



Dear Friends,

First of all I want to express how honoured I feel to have been elected President of EDF. Thank you to all of you for believing in me! As you probably all know, family matters kept me from being in Vienna but from what I have heard, „it was one of the best General Assembly's ever“! A great thank you to my predecessor Didi Jackson and the Austrian Dystonia Society for all their efforts in making this such a successful event.

A few weeks ago I travelled to Hamburg to spend a few days with Didi in order to learn as much as possible about EDF and the work involved in being President. Our Executive Director Alistair Newton also joined us for a day. We had some intense days of work and I understand that there is still a lot more for me to learn. I am now looking forward to meeting my „colleagues“ of the Board for our first meeting in January. I am very happy to be part of the same group as them and I am confident that, all together, we will do our best to do a good job for Dystonia patients all over Europe.

I am very grateful, though, that Didi is still around as „Immediate Past President“ so that I can contact her if I might need advice. I wish her all the best as she moves on in her life and most of all: a great thank you from all of us to her for everything she has done for Dystonia in Europe.

As I am writing this, it is the time of Dystonia Week all around Europe. I know that there are some national groups who are very active this week. I wish you all good luck in your various projects, and we look forward to hearing about them in the next issue of Update. Here in Sweden we have nothing special planned for this week but hopefully we'll have an event this spring.

A few days ago, another Swedish board member and I attended a meeting at the library of the University Hospital of Lund. The library is in charge of organising a series of lectures about different diseases and health problems, and now they have suggested to organise an evening about Dystonia! Neurologists and physiotherapists will be invited to make presentations about dystonia and our Swedish Dystonia Society will also be able to present themselves.

Invitations will be sent out to medical students, nursing schools, all departments of hospitals in the region etc., and the public will be invited through ads in the main newspapers of the area. My friend and I were so filled with happiness and excitement that we practically jumped for joy. Finally something is happening for us! This is a fantastic opportunity to be seen and heard! And, as we all know, it is only by the work of informing and raising the awareness of people, students and professionals that we can make dystonia not so unusual, not so unknown. I hope that everything will work out as planned so that I can let you know more about this event in the next Update!

Autumn is now slowly moving into winter here in southern Sweden. One morning this week I woke up and looked out the window and the ground was covered with a thin layer of snow. Such a peaceful sight! It hit me then that we are rapidly approaching Christmas, this very special time of the year for sharing and giving. I am looking forward for some time off from work and a relaxing Christmas holiday with my family in the Swedish mountains.

I hope that wherever you are you will have a Christmas season filled with peace and joy!

From all my heart to all of you

A Very Merry Christmas and All The Best for 2008!

Monika Benson, President



My name is Monika Benson. Six years ago, at the age of 40, I was diagnosed with torticollis, my head turning left. Since autumn of 2001 I get Botox injections every four months.

In combination with physiotherapy my physical condition has improved so much that I am now able to work approximately 25%.

I work as an administrator at a school in the city of Lund, in southern Sweden, responsible for the school's lecture programme.

I am also a full-time mother to my four wonderful daughters

21, 19, 17 and 13 years old. The three young ones still live at home.

Five years ago I joined the Swedish Dystonia Society and in 2006 I was elected Deputy Chairwoman. Due to my language skills (French and English) I also keep an eye on what is happening on the international level in order to share it with our Swedish members.

I am very fortunate to live in a little village surrounded by wonderful nature where I enjoy to go for long walks all year round. I also enjoy swimming and dancing. My girls and I are very much into cooking and baking. I love the smell of freshly baked bread in my house! I find it very relaxing to read a good book or sit down with my knitting.

Didi Jackson retires from EDF Presidency

Didi Jackson, who has been at the helm of EDF since 2001, retired from the Presidency at the 2007 General Assembly in Vienna in September. She has worked tirelessly for dystonia patients since the early 1990s and has been involved with EDF since its foundation in 1993, first as a delegate, then Treasurer, Vice President and President.

The EDF Board made a presentation to Didi in recognition of her tremendous efforts, and she also received a number of personal gifts from her many friends among the Board members and delegates.

Didi has committed much time and energy to her

work as President and she has been a fine advocate for dystonia patients nationally and internationally. EDF will miss her in her Presidential role, but we are very pleased to know that she will remain involved with the Board in her new position as Immediate Past President. She will also continue to work hard on the arrangements for Dystonia-Europe 2008, the international medical conference organised by EDF which will take place in her hometown of Hamburg next October.

We take this opportunity to thank Didi for all she has done to support EDF and wish her and her husband Barry health and happiness for the future.

General Assembly 2007 in Vienna

EDF's 2007 General Assembly in Vienna in September was voted a great success and delegates enjoyed not only an excellent programme of medical presentations, but also two superb evening social events. The location and services of the hotel, in one of Europe's most beautiful and interesting cities, were excellent and the Board is grateful to Didi Jackson who organised and co-ordinated the conference. We would also like to thank Christa Hafenscher and Richard Schierl, our friends and colleagues of the Austrian group, for their generous local assistance which helped so much to make this year's event so memorable.

Election of New President – The delegates voted unanimously to elect Monika Benson from Sweden as EDF President. Monika introduces herself in the editorial of this edition.

Election of new Board Members – Philip Eckstein, Executive Director of The Dystonia Society (UK), was elected in Vienna as EDF Secretary. Philip has been 'acting' Secretary since the 2006 GA and is already known to many people throughout the Federation. Philip has extensive experience working in various jobs for other UK medical charities before he joined TDS and has played a central role in the development of The Dystonia Society over the past few years.

Anna Moiana, a past Vice-Chairwoman of ARD, the Italian Dystonia Society, was elected as an EDF Board member. Anna has been a member of ARD since 1999 and their delegate to EDF meetings for a number of years. Her blepharospasm has been in remission since 2003 and now she works as assistant to a professor at an Italian national neurological institution in Milan, on various research projects into disability. Anna

teaches foreign languages (she speaks four) and she also works as a freelance translator.

We welcome both Philip and Anna and look forward to working with them to extend the work of EDF in the future.

GA Programme - For the Vienna meeting, Didi excelled herself in organising the programme of speakers and the delegates heard very interesting lectures on a broad variety of topics. The texts of the presentations are printed later in this issue of Update, with

the CVs of the presenters.

A special guest at the GA was Janet Hieshetter, Executive Director of Dystonia Medical Research Foundation (DMRF), USA. As most of you already know, DMRF is the longest established and (by a long way) the largest dystonia patient group in the world. DMRF is an extremely active organisation and raises very large amounts of funding for research every year. EDF has enjoyed good relationships with DMRF in the past and we are very glad that we are again working closely with Janet and her organisation.

Presentations at the General Assembly 2007 in Vienna

DMRF and Dystonia Activities in USA

Janet Hieshetter became Executive Director of DMRF in 2004 and her numerous responsibilities revolve around leading DMRF in USA and Canada in supporting research, with combined budgets of \$ 4 million annually. Janet studied Health Sciences in Kalamazoo, USA, and received her BA in 1980. She also studied one year at the

University of Hannover, Germany in 1979. She has been dedicated to health sciences ever since, with appointments and Board positions in such US organisations as the Osteoporosis Advisory Committee in Texas, Illinois and Massachusetts, the Alzheimer Association and the Baker Demonstration School.

Janet Hieshetter opened her remarks with an overview of the US Foundation. She gave the mission of the DMRF: *To advance research for more effective treatments and ultimately a cure; to promote awareness and education; and to support the needs and well being of affected individuals and their families.*

All dystonia patient organizations are focused on promoting awareness. Janet Hieshetter noted that the DMRF struggles with this and DMRF wants to reach those affected by dystonia but also researchers, especially young investigators, to engage them in dystonia research. She reported on the success of the documentary film, "TWISTED". This 54-minute film, depicting the lives of three individuals living with dystonia, has been a great success in helping unaffected persons in the USA to better understand dystonia. It has also been helpful in reaching those affected by the disorder to let them know they are not alone.

Research continues to be a major focus of the DMRF. The Foundation has provided more than \$22 million (US) for over 400 grants in the past 30 years. "Our goal is – eventually - to close our doors and go out of business," said Janet Hieshetter. "We will only do that by working together – globally – to promote advancements in research."

The DMRF has two research programs: the basic research that supports investigations leading to a bet-

ter understanding of the mechanics of dystonia, and the recently launched translational (*) research program, Cure Dystonia Initiative (CDI), designed to expedite development of a pipeline of more effective therapeutics for the dystonias in the next five years. Janet Hieshetter expressed the Foundation's appreciation to the dedicated people who serve on the Advisory Councils for these programs and to Mahlon DeLong, MD for his leadership in serving as the Medical & Scientific Director of the DMRF.

"The CDI program is a more directed research effort" said Ms. Hieshetter. "The Foundation's Science Officer, Dr. Jan Teller is contracting with investigators to address specific areas of dystonia research, such as assay development and target validation."

The DMRF would like to be able to seize every opportunity to advance the field and is limited only by the amount of funds available. "We want to keep the momentum going," she said.

The DMRF is working with partners in this effort. The US Dystonia Study Group is working with the Foundation in the development of a clinical trial to be implemented in 2008. The DMRF is also looking to the US Government's National Institutes of Health, biotech companies and other dystonia patient organizations as partners. Janet Hieshetter encouraged cooperation among dystonia organizations globally and congratulated the EDF on plans for the October 2008

medical conference in Hamburg.

The DMRF has recently developed a business case to demonstrate to the pharmaceutical industry that dystonia is a disorder that is worthy of their attention. The business case outlines the potential dystonia market. The Foundation has utilized this resource with several small companies to initiate discussions regarding their interest in dystonia research and product development.

"No single organization alone is going to unlock the mysteries of dystonia. We must work together! I ap-

preciate this opportunity to meet with EDF representatives to encourage collaboration. The DMRF is pleased to be a part of this meeting"; she concluded.

(*) "Translational research" describes the focus on 'translating' basic scientific research in the laboratory into practical and useful treatments for patients.

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Deep Brain Stimulation for the Treatment of Dystonia

Professor Volkmann graduated in medicine at the University of Düsseldorf in 1993 and received a degree in medical sciences in 1995, also working on research projects at New York University from 1991 – 1993. From 1994 – 2001 he specialised in neurology and psychiatry, his main interest being in the pathophysiology of movement disorders.

He was among the first neurologists in Germany to build up a programme for the surgical treatment of movement

disorder together with Prof. Sturm in Cologne. In 2002 he joined the Dept. of Neurology at the University of Kiel, where his main project was to establish a research group, mainly on DBS. Recently, he became Professor at the University of Kiel and he has published many important papers on DBS and other topics.

Prof. Volkmann was the neurologist involved in the surgical case described in the article by the patient – Gabriel Horseng.

Primary dystonia comprises a heterogeneous group of incurable movement disorders with variable age of onset, body distribution, and genetic background. All are characterised by twisting, repetitive movements or abnormal postures caused by involuntary muscle contractions. The mainstay treatment of focal or segmental dystonia is botulinum toxin injection to denervate the affected muscles. When this approach fails, because too many muscles are involved, the movement pattern is too complex, or neutralizing antibodies develop, dystonia management becomes notoriously difficult. Drug therapy is mostly unsatisfactory, leaving many patients with profound motor incapacity and social stigma. Anticholinergics have been shown to be of some benefit in childhood-onset primary or secondary dystonia, but are of little use in adult-onset disease. Antiepileptics and antidopaminergic drugs have been used to treat dystonia; however, few clinical studies with small numbers of patients are available and the limited experience does not allow a widespread use. Therefore, surgical approaches have been investigated since the early 1950s for severe and medically intractable dystonia. Based on the idea, that dystonia is driven by abnormal neuronal activity of deep brain structures, circumscribed lesions were placed inside the motor thalamus and the basal

ganglia. These surgeries known as thalamotomy or pallidotomy were successfully used to treat different types of dystonia. However, the procedures were not without risk: Patients could experience short-term contralateral weakness, confusion and impairments in speech, especially if surgery was performed on both sides of the brain. Pallidotomy has not been widely used in dystonia to date. When patients undergo a pallidotomy, part of the internal globus pallidus is destroyed. The globus pallidus is part of the basal ganglia, a system of interconnected brain structures that regulate the information flow of motor signals in the brain. This may help restore the balance that normal movement requires.

A major drawback with both thalamotomy and pallidotomy is that these procedures are irreversible and that tailoring the size of the required lesion is difficult. If the lesion is made too small, symptom control may be incomplete or transient. If the lesion is too large and encroaches upon surrounding brain structures, irreversible side effects may appear.

In the 1990s, researchers began studying deep brain stimulation (DBS) as an alternative to lesioning of deep brain structures. DBS was first applied to control

tremor or symptoms of Parkinson's disease. Meanwhile more than 40,000 patients have been treated with this therapy worldwide for a variety of movement disorders.

DBS is accomplished by implanting an electrode with four contacts into the target area within the brain and connecting it to an internal pulse generator usually located in the chest area. The stimulator settings can be adjusted telemetrically with respect to electrode configuration, current amplitude, pulse width and pulse frequency. DBS has rapidly replaced ablative stereotactic surgery in movement disorders for several advantages: (a) DBS does not require destroying brain tissue, (b) it can be performed bilaterally with relative safety in contrast to most lesioning procedures, (c) stimulation parameters can be adjusted postoperatively to improve efficacy, to reduce adverse effects and to adapt DBS to the course of disease, and (d) DBS is in principle reversible and does not preclude the use of possible future therapies in neurological movement disorders requiring integrity of the basal ganglia circuitry.

Since 1999, almost 100 patients with dystonia had been reported to have a favorable response to DBS, which led the US Food and Drug Administration to approve DBS for dystonia and Parkinson's disease in 2003. The use of DBS in Europe is supported by a CE mark (representing conformity to health and safety standards), which was awarded in 2004.

In patients with dystonia, DBS surgery is usually carried out under general anesthesia, because of the difficulties of immobilizing these patients for the duration of the surgery and because test stimulation does not result in immediate beneficial effects that allow guidance of the implantation. The surgery involves implanting stimulating leads into the therapeutic targets, usually the GPi. A stereotactic frame is used to help immobilize the patient and a stereotactic guidance system is used to insert the leads through one or two small burr holes which have been drilled behind the front hairline. Prior to surgery, magnetic resonance imaging (MRI) is used to identify the precise coordinates of the target areas with reference to the frame based coordinate system. These initial coordinates are further refined by intraoperative micro-electrode recordings and test stimulation. The next stage of the procedure involves connecting the leads to a small device (the neurostimulator) implanted under the collarbone or, occasionally, under the skin on the lower abdominal wall. The neurostimulator sends electrical impulses through the leads into the brain. The physician later uses a hand-held programmer to set the parameters of the neurostimulator to the individual patient's needs. A patient remote control allows modulation of the stimulation strength within limits set by the physician, to turn the device on and off and to control the battery status.

Selecting patients for DBS

DBS is usually reserved for children and adults suffering from primary dystonia (dystonia not caused by birth injury, head trauma, stroke or other brain lesions) which cannot be treated adequately by medication and suffer from marked motor impairment or pain. Disability resulting from dystonia must be severe enough to justify the risk of brain surgery. The implantation of brain electrodes carries a risk of 1-2 % of suffering from serious side effects such as infection, intracranial bleeding or stroke causing permanent neurological deterioration. Stimulation itself is usually well tolerated, except for mild impairments of speech, which can be managed by lowering the stimulation amplitude in most cases. Potential candidates will need to undergo a neurological workup to assess if DBS is an appropriate therapy option. This is usually performed at a centre experienced in DBS for movement disorders on an in-patient basis.

Clinical results

An early case report of a patient with severe primary generalized dystonia noted that bilateral stimulation of the GPi with DBS resulted in progressive clinical improvement of dystonia over a variable period of weeks to months. Two larger controlled clinical studies performed in France and in Germany found significant improvements in dystonia severity within the first 6 to 12 months after surgery. The reduction of symptoms measured by a dystonia movement score was, on average, 50 to 60%. This led to a marked benefit in daily activities (dressing, eating and swallowing, feeding, hygiene, walking, etc.) and quality of life.

The German study was unique, because it compared active DBS to an untreated control group to rule out possible placebo effects: 40 patients with primary segmental or generalized dystonia received a DBS device and were then randomly assigned in a blinded fashion to receive either active neurostimulation or sham stimulation for 3 months. At 3 months, patients in the active stimulation group experienced a significantly greater change in movement score than those in the sham group (39%; $P < 0.001$). This improvement was sustained at 6 months. In addition, patients in the sham group who were switched to active stimulation at 3 month achieved similar benefits after six months of active stimulation. Patients treated with active neurostimulation experienced substantial improvement in all movement symptoms (except speech and swallowing), disability and quality of life. Neurostimulation also significantly reduced depression.

DBS has been used in individual patients with focal dystonias, such as spasmodic torticollis (cervical dystonia) or Meige syndrome. The results have been favourable, but the experience is still limited and controlled clinical studies are currently under way. The role of DBS in secondary dystonia is less apparent. Tardive dystonia (dystonia as a side effect of treatment with antidopaminergic drugs, usually in pa-

tients with psychiatric disorders) seems to respond well to DBS. The experience with dystonia resulting from hypoxia during birth or other forms of brain injury is less consistent and usually inferior to DBS for primary dystonia.

Improvement of symptoms following DBS may occur immediately or within hours or days after surgery. However, the full benefit after DBS may only be observed with adequate neurorehabilitation to mobilize the patient and gradually develop over months. In particular, fixed dystonia may show a delayed and gradual improvement. Sometimes, orthopedic surgery may be required to improve fixed limb deformities or scoliosis.

Conclusion

Deep brain stimulation of the internal pallidum is an effective and relatively safe surgical treatment for patients with severe primary dystonia. The therapy is

still evolving and future studies may help to better define ideal candidates, optimal target location and optimal stimulation parameters. If marked disability results from dystonia and medical therapy has been ineffective, DBS surgery should be considered before secondary complications (orthopedic deformities) limit the potential benefit of DBS. Only a few centres offer this therapy on a routine basis, because pallidal deep brain stimulation for dystonia requires special experience in the neurological management of dystonia and the surgical treatment of movement disorders. Patients interested in this therapy should discuss the option with their home neurologist and ask to be transferred to a specialised centre for further evaluation.

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Back to Normal Life



Gabriel Horseng is a member of the Norwegian Dystonia Society and he travelled to Vienna with his wife Christin to share his experience with the EDF Delegates. His story tells us about him and his journey with dystonia.

Fellow Dystonians and other good folks!
I am glad to be here in Vienna, and I'm very proud to be invited to speak at this EDF-meeting. The reason why I am here is because I have had Deep Brain Stimulation surgery. It is not only the reason why I have been invited – it is also the reason why I am able to attend! Because without the operation I would not have been able to come here and talk to you; my physical and psychological situation would simply not have allowed it.

I got dystonia at the age of 15. It started as torticollis, but within a few years it spread to the whole body and the diagnosis was generalised dystonia. The following ten years, which should have been my golden years, became a nightmare. I had to quit school, quit playing handball and the band I was playing in was suddenly without their guitarist.

Botox-injections had no effect at all and I started drug therapy: Artane, Sinemet, Valium etc. In the beginning the pills had a pretty good effect, but soon I had to increase the daily dose – and again and again and

again, until it was totally out of control. I don't want to complain about pharmaceutical companies here. They do a very good job and help many people.

Then, in the spring of 2002, my doctor at Rikshospitalet" in Oslo told me about the DBS- surgery. Would I try it if I got the opportunity? I said yes there and then. There was no doubt whatsoever in my mind. It was the first glimpse of hope for me since I got the disease. The way I saw it, I had nothing to lose.

DBS-surgery had never been performed on a dystonia patient in Norway before, so it had to be done abroad. It was arranged in Kiel, Germany. So after a year of waiting, my parents and I flew down to Kiel. It was the first day of February 2003, and the operation was scheduled to take place on February 6th. The week before 'D-day' was filled with different tests and examinations. I was also asked to take part in a study – a study which still continues. One element of this study was that the patient would not be told if the power of the stimulator was turned on or not until 3

months after the surgery.

Then, on the day of the operation, I was anaesthetised and I woke up about ten hours later. I felt it right away: something had changed. My body was lying there perfectly still, and I knew right away that the power of the stimulator was switched on! I just started to weep – the most happy tears in my life. An interesting phenomenon, by the way, was that I only spoke in English immediately after I woke up – even to my parents. The first thing I said was: “Daddy, I have to pee...”

During the days after the surgery, I felt like I could fly. My body felt so light. Something heavy was removed. I remember an episode: My mother and I were taking the bus in to the centre of Kiel, it was some days before we returned to Norway. I could sit on the bus-seat like a normal person. Not thinking of my disease. Not using up energy on spasms. Not thinking of ways to hold my head straight. Wonderful!! The following weeks I started to reduce the drug dose, and 2 months later I was off the pills for good.

It has been 4 years now, and the stimulator works perfectly well. I go to the hospital twice a year for control and to adjust settings if needed. In February this year the battery for the stimulator was changed. It was only a small surgical procedure, done under local an-

aesthesia, and the next day I could return home!

So - to the important question: How is my everyday life now, compared with how it was before? It's like night and day. Before I couldn't do much, and everything I did was “sapping” me of energy. It was my own little private hell. Now I can do pretty much what I want to do; I take care of the housework and I act like a normal family father.

To conclude, there are a few things I want to share with you: People often contact me, asking about the DBS-surgery. “Is it terrifying?” “Will the battery be obvious under my skin?” etc. I always end up saying to them: “Do it! Don't hesitate. You got nothing to lose!...”

Ps. There can be nice, but unexpected side-effects coming from DBS surgery.

I found, or more correctly, my wife found me after seeing me on a science program on Norwegian television. She sent me a text-message, and now we are married. I love you, Christin and thank you for being here with me.

To you other good people, thank you for listening!!

Gabriel Horseng
Norwegian Dystonia Society

Drug-induced Dystonia

Professor Ceballos-Baumann was born in Spain, studied medicine in Heidelberg, where he received his medical Licence in 1987. Further studies followed in Nottingham, UK and Illinois, USA. Later, he was involved in the PET scanning project during postgraduate medical training at the National Hospital for Neurology in London.

In 1995 he took up a position in the Technische Universität München and in 1997 he became a consultant neurologist there. Since 2002 he has been associate Pro-

fessor of Neurology. His main interests are dystonia and Parkinson's syndromes, botulinum toxin therapy and DBS.

Since 2004 he has been Director of the Neurological Hospital in München, specialising in chronic neurological conditions such as dystonia, Parkinson's Disease, stroke and rehabilitation with 48 full time staff in physiotherapy, voice therapy and ergotherapy.

He is a member of the EDF Medical Advisory Board.

Drug induced dystonia can be classified into either acute or tardive. Acute dystonic reactions after drugs are usually self-limiting and easy to treat. Acute dystonias can appear after a single dose of certain drugs, but they do not persist and disappear once the offending drug is withdrawn. Acute and tardive dystonia are caused by neuroleptics, which are needed for the treatment of psychoses. Unfortunately they have also been used for treating anxiety and depression and are still prescribed today, even though there are much more appropriate therapies available now. Fluspirilen is a neuroleptic which is given in many

countries quite often as a tranquiliser or antidepressant in injection form associated with the development of tardive dystonias. The odd thing is that many patients are not informed about the sort of drug which is being injected.

Sometimes, drugs similar to the neuroleptics also blocking dopamine receptors in the brain are used for gastrointestinal problems, nausea, dizziness, motion sickness and vomiting. Metoclopramide is the most common dopamine receptor blocker used outside psychiatric indications. Flunarizine and cinarizine are

two other drugs commonly used in southern Europe for all sort of ailments, although there can be side-effects similar to those of the neuroleptics.

Tardive dystonia is a special kind of tardive dyskinesia. Dyskinesia only means abnormal movement, but traditionally implies abnormal movement due to drugs that someone has been taking for some length of time. The most common tardive dyskinesias are chewing smacking movements of the mouth – so called orobuccolingual dyskinesias – and restlessness of the legs or inability to keep still (called akathisia). Stopping or reducing the dose in the course of treatment with neuroleptic drugs typically worsens or even brings about tardive dyskinesia.

“Tardive” means late and implies that these phenomena appear late after exposure to the offending drug. By convention tardive dystonias can appear after treatment with any neuroleptic (dopamine receptor-blocking) agent for as little as three months, even if the drug is stopped, and with a latency of up to three months after the drug has been stopped. However, there are reports of tardive dystonia describing patients who were exposed for less than 3 months with the drugs, i.e. tardive dystonia, can develop at any time, ranging from days to years after the introduction of neuroleptics; there is no ‘safe’ period.

Overall, the phenomenology of tardive dystonia is indistinguishable from that of primary (idiopathic) dystonia, although retrocollis and anterocollis are more

common in tardive dystonia. Tardive dystonia is a very persistent disorder; according to the largest patient series reported so far only 14% of patients had a remission over a mean follow-up period of 8.5 years and discontinuation of neuroleptics increased the chances of remission fourfold (Kiriakakis et al. 1998, Brain).

What can be done? Prevention is most important. Neuroleptics should not be given on a regular basis unless it is absolutely necessary - as in the treatment of psychoses. Once it has become clear that the dystonia is tardive in nature the causative drug should – if possible - be discontinued even if the dystonic symptoms initially worsen. In extreme cases it may take up to 5 years for the symptoms to resolve. After that time remission is very unlikely and with respect to age and circumstances it may be advisable to continue the “dystonic drug”, which is then the only possible treatment of tardive dystonia. None of the numerous treatments tried in patients with tardive dystonia, including clozapine and botulinum toxin injections, seemed to relate to overall outcome. Otherwise the treatment is similar to the primary (idiopathic) dystonia with botulinum toxin injections for focal dystonias, anticholinergics (if tolerated) for more generalised forms. Deep brain stimulation is tried in some patients with promising results.

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Coping with Dystonia



Professor Gündel studied Psychiatry in Münster, Germany and Vienna, then worked at the University Clinic in Münster from 1990 – 1993 and at the Dept. of Neurology/Städtische Kliniken Dortmund from 1993 – 1996, and from 1996 – 2006 at the Technische University in Munich. In 2003 he became a Professor at the TU Munich and in 2005 received the German Board Certification in Psychoanalysis. Since 2006 he is Director and Professor at the Dept. of Psychosomatic Medicine and Psychotherapy at the Medical School Hannover, Germany.

Dystonia often is a really serious disorder that can produce occupational dislocation, social withdrawal, anxiety, depression and so-called adjustment disorders. Also there is a relevant subgroup of dystonia patients who believe to a certain extent - from the patient viewpoint - that their disease may have a psychogenic origin. In contrast to that, today’s medicine is quite sure that dystonia is a neurological disease, but in addition may be exacerbated by a variety of psychological and psychosocial factors, like negative mood (anxiety, depression, catastrophizing) and

social isolation. Therefore neglecting psychological disturbances and psychiatric comorbidity may lead patients and clinicians, under the misconception that emotional disturbances may be an expected concomitant of dystonia, to ignore individual distress. This may also hinder the access of these patients to appropriate treatment or result in denial of coverage by their health insurers, thus making it harder to get back to a more positive mood and mode of coping with dystonia.

However, patients by themselves and their relatives should be informed and trained in how to do their best in this respect (coping with dystonia), and often this really may prove to be helpful. On invitation by the EDF President Didi Jackson, I presented some material at this years meeting of the EDF in Vienna, and will briefly summarize the contents of this presentation:

Let's start with the following questions:

If you would have to tell someone how your life changed after dystonia began and how your life is influenced by dystonia right now, what would you say? And when you heard the diagnosis for the first time, what did you feel and think at that time? Or, if dystonia started in early childhood, what did you feel and think as you became aware for the first time what is different with you?

Often, patients with dystonia may initially or repeatedly during the course of the disease experience some sort of a shock, reproaches against oneself, anxiety, help- and hopelessness, anger, despair and isolation. There is a huge variety of so-called "vicious circles"

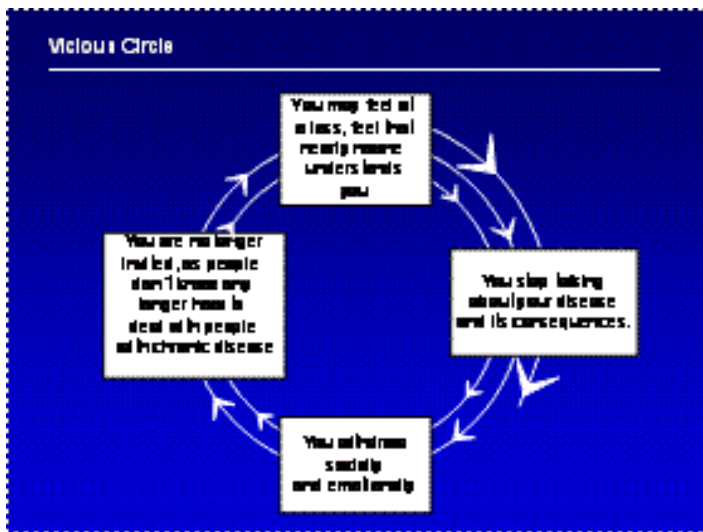


Fig 1.

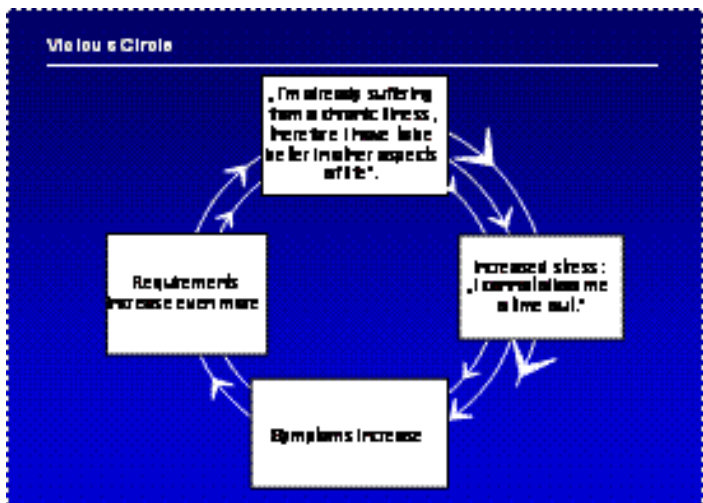


Fig 2.

which may lead to additional personal distress and also support chronification of disease and psychosocial isolation (see examples in Figure 1 and 2):

What can you do in order to get better again? There are some basic rules ("steps of coping") which may give some orientation.

Steps of Coping with Dystonia

First, realise that some of these negative feelings are a normal reaction to the onset or exacerbation of disease. After having realised this, it may be necessary to say goodbye to some aspects of your previous life, but also it's even more important to give other aspects of life space to live and develop again, and to open up to the world again.

There often may be some sort of grief or sadness due to some capabilities or aspects of life you may have lost, but it is much more important to look consciously on what is still remaining, and perhaps, even if or what you may have "gained" by dystonia. Indeed, there are many people suffering from chronic diseases who experience life in a much more precious and worthwhile way, compared to the time before the onset of the illness, when their personal health was taken for granted.

In this context, it is important and helpful, in most cases, to collect more information about your disease and about how to cope as best you can. Also, start to differentiate symptoms of disease from your person and personality. Find a balance between respect for dystonia and your will to enjoy life as much as possible, without being overwhelmed by disease.

And, use your creativity and the creativity of people close to you, in order to do as best as you can in the job, in your leisure time, within the family and with your friends in order to get as much as you need. Decide by yourself, how you want to live with dystonia, and look for additional help, including professional support if you feel you cannot achieve your goals by yourself.

Basically, the main aim of coping with dystonia, which can be applied for most chronic diseases, is to maintain or develop "as much normality as possible" in your individual life.

Generally, it is helpful to pay attention to the following points, which are simple, but have been identified in recent stress research as some of the main human resources individuals can influence or generate by themselves:

- Improving sleep quality and quantity
- Cultivating a positive outlook on life
- Exercise (regular, moderate physical activity)
- Social support

As every individual has a single and specific situation, it's often very worthwhile to consult other people in order to talk about difficulties in coping with dystonia. Sometimes, it may feel even a bit shameful to talk about often disease-reactive psychological problems or conflictual changes in close relationships in private and/or the job life. But research shows that especially these themes may have an enormous impact

on quality of life, and the best outcome may be influenced and changed by your own will and activity.

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The Role of Rehabilitation in the Management of Dystonia



Professor Schneider, born in Graz, has not only devoted his time to Neurology, but has practised in many other medical fields such as gynaecology, surgery and as a doctor travelled worldwide, practising in South Africa and Iran. In 1991 he settled down at the AKH Vienna in the Dept. of Neurology and later on as Head of the Dept. of Neurological Rehabilitation at the AKH Vienna. Since 2002 he has been head of the Dept. of Neurology at the LKH Grimmerstein and Director of the LHK since 2003. He has supported the Austrian Dystonia Society from its formation in 1995.

The role of rehabilitation in the treatment of dystonia is not well defined. There are three well known strategies in the treatment of dystonia. The use of anticholinergics has been well known for decades and offers some affected patients partial relief even in the presence of adverse events. Secondly, botulinum toxin is the only licensed drug for the treatment of cervical and cranial dystonia and has been used also effectively for other focal dystonias. Thirdly, the availability of deep brain stimulation completes the field of established treatments mainly for patients with generalized dystonias. However dystonia is a chronic disease, which can not be cured by known treatment options. Patients still have a large number of different symptoms due to the disease and therefore look out for further treatment options. Rehabilitation can help to support the known medical therapies and improve, therefore, quality of life.

Physiotherapy does address pain, exercise and posture, and provides understanding of the condition while developing strategies for management. After sensomotor analysis, goals are the reduction of dystonic movements and the re-education of physiological movements. Sensory tricks, EMG-Biofeedback and many repetitions may support the concept of "Learning by self experience". Special home programmes have been developed for patients with dystonia by J-P Bleton. Recent studies showed that botulinum toxin treatment plus a specific programme of physical therapy in the treatment of cervical dystonia is more effective than botulinum toxin alone. A longer duration of clinical effect and a lower dose of botulinum

toxin at re-injection has been described (Tassorelli et al. 2006). Various experts report on the positive effect of hippotherapy and balneotherapy in patients with dystonia.

Stress plays a major role in the life of patients with dystonia. Often, an increase of uncontrolled movements under stress is observed. Successful strategies in handling stress such as thought stopping, visualisation and progressive relaxation may help to control stressful events. Also Yoga, Alexander technique and others can help to reduce stress.

Activity and sport has an excellent effect for general health. Improvement of vascular risk factors (high blood pressure, cholesterol, body weight) but also improvement of constipation, fatigue and depression can be achieved. However sport and physical activity are rarely discussed in the management of dystonia. Not all patients report on an increase of dystonia during activity. According to Jahanshahi, running improves dystonia in 20 % and does not affect dystonia in 20 %. Also, patients report on improvement after various sporting activities such as dancing, walking and swimming. Rehabilitation strategies should also include efforts to find proper activities for every patient to support general health.

Management of pain is another important issue in the rehabilitation of patients with dystonia. Any interventions providing even short-term relief can be valuable to the patients, hot and/or cold packs can be tried and pain relieving modalities (Ultrasound and

interferential therapy, TENS) may be useful. Generally any activity or posture that aggravates a patient's symptoms should be avoided.

Recent observations suggest that abnormal sensory processing is a fundamental disturbance in patients with focal dystonia (Molloy et al. 2003). Additionally it has been shown that Braille reading in patients with focal hand dystonia improves their spatial acuity and also their dystonia (Zeuner and Hallet 2003).

In conclusion, Neurorehabilitation is a useful tool to treat patients with dystonia. Various studies show

that physiotherapy and other forms of sensory and motor training may be useful alone or in combination with other treatment strategies

Physical activity and stress management should be advised on a regular base to improve physical and mental capacities and to prevent secondary complications.

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EDF – David Marsden Award 2007, sponsored by Merz Pharma

The David Marsden Award is intended to encourage research into Dystonia in all European countries, especially by young scientists. It is awarded every two years by EDF, in honour of the late Prof. David Marsden who did so much to promote the cause of dystonia and treatment for dystonia patients. The 2007 Award was sponsored by Merz Pharma and we are very grateful for their support.

The EDF Medical Advisory Board and the Board of the EDF evaluated 10 papers submitted for the David Marsden Award 2007 on the basis of originality, clinical and scientific content, experimental design, clarity of presentation and - most important - significance in the field of dystonia.

All papers were of high quality and we congratulate

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on being chosen as Winner of the David Marsden Award 2007 for her paper:

'Defective Temporal Processing of Sensory Stimuli in DYT1 Mutation Carriers: a New Endophenotype of Dystonia?'



The Award of EUR 2.500 was presented by the EDF President Didi Jackson to Mirta Fiorio during the EFNS Congress in Brussels at the Basal Ganglia Club Meeting on Sunday, 26th August 2007 and again at the EDF General Assembly 2007 in Vienna in September.

Mirta is the third winner of the David Marsden Award. In 2003, the first was Mark Edwards from London, with his paper on the DYT1 gene, and the second (2005) was Victor Candia of Zürich, with his paper on Musicians with Dystonia.

Involvement of the Sensory System in Dystonia



Dr. Mirta Fiorio was born in Verona, studied there from 2001 – 2004 and also worked in London at the National Hospital for Neurology, specialising on research in the field of sensory and cognitive brain functions related to movement disorders, in particular dystonia and Parkinson's Disease.

Why do we study the sensory systems (touch and vision) in a movement disorder, like dystonia, where the common clinical observations are motor symptoms affecting a specific or different parts of the body? The main reason to do it is that the pathophysiology of dystonia has been related to basal ganglia dysfunctions. These structures of the brain are important not only for motor control, but also for the correct functioning of the sensory signals coming from the periphery of the body. In particular, basal ganglia can integrate sensory inputs belonging to different modalities (i.e. tactile and visual) and can process sensory information useful for motor execution. The investigation of sensory abilities in different forms of dystonia (generalised, cervical and focal-hand) has revealed the presence of sensory deficits. The question is whether these sensory deficits are strictly related to the pathophysiology of dystonia or whether they are a consequence of motor symptoms.

To answer this question we studied whether sensory dysfunctions in dystonia may be related to an abnormal genetic substrate. To this aim, we studied sensory abilities of patients with the DYT1 gene mutation, which causes early onset of dystonia. We also studied the sensory functions in patients' family members with the same genetic mutation (DYT1), but without

dystonic motor symptoms.

We found sensory deficits not only in patients with DYT1-dystonia, but also in their asymptomatic DYT1-carrier relatives. This result suggests that the sensory abnormalities are related to the presence of the gene mutation and not to the presence of motor symptoms. Therefore, the sensory system might be impaired even before the occurrence of motor symptoms.

In the light of the current results, we can suggest that a sensory liability is present both in patients and asymptomatic DYT1 carriers. Factors overloading a defective sensory system might increase the risk of developing dystonia in predisposed subjects.

(The scientific paper '*Defective Temporal Processing of Sensory Stimuli in DYT1 Mutation Carriers: a New Endophenotype of Dystonia?*' was published in **Brain** (November 14, 2006)

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Medical Advisory Board: Dr. J. D. Speelman retires

At the end of this year, Dr. J. D. (Hans) Speelman will retire from his position at the Academic Medical Centre of the University of Amsterdam and, also from his six year membership of EDF's Medical Advisory Board.

During his career, Dr. Speelman has given much of his time and energy to help patient advocacy organisations, especially in the Netherlands as a medical ad-

viser for Nederlandse Vereniging van Dystoniepatiënten. It has been a great pleasure for the EDF Managing Board and the Medical Advisory Board to work with Dr. Speelman and we take this opportunity to thank him most sincerely for all he has done to help dystonia patients.

THE MEDICAL ADVISORY BOARD OF THE EUROPEAN DYSTONIA FEDERATION presents



This will be the first large-scale medical conference solely on dystonia held in Europe for more than a decade and many of the foremost experts in Europe and North America have accepted an invitation to make presentations. This is a very exciting project and EDF and the Medical Advisory Board are proud to be able to organise such a worthwhile event which will bring eventual benefits for dystonia patients and their families around Europe (<http://www.dystonia-europe-2008.org>).

The conference programme will be sent to member groups very soon. The target audience will be clinicians, researchers, general practitioners, health professionals and medical students. However, **one** delegate from each EDF member group will be invited to attend the conference and registration will be free for those delegates.

EDF General Assembly 2008

The EDF General Assembly 2008 will be held in Hamburg immediately after the medical conference ends, on the afternoon of Sunday, October 19, 2008. More details of the GA will be issued during 2008, but keep these dates free!

New Companies Marketing Botulinum Toxin in Europe

Over the past two or three years, Merz Pharma (based in Frankfurt, Germany) has been developing its marketing strategy for a new version of type "A" botulinum toxin "Xeomin" and approval has been gained from the EMEA (European Medicines Evaluation Agency) for release of the product in 10 European countries.

More recently, Eisai Europe, whose parent company is based in Japan, has bought the European marketing rights for "Neurobloc", the type "B" botulinum toxin formerly sold by Elan Pharma. The Eisai parent company already markets the product in Japan and Eisai Europe expects to begin operations during 2008.

These new activities have breathed life into the botulinum toxin market and the long established

companies – Allergan and Ipsen – now have more competition.

In addition to these pharmaceutical treatments, of course, we also recognise the "Activa" therapy of Medtronic, which is bringing amazing improvements to the lives of an increasing number of dystonia patients with severe symptoms, where other treatments have failed.

We are grateful to all of our sponsors for all that they have done to help us, especially those who have given support throughout the past 14 years since EDF was founded. We look forward to working with them all in the future to promote the cause of people who live with dystonia across Europe.

International Conferences

EDF attends international medical conferences every year, with an information booth to respond to doctors who enquire about dystonia, or who want to find information to give to patients, back home'. EDF also uses these conferences as an opportunity to 'network' with dystonia specialists, who may be active in international research and other areas of interest among medical scientists.

In the past, attendance at conferences has tended to concentrate on the annual congresses of the Movement Disorder Society and the European Federation of Neurological Societies. Two new meetings are now becoming important, however. These are the World Federation of Neurology biennial meeting on „Parkinson's Disease and Movement Disorders“, and the „Toxins“ conference which now occurs every three years (alternately in Europe and the USA). Increasingly, these newer meetings have more dystonia content and this improvement in the level of scientific interest in our condition encourages EDF to provide a conference presence to promote EDF and its members' interests.

The Toxins-2008 meeting will take place in Baveno, Italy and will be organised by Prof. Alberto Albanese of Milan, a member and long-term supporter of our Federation. It is hoped that EDF will participate.

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