about dystonia and asked people to jump and we took photos of them.

For this event, we had invited Monika Benson, Dystonia Europe Executive Director, who also joined for lunch with Dr. Josef Kraus from the department of Paediatric Neurology at the University Hospital Motol together with other members Kateřina Svobodová and Aliya Bokash.

For the afternoon programme dystonia patients and their families came to listen to various presentations on dystonia such as: "Genetics and dystonia" with prof Robert Jech; "Rehabilitation in dystonia" with Dr. Petra Havráňková; "Anxiety and Panic Disorder" with Dr. Tereza Uhrová.



Happy Dystonia Advocates in Czech Republic



Dystonia Patients meeting in Prague

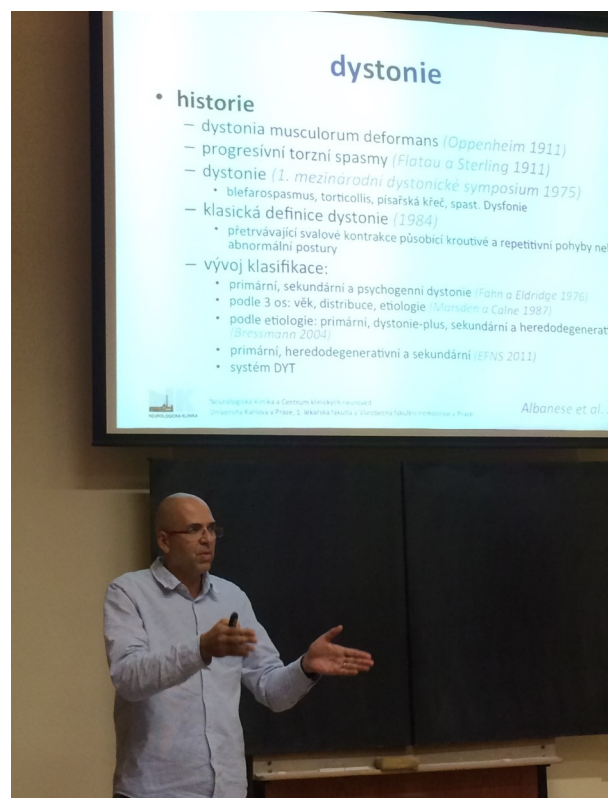
Around Europe & Beyond

For pediatric patients, our organisation has donated special toys made of fabrics which we call “Families together for dystonia”. A member of our organization sews the toys in the workshop “Šošana”.

These products are also prizes in the competition for the best jump for dystonia. Participants of the contest must answer the question: what is dystonia? You find the answer on our website and in the leaflets of our organisation. Unfortunately we don't have permission from the authorities to sell our products yet.

We are very pleased with our meeting last September in Prague and we thank speakers, participants and Monika for being here. Another meeting is scheduled for December 8, 2016 in Olomouc.

Jana Vičarová
President
Czech Dystonia Association



Prof. Robert Jech



Jump for
Dystonia in
Czech Republic

2017

January

- 11-13 2017 BPNA Annual Conference, Cambridge, UK
18-21 Toxins 2017, Madrid, Spain
26 Dystonia Europe Board Meeting, Lund, Sweden
26-27 Dystonia Europe Winter Think Tank 2017, Lund, Sweden

February

- 2-3 World Paediatric Movement Disorders Congress, Barcelona, Spain
TBC Brain, Mind & Pain Working Group Meeting, EU Parliament, Brussels, Belgium

April

- 10-11 EPF, General Assembly, Brussels, Belgium

May

- 11-14 AOCCN 2017, 14th Asian and Oceanian Congress of Child Neurology, Fukuoka, Japan
11 Dystonia Europe Board Meeting, Rome, Italy
12-14 Dystonia Europe D-DAYS 2017 & the 24th Annual Conference & General Assembly, Rome, Italy
12-13 6th Biennial Workshop on Dystonia: "Dystonia-the link between hypo- and hyperkinetic movement disorders", Rome, Italy

June

- 4-8 21st International Congress of Parkinson's Disease and Movement Disorders, Vancouver, Canada
19-20 2nd EPNS International Satellite Symposium on Advances in Neuromodulation in Children,
20-24 12th EPNS Congress, European Paediatric Neurology Society, Lyon, France
24-27 3rd Congress of the European Academy of Neurology, Amsterdam, The Netherlands
EFNA General Assembly



D-DAYS

24th ANNUAL CONFERENCE

12 & 14 MAY, 2017 ROME,
ITALY

SAVE THE DATES

D-DAYS 2017 will take place at the Hotel IBIS Styles Roma in Rome, Italy: <http://www.ibis.com/it/hotel-9301-ibis-styles-roma-eur/index.shtml>

The hotel is located in Rome's EUR business district, only 20 km from Fiumicino airport, and is just 15 minutes' walk from the EUR Palasport metro stop, with direct connections to the Colosseum.

You can already now book your room directly with the hotel and get a special room rate. Download the booking form from our website and send it to: H9301@ACCOR.COM
To guarantee the special room rate and availability please book before 31 March.

PROGRAMME

Programme and more information about the event will soon be available on the Dystonia Europe website, where you can also register for the congress. <http://dystonia-europe.org/activities/events/dystonia-europe-2017-rome/>

Main topics on the programme will include: Overview of Dystonia, Research, Treatment of Dystonia, and Rehabilitation.

DYSTONIA
EUROPE



We welcome patient leaders, dystonia patients and their families, physiotherapists, nurses, junior doctors and everybody else with an interest in dystonia.

Hope to see you in Rome!

Members

Dystonia Europe consists of 21 national member groups from 18 European countries and they are:

Austria, Belgium, Croatia, Czech Republic, Denmark, Finland, France, Germany, Ireland, Italy, Norway, Poland, Portugal, Romania, Spain, Sweden, Switzerland and United Kingdom.

Donation & Support

Dystonia Europe welcomes and greatly appreciates any donations/support from organisations and individuals to further develop all the work for dystonia in Europe by funding research, awareness and education. If you would like to support the work of Dystonia Europe please contact us at sec@dystonia-europe.org to discuss the possibilities.

To donate directly please use the following on-line banking details:

Accountholder: Dystonia Europe

BANK:	KBC Bank, 16a Tervurenlaan, 1040 Brussels
IBAN number:	BE83 7350 0508 5515
SWIFT/BIC	KREDBEBB

When making a payment please include a reference to allow us to identify the donor.

You can also use the donate button on the Dystonia Europe website, which will take you to the following page: <https://dystonia-europe.org/donate/>

Thank you for supporting Dystonia Europe!

Medical & Scientific Advisory Board

Prof Alberto Albanese – Milan
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Prof Marie Vidailhet – Paris



Management & Staff

Board of Directors - Dystonia Europe is governed by a volunteer Board of Directors.



Merete Avery, President,

Norway

Merete was appointed to the board of Dystonia Europe in 2013 and has held the position as Secretary from 2013-2015. Merete was appointed President at the Board Meeting, following the GA, in Rotterdam 2015. She is from Norway. She was diagnosed with cervical dystonia in 2006 and she was Chairwoman of the Norwegian Dystonia Association between 2010-2013. Merete is working with customer service, accounting and administration for a company in Molde, Norway.



Monika Benson

Board Member and Executive Director, Sweden

Monika was elected president of EDF, now Dystonia Europe, in 2007. She was re-elected for a second term in 2010. Monika stepped down as President in 2013 after serving the maximum period of 6 years. She took over as Executive Director after Alistair Newton's retirement. Monika has cervical dystonia and is also a board member of the Swedish Dystonia Association. Monika has been working as a coordinator of work-shops, courses and lectures at a school in Lund, Sweden.



Sorin Ionescu, Board Member, Romania

Sorin is the founder and President of the Romanian Dystonia Association, Asociația Dystonia. The objectives of the association are to raise public awareness, provide support and advice to dystonia patients and their families and to promote research. Sorin has a degree in social science and is a writer, painter and has been involved in social activities since 2010. He was diagnosed with Generalised Dystonia in 2012.



Maja Relja, Vice President,

Croatia

Maja is a professor of neurology, in charge of the movement disorders section at Zagreb University Hospital. She founded the Croatian Dystonia Association and has served as its president for over 20 years. In 2011, she was elected to the board of Dystonia Europe, and appointed Vice President in 2012. She also serves on the DE Medical and Scientific Advisory Board and is heavily involved in the COST-funded Dystonia Research Network.



Erhard Mätzener, Treasurer, Switzerland

Erhard was appointed Treasurer by the Dystonia Europe Board in November 2015. He is a member of the Swiss Dystonia Association SDG since 2008, and from 2010 he is their auditor/controller. In 2012 he was appointed the SDG delegate to Dystonia Europe. Erhard is an economist and was working as an investment advisor for a Swiss Private Bank for more than 20 years until he retired in 2002. Erhard suffers from blepharospasm.



Cristina Frosini, Board Member, Italy

Cristina was appointed to the Board of Dystonia Europe in 2016. She is a pianist and deputy Director of the Milan Conservatory. She has won many international competitions and played hundreds of concerts including orchestral recitals radio and television recordings. Since 2005 Cristina has been suffering from focal dystonia which forced her to stop her concert activities. After being treated in Hannover, Germany, she is now almost completely recovered. She therefore is devoting her focus on pianophysiology.



Alistair Newton, Advisor, UK

After several years as Vice Chair of the Dystonia Society in the UK, Alistair founded European Dystonia Federation, now Dystonia Europe, in 1993. He served 8 years as President and was the Executive Director between 2001 and 2013. He was appointed to the Dystonia Europe Board with special responsibility of the Dystonia Research Network. Alistair also co-founded EFNA – the European Federation of Neurological Associations – in 2000, where he acted as Treasurer and Secretary-General until 2011. In 2003, he was involved in the founding of the EBC – the European Brain Council, where he was a board member and a treasurer for many years. He is now an advisor to DE on special projects. Alistair has had cervical dystonia for 30 years.



Eelco Uytterhoeven, Advisor, The Netherlands

Eelco has been a professional IT-consultant and developer since 1999. For the last 2 years he has been working as a freelance developer on several internet projects related to Dystonia Europe. Since beginning of 2016 he is responsible for the maintenance and further development of the myDystonia platform. Eelco was asked to become a special IT advisor to the Dystonia Europe board at the D-Days 2016 in Oslo. Together with the board Eelco wants to raise the professional level of the IT projects of Dystonia Europe and assist in creating new possibilities for the future.

Partners & Sponsors

We have good working relations on a variety of topics with:

DMRF – Dystonia Medical Research Foundation, FDR – Foundation for Dystonia Research, Dystonia Coalition, EFNA – European Federation of Neurological Associations, EBC – the European Brain Council, EAN – European Academy of Neurology, MDS – International Parkinson and Movement Disorder Society.



Live better. Feel better. Look better.



www.dystonia-europe.org