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DYSTONIA EUROPE

Norsk Dystoniforening

Dystonia Europe would like to thank the following for their involvement in making this conference possible:

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DYSTONIA EUROPE

Norsk Dystoniforening

Oslo 8 April 2016

Dear Participant,

Welcome to Oslo and the Annual European Dystonia Days – D-DAYS 2016.

On behalf of Dystonia Europe and in collaboration with the Norwegian Dystonia Association, we are delighted to welcome you to Oslo, the capital city of Norway.

During these D(ystonia)-DAYS you will hear presentations on dystonia and learn about what is going on in the field of treatment, research and rehabilitation. We are very grateful to all the speakers and chairs who have joined us to share their expertise within dystonia.

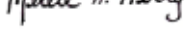
As well as learning more about dystonia we also hope that you will enjoy meeting old friends, and connect with new ones, and above all, have a pleasant weekend here in Oslo.

If you have any questions about anything do not hesitate to ask us and we will try and help as best we can.

We also take the opportunity to thank our sponsors for making this meeting possible. Dystonia Europe is very grateful for your support and we are glad that so many of you are showing direct interest in our work by being represented here.

We thank you all for joining us in Oslo and we wish you an enjoyable, interactive and fruitful conference!

Warm regards



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Physiotherapist Johanna Blom

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Workshop: "Physiotherapy and Dystonia"

The main focus in this workshop will be cervical dystonia. Physiotherapy is an important complement to other treatments, for example Botulinum toxin and DBS. There are some studies on physiotherapy in Dystonia with good results, but there is a lack of controlled and randomized studies with larger groups of patients. Currently, there is an on-going study by Van den Dool et al including 100 cervical dystonia patients, testing a standardized therapy programme compared to regular physical therapy. From my experience,

The role of the physiotherapist is important for the training-practicing-relearning, the strategies and help to cope with dystonia, but also to optimize the benefit from the Botulinum toxin injections by cooperation with the clinicians.

All treatment should start with an accurate assessment of the patient. This includes my special guide to see if the dystonia is stable or unstable. It´s also important to know how the dystonia affects daily life and what the patient thinks is the biggest problem caused by the symptoms. This will result in an individualized treatment programme based on some specific strategies. For example, training of motor and postural control, suppression of excessive muscle tension and involuntary movements, activating and reinforcing the antagonists, correction of the posture, finding relaxation in posture and movements, using feed-back systems (mirror for example) and - for the patients with stable dystonia - reciprocal rocking-sweeping arm movements. A home programme of exercises is necessary to improve the symptoms and the situation.

In the workshop I will explain more about why training is important, how to train, give examples of exercises and also talk about common consequences of dystonia and how to handle them. I will use some video cases.

Professor Espen Dietrichs, MD, PhD

Head, Department of Neurology
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Dystonia and Plasticity

The pathophysiology behind various forms of dystonia is still largely unknown. However, both the results from experimental studies in animal models and observations in dystonia patients indicate that abnormal brain plasticity may be important. While nerve impulses are electrical signals that travel along nerve fibers, the signals from neuron to neuron are transmitted by chemical substances in the synapses.

Plastic changes occur in these synapses. New synapses may be formed, and existing synapses may be withdrawn. Synaptic efficacy may also change, and synapses may increase or reduce their signal transmission. According to current theories plastic changes in the cerebral cortex as well as in the basal ganglia may result in dystonia.

Anniken H Hagen, Chairwoman NDF

Physiotherapist,
Chairwoman of Norsk Dystoniförening,
Bergen, Norway

Project on Physiotherapy for Cervical Dystonia

The Norwegian Dystonia Association (NDF) has received 1.200.000 NOK (approximately € 118 000) during the last three years, from the Norwegian Foundation for Health and Rehabilitation ("ExtraStiftelsen Helse og Rehabilitering"). The funding is for a project to train physiotherapists in treatment of cervical dystonia. The Norwegian "ExtraFoundation for Health and Rehabilitation" is an institution consisting of 28 non-profit organizations within health and rehabilitation.

The main goal of the project is to educate 60 physiotherapists in Norway within 3 years. The project started in 2014 and will end in 2016. NDF sees the need for a continuation of the project. We hope we can finance the project onwards from 2017. We want to contribute to upgrade skills of the movement disorder, and improved treatment options for patients with dystonia. Through the project we want to contribute to increased coping and to improve the quality of life for those affected by dystonia.

With proper guidance and individualized exercises, we will be able to reduce tension in the dystonic muscles and reduce pain.

The physiotherapists who have been certified through NDF, are published on our website. NDF has been aiming at a high level of expertise in the project, including neurologists and physiotherapists.

Lena E. Hjermind

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Genetics in Dystonia

It has been known for more than a hundred years that dystonia might be hereditary. The German physician MW Schwalbe probably made the first description of dystonia as a familial disorder (1908) in "Eine eigentümliche tonische Krampfform mit hysterischen Symptomen". He reported hysterical symptoms in three siblings of Jewish ethnicity, later recognized as having early-onset generalized dystonia. Few years later (1911) his compatriot H Oppenheim reported the same symptoms in patients, but he was, in contrast to Schwalbe and other colleagues, convinced that the symptoms were not psychiatric, and named the disorder "dystonia musclurom deformans".

It has been estimated by NA Fletcher, HM Waddy, and co-workers (1990 and 1991) that 85% of all cases of generalised, multifocal, and segmental dystonia and 25% of all cases of focal dystonia are hereditary. These estimates can be and have been discussed many times since. Additionally, as we do not know the exact prevalence of dystonia, it is seemingly difficult to estimate the frequency of hereditary dystonia, which, furthermore, varies worldwide due to different origin.

We now know a still growing number of loci and genes in which mutation can cause dystonia, either alone or as the main symptom. The first dystonia type gene, DYT1/Tor1A was described almost twenty years ago (1997) by LJ Ozelius and co-workers and the latest in this nomenclature, DYT27/COL6A3, was described recently (2015) by M Zech and co-workers. But the numbers of genes, in which mutations can cause dystonia, and the eventual influence between genes and environmental factors are still a puzzle.

We will not have time to go through all the known forms of hereditary dystonia, but focus on the most common forms according to our knowledge so far and the relevance to the families with dystonia in one or several members; for whom the question might be: Is genetic test a possible option? Does it have any consequences for me? or my children/siblings?

Professor Marjan Jahanshahi

Professor of Neuropsychology
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Living Well with Dystonia – A Self-Management Approach

Dystonia is a chronic disorder. There are striking differences between people with dystonia in terms of how well they live and cope with dystonia. This fact together with research evidence suggests that personal factors and social resources influence psychosocial adjustment to dystonia, which will be the focus of the first part of my talk. Then I will consider a list of strategies that can be employed as part of a self-management approach to live and cope well with a chronic illness such as dystonia.

Jeanette Koht, MD, PhD

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Dystonia and Physical Activity

Background:

Physical inactivity has been identified as the fourth leading risk factor for global mortality. In addition, the most important risk factors for chronic diseases include; high blood pressure, high concentrations of cholesterol, inadequate intake of fruit and vegetables, overweight and obesity, physical inactivity and tobacco use. Many of these risk factors are closely related to physical activity. Physical activity is defined as any bodily movement produced by skeletal muscles that require energy expenditure. WHO recommend regular and adequate levels of physical activity to prevent disorders, but also to increase quality of life. Physical activity includes exercise as well as other activities as part of playing, working, active transportation, house chores and recreational activities. For adults at least 150 minutes of moderate-intensity physical activity is recommended throughout the week to prevent chronic disease. So the arguments to be physically active are many and good, but how to do it when affected by dystonia?

Physical activity and dystonia

Little research in this field is done, but generally people with disabilities report worse health status compared to people without disabilities and they also receive fewer preventive health services such as counseling around exercise habits. There are many different kinds of dystonia described, even some that are exercise-induced or activity-induced.

Therefore it is important that you know which type of dystonia you have, which types of exercises/movements make it worse/better and which type of exercise is impossible to do. Every patient should therefore have a thorough understanding of their unique dystonia together with their doctor and physiotherapist.

Those activities that might trigger negative feelings, negative self-esteem or pain should be avoided. Some patients have to avoid certain activities. Especially sports involving asymmetrical movements of the trunk, can increase the symptoms, especially for individuals with cervical dystonia. Lower stress exercises such as water exercise, walking, yoga, low-impact aerobics are less likely to worsen symptoms. To find the best activities it is important to get some guiding, advice and last but not least motivation.

Conclusions

The benefits of being physically active for physical and mental health are huge, also if you have dystonia. To encourage physical activity in general as part of the health promoting behavior a precise assessment is important. In addition most patients need guiding, facilitation, support and motivation on their way and trained physiotherapists are of great importance helping in this work.

Professor Maja Relja

Movement Disorders and Neurodegeneration Center
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Non-Motor Symptoms in Dystonia

Dystonia is a hyperkinetic movement disorder typically characterized by involuntary sustained or intermittent muscle contractions causing abnormal postures. Contrary to common views the non-motor symptoms are present in dystonia patients. The non-motor symptoms may complicate motor presentation in dystonia. Evidence is emerging that non-motor symptoms such as neuropsychiatric, cognitive, sleep, sensory and pain play an important role in dystonia. Non-motor symptoms may occur in dystonic patients related to the primary pathophysiology of dystonia and/or as a secondary consequence of dystonia.

It is well known that non-motor symptoms in Parkinson's disease is stronger determinants of Quality of Life (QoL) than the motor symptoms. Thus, non-motor symptoms in patients with dystonia can be a key determinant of health related QoL. Consequentlly, higher awareness of non-motor symptoms in dystonic patients' needs to be raised. Routine clinical examination in patients with dytonia should include motor as well as non-motor status examination.

Inger Marie Skogseid, MD, Phd

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Dystonia and DBS

Deep brain stimulation (DBS) in the internal globus pallidus (GPi-DBS) was first established as an effective treatment for primary/pure generalized and segmental dystonia. It was pioneered by French neurosurgeons and neurologists in Montpellier, and through the first controlled, single-blinded, French multicenter study, lead by Marie Vidailhet. Further proof of good efficacy and safety was provided by the German-lead randomized, double-blind and sham-stimulation-controlled study, in which our center in Oslo also participated. This study showed significant improvement in quality of life already after 3–6 months of GPi-DBS. All of these groups have during recent years also published long-term outcomes of their patients.

At 3 years follow-up, mean improvement in the motor/dystonia score of the Burke–Fahn–Marsden Dystonia Rating Scale (BFMDRS) was 58% in the French study and 61% in the German study, thus a further significant improvement compared with results at the 6 months follow-up. In the German study, 5 years follow-up data showed a sustained efficacy compared to 3 years, both in the generalized and segmental dystonia group. Thus, in most patients the efficacy of GPi-DBS in primary dystonia is sustained over many years, and in our own experience this now includes patients treated for up to 12 years. However, the Montpellier group have reported significant worsening in some of their primary generalized dystonia patients after many years of effective GPi-stimulation, and have shown that implanting a second pair of electrodes can improve outcome in some of these patients.

In primary/pure focal or segmental cervical dystonia (CD), several patient series of GPi-DBS have shown that this treatment can be very effective also for this dystonia subgroup. In a published series of our first eight patients, median improvement in the severity score of the TWSTRS (Toronto Western Spasmodic Torticollis Rating Scale) was 70% at median follow-up of 21 (12–48) months. The group in Toronto has reported a 54% (SD 27%) improvement (n= 10), at mean 7.7 years. A sham-stimulation controlled study of 58 CD patients published in 2014, was positive and thus established severe and medically intractable primary CD as an indication for GPi-DBS.

GPi-DBS has also been used with very good benefit in myoclonus–dystonia and in patients with drug (neuroleptic)-induced tardive dystonia. Functionally important benefit from GPi-DBS has also been observed in other forms of secondary/complex dystonias, such as dyskinetic cerebral palsy, PKAN, Lesch–Nyhan syndrome, and in some other dystonia syndromes.

Finally, beneficial effect for dystonia has also been reported for DBS in other targets, such as the subthalamic nucleus, the zona incerta (for dystonic tremor), and ventral thalamic nuclei, such as the ventral-oralis nuclei for task-specific hand dystonia. These targets have much less robust scientific basis than GPi-DBS in dystonia, however.

Charalampos Tzoulis, MD, PhD

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What is Dystonia?

Dystonia may be defined as a movement disorder consisting of involuntary sustained or intermittent muscle contractions, which cause twisting and repetitive movements and/or abnormal postures. Dystonia has a complex and heterogeneous etiology ranging from symptomatic (i.e. secondary to other brain disease or injury), to monogenic (i.e. caused by specific genetic mutations) and idiopathic (i.e. of unknown cause).

In this presentation, I will give an update on the different forms and types of dystonia classified according to phenotype or cause.

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