



update

European Dystonia Federation

December 2009

News • Facts • Dates

Dear Friends

It is already mid December and just a week until Christmas, this lovely time of the year for gatherings of friends and families, and the season for sharing and giving. It is therefore a great pleasure for me to share with you this issue of the EDF December Update 2009, filled with interesting articles about ongoing research, developments, activities and dedicated work on dystonia by doctors as well as your fellow dystonia patients – especially those who lead your organisations.

The highlight of the year was, of course, the EDF General Assembly - held in Brussels at the beginning of October. The annual meeting is the big occasion for delegates from all over Europe to get together and share thoughts and ideas, and to socialise. And it is also an excellent opportunity to hear new information about what is going on in the field of dystonia from doctors, companies and your colleagues from other countries. In 2009, the number of people attending the event was over 40, equalling the excellent GA in Vienna 2007. Fifteen member groups out of a possible eighteen were represented, and we missed seeing our colleagues from Belgium, Croatia and Denmark. This year, nine speakers including the two joint David Marsden Award winners, contributed to a scientific program of very high quality. It is my hope that their articles will be of interest and help you in spreading the latest information on dystonia in your country. Some EDF delegates also shared and presented on-going activities in their home countries that you also can read about in this newsletter.

2009 was also the year of the 4th David Marsden Award, which was first presented in 2003. In beginning of September I attended the EFNS (European Federation of Neurological Societies) congress in Florence in order to present the award to Dr. Ana Djarmati and Dr. Susanne Schneider, during the congress session for movement disorder specialists. They were very grateful to EDF for this acknowledgement. It is very encouraging to meet such young doctors already having achieved so much, and being very eager to continue their research for unravelling the mysteries of dystonia. The opportunity to have a central position at this important neurological congress allows EDF to publicise the very positive benefits for doctors who work in partnership with dystonia patients.

In Florence I was also able to participate in some EFNA events. EFNA is the Federation of several European neurological patient organisations where EDF is a member. The co-operation between EFNA and EFNS has grown over the past 10 years and has developed into "Partnership for progress". Since 2006, EFNA has every year at the EFNS congress, organised a session called "The Good Life", with the intention of showing how important the arts, personal interests and education can be in the recovery of patients with neurological disorders.

Being involved in working for people with dystonia is part of my Good Life and I am very grateful to all of you who also dedicate yourselves and your time to this important work. To our Executive Director, members of the EDF Board, delegates, member groups, doctors, advisers, sponsors and donors, I would like to say: Thank you for everything you do for dystonia patients. I appreciate very much the friendship and good partnership between all of us and I look forward to continue working with you in the future. On behalf of the board of EDF I wish you all **A Merry Christmas and a Happy New Year 2010!**

My warmest regards

Monika Benson, President



The New EDF Board

EDF Board 2010

Monika Benson, Sweden
Anthony Butler, UK
Alan Tamlyn, UK
Sølvi Engeland, Norway
Göran Bylund, Finland
Herman De Craecker, co-opted, Belgium

Advisers:

Fiona Ross, UK
Lieve Van Gorp, Belgium
Greet Ruelens, Belgium

European Dystonia Federation

Registered Address: Zweep 14,
B-9991 ADEGEM, Belgium

Executive Director: Alistair Newton

Secretariat: 69 East King Street,
Helensburgh, G84 7RE, United Kingdom

Tel/fax: +44 1436 678799

e-mail: sec@dystonia-europe.org

internet: www.dystonia-europe.org

Recent changes in EDF Board membership

There have been several changes recently in the EDF Board membership and EDF would like to pay tribute to the following, who retired at the General Assembly in October 2009. Annar Hansen came to the end of his term of office after six years of committed effort, for which we thank him most sincerely. He has already offered to assist with individual activities and we will certainly make sure he continues to be involved whenever possible!

Two other Board members found it necessary to resign in October. Anna Moiana, who joined the Board in 2007, has increasing family responsibilities which require her time and attention. We regret her going and wish her well, with our grateful thanks for her work during the past two years. Philip Eckstein, our Secretary, was co-opted to the Board in 2006 and elected in 2007, and has given absolutely invaluable input and support for our activities during that time. Unfortunately, Philip has been forced to resign from the Board because of illness and we send him our sincere thanks and warmest wishes for the future. His efforts continue, however, and he has been instrumental in the preparation of this edition of "Update".

Four new members were elected to the Board at the 2009 GA: Sølvi Engeland, Norway; Dr Göran Bylund, Finland; Alan Tamlyn, The Dystonia Society, UK; and Dr Anthony (Ginger) Butler, ADDER, UK. Sølvi, who is involved in the IT industry, has already taken responsibility for the EDF website and Göran brings to the Board his long experience as a scientist, who has lived and worked abroad. Alan and Ginger have given their services as volunteers for many years, bringing their working career knowledge to the two very successful dystonia patient groups in the UK -

Alan as Vice Chairperson of TDS, which is the oldest and very much the largest and most active dystonia group in Europe. Ginger is, of course, a former EDF Vice President, as well as having gained his PhD in the very specialised field of dystonia epidemiology.

These appointments have re-energised the Board just at the right time, and will help us create the approach we need for the next stage of EDF's development. Monika Benson continues in office as President for another year and then has the possibility of re-election for one more term. Herman De Craecker was again appointed as Treasurer. The Board decided that no other officers should be appointed at the moment, while EDF strategy for the future is being considered.

Advisors - During the past year, the Board invited Greet Ruelens and Lieve Van Gorp to become advisers, particularly in the area of promoting dystonia research. Both ladies have extensive experience in business and are closely involved with our colleagues in the United States, as Board members of the Dystonia Medical Research Foundation. We are already grateful for their extremely useful input at Board meetings and we look forward to developing and strengthening that relationship. At the Board meeting immediately after the 2009 General Assembly, Fiona Ross, Chairperson of The Dystonia Society, UK, was invited to act as the Board's adviser for strategy. This covers a very broad area to assist the Board to identify the key issues which EDF faces, especially in today's challenging financial environment. Fiona is an experienced journalist and TV broadcaster, in the field of national and international politics.

Medical presentations

What is actually happening in the cells that cause dystonia?

By Dr Tom Warner, London, UK

Most cases of dystonia are 'primary' which means that there is no obvious cause and no loss or death of cells in the brain. This is in contrast to many other neurological movement disorders including Parkinson's disease and Huntington's disease, where specific groups of nerve cells die off leading to the symptoms. So in dystonia, it is dysfunction of nerve cells that lead to abnormal processing of movement commands in the brain, and hence dystonia.

A good example of this is dopa-responsive dystonia, a very rare genetic form where the abnormal gene means that certain nerve cells cannot produce enough of the chemical messenger dopamine and this results in dystonic movements. For most other cases the problem is not so clear. There has been much study of DYT1 dystonia, another genetic form of primary dystonia that causes familial childhood onset, generalised dystonia, but also can cause focal adult onset dystonia. The DYT1 gene is the code for the protein torsinA, and studies in cells show that this protein is widely distributed, especially in the area of the nuclear envelope and endoplasmic reticulum (where proteins are produced) in the cells. Abnormal (or mutant) torsinA is redistributed to the nuclear envelope and abnormal membrane whorls or inclusions. These inclusions have also been identified under the microscope in the brains of people with DYT1 dystonia, postmortem.

Another way of understanding the function of a protein in cells is to look at what other proteins it interacts with. We have identified a protein called snapin that interacts with torsinA. Snapin is important for the fusion of synaptic vesicles (packets of chemical messengers) with the cell membrane. The release of these messengers from vesicles is the way that one nerve cell talks to another. Our work has shown that in the normal situation torsinA plays a role in regulating synaptic vesicle turnover, and when there is mutant torsinA, this regulation is lost and this leads to abnormal synaptic vesicle turnover. This is likely to affect the way nerve cells communicate and we believe that it is this loss of regulated neurotransmission that, at a cellular level, underlies the disruption of motor pathways and lead to dystonic movements.

Study in nerve cells is beginning to unravel the mystery of dystonia and further work is needed to clarify exactly how this happens. This will be the first step in designing new ways to correct these problems and find novel therapies for dystonia.

David Marsden Award 2009 – winner: Mutations in THAP1 (DYT6) and generalised dystonia with prominent spasmodic dysphonia: a genetic screening study.

By Dr Ana Djarmati and Dr Susanne Schneider, Lübeck, Germany

Recently, there have been new developments in the field of genetic aspects of dystonia with discovering a new gene that underlies generalised dystonia. This particular form of primary, early-onset torsion dystonia is referred to as DYT6. Unlike in DYT1 dystonia, the symptoms of DYT6 dystonia frequently involve the craniocervical region. DYT was originally clinically described in some Mennonite families.

At the beginning of this year, two mutations in the so-called THAP1 gene have been identified as a cause of DYT6 dystonia. This THAP1 gene encodes the THAP (thanatos-associated protein) domain-containing apoptosis-associated protein 1 (THAP1) that belongs to a family of sequence-specific DNA-binding, cellular factors.

In a recent study by Djarmati et al., published in *Lancet Neurology* in May 2009, the authors investigated the frequency and spectrum of genetic changes in the THAP1 gene and the associated clinical picture (phenotypic spectrum) in their dystonia patients. To do so, the THAP1 coding region was analyzed with latest genetic techniques (qualitative and quantitative screening methods) in a large group of neurologically well-characterised mostly German patients with various forms of dystonia.

Three novel heterozygous changes were identified: two small deletions in the coding region of the THAP1 gene and one double substitution upstream of the coding region. There were no whole gene deletions or duplications. Of these, the two small deletions we detected are likely to be pathogenic, i.e. they are

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predicted to cause a shift in the reading frame and thus lead to premature truncation of the protein. Because the truncated protein is most likely unable to be transported into nucleus and perform there its DNA-binding activity, the mutations are thought to be the cause of dystonia. The clinical picture in these patients carrying the two small deletions in the THAP1 coding region was characterized by early onset. Both had laryngeal dystonia that started in childhood and both went on to develop generalised dystonia. The gene is inherited in an autosomal dominant fashion; and one of the two patients had two family members who also carried the same mutation.

The conclusion of the study was that, although mutations in THAP1 might have only a minor role in patients with different, but mainly focal, forms of dystonia, they do seem to be associated with early-onset generalised dystonia with spasmodic dysphonia. This combination of symptoms might be a characteristic feature of DYT6 dystonia and could be useful in the differential diagnosis of DYT1, DYT4, DYT12, and DYT17 dystonia. In addition to the identified mutations, a rare non-coding substitution in THAP1 might increase the risk of dystonia.

The Reality of Psychological Factors in Dystonia: Not “all in the mind”!

By Dr Mark Edwards, London, UK

A strange assumption has run through much scientific and medical thought over the past few centuries: that the mind and the body are separate and should be studied as such. In fact, evidence is growing that the mind and body are intimately connected (which is not so surprising really). For those interested in how the brain controls movement (and how it may go wrong in patients with movement disorders) the study of “motor cognition” – the way that movement is integrated with the mind, is an area

of growing study. For example, research studies show that thinking about a movement (for example playing the piano) can produce changes in the brain similar to those produced by real practice. We are understanding more about how normal feelings such as the intention to move, and the feeling that a movement was produced by ourselves and not something else (a sense of agency) are generated by specific mechanisms in the brain. When these go wrong, unusual syndromes can occur, for example patients who have suffered a stroke and cannot move one side of the body but deny that they have any problem with movement at all. There are groups of people who experience psychological trauma and then develop physical symptoms (which can be abnormal movements such as tremors, jerks or dystonic postures), but where treating the underlying psychological disturbance with psychological therapy can stop the movement. These so called “psychogenic” movement disorders show how strong the influence of the mind can be on the control of movement.

Dystonia has a difficult history with many examples of patients told that it was “all in the mind”, before dystonia was recognised as a neurological condition. But given the discussion above, we need to recognise that, in common with all neurological conditions, psychological factors do influence the expression of physical symptoms in dystonia, and their management can help improve overall quality of life. If we fail to treat people in a holistic manner, by focusing solely on physical symptoms, then we will fail to treat people in the best way in which we can. We will also be closing our minds to the full understanding of how neurological conditions such as dystonia influence cognitive as well as physical aspects of life for patients. It is likely to be in this deeper and more complex understanding that the true nature of the brain dysfunction causing dystonia will be found, and will move us forward towards improving treatments for the condition.

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Professor Altenmueller



Speakers at the conference

Apollo's Curse: Musician's Dystonia: recent results and new developments

By Prof Eckart Altenmueller, H.-C. Jabusch, Hannover, Germany

Introduction: Musician's dystonia is a task-specific movement disorder which presents as a loss of voluntary motor control in extensively trained movements. In many cases, the disorder terminates the careers of affected musicians. The first historical record, from 1830, appears in the diaries of the ambitious pianist and composer Robert Schumann. As was probably the case for Schumann, prolonged

practice and pain syndromes due to overuse can precipitate dystonia, which is developed by about 1% of professional musicians and in many cases ends their career. Neuroimaging studies point to dysfunctional (or maladaptive) neuroplasticity as one of the relevant pathomechanisms.

Epidemiology: Epidemiological data demonstrated a higher risk for those musicians who are playing instruments requiring a maximum of fine-motor skills. In instruments with different work load for both hands, focal dystonia appears more often in the more heavily used hand. These findings strengthen the assumption that behavioral factors may be involved in the etiology of musician's dystonia. Recent data suggest that hereditary factors may play a greater role than previously assumed. In a pilot study, three families were identified with three index patients affected by musician's dystonia, and a total of seven relatives affected by other forms of focal task specific dystonia. These preliminary findings suggest a genetic contribution to focal task specific dystonia with phenotypic variations including musician's dystonia.

Treatment: Treatment options for musician's dystonia include pharmacological interventions such as administration of trihexyphenidyl or botulinum toxin-A as well as retraining programs and ergonomic changes in the instrument. In a long-term follow-up study, outcome was revealed on average 8.4 years after onset of symptoms in 144 patients with musician's dystonia. The treatment protocol included the following guidelines: Trihexyphenidyl was offered as first-line medical treatment in patients without contraindication. Botulinum toxin-A injections were recommended to patients in whom primary dystonic movements could be clearly distinguished from secondary compensatory movements. Pedagogical retraining comprised a variety of supervised behavioral approaches and was offered to patients with all forms of musician's dystonia. Ergonomic changes were recommended whenever applicable. The aim of ergonomic changes was blocking or circumvention of dystonic movements, e.g., by modifications to the instrument. Outcome was assessed by patients' subjective rating of cumulative treatment response and response to individual therapies. Seventy-seven patients (54%) reported an alleviation of symptoms: 33% of the patients with trihexyphenidyl, 49% with botulinum toxin, 50% with

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pedagogical retraining, 56% with unmonitored technical exercises, and 63% with ergonomic changes. In ebouchure dystonia, only 15% of patients reported improvement.

Conclusion: The results demonstrate that the situation of musicians with focal hand dystonia may be improved significantly. Positive results after retraining and unmonitored technical exercises underline the benefit of an active involvement of patients in the treatment process. Only exceptionally, however, musicians with focal dystonia return to normal motor control using the currently available therapies.

Breaking new ground in DBS

By Jim McGivern, Niel, Belgium

It was a privilege for 3Win, N.V. to be invited to the European Dystonia Federation's 2009 General Meeting in Brussels to present our concept on enhancing the outcomes of deep brain stimulation (DBS) for dystonia. DBS as a therapy for dystonia is a relatively recent, yet exciting treatment. Some patients have experienced astonishing outcomes, others good but not remarkable and yet others little help at all. The cause for the wide gamut of results is not fully understood, however, many researchers believe that increased accuracy in the establishment of the stimulation fields may well be the answer.

Current DBS technology is based on single source pacemaker technology and it has not shown the consistent ability to deliver therapeutic stimulation fields or algorithms to target structures. It is our contention that the inconsistency in results could be minimized if a more sophisticated stimulation system, such as one based on cochlear implant technology were utilized.

3Win is designing such a DBS system based on the advances in stimulation field generation that cochlear implant technology has developed over the past thirty years. We hope to illustrate that the same technology that allows for restoration of hearing to the profoundly deaf to the point where they can enjoy music and easily understand speech, can be transferred to the shortcomings associated with contemporary deep brain stimulation. We feel that by using multiple, independently powered and controlled electrodes, we will be able to create the appropriate stimulation geometries to precisely target the neural structures responsible for dystonia, Parkinson's disease and other neurological disorders. We plan to have our first implant completed before around the middle of next year and look forward to the prospect of improving the field of DBS for those individuals suffering from the ravages of dystonia and other such disorders.

Member group presentations

Austria



Systematic care of Dystonia – self-help work in all Provinces of Austria

This is probably our most important growth project. It is the project on which we have learned a lot. And it is the project that brings the most work.

Austria is a relatively small country of 8 million inhabitants. So we must expect around 16000 people affected by Dystonia.

How to reach them?

Austria extends from Lake Constance to Lake Neusiedl. These are very different landscapes, reaching from 3000 metres high mountains up to Puszta which is already part of the Hungarian Plain. As diverse as the landscapes- also we have learned - are the people. Altogether there are in Austria 9 Provinces (from west to east: Vorarlberg, Tyrol, Salzburg, Upper Austria, Carinthia, Styria, Lower Austria, Vienna, Burgenland)

In 1995 we started in Vienna, then we ventured to Lower Austria and Upper Austria. Then we were invited to the Tyrol. Today, we are planning so that we visit each province at least once every two years.

Theme of these visits is information and motivation.

Namely for the target groups: **Patients** and **doctors** and **healthcare practitioners** (physiotherapists, SLTs, etc.).

The most important part is always a Patients' meeting of about half a day, mostly in the seminar room of a neurological clinic. The number of participants varies here between 40 and 120 people.

The speakers are local doctors, psychologists, representatives of health authorities etc., and of course representatives from the Austrian Dystonia Society. **Patients also give presentations:** they ask questions, they can exchange their ideas and views in a kind of small patient 'party'. They get in contact and share plans. The meetings are friendly and relaxed which reinforces the motto: **YOU are not alone!**

After the meeting, however, our work is not finished. As we are already in that city, we also visit:

- Local radio and TV stations,
- Editors of local newspapers,
- The offices of health insurers, medical associations etc.

When we go home again, our notebooks are stuffed with projects and suggestions, new names, new patients, new people we have found and they want to work with us - and again with new experiences and insights.

To support this work was **in fact one of our most important task** and by itself everything else follows.

Presented and written by Richard Schierl

Finland

The Finnish Dystonia Society has presently about 760 members. There are 19 regional supporting groups covering all parts of the country. A number of special working groups are focusing on different aspects of dystonia all of them aiming at promoting support for dystonia patients and increasing the awareness of the disease in general and among health professionals. An information newsletter is published four times a year.

Training courses for physiotherapists are regularly arranged in connection with the annual meetings of the Society. A list of physiotherapists with competence in dystonia treatment was recently completed. This means that, in all parts of the country, dystonia patients will have information available on where they can find this treatment in their own region. A handout with a broad spectrum of information about dystonia is presently being prepared by a working group led by a neurologist.

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This handout will be distributed to all hospitals with neurological departments. The target group is health care personnel, including doctors.

A programme with dance performances was organised by a young dystonia patient in order to spread information about dystonia. The programme has gained much interest and will apparently be broadcast on one of our main TV-channels. A form of soft water therapy developed and successfully used by a patient with cervical dystonia is gaining increased interest and will hopefully be described on a website soon.

Considerable efforts have been made during the last year in Finland to increase the awareness of EDF and its activities.

Presented and written by Dr Göran Bylund, Finland

Dutch Dystonia Society



Koos Mistrat Haarhuis

In June 2009 the Dutch Dystonia Society presented a document with the terms of quality of care for dystonia patients. These terms were gathered by different members of our Society in several workshops. We did this together with the Parkinson Society, a consultancy company and the Dutch Patient and Consumer Federation (NPCF). This project was sponsored by a governmental organization.

We want to use these terms in our contacts and negotiations with health professionals and organizations, for example public authorities or health insurance companies. This document with the terms of quality of care is a 'living' document and has to be updated continuously.

Another project we started this year is the making of a film on DVD. This DVD we want to use for publicity

and as a source of information for health professionals and dystonia patients. In this film we present, among other things, patients who talk about their experiences, a patient before and after his DBS surgery and doctors who talk about dystonia.

If possible we will also make short films to place on our website and we hope to complete this project in September 2010 or earlier.

Presented and written by Koos Mistrat Haarhuis

Sweden

The Swedish Dystonia Society presented its success with regional networks. Due to the unique shape of Sweden, making it hard for members of our society to meet with any regularity, the board made the decision in 2004 that "organising local networks should take the highest priority".

Since then, nine regional networks have been established. A regional network is managed by one person who arranges and invites the regional members to participate in meetings. There are usually at least two such meetings every year. The meetings are often organised around a presentation by a medical professional, a member's talk about his or her treatment, or just a get together in some local place of interest.

Presented and written by Mats Carlsson

Norway

Mapping children with dystonia (CWD)

Mapping children with dystonia is a cooperative project between Norway, Sweden (Britt Forsberg), Denmark (Mette Spangsberg) and Finland (Christel Bergman).

Background

Children are our future. If they have excellent opportunities for education and free choice as they progress through their lives, most of them will be able to achieve their goals and contribute positively to society as a whole. Unfortunately, this is not always possible.

Member group presentations

When we consider children with dystonia, in relation to opportunities and free choice, it is clear that there can be difficulties. The likelihood of being able to complete primary school and further education MAY be reduced.

Objectives

It appears that no previous mapping of Children With Dystonia (CWD) has been done in Norway or any of the other Scandinavian countries. So, the Norwegian Dystonia Society, in co-operation with dystonia patient groups from Sweden, Denmark and Finland, decided to focus on this relatively rare form of dystonia to increase knowledge about the children who live with this illness.

Main goals

- Mapping children with dystonia
- Increasing knowledge about dystonia
- Increasing the opportunities to make a diagnosis
- Increasing follow-up of CWD
- Cross-border co-operation

Process

All main hospitals as well as the Health and Rehabilitation centres for children are being contacted by e-mail. An information letter is included and a questionnaire is attached to be completed and returned. The data will then be tabulated and conclusions made.

It is expected that the final report will be ready by the end of 2010. In addition to the mapping results, the reports will include professional papers published by Jon Barlinn and Inger Marie Skogseid, Norway, as well as a paper on physiotherapy as a treatment for dystonia by Lena Zetterberg, Sweden, and one on deep brain stimulation by Medtronic Norway.

Sølvi Engeland *Project Manager, Norway*

Presented and written by Sølvi Engeland

COOPERATION CROSS-BORDER



Switzerland



Schweizerische Dystonie-Gesellschaft
Association Suisse contre la Dystonie
Associazione Svizzera contro la Dystonia
Member of the European Dystonia Federation
www.dystonie.ch



Swiss Special Project: Organ Concert for the Benefit of Musicians with Dystonia

The first special thing about the project I want to present here is that it was initiated by someone outside the SDG. In December 2008, Barbara Gygli was informed by Tobias Willi, organist of the protestant church of Pfäffikon ZH, that he had obtained permission from his parish to give a concert the revenue of which was to be used for a project about musician's dystonia. It was already the second time that he decided to make this effort. A similar concert in Windisch (AG) in June 2006 had yielded the sum of almost 2000 €, which was donated to a research project. The reason for Tobias Willi's commitment is that he has seen a close friend and colleague suffer for years - not only from musician's dystonia itself but also, and sometimes even worse, from all the problems it causes, directly or indirectly. His second concert devoted to dystonia took place in 'his' church on 21st June 2009.

To make it even more special, he had decided it should be a musical request programme, or – the German word is more pleasant - a Wunschkonzert, consisting entirely of pieces wished for by the audience, who were invited to write to him what they would like to hear at the concert. Within days after this announcement, he got twice as many wishes as

Member group presentations

he could possibly fulfil! With the assistance of Barbara, he prepared a programme that included explanations not only about the music he would play, but also about musician's dystonia as well as dystonia in general. The church was almost full that evening, and the audience was moved just as much by Tobias Willi's words about his friend and his condition as they were by the wonderful music. Because his parish also made a very generous contribution in the form of an aperitif, it was possible for those who wanted to stay on and get more information. Barbara had various conversations with people who took a genuine interest in dystonia and wanted further information material. The sum of the collection was about the same as in 2006: almost 3000 Fr.

We all felt that this is a very special sum of money, and asked ourselves what would be the best way for as many musicians as possible to benefit from it. After considering possible ideas, we decided that the best was to have a brochure written and published about musician's dystonia, containing also addresses to turn to, and to distribute it as widely as possible among musicians and, perhaps even more important, students of music. Given the international character of the world of music, it is to contain information for all the German-speaking countries so it could be reprinted and circulated more widely than just in Switzerland.

We hope to be able to present it to you next year – but for the moment, above all we want to thank again Tobias Willi for his tremendous effort and generosity. Thank you very much for your attention.

Presented and written by Clara Forero

UK – ADDER (Action for Dystonia, Diagnosis, Education and Research)

This year, ADDER is celebrating its 10th anniversary and, as part of that celebration, has organised several different medical symposia, helping to provide better dystonia information to the various associated medical professions. For a number of years, in the North east of England, ADDER and the local Neurological Patient Alliance have jointly arranged access to dystonia clinics on a regular

basis, for first year medical students. This allows students to recognise the different types of dystonia and has been a very successful co-operation with the Neurological Alliance. It appears that all neurological conditions have about 80% of similar problems and treatments and they only differ in the last 20%.

Regarding diagnosis, we learned a very long time ago that dystonia awareness is much lower among the general public and even among some medical professionals. Despite a prevalence level second only to that of Parkinson's disease, the fact that patients do not die because of dystonia is a prime factor in awareness. To aid faster and more accurate diagnosis, we have specifically targeted the physiotherapists and their speech and occupational colleagues, as dystonia patients are often referred to them after misdiagnosis elsewhere. A number of whole-day training sessions have been organised for them, in association with the local hospitals and universities. As part of an ongoing educational programme we have arranged a number of visits to local secondary schools to tell them about neurological conditions in general and dystonia in particular. This is presented very carefully and does not use any medical terminology but it has been very successful in recent months.

In research, the Epidemiological Survey of Dystonia (ESD) is still progressing although, because the numbers have now reached almost 2,400, the final publication of the book describing these conditions over the past 15 years has been somewhat slower than hoped for. The Genetic Survey of Dystonia (GSD) is progressing well, however, and it is hoped that further results will be published during 2010.

Presented and written by Dr Anthony Butler

UK – the Dystonia Society

The Dystonia Society was formed 26 years ago. We celebrated our 25th anniversary last year in the presence of our Royal patron Princess Alexandra. Since humble beginnings we have grown to an organisation of 3,000 plus members.

The Dystonia Society presently has a Board of 8 Directors. In the UK they are called trustees. In

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In addition we have a Chief Executive, Company Secretary and a full-time Fund-Raiser. We employ 13 people in total, primarily in London, and 2 in Scotland. Most are in part-time employment.

We have 45 Branches throughout the UK which are run by local voluntary committees. The committees organise local events such as garden parties and visits to places of interest which are financed by local fundraising. Local 'mini marathons' are also a popular method of fundraising and there is never a shortage of volunteers.

Local fundraising also helps to support the Society at national level. Examples of such fundraising are the 40 kilometres Rutland Water bicycle ride and the London Marathon 40 kilometres run. Every year we have a national Living with Dystonia Day and an Annual Members Meeting held in London. The Society organises an Annual Prize Draw and the winner is announced during the Members Meeting. The Society also has a range of corporate items for sale which includes Christmas cards.

The Society operates a Helpline and One-To-One Service. The Helpline operates 10am to 4pm Monday to Friday and is available to all who wish to contact the Society. The One-to-One Service is a telephone service which links people who wish to talk to someone with members who have a personal understanding of their condition and problems. There are almost 50 volunteer members who are available to provide this service. The Society provides training courses for those who wish to volunteer for this work.

We also have a Welfare Committee which gives financial support to members who have particular needs which cannot be met from usual sources. Providing certain criteria are met help is usually available as grants from €100 to €1000.

The Society also has a Research Committee that reviews requests for financial support from medical researchers. Such research has to be in the field of dystonia or relevant genetics. If necessary the Society will seek targeted financial support for such a venture.

Every year the Dystonia Society offers 3rd, 4th and 5th year UK medical students the opportunity to learn more about dystonia and win serious prizes. Students are invited to write and submit articles about dystonia to our Research Committee which includes a panel of medical experts. The Jackie Deakin essay competition offers a 1st prize of €1,500 a 2nd prize of €750 and five 3rd prizes of €300. The competition is popular and many worthy entries are received.

Presently, we are financing a research programme in Scotland which will compare standard physiotherapy techniques with the Jean-Pierre Bleton method. This programme will assess both methods applied to the treatment of torticollis and provide information about the comparative success of each method. We are also financially supporting the Oxford Blepharospasm Project which is investigating various visual devices which might alleviate the symptoms of blepharospasm.

Recently we conducted a membership survey to determine the range of concerns and areas of interest of the Dystonia Society's members. We believe this information might be of interest to others, so it is our intention to make the results of this survey available to all member organisations of the EDF.

The Society's website has recently been redesigned to provide easier reading and more information. It can be accessed on: www.dystonia.org.uk

Presented and written by Alan Tarnyn

Events for 2010

EDF General Assembly 2010

September 10-12 *As soon as the city and venue are decided you will be informed.*

- *Next board meeting of EDF*
January 30-31 2010 in Brussels

*A new **EDF Members Directory** was published in spring of 2009. Please contact **Monika Benson** on edf.benson@telia.com if you need any copies and she will send them to you.*

Snapshots from delegates dinner

