Connecting People for Dystonia

DYSTONIA EUROPE NEWS

Winter 2014

"Welcome to Edinburgh and the Dystonia Europe 20th Anniversary"!

Former Dystonia Europe President and Executive Director Alistair Newton receiving the guests for the Anniversary Dinner at the Royal College of Surgeons in Edinburgh.

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

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President's Message

Robert Scholten

Dear all,

In September 2013 I had the great honour of being elected new President of Dystonia Europe. It is my wish that during my presidency I will in some way contribute to the improvement of the daily lives of dystonia patients. I look forward to cooperating with you.

Fortunately, Alistair Newton and Monika Benson, together with the Dystonia Europe Board, achieved a lot over the past years. I am very grateful for this and I especially would like to thank Alistair Newton for all the work he has done for dystonia in Europe over the past 20 years. In September 2013, he withdrew as the Executive Director of Dystonia Europe and is now a Board member with special responsibility to coordinate the Dystonia Research Network. I also would like to thank Monika for the hard work she did in the past six years as President of Dystonia Europe. She did an outstanding job and she has now moved on to the position as Executive Director. I hope to learn a lot from Monika and during my first months as President she has been of great support. It is wonderful that there is already a solid foundation that we can continue to build upon. In my maiden speech at the General Assembly in Edinburgh I declared already that I want to focus on a theme for Dystonia Europe over the next few years:
Connecting People for Dystonia.

Vision and strategy: Connecting People for Dystonia 2014-2020
A new vision and strategy plan for the next years 2014-2020 is being developed. How do we want Dystonia Europe to move forward? This plan will be presented and discussed with DE delegates at the next General Assembly, in October this year in Paris.

A Digital Platform for Dystonia
In today's society we are increasingly sharing information and experiences through internet and social media. It is therefore important that all people with an interest in dystonia easily can be connected. We want to facilitate these activities by creating **a digital platform for dystonia** over the next few years.

Face-to-Face Contacts
It is of course also important with face-to-face contacts. The DE General Assembly is a very important occasion for these kinds of meetings and interactions between member representatives. To increase these interactions one idea for the future could be to combine the DE Annual Meeting with national member meetings.

President's Message

Awareness Campaign 2014: Jump for Dystonia
There is still a lot of misunderstanding about dystonia and it is crucial to continue the work of raising awareness and spreading information about the disease. Our next awareness campaign called ' Jump for dystonia ' will be launched during the year.

Sponsors
The collaboration with our various partners and sponsors has developed and grown stronger.
I would like to take this opportunity to thank everybody for their generous support and their belief in Dystonia Europe, our vision, ideas and activities. It will be exciting to work together on our next projects: the Digital Platform and Jump for Dystonia.

Dystonia Training School in London
Last November the Dystonia Training School 2013 (within the COST project The European Network for the Study of Dystonia Syndromes) took place at Queens Square Hospital in London. I was very happy to attend some of the meetings. The 43 young neurologists from all over Europe, attended the 3-day programme that included presentations on dystonia research, treatment, diagnoses etc, held by some of the most distinguished dystonia experts in Europe. It is with great pleasure I see Dystonia Europe being part of the important work to inspire and train future dystonia experts.

Robert Scholten taking the first steps as DE President together with Monika Benson, Sissel Buskerud, Heike Wolf and Merete Avery.

New members
In 2013 we welcomed two national dystonia associations from Romania as members of Dystonia Europe: Asociata Children's Joy and Asociatia Dystonia. We wish them all the best in their work for dystonia patients in Romania.
We hope that new dystonia member groups will continue to be established in countries where there are none. It is the wish of Dystonia Europe that all European dystonia patients have a support group somewhere near them. The more members we are in Dystonia Europe the stronger we will be and hopefully our voice will be better heard and dystonia more known.

Annual Meeting & General Assembly Paris October 2014
I look forward to seeing everybody again at the Annual Meeting and General Assembly in October this year in Paris. A preliminary programme has been drafted and the venue is decided, see more information on page 33.

I would like to take this opportunity to thank AMADYS and its President Stéphanie Fréhel as well as Professor Marie Vidailhet for all their help in planning this meeting so far, which, I am sure, will be a great success.

Finally I would like to thank you once again for trusting me to lead Dystonia Europe towards 2020 and I look forward to working with you.

I wish you all the best for 2014!

Best regards
Robert Scholten
President

The Dystonia Europe 20th Anniversary Conference "Achievement & Hope"

Last September 6 & 7 Dystonia Europe celebrated its 20th Anniversary by holding the conference "Achievement & Hope" in Edinburgh, Scotland. About 85 participants from all over Europe had gathered at the James Thistle Hotel in central Edinburgh. There were dystonia experts and national dystonia patient group leaders from all over Europe as well as dystonia patients from the UK.

Dystonia Europe President for the last 6 years, Monika Benson, greeted the audience with a warm welcome speech. Monika spoke about the ACHIVEMENT throughout the years, since the European umbrella organisation EDF, now Dystonia Europe, was founded in 1993, and she also spoke about the HOPE ahead of us. Monika expressed it in these words: «My dream is that one day I can say that I have dystonia and people will know what it is". Unfortunately, too often when people talk about dystonia, there are very few who have heard about it. I think Monika expressed the HOPE that so many people affected by this movement disorder feel. In addition there is HOPE for the development of better treatment, and maybe one day a cure.

Then followed Dystonia Europe Executive Director Alistair Newton whit his presentation "Setting the Scene". Alistair was one of those who took the initiative and founded the organisation in 1993. He talked about the history and the achievements of Dystonia Europe over the last 20 years. In 1992 there was no known research network within Dystonia.

In 2003 there was "the European Dystonia Research Group", and in 2011 "the European Network for the Study of Dystonia Syndromes" was founded. Dystonia Europe has played a major role in arranging conferences solely on dystonia and thereby contributed to increase the interest in dystonia among the medical profession and researchers.

Anniversary

The achievements throughout the years have been considerable. Dystonia Europe has carried out an excellent job and created an arena for patient groups and the medical profession to work together. Many new ideas and various projects all around Europe have developed from the Dystonia Europe network, the Anniversary was well worth celebrating!

The Anniversary dinner was held at the Royal College of Surgeons in Edinburgh. Bagpipe musicians and traditional Scottish folk dancers entertained the guests, and on the menu was haggis, a typical Scottish dish. It was a great Anniversary dinner in a magnificent Scottish setting. Alistair Newton was honored with speeches and acknowledgments for his dedicated work for Dystonia Europe over the past 20 years.

Conference

The 2-day program contained many interesting presentations given by twenty dystonia experts from all over Europe. It is exciting to hear about many different topics of dystonia, such as: genetics, pain, treatment, research, physical therapy and much more. In addition there were speakers from patient organisations sharing their work and how they collaborate with the medical profession throughout Europe. At the conference "The David Marsden Award 2013" was given to Dr. Katja Lohmann from Lübeck in Germany, for her research on hereditary whispering dysphonia. Read more about it on page 16. To have the opportunity to meet and learn about the work of so many dedicated and enthusiastic dystonia experts: neurologists, researchers, surgeons, nurses, physiotherapists, is both comforting and promising for all of us affected by dystonia, and it gives a lot of HOPE for the future!

Merete Avery
Secretary, Dystonia Europe
Board Member,
The Norwegian Dystonia Association.

"Achievement and Hope" - Dystonia Europe 20th Anniversary Conference - Edinburgh, 6/7 September 2013

Alistair Newton

Somehow, the 20th anniversary celebration of a pan-European network like Dystonia Europe is a special moment which should be celebrated in style, and our efforts in 2013 seem to have achieved just that!
The Dystonia Europe Board made the choice of venue - Edinburgh, Scotland, and this was, for me, a fitting close to my twenty years of central involvement in our organisation. I am grateful to Monika Benson, who proposed the city and worked so hard to make the whole event such a success, and to all the patients, supporters from the industry and the healthcare and medical specialists who attended.
Our session chairs, speakers and also our sponsors deserve particular thanks. So many very senior specialists and equally busy friends from the industry gave their time and effort to join us and create a meeting which brought the latest information on dystonia to the audience. Without that real partnership of so many clinicians, researchers and sponsors, in a transparent and ethical working relationship, Dystonia Europe could not have achieved many of its goals. Their special support of the 20th Anniversary Conference is much appreciated.
The conference theme - "Achievement and Hope" was intended to showcase the very positive changes in the world of dystonia since Dystonia Europe was founded in 1993, and the hope that these improvements can bring for patients in the years ahead. An audience of almost 90 people from across our continent enjoyed the atmosphere of the Dystonia Europe family, the excellence of the speakers and the social events.

The speakers and the programme
During the conference, more than twenty internationally-respected dystonia experts from across Europe gave presentations spanning those twenty years. The conference extended over two days and, in the first session, we heard about the history - the prevalence of the illness and the development of medications, surgery and botulinum toxin. In the second, presentations described the considerable advances of research into areas such as genetics, broader aspects - like rehabilitation - in managing the illness, paediatric dystonia and more effective use of botulinum toxin by better training of injectors.
A third session, covering more practical topics for patients, dealt with issues of pain, psychological factors, the benefits of teamwork in the clinical setting, patient advocacy, physiotherapy and the patients' expectations of the doctors who are treating them. A programme with interest for all.
Speakers' abstracts and programme can be found on the DE website:http://dystonia-europe.org/activities/events/past-events/

The 20th Anniversary Dinner
All guests were given a real Scottish welcome, with bagpipes and Scottish country dancers at a reception, followed by a very fine dinner in the impressive main hall of the Royal College of Surgeons of Edinburgh. As a 'digestif', post dinner, we had a private visit to the famous Surgeons museum. Fascinating, even if a little grisly.....

On a personal note.....
These events took place on 'home' ground for me, and I was very happy that my wife Pat and our two daughters, sons-in-law and grandson could attend the reception, to share such a wonderful evening with so many of my friends from the world of dystonia.

Former Executive Director and founder of EDF; Alistair Newton, together with his wife Pat, daughters Claire and Jill and grandson Harris.

At the dinner, I was greatly privileged to be presented with the certificate of Honorary Membership of Dystonia Europe and a generous gift subscribed to by Dystonia Europe members and individuals. I'm very grateful to all who contributed to this, and for the several individual gifts I received in Edinburgh.

I particularly want to thank all the Dystonia Europe Board members and officers, past and present, who have given their time and energy to support and guide the organisation during its twenty years. The 20th Anniversary events showed clearly that Dystonia Europe is in good shape and I hope that there will be an invitation to former Presidents for the next anniversary celebrations in 10 years' time!

Alistair Newton
Board Member
Dystonia Europe

Photo Gallery of the 20th Anniversary Conference in Edinburgh

Latest News


DYSTONIA
EUROPE
20th Anniversary
Congratulations
1993~2013

Latest News from the GA in Edinburgh

Dystonia Europe Board 2014

At the last GA in Edinburgh there were some changes to the Dystonia Europe board. Monika Benson, stepped down as President after serving the maximum period of 6 years. Robert Scholten, President of the Dutch Dystonia Association, was elected new President of Dystonia Europe. Other new members to the board are: Treasurer Heike Wolf from Germany and Secretary Merete Avery from Norway. Alistair Newton retired from the position as Executive Director and is now on the DE Board with special responsibility for the Dystonia Research Network. Board members who continue are Vice President Maja Relja from Croatia, Marina de Koning-TIjssen from the Netherlands and Sölvi Engeland from Norway. Monika Benson has been appointed Executive Director.
Read more about the board members on page 35.

First row from left: Robert Scholten, Maja Relja, Monika Benson, Alistair Newton and Sölvi Engeland. Second row from left: Marina de Koning-Tijssen, Merete Avery and Heike Wolf.

Flowers for Monika Benson after her last Board Meeting after 6 years as President of DE.

Honorary Membership to Alistair Newton Founder of EDF

At the GA in Edinburgh Alistair Newton was honored a Honorary Membership of Dystonia Europe for his long-term dedication and commitment to work for dystonia patients and their families.

It all began more than two decades ago. In the spring of 1992 Allergan approached Alistair, who was then on the board of TDS (the Dystonia Society), to take part in a satellite symposium of ”the Second International Congress of Movement Disorders” with the title ”Reach Out – a practical workshop for those in patient support”.

Participants were able to select and attend workshops sessions on: ”Media/Publicity”, ”Communications/Newsletters/Keeping in Touch”, ”Running a Dynamic Meeting” and ”Telephone Counseling.” The meeting took place in Munich in June and was the starting point for trying to create a European Dystonia Patient Organisation. Half a year later another meeting was held in Barcelona where seven dystonia patient groups met and discussed how to move forward with the idea of a European Federation. The countries that attended were France, Germany, Italy, the Netherlands, Spain, Sweden and the UK. Later in 1993 the European Dystonia Federation was founded in Spoleto in Italy and Alistair Newton was the first President.

20 years - 20 Member Organisations

At the 20th Anniversary we achieved the goal of representing 20 European dystonia patient organisations. Two Romanian groups had applied for membership of DE and by the approval of the GA we reached 20 member groups. There is already a membership application submitted from the Finnish Dystonia Association for this year's GA. It is exciting to see how the Dystonia Patient Network is growing in Europe. Can we be 30 member organisations for the 30th Anniversary?

Membership history

1993 – 11 dystonia patient organisations from 11 countries (formed EDF)
1999 – 16 dystonia patient organisations from 15 countries
2014 – 20 dystonia patient organisations from 17 countries

MEMBERS OF DYSTONIA EUROPE 2014

- Austria
- Belgium
- Croatia
- Denmark
- France
- Germany
- Ireland
- Italy
- Netherlands
- Norway
- Poland
- Portugal
- Romania
- Spain
- Sweden
- Switzerland
- United Kingdom

DYSTONIA EUROPE

Some figures and facts from the past

Country	Founded	Members
France	1984	350
Germany	1990	340
Italy	1992	200
Spain	1991	70
Sweden	1990	79
United Kingdom	1983	1400

From Meeting minutes of the Swedish Dystonia Association March 1993.

Welcome to the Two Romanian Patient Organisations

The last member organisations to join Dystonia Europe are: **"Asociatia Children's Joy"** led by Chairwoman Sabina Gall and **"Asociatia Distonia"** led by Chairman Sorin Ionescu from Romania. Both organisations work to raise public awareness of dystonia, to provide support and advice to dystonia patients and their families, and to promote research.

We welcome both organisations to the Dystonia Europe Family, and we look forward to working with them.

Monika Benson with Presidents Sabina Gall and Sorin Ionescu.

Dystonia Europe at WCN 2013 & EFNA General Assembly in Vienna

The 21st World Congress of Neurology was held in Vienna last September. 6300 participants from 135 countries had come to the Austrian capital for the world biggest neurology event to take part in presentations, meetings and work-shops on various topics within neurology. Dystonia Europe had a booth in the exhibition hall where President Robert Scholten and Executive Director Monika Benson received many visitors who were interested in hearing about the work and activities of Dystonia Europe. Leaflets and printed copies of the DE News Summer 2013 were handed out. Robert Scholten had brought his camera and people were asked to make a "Jump for Dystonia" in front of the Dystonia Europe banner.

President Robert Scholten and Executive Director Monika Benson at the WCN exhibition hall in Vienna.

The activity created many happy and energetic moments that will be part of a future dystonia awareness campaign planned for 2014-2015. The next World Congress of Neurology will be held in Chile in 2015.

Alongside the congress EFNA held its annual General Assembly 2013. DE is a member of EFNA and was represented by President Robert Scholten and Executive Director Monika Benson. New members that were accepted by the GA were: International Bureau for Epilepsy, European Mysathenia Gravis Association, Pain Alliance Europe (associate), and Danish Trigeminal Neuralagia Association (associate). EFNA now has a total of 19 member organisations within the field of neurolgoy. The next EFNA GA will be held alongside the EFNS Congress in Istanbul in May this year.

Boston Scientific receives CE Mark approval for deep brain stimulation in treating dystonia

The American medical device company Boston Scientific has received regulatory approval for the use of deep brain stimulation (DBS) for managing primary and secondary dystonia. For further information see the following link: http://www.prnewswire.com/news-releases/232457721.html
Boston Scientific is an American medical device company and their products and technologies are used to diagnose or treat medical conditions, including heart, digestive, pulmonary, vascular, urological, women's health, neurological and chronic pain conditions.
Dystonia Europe and Boston Scientific met outside of Paris shortly after the announcement of the CE Mark approval, and an agreement for a 2-year partnership was signed.

Dystonia Europe and Boston Scientific Teams meeting in Paris.

Dystonia Europe in Norway

The Norwegian Dystonia Association organizes an ”autumn meeting” for their members every year during a weekend in October. This year the meeting took place in Trondheim. Over 80 members had come to the beautiful city in the north to take part in the one-day event. The President Anniken Hagen welcomed everybody and informed about the latest activities of the organisation.

Anniken Hagen, President of the Norwegian Dystonia Association.

The highlight of the year was the ”Run for Dystonia” event that took place in Oslo end of September.

The Norwegian dystonia organisation had gathered about 30-40 people who all were wearing t-shirts with the dystonia logo while running or walking.

The positive thing was that there was quite a lot of attention given to the event in various newspapers and magazines, creating more awareness of dystonia in Norway. Plans are already made to participate this year’s Oslo Marathon.

Monika Benson was invited to the meeting to speak about Dystonia Europe and its activities. Other speakers were a Norwegian speech therapist who spoke about how the vocal cords can be affected when having cervical dystonia and how important it is with correct breathing. A member shared her story of dystonia and how she has dealt with it.

In the afternoon the members were given the possibility to participate in a Pilates session held by the NDF Chairwoman Anniken Hagen or to go on a guided tour in Trondheim. In the evening there was a dinner for everybody at the hotel, a nice opportunity for people to meet and talk about dystonia and so many other interesting things.

Monika Benson

Dystonia Europe Training School 2013

The Dystonia Training School 2013 was held at the National Hospital for Neurology and Neurosurgery at Queens Square in London. Read more on page 14 and 15.

Managing Dystonia in Children

Over the last 20 years there has been a growing perception that dystonia is under-recognised in children and that dystonia has strong developmental origins involving basal ganglia, cerebral cortex and cerebellum.

Edinburgh is one of the paediatric centres with a long tradition of attempting to characterise movement disorders in children, recognising existing limitations of our understanding of the mechanisms of hyprtonus and measure the contributions of dystonia and spasticity to movement patterns in children. It is therefore fitting that the Dystonia Europe 20 year celebrations, reflecting on what has changed in our teatment of dystonia in children should take place in Edinburgh.

TTS Ingram was a pre-eminent Edinburgh clinician who made a major contribution to our understanding of cerebral palsy and like his successor, J Keith Brown was deeply influenced by the work of Denny Brown and the growing interest in the functions of the basal ganglia.

The first half of the 20th century saw the rise of orthotics and braces to attempt control of body alignment and function in children with movement disorders. Yet none of these methods have been shown to prevent fixed deformity. Consequently, it has been claimed that the second half of the 20th century was the 'orthopaedic era' of management of cerebral palsy, culminating in the development of gait analysis laboratories to help in 'planning' for multi-level orthopaedic procedures, often involving bilateral femoral and tibial osteotomies and muscle lengthening procedures, requiring simultaneous adjustment of 8-12 muscles chiefly for the 'spastic phenotype' due to the recognition that orthpaedic procedures are less predictable and thus less successful in children with dystonia or dystonic chorea.

Selective dorsal rhizotomy (SDR) has been hailed as very successful for children with spastic diplegia, but not dystonia. Paradoxically many centres abroad are quietly abandonning the procedure because of the profound permanent weakness which the procedure uncovers, whereas in the UK, SDR is enjying a 'renaissance'.

These orthopaedic and neuro-ablative surgical procedures have, at the very least, required increasingly careful evaluation of the nature of the underlying movement disorder, measurements of function and quality of life.

Botulinum toxin became a very important tool for relieving muscle spasms in the early 1990s. Although heralded originally as a means of preventing contrature in 'spastic ' (probably hyperexplectic) mice, randomized controlled trials of botulinum toxin injections with and without splints or orthoses have not demonstrated prevention of contracture in children with cerebral palsy.

Deep brain stimulation (DBS) has come at an interesting time in this field, when all these 'older methods' of management have been found wanting.

The singular success of DBS for genetic dystonias has led to a renewed interest in the motor neurophysiology of the brain and concepts of refining motor focus with high frequency DBS. Defining the timing of intervention as well as new targets for DBS may help a larger number of children affected by cerebral palsy, by reducing the proportion of life lived with the severest forms of dystonia. Thus a convergence of strategies designed for very selected motor disorders may be relevant for a wider group of dystonic disorders.
This should be interpreted in the context that recovering lost motor skills is quite a different process from developing previously non-existent motor function.

Other promising avenues are the effective management of pain in dystonic disorders and managing the neuroinflammatory component of encephalitis which produces receptor antibodies often associated with severe movement disorders.

The real challenge to these earlier palliative strategies is to harness advances in neuroimaging, clinical genetics, neuropharmacology and neurosurgery to improve our early therapeutic strategies for children with movement disorders dominated by dystonia with the aim of altering an otherwise grim and un-remitting natural history.

Perhaps we can look forward to the 21st century as the neuroplasticity, neurophysiological and neurosurgical era of management of dystonia in children.

Jean-Pierre Lin
MRCP(UK) PhD
Consultant Paediatric Neurologist
Complex Motor Disorders Service
Evelina Children's Hospital
Guy's and St Thomas' NHS Trust and King's Health Partners, London.

PhD Jean-Pierre Lin and Prof. Alfredo Berardelli

News from the Dystonia Research Network

COST Action BM 1101 - The European Network for the Study of Dystonia Syndromes

Most of our readers will have heard of the research network which has been formed recently in Europe, with the support of a grant from the inter-governmental organisation "COST". The name is derived from "Co-operation in Science & Technology" (www.cost.eu)
COST was formed by interested Governments in 1971. Currently, 35 countries are COST members, although other countries regularly become involved in the activities of the networks.

COST provides grants to help researchers from different European countries to work together and achieve better collaboration in focussed international networks. It does not, however, provide grants for research projects, and the rules for applying the funding are very strict. Most of the principal dystonia specialists in Europe are actively involved in the activities of the Network, and well over 200 scientists and clinicians are associated with the different Workgroups.

A Management Committee (MC), chaired by Prof Alberto Albanese, Vice Chair Prof Marie Vidailhet, meets once each year, while a smaller Steering

Committee holds more frequent discussions to guide the functioning of the Network within the framework agreed by the MC. Dystonia Europe was elected by the MC to act as the Grant holder and administrator of the network.

The core of the network is centred on the 4 Work Groups:

WG1 – Genetics
WG2 - Animal models
WG3 - Clinical studies
WG4 - E-Infrastucture

These groups also interact with one another on topics of joint interest.

Currently, the dystonia network is near the end of the second year of a four-year grant period, and the successes so far have been in two main areas of activity - Training Schools and "Short-term Scientific Missions".

Training Schools - Two very successful training schools have been held, with 35 young scientists and clinicians in Bol, Croatia - alongside Dystonia Europe's 2012 Annual General Assembly, and in London in November, 2013, with over 40 trainees. The programmes at these events have included lectures on aetiology, genetics, clinical aspects, basics of organising research projects, psychological effects on patients, rehabilitation, etc.., as well as 'hands-on' experience in examination of patients and diagnosis of the many different presentations of dystonia symptoms. The trainers are dystonia experts from among the most respected specialists across Europe, and their time and effort, as always, are given freely The COST funding allows us to provide grants to young clinicians and researchers as support for their travel and living costs in attending these schools. Already, the opportunity to learn about dystonia and its research is attracting a lot of attention, and 120 applications were received for the 48 places available in London last year.

Short-term Scientific Missions (STSMs) - The Network can also offer grants to young researchers for short-term assignments on specific projects at research laboratories in other countries, where a senior dystonia specialist provides facilities and supervision. Six grants were awarded in 2012 and 7 more in 2013. A call for grant applications will be made in 2014.

European Dystonia Registry - After considerable early planning, work is starting in earnest now in the Network, to create and develop a unified European Dystonia Registry. This will have two objectives: the implementation of a minimum clinical data set to serve as a basis for genetic research; and the creation of a more extensive clinical data set, to serve as a basis for clinical care and data for future studies. These studies will focus on varied clinical phenotypes, information on treatment procedures and pathogenesis of dystonia.

Through these initiatives, many young scientists are being encouraged to work on projects in dystonia and to develop a long-term interest in the condition. This can only improve the opportunities for dystonia patients to be able to access better treatment in the years ahead.

More information can be found on the project website: www.euro-dystonia.org and on the Dystonia Europe website: http://dystonia-europe.org/research/the-european-network-for-the-study-of-dystonia-syndromes/

Alistair Newton

David Marsden Award Winner 2013

Monika Benson and the DMA Winner Dr Katja Lohmann.

Dr Katja Lohmann of Institute of Neurogenetics at the University of Lübeck in Germany was awarded the 6th David Marsden Award at the Dystonia Europe 20th Anniversary Conference last September in Edinburgh.

The Award of € 10,000 is intended to encourage research into dystonia in all European countries, especially by young scientists. Dystonia Europe is grateful for this award so generously sponsored by Ipsen.

After receiving the award from Dystonia Europe President Monika Benson, Dr. Katja Lohmann presented the results of her research.

We wish Katja and her team all the best in their continued research within the field of dystonia.

Monika Benson
Executive Director

Elucidation of the Genetic Cause of DYT4 Dystonia - "Whispering Dysphonia"

Abstract

Some forms of dystonia have an inherited cause, especially when dystonia starts early in life. To date, a list of 25 genetic forms (referred to as DYT1 to DYT25) has been established and causative genetic variants (mutations) have been identified in 15 forms. The latest member of this gene list represents the beta tubulin 4 gene (TUBB4/TUBB4A) in which mutations lead to DYT4 dystonia. DYT4 dystonia is characterized by generalized dystonia and whispering dysphonia. It was originally described in an Australian family in whom patients were able to speak and shout during sleep but not when awake. In collaboration with an Australian neurologist, we evaluated this family, mapped the disease causing gene and finally identified the disease causing mutation. The same mutation was also found by researchers in London who had access to other samples from the same Australian family. We are now investigating the role of TUBB4 which codes for an important component of the cytoskeleton. Our research will further contribute to a better understanding of the disease mechanisms underlying dystonia.

In 1886, a young man and his two sisters originating from a small village in Norfolk (England) immigrated to Townsville (Queensland, Australia). Over the years, the two sisters developed an unusual disease and got many offspring, several of whom also developed a disorder. James Parker, an Australian psychiatrist came across this family in ~1960 and noticed that there was an inherited "most unusual speech disorder" in this family. Patients were able to shout and yell when emotional, had no trouble communicating after drinking alcohol and talked normally in their sleep, yet when they tried to speak their voices came out only in a faint whisper.[1]

About ten years ago, Dr. Robert Wilcox, a neurologist located in Adelaide by chance saw one of the living family members with whispering dysphonia in his botulinum toxin clinic (Figure 1). Doing a detective work, he got in contact with almost 20 family members spread all over the east coast of Australia.[2] He shared the clinical information and the valuable DNA samples with the research team of Prof. Christine Klein and me at the University of Lübeck in Northern Germany.

The search for the disease causing mutation in such a family is quite challenging since the human genome consists of 3,000,000,000 base pairs. To find the one altered variant among all this bases is as difficult as finding the needle in the haystack. In search for the disease-causing gene, we first performed a genome-wide linkage analysis. Using this approach, we figured out that there was a region on chromosome 19 shared by all affected family members but not present in any of the healthy relatives. This gave us some hint that the genetic variant causing the disease was somewhere located on the top of chromosome 19. However, there were so many genes in this region that it was like having found the right street in London but still looking for the apartment of Mr. and Mrs. Smith. Luckily, genetic methods to detect genetic mutations had recently improved and so called next generation sequencing had become available. Combining this method together with our knowledge about the location of the mutation, we were able to identify a single nucleotide substitution in a gene called beta tubulin 4 (TUBB4/TUBB4A) as the cause of the disease in this large Australian family (Figure 2).[3] The same mutation was also found by researchers at the Queen´s in London.[4] The mutation resulted in less expression of the gene leading to less functional gene product.[3]

Next, we screened about 400 additional patients with diverse forms of dystonia. In one patient referred to us by Dr. Marina de Koning-Tijssen from Groningen, The Netherland, we detected another mutation. Interestingly, the patient also had severe problems with her voice (spasmodic dysphonia).

Beta tubulins (there are several of this in a cell) are part of the cellular scaffolding (cytoskeleton) and are mainly involved in transport of certain cargos within the cell. The exact function of beta tubulin 4 is largely elusive but it is conceivable that it is involved in maintaining the shape of the cells, guiding cell migration, division, and differentiation during development, or being important for transport processes within the cell.

To find a mutation in such an important gene in DYT4 dystonia was quite surprisingly and expanded our knowledge on the pathophysiology of dystonia. The pathophysiology of dystonia is still poorly understood due to different reasons including limited knowledge on the genetic basis of the disease and lack of models. Identification of a mutation in a beta tubulin adds a new perspective to the pathophysiology of dystonia. Further research is now necessary to understand the pathways in the cell that are disturbed by the TUBB4 mutation.

We have started to investigate how the detected mutations disturb the proper function of the cell. In addition to established cell lines, we will use easily available skin cells from patients to study the mutation in their “natural” surrounding. However, since dystonia is a neuronal disorder, skin cells may not fully reflect the processes that take place in the brain. Therefore, we will use a newly developed method that was awarded with the Nobel Prize last year to the Japanese scientist Dr. Yamanaka, i. e. reprogramming of skin cells in so called induced pluripotent stem (iPS) cells. His method has been well established in our institute at Lübeck University. These cells can then be differentiated in neurons. We will test for the stability of TUBB4 and investigate its localization within the cells. Further, we will study the

structure and function of the entire cytoskeleton. The overall goal of our project is to reveal the disease mechanism(s) underlying this new genetic form of generalized dystonia recently identified by our group. Given possible shared underlying pathways, these results may also impact on other forms of dystonia.

This ongoing research is being performed in an excellent scientific environment (www.neurogenetics-luebeck.org). With an interdisciplinary team of movement disorder specialists, clinician and basic scientists, study nurses and research technologists, PhD and medical students closely collaborating, the research at the Institute of Neurogenetics at the University of Lübeck is translational in nature and directed from the patient to the bench and back (Figure 3). In close collaboration with the Departments of Neurology, Pediatrics and Psychiatry and the Institute of Human Genetics, we offer specialized outpatient and inpatient care. Main research methods include molecular neurogenetics, functional analyses of proteins and molecular pathways using cellular models and, most recently, animal models. Patients with genetic movement disorders undergo multimodal neuroimaging and electrophysiology studies and frequently participate in long-term follow-up or prospective clinical-genetic studies. The Neuropsychiatric Epidemiology Working Group takes research questions to the population level, designs and maintains the Institute´s databases, and provides statistical support. The Institute is engaged in a large network of national and international collaborations and actively participates in data and sample exchange. With visiting scientists and students from many different countries and members of the Institute gaining experience abroad, we are well connected on the world map.

Katja Lohmann
Institute of Neurogenetics, University of Lübeck, Lübeck,
Germany

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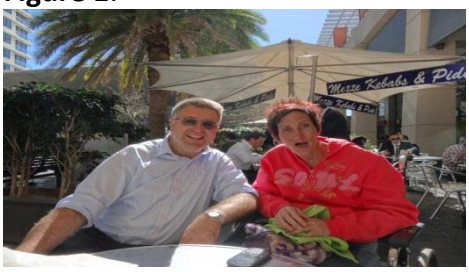
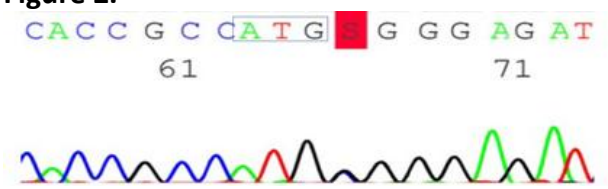
Figures

Figure 1.



Dr. Robert Wilcox (left), the Australian neurologist who clinically evaluated the DYT4 family together with a patient from the family.

Figure 2.



The DNA sequence of the mutation in the beta tubulin 4 gene. Each colored peak represents one base pair and each color represents one of the four different units of the DNA (A, T. G. or C). The gene starts with the ATG (boxed). The mutation is indicated by the red boxed letter “S”. At this position, there are actually two overlapping peaks: A blue one for “C” and a black one for “G”. In healthy individuals there is only a “G”.

Figure 3.

The team of the Institute of Neurogenetics at the University of Lübeck.

Dystonia Coalition - 5th Annual Meeting

Research Priorities for Laryngeal Dystonia
The 5th annual meeting of the Dystonia Coalition was held in Atlanta in October. The coalition focuses on a different type of dystonia at each meeting and this time it was the turn of laryngeal (voice) dystonia. The Dystonia Coalition is an international partnership between patient groups and dystonia treatment centres – so far 17 patients groups and 46 treatment centres are involved.

Diagnosis of Laryngeal Dystonia (LD)
One of the most important challenges in the treatment of dysphonia (voice problems) is the correct diagnosis of what is causing the problem. Dysphonia can result from a number of conditions of which **laryngeal dystonia (LD)** is one. Another is **muscle tension dysphonia (MTD)** which is a functional problem with an emotional or behavioural rather than a neurological cause. There are a number of methods used to distinguish LD and MTD of which one is to listen to the voice - dystonia has been thought to have a spasmodic effect on the voice, causing voice breaks, while MTD has a more continuous effect.

The ability to diagnose these conditions is important for both treatment and research. Most cases of LD can be treated using botulinum toxin but usually this is not effective for MTD which is mostly treated using voice therapy. Also, for research, it is important to ensure all participants have the same diagnosis to avoid confusing the results. For these reasons, the Dystonia Coalition has run a project to develop a new diagnostic tool. The results were presented by Dr Christy Ludlow.

They initially identified that there is a lack of consistency in diagnosis with different doctors diagnosing the same symptoms in different ways. One possible reason is that people with LD develop MTD as a result of trying to cope with their dystonia during the long period pre-diagnosis making the conditions hard to distinguish. Another is that looking for "spasmodic" symptoms to identify LD is not always reliable – for instance, some patients who are known to have LD caused by a genetic form of dystonia do not have voice breaks.

Dr Buz Jinnah suggested the problem may be analogous to neck dystonia, where there is a spectrum of cases depending on how much movement they display – at one end of the spectrum are cases with a lot of tremor and at the other cases that are "tonic" which means the posture is abnormal but without movement. In between are cases that are a mixture of these two types. The same may apply in LD – so some cases may be "spasmodic" while others have a continuous effect on the voice – and this latter type may be very hard to distinguish from MTD.

The Coalition has used a process called DELPHI to develop an improved diagnostic tool. This tool has yet to be fully tested but initial results suggest that, while many cases can be identified as clearly LD or MTD, some cases remain difficult to diagnose. Unfortunately, there is no immediate solution to this. The hope is that brain imaging or other indicators such as temporal discrimination (see below) may provide other means of distinguishing dystonia from other conditions. Further work on this is ongoing.

Treatment of LD

Dr Andrew Blitzer presented information on using botulinum toxin to treat **adductor LD** (the type of dystonia that makes the voice sound strained or strangled). His results showed very good relief in the vast majority of his patients across the botulinum cycle – he emphasised that to get such

good results it is important to be very flexible both in the injection gap and dose. He injects some patients as often as every 6 weeks with very small doses – others have much longer gaps. The size of the effective dose varies enormously between patients. It is not known why this is – one theory is that some people only require a very small dose as it creates a feedback loop with the central nervous system similar to the mechanism of the geste antagoniste.

For those with adductor LD who do not get good response from botulinum toxin a number of surgical options have been tried. Some people get very good results – but the results are mixed and it is not yet known how often the dystonia returns especially for the newer treatments.

Another concern that was highlighted was that, while both botulinum toxin and surgery can be helpful in treating **abductor LD** (the type that causes a whispery or breathy voice), they are not as effective as when treating adductor LD. It was agreed this was a priority for future research.

Causes of LD

Laurie Ozelius presented the latest findings on the role of genetics in causing LD. A number of genes have been identified that can cause dystonia and some of these cause LD as a symptom – however in cases of dystonia caused by the genes identified to date, LD rarely appears in isolation from dystonia affecting other parts of the body. In addition, the proportion of LD cases explained by such genes is tiny.

Sometimes more than one unexplained pure LD case (where LD is the only symptom) appears in the same family tree so there must be other genes causing LD that are yet to be identified. However, no family has yet been found with more than two pure LD cases – and this is a problem as, to isolate the responsible gene, Researchers need several gene carriers in the same family. As often with genetic dystonia research the problem may be one of low "penetrance" - this means that only a small proportion of those who carry the gene actually develop symptoms of dystonia and, as a result, it is hard to find family trees with many cases.

A promising solution to this is to use a test called a temporal discrimination threshold. This is a simple cognitive test in which people with focal dystonia respond less quickly than a healthy control group. Helpfully, it appears that those who carry dystonia-causing genes but who have not developed symptoms of dystonia (this is called "non-manifesting") also perform more slowly on this test – so it may be a quick and cost efficient method of identifying more carriers of the LD gene. This has yet to be proven but, if it is right, then the test may be able to identify enough carriers of the LD causing gene to isolate it. Once the gene is identified, researchers can look what the gene does and explore the mechanisms that lead to LD with the hope this may lead in time to new treatments.

One question is, if many people carry the LD causing gene without developing LD, then what causes LD to appear in some cases. Dr Caroline Tanner has used employment health insurance records to compare the employment and medical histories of people with LD with healthy controls. She found that people with LD were more likely to have worked in professions which require continual use of the voice and also to have been

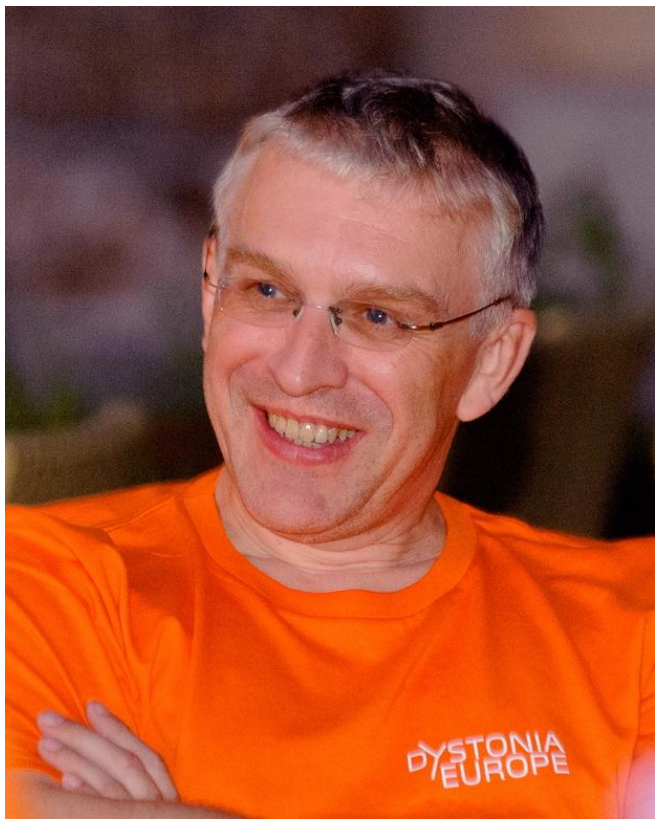
to the doctor with a throat infection. More work needs to be done in this area – one possible route is to identify non-manifesting gene carriers using the temporal discrimination threshold and then compare their histories to those with LD.

Another approach to looking at causes of dystonia is to look for unusual activity in the brain using imaging technology such as MRI scanners. This work has found unusual activity in a number of brain areas: the basal ganglia, thalamus, cerebellum and cortex. These are all areas of the brain involved in the selection and implementation of movement. However, it remains a problem to distinguish unusual activity in the brain that is causing dystonia from unusual activity resulting from the brain trying to correct for dystonia caused in other areas.

Projects of the Dystonia Coalition
Key projects the coalition has been working on include:

- Setting up a **Patient Registry**. This is a registry of patients that can then be accessed by researchers looking to organise research projects. More than 3200 patients have signed up so far – if you interested in signing up you can access the registry in the "About dystonia" menu on our website.
- Developing a **bio-repository**. This is a more detailed database of information about patients and includes blood samples and videos as well as medical history. Information is provided through participating clinics.
- Developing a **new rating scale** to assess the severity of cervical dystonia. This is a development of the current scale. (TWSTRS) to incorporate factors currently not taken into account such as tremor, retrocollis, antecollis and psychological impacts. The new scale performed well in initial tests and now needs to be validated for reliability.
- Providing **grants** for researchers to develop projects.

Twenty research papers have so far resulted from the work of the coalition.



Paul King
The Dystonia Society
United Kingdom

The Netherlands

Four-Minute DVD Speeds up Diagnosis of Dystonia

General Practitioners and physiotherapists often have difficulties recognising and therefore diagnosing dystonia. Very often a potential dystonia patient is referred to a psychotherapist, a physiotherapist or a specialist in pain management, before the proper diagnosis has been made. Unfortunately there are still many patients who have been around for more than 35 years within the medical care without getting a proper diagnosis, which causes great suffering for the individual patient, very often leading to unnecessary pain and other physical discomfort. Dystonia patients often experience a lack of understanding from family and friends. Sometimes dystonia patients even lose their jobs due to misunderstanding by their employer.

To overcome these difficulties in recognizing dystonia the Dutch Dystonia Association has made a four-minute DVD aimed to inform general practitioners and physiotherapists. In four minutes Professor Marina the Koning-Tijssen explains what dystonia is, what types there are and what treatments are available.

In 2013 all 14,000 general practitioners and physiotherapists in the Netherlands received the DVD. In general we got a lot of positive response.

For the Dutch Dystonia Association we consider the campaign successful even if it is only one dystonia patient who is diagnosed by the help of the DVD. Furthermore, in 2014, we plan to send the DVD to rehabilitation doctors and to company doctors. For more information go to: www.dystonievereniging.nl

Recruiting New Members

In The Netherlands there are approximately 18.000 dystonia patients. The Dutch Dystonia Association has around 1.800 members, which is 10% of all dystonia patients. We were wondering if it would be possible to increase the number of members. We launched a recruiting display. At the end of December 2013 we sent the display to all neurologists treating dystonia patients in the Netherlands. We asked if they could place the display in the examination room as well as in the waiting room. Although the campaign just started we got several positive comments already. Some neurologists even offer to hand out the recruiting cards to their patients, which we are very grateful for.

We've already got the first responses from new members. In the next DE News there will be a report on the result of this campaign.

Robert Scholten, President
Dutch Dystonia Association

Germany

Latest News from DDG – the German Dystonia Association

Heike Wolf

In October 2013 the German Dystonia Association hosted a small roundtable talk in Hamburg about so called *"psychogenic dystonia".* Chairwoman Ute Kühn and board member Marion Weiser had invited five medical specialists including one psychologist and one neuro-orthopaedist to discuss this problematic diagnosis which is often considered stigmatizing. In Germany patients with this diagnosis are denied botulinum toxin treatments by health insurances. Many patients get this diagnosis only because no physical cause can be found. However, this is true for most dystonia patients and for almost all of them a more precise diagnosis could be found! The conversations of our guests on this subject were very intense and committed . The doctors reached a common conclusion which will soon be presented in print, so therefore I don't want to anticipate their agreement at this point. However, we will be provided with an abstract in English for a later newsletter.

In July patients suffering from *Segawa-syndrome* met in Freising. This relatively small group suffers from a rare kind of dystonia caused by a genetic lack of dopamine. Children with this disease are often misdiagnosed with cerebral palsy. Patients need life-long medication with levadopa, but usually respond very well to the treatment. A new support group for these patients has now been established with the common goal of collecting data about this rare condition. A questionnaire has been developed by the German Dystonia Association and sent to other national patient groups in English. So far we haven't gotten any replies. If you would like to receive the questionnaire please write to info@dystonie.de. All patients giving us their information will of course remain anonymous. Thank you very much in advance for your help.

On behalf of the German Dystonia Association I would like to wish all our readers a happy new year.

Heike Wolf
Treasurer Dystonia Europe

Professor Dirk Dressler and Ute Kühn

Romania

The Association for Children's Joy and The Neurology Society in Romania appeal to the Romanian Health Ministry to facilitate treatment access for dystonia patients

The fact that dystonia remains an underdiagnosed disease, as well as the difficulties patients have to overcome in order to get access to treatment, were the main topics of the national debate "Dystonia, the disease of the body that won't listen to you" held in Bucharest, last October 1st. The event was organized by Houston NPA for the Romanian Association of People with Dystonia (Children's Joy) and was endorsed by the Romanian Neurology Society.

Dystonia - the 3rd most common movement disorder

Dystonia is the 3rd most common movement disorder (after essential tremor and Parkinson's disease) and it affects the lives of 500.000 people in Europe and more than 300.000 Americans. At the moment there is no cure. The existing treatments; including oral medication, botulinum toxin (for focal dystonias) and deep brain stimulation (for more severe cases of dystonias such as generalized dystonia) only manage to keep the symptoms under control and to offer the patient a certain level of comfort.

In Romania, dystonia is still a highly underdiagnosed disease. Even though the patient association only counts 50 members, experts in neurology claim that the number of patients is much higher. Associate Professor Bogdan Ovidiu Popescu says that he has seen more than 80 patients in the last 8 years: "*And I am definitely not the only doctor in Romania who treats dystonia patients.*" The experts estimate that the incidence of dystonia is 1 patient to 100.000 inhabitants which means that there are approximately 2000 patients in Romania. Unfortunately at the moment there exists no registry of people suffering from dystonia in Romania.

Even among many doctors, dystonia is not a very well-known disease. Even though most focal dystonia patients respond to injections of botulinum toxin, The National House of Health Insurance does not cover the costs of this type of treatment. A single injection costs € 350-400, and the treatment must be repeated every 3 to 5 months.

For generalized dystonias (which affect most muscle groups in the body) the main treatment is DBS - deep brain stimulation. In Romania, this type of surgery is only performed in a couple of specialized medical centers and for Parkinson's disease only, the indication of dystonia has not yet been approved.

Patients, neurology experts and representatives of the authorities around the debate table

The conference titled "Dystonia, the disease of the body that won't listen to you" focused on the administrative, legislative and social obstacles that dystonia patients face, and what treatment options are available. The scientific coordination was provided by Assistant Professor Bogdan Popescu (The Carol Davila Medicine and Pharmacy University), who represented both the point of view of the Neurology Society, as well as the position of the Neurology Comitte of the Health Ministry.

More than 80 people attended the event and 66 of them were health professionals (neurologists, family doctors, psychologists, pediatricians and other specialists). There were 15 dystonia patients and students.

Special guest of the conference was Monika Benson, Executive Director of Dystonia Europe. She spoke about the activities of Dystonia Europe, and also about how the medical system in Sweden works for dystonia patients. Mrs. Benson insisted on the importance of partnership between all stakeholders: the medical profession, patients and their families, the pharmaceutical and medical device industries, and the decision-makers within the public health sector.
The health authorities were represented by Assistant Professor Camelia Margareta Bogdanici, the Vice-President of the National Health and Family Committee in the Romanian Parliament.

Speakers at the conference

Solutions for making it easier for patients to get treatment access
The central theme of the debate was how to facilitate treatment access. Assistant Professor Bogdan Ovidiu Popescu underlined the importance of having the focal dystonia treatment be covered by the National Health Insurance House. He also emphasized the role that the Neurology Committee should play in the administration of this type of treatment: *"Of course, there is the justified fear that one day doctors will prescribe botulinum toxin in appearance for dystonia, but in reality for wrinkles. Most definitely, no ministry and no Health Insurance Company can afford that. But the Neurology Committee can both identify and approve the doctors/neurologists who are able to responsibly use botulinum toxin for treatment of dystonia patients."* In cases of generalized dystonia, when the patient no longer responds to botulinum toxin treatment, the only hope is the DBS procedure. Only 8 dystonia patients from Romania have undergone this procedure so far, and they have all had to go abroad in order to receive the treatment. Both the Neurology Society and the Association for Children's Joy proposed that the national programme for DBS in Parkinson's disease is extended to include dystonia as well.


More than 80 people attended the event and 66 of them were health professionals (neurologists, family doctors, psychologists, pediatricians and other specialists). There were 15 dystonia patients and students.

Appeal to the Health Ministry
Most of the participants signed an appeal written by Children's Joy and addressed to the Health Ministry in Romania. The document emphasizes on the medical and social problems of dystonia patients, and appeals the Health Ministry to support the patients in getting appropriate treatment: botulinum toxin for focal dystonia, by covering the costs, and deep brain stimulation for generalized dystonias, creating the possibility for the procedure to be performed in Romania.

Alexandra Nistoroiu,
Houston NPA, Romania.

United Kingdom

Dealing with Dystonia

Bettina Starke

My experience with dystonia started in early 2008, when I noticed a twist and tilt in my head posture, which I could correct only momentarily before the pull started again. With the help of an osteopath friend I soon found a name for the symptoms online. A few weeks later a neurologist confirmed my suspicion: I have neck dystonia, to be treated with botulinum toxin, with the possible addition of medication. I was told that exercises (my first hope) would make no difference to my condition. I also heard that there was no local patient network which might provide me with answers to so many questions I had. As must happen with many patients when they are first diagnosed with an unusual condition, I felt a frustrating sense of isolation, not being able to have an exchange with fellow patients who were perhaps a few years down the road from me. Soon I travelled to London for the occasional support group meeting, and in 2010 I started a local Dystonia Support Group.

Two years after the onset of my symptoms I found a neurologist whose injection treatment turned out to be effective time after time, but of course only within the usual limitations, i.e. the waxing and waning of the toxin effect over the 13 week treatment cycle.

Around the time of my diagnosis I decided to start swimming again, a form of exercise I had always enjoyed, and a year later I added daily physio exercises, instructed one by one, by a succession of physiotherapists, two of whom had had experience of treating patients with dystonia. Sadly I found no real change, even after two years. Around that time, in my quest to find an added benefit through physio, I met a small number of people with neck dystonia who each reported drastic improvement through daily physical exercise that stabilises the posture muscles of the trunk.

At one of our group events I met a physiotherapist who has first-hand experience with Dystonia: her husband, a fellow neck dystonia sufferer, no longer needs injections through doing daily exercises. She offers one to one consultations and gives weekly Pilates classes for patients, many of whom live with chronic pain. I was told that regaining trunk muscle control is the basis for getting strength back into those muscles which have been weakened out of action and for quietening the dystonic muscles which over-fire. I was also given to understand that, to get any lasting benefit, I would have to commit myself to exercising for two sessions of five to fifteen minutes a day. This involves slow, fluid, gentle but specific movements, often with small muscle effort, initially lying on the back or front, then four-point kneeling, sitting on the floor and a chair, with and without back support, and finally standing. What most of the exercises have in common is a straightness in spinal profile (in sitting or standing, think TALL), concentrating on a correct posture in the neck, chest and lumbar regions, with added attention given to shoulder blade engagement. I also learned that, apart from the daily exercises, I have to bring this new trunk posture into my daily life. This was easily said, and it all made sense as I had never had perfect posture, but I was well aware of how much effort goes into reminding myself continually to correct my sitting, standing and moving about.

This is the most difficult but also the most effective part of my new regime, something that interferes but also accompanies me throughout the day.

A few months after starting the new programme, I noticed a slight easing in the muscles affected by dystonia, and it felt like a fantastic achievement to be given a reduced injection dose as a consequence. Despite this reduction I managed to stretch the subsequent 13 weeks' injection interval by another 6 weeks, with another reduced dose, and more recently I managed to stretch the interval by 10 weeks, again on that low dose. My great hope is to ease the dystonia symptoms to the point where I'll no longer depend on botulinum injections, having regained control over my movement coordination.

What gives me huge motivation is the improvement I have experienced so far, as well as the success stories of some of my fellow sufferers, to whom I feel very grateful. In the class I had joined I am now one of a group of three dystonia patients- an added bonus for each of us, providing a tiny weekly network, receiving support and motivation from our teacher and from each other.

Bettina Starke
United Kingdom

Dystonia Stories

The chair might be empty but the stories are up and running!

Dystonia Stories is the 20th Anniversary Dystonia Awareness Campaign that was launched beginning of 2013. It consists of 20 short video clips featuring interviews with dystonia patients, neurologists and researchers and their different views on dystonia. The last clip **"Twenty Years of Working Together"** was published at the time for the 20th Anniversary. Hear dystonia experts and patients share their views on the past and the future!

The clips are available in English on the Dystonia Europe Website: www.dystonia-europe.org but if you go to the Dystonia Europe Youtube Channel you will also find them subtitled in French, German, Italian and Spanish.

A Dystonia Patient Event in Melbourne, Australia

At the Dystonia Treatment Congress in Hannover in May last year, Dr Lynley Bradnam, from Discipline of Physiotherapy at Flinder's University in Aidelaide, met Dystonia Europe President Monika Benson. Over a conversation about the importance of physiotherapy for dystonia patients and dystonia patient advocacy work, Monika shared that she would be coming to Melbourne later in the year for her daughter's graduation. Verys soon after Lynley's return to Aidelaide the first connections between Dystonia Europe and Australian dystonia patients were made. Lee Pagan and Hariklia Nguyen took the first steps to try and organize a meeting and very soon there were more volunteers involved.

Saturday January 11th about 50 dystonia patients and family members from various places of Australia (Melbourne, Aidelaide, Perth, Blue Mountains, Canberra, Brisbane and Cairns) gathered at the Cerebral Palsy Education Centre in Glen Waverly, a suburb of Melbourne. Sue Kennedy who works for the centre and whose daughter has generalised dystonia, welcomed everybody and introduced the speakers.

Dystonia Europe Executive Director Monika Benson presented the work of Dystonia Europe and the activities of a national dystonia patient organisation giving the Swedish Dystonia Association as an example. Monika concluded with a greeting from Dystonia Europe and its President Robert Scholten with an invitation to the recently established DNA – the Dystonia Network of Australia, to become an associate member of DE.

Dr. Lynley Bradnam, presented her research on dystonia and physio, using and measuring the effectiveness of TMS (Transcranial Magnetic Stimulation) as a method for treatment of cervical dystonia. Lynley is a strong advocate for combining specialised physiotherapy for the best treatment results of cervical dystonia and she showed an interest in developing a programme to educate general practitioners and physiotherapists in Australia about dystonia.

After a short break for coffees and teas and time to meet and chat, the programme continued with Kerry Jackson from the Blue Mountain Support Group, presenting her story of living with dystonia. She shared her painful struggle during many years before getting correct diagnosis and treatment. Laraine McAnally, a nurse with an interest in dystonia, continued and explained the history and background of the various dystonia support groups/organisations in Australia. Kerry and Laraine have now taken the initiative to form a national dystonia patient organisation and just recently, on January 6th the Dystonia Network of Australia – DNA was registered.

The administration Group: Sue Kennedy, Lee Pagan, Nadine Schnabel, Lisa McInnes and Hariklia Nguyen and Monika Benson.

Around Europe & Beyond

Jan Hoffmann of the Melbourne support group and Suzanne Bayliss of the Cairns support group both shared some information about their activities and encouraged the participants to get involved. Lee Pagan who started the Australian national Facebook page encouraged the participants to join the group.

Kerry Jackson, Lynley Bradnam and Laraine McAnally

Thank you to all for coming to the meeting in Glen Waverly and especially thank you to the "Admin group": Sue Kennedy, Lee Pagan, Lisa McInnes, Hariklia Nguyen, Nadine Schnabel for all their hard work and a well organised meeting, which will hopefully encourage and inspire to new and similar events around Australia. We wish the DNA lots of good luck in their work and we look forward to follow the development. Dystonia Europe is very happy to see how dystonia patients in various countries get together to establish dystonia organisation and if we can be of help in this process we will always try and support in the best way possible.

Monika Benson

Other News

Connect with Dystonia Europe

Over the last year more work has been done to further improve the Dystonia Europe website. Our goal is to keep the site updated with the latest news as well as information on dystonia and treatments, past and future activities. Please visit us at www.dystonia-europe.org

For the latest news and to connect with us and others who have an interest in dystonia visit our Facebook page at www.facebook.com/dystonia.europe which today has more than 1100 followers. Welcome!

Belgian Foundation Supports Dystonia Research

In 2013 The Belgian Foundation FDR - Foundation for Dystonia Research - made grants available for collaborative research on primary dystonia and dystonia-plus syndromes. The research should lead to a better understanding of the pathogenesis of dystonia and contribute to improve the diagnosis of the disease and the treatment of persons with dystonia. The research proposals had to be submitted by at least two collaborating research groups with an overall maximum budget of €200 000. Out of 19 research proposals, submitted by a total of 52 research teams from 15 different countries, 4 research projects were selected for funding. Read more about these projects at the Foundation's website: http://www.foundationdystoniaresearch.org

Meet the new President: Robert Scholten

Last September Robert Scholten from the Netherlands was elected new President of Dystonia Europe. Below he tells you a little more about himself, his family and his interests.

"I live in the centre of the Netherlands in the city of Apeldoorn with approximately 155,000 inhabitants. Apeldoorn is located near the woods, where I like to take my dog for walks and to look for deers, wild boars and foxes.

I am the father of two daughters 23 and 19 years old: Ante and Myrthe. My lovely wife Antoinette has also two children: a girl and boy, Frederique and Maurice.

Robert together with his daughters Myrthe and Ante.

My hobbies are playing guitar and guitar building. Unfortunately I have for the time being not enough time to build guitars. Additionally I like to read and study the history of the region I come from. It is my belief that by understanding the past you understand the present better.
I have had cervical dystonia for 14 years. Every three months I get botulinum toxin treatment. In addition I see my physiotherapist every week. I have pain every day and living with dystonia takes a lot of my energy. I also meet a lot of misunderstanding, since a lot of people don't understand what dystonia is. Thankfully I can still work. My work is in the field of marketing and communications. Originally I have an agricultural training and eventually I graduated as a business engineer. I am also the President of the Dutch Dystonia Association which today has about 1800 members.

Since 'Connecting people for dystonia' is my theme for Dystonia Europe for the next few years, it also means that you should easily be able to connect with me!

You can always reach me at:
robert.scholten@dystonia-europe.org

Robert Scholten

Robert and his wife Antoinette.

Antoinette together with her children Frederique and Maurice.

EFNA – Who are they?

When I started getting involved in Dystonia Europe I got quite confused by all the abbreviations of organisations and projects connected to our work at the European level. I shared this with Monika and we decided to start a series in our newsletter in order to demystify some of these abbreviations, especially for our new members, starting with EFNA.
Before answering the question of the headline please allow me to start with a personal, slightly ironic view:

When you are newly diagnosed with this rarely heard of disease, one of the first things you learn is that dystonia does not affect your mental capabilities. Nevertheless you feel like you enter the proverbial land of confusion. Consequently you complete your own special course in neurology and you get credit for knowing all your muscles, the various brands of botulinum toxins and how they differ from one another, the medications, DBS, all known dystonia genes and how they express themselves, to name only a few of our specialties. Why do we do this? We do it not just in order to understand our condition but also to be able to educate the public about our not very well-known disease as well as caregivers, physiotherapists, doctors, and even neurologists. Dr. House would be so proud of us!
In our national patient organisations we are doing our best to help patients in medical, psychological and social matters. We are also expected to spread awareness and to promote research. We have to learn how to influence legislation processes and to raise funds for our activities. The list of responsibilities is endless and too big for people who are most of the time working on a voluntary basis meanwhile battling dystonia in their own private lives. So we're therefore looking outside for help and cooperation.

After the European Dystonia Foundation was founded in 1993, national patient organisations have joined in order to share experiences and to work together at a European level, which made even more sense after the EU had been founded in 1992. But we realize that there is never enough expertise and funding available on a national level as well as European. We are all competing for public recognition, donations and public funding with several other patient organisations both neurological and other kinds of diseases.

In 2000 the European Federation of Neurological Associations (EFNA) was founded as a non-profit umbrella organisation representing pan-European patient organisations within neurology. Their slogan "Empowering Patient Neurology Groups" reflects the work of EFNA as an organisation committed to adding capacity and value to the work of its members, among them Dystonia Europe. EFNA is registered under Belgian law.

EFNA Board together with President Audrey Craven to the left and Executive Director Donna Walsh to the right.

According to the statutes the aims of EFNA are:

- to improve the quality of life of those affected by neurological problems or disorders;
- to promote rapid and accurate diagnosis, appropriate treatment and care for people with a neurological illness;
- to promote better access to information which is accurate and readily understandable;
- to promote awareness and understanding of neurological conditions to the public, the authorities and medical corps;
- to eliminate prejudice and discrimination associated with neurological diseases;
- the representation and defence of moral and material interests of the members of the organisation and this, subsidiary and without prejudice to the principal pedagogical and scientific aims of the association;

The association may realise its objectives in whichever way it deems to be most appropriate. It can, inter alia, organise all activities which contribute to the advancement of neurology and related areas through: the European Commission and the European Parliament; collaboration with health professionals; collaboration with related organisations; realisation of conferences; preparation of guidelines on the optimum management of neurological conditions; collaboration in the organisation and promulgation of international research projects; collaboration in the establishment of registers and other databases of centres of treatment, teaching and research in the neurological conditions; publication of newsletters, a directory of members and other publications including the development of electronic media; campaigning at a European level, assisting in the development of patient support organisations; any other activities the General Assembly may deem appropriate; increase the priority given to neurology by policy and decision-makers and by health care providers.

The association will closely co-operate with the EFNS (European Federation of Neurological Societies), whilst remaining independent. Like in Dystonia Europe, member organisations nominate a delegate to represent its organisation at the General Assembly and to vote on all decisions. EFNA funding arises from a small number of sources, including the pharmaceutical and medical device industries, the European Commission and others.

In February 2013 Monika and I attended an EFNA-workshop in Brussels at which member organisations were informed about the initiative „European Month Of the Brain“. We were given a lot of background information and ideas about what we could do to support our common goals like releasing press statements, branding our own events as part of EMOB and how to use Social Media. A lot of the information was also useful for our work in general. We learnt for example how to write a short and effective article, how to campaign for donations and how to go about lobbying.

To find out more about EFNA, headed by President Audrey L. Craven and Executive Director Donna Walsh as well as their upcoming events also open to member organisations of Dystonia Europe, please go to www.efna.net where you also can sign up for the EFNA E-newsletter.

Heike Wolf
Treasurer Dystonia Europe

Other News

EFNA Hosts Patient Workshops at LSE

Mid September EFNA, the European Federation of Neurological Associations, hosted a 2-day workshop on "Pharmaceutical Pricing, Access and Reimbursement" at the London School of Economics. This important initiative is aimed at equipping participants with the knowledge and skills they need to be actively involved in influencing regulatory decision-making in relation to medical technologies/pharmaceuticals.
The 25 participants representing patient associations from all over Europe, obtained insights on: (a) how pharmaceuticals fit within the overall health care system; (b) how different countries in Europe and beyond pay for pharmaceuticals; (c) what are the most salient regulations addressing coverage/access and how these affect patient access; (d) how stakeholders – including patients – participate in the decision-making process.

Participants at the EFNA/LSE workshop in London.

The course is run by Prof. Panos Kanavos of the LSE. Prof. Kanavos is Reader in International Health Policy and is an economist by training. He coordinates the activities of the Medical Technology Research Group at LSE and works closely with the wider stakeholder community (government, patients, industry). His research interests comprise health systems analysis and reform, health technology assessment and the economics of medical technology. EFNA is planning to organize this workshop again in 2014. For more information on these workshops check out the EFNA website: www.efna.net.

Monika Benson

Events

2014
1-2 February, Dystonia Europe Board meeting, Amsterdam, The Netherlands
21 February, Research Network Working Group meeting in Amsterdam, The Netherlands
31 May – 3 June, EFNS and ENS joint congress, Istanbul, Turkey
7 June, Dystonia Europe Board meeting, Stockholm, Sweden
8-12 June, MDS Congress, Stockholm, Sweden
10-12 October, Dystonia Europe 21st Annual Conference & General Assembly, Paris, France
6-7 November, Dystonia Coalition Annual Meeting, St Louis, USA

2015
14-17 January, Toxins 2015, Lisbon, Portugal

Dystonia Europe 2014

the 21st Annual Conference and General Assembly will take place on October 10-12

at the Hotel Ibis Berthier Porte de Clichy in Paris, France

More information about program and registration will soon be on the Dystonia Europe website: www.dystonia-europe.org

Members

Dystonia Europe consists of 20 national member groups from 17 European countries and they are: Austria, Belgium, Croatia, Denmark, France, Germany, Ireland, Italy, Netherlands, Norway, Poland, Portugal, Romania, Spain, Sweden, Switzerland and United Kingdom.

Donation & Support

All the work and activities of Dystonia Europe is funded by membership fees, individual donations, various grants and organisational 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.

Thank you for supporting Dystonia Europe!

Medical & Scientific Advisory Board

Prof Alberto Albanese – Milan
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Prof Andres Ceballos-Baumann – Munich
Prof Rose Goodchild – Leuven
Prof Marina de Koning-Tijssen – Groningen
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Prof Tom Warner – London
Dr Jean-Pierre Lin – London
Prof Maja Relja – Zagreb
Prof Eduardo Tolosa – Barcelona
Prof Marie Vidailhet – Paris

Board of Directors

Dystonia Europe is governed by a volunteer Board of Directors.

Robert Scholten, President, Netherlands

Robert was elected President of Dystonia Europe at the General Assembly in Edinburgh in 2013. He is from the Netherlands and he is also the President of the Dutch Dystonia Association. Robert is working in the field of communication, marketing and public affairs. He has had cervical dystonia since 2000.

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.

Marina de Koning-Tijssen, Board Member, Netherlands

Marina is a professor in charge of the movement disorders department at Groningen University Hospital and has been committed to supporting the Netherlands dystonia patient organisation for a number of years as a member (and chair) of its Scientific Advisory Board. She was elected to the board of Dystonia Europe in 2011, is a member of our Medical and Scientific Advisory Board and heavily involved in the COST-funded Dystonia Research Network.

Sölvi Engeland, Board Member, Newsletter, Norway

Sölvi was elected to the board of EDF, now Dystonia Europe, in 2009 and she was re-elected for a second term in 2012. Sölvi was diagnosed with cervical dystonia and blepharospasm in 1989. She was a board member of the Norwegian Dystonia Association for 11 years. Sölvi's professional training is within management, strategy, sales and marketing. Sölvi is responsible for the Dystonia Europe Newsletter.

Merete Avery, Board Member, Norway

Merete was appointed to the board of Dystonia Europe in 2013. She was diagnosed with cervical dystonia in 2006 and she was Chairwoman of the Norwegian Dystonia Association from 2010-2013. In 2013 she was elected Secretary to the Board of the Norwegian Dystonia Association. Merete works with administration and customer service for a company in Molde, Norway.

Alistair Newton, Board Member, Scotland

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 has been appointed to the Dystonia Europe Board with special responsibility of the Dystonia Research Network. Alistair also co-funded 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, and has been a board member and Treasurer ever since. Alistair has had cervical dystonia for 30 yrs.

Heike Wolf, Board Member, Treasure, Germany

Heike is a member of the German Dystonia Association and a delegate to Dystonia Europe since 2012. She was elected to the board of Dystonia Europe in 2013. After three years of professional training as a tax consultant assistant Heike has been working within this field in Berlin since 1997. Heike has cervical dystonia.

Monika Benson, 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.

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, EFNS – European Federation of Neurological Societies, EBC – the European Brain Council, St Jude Medical and Allergan.

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