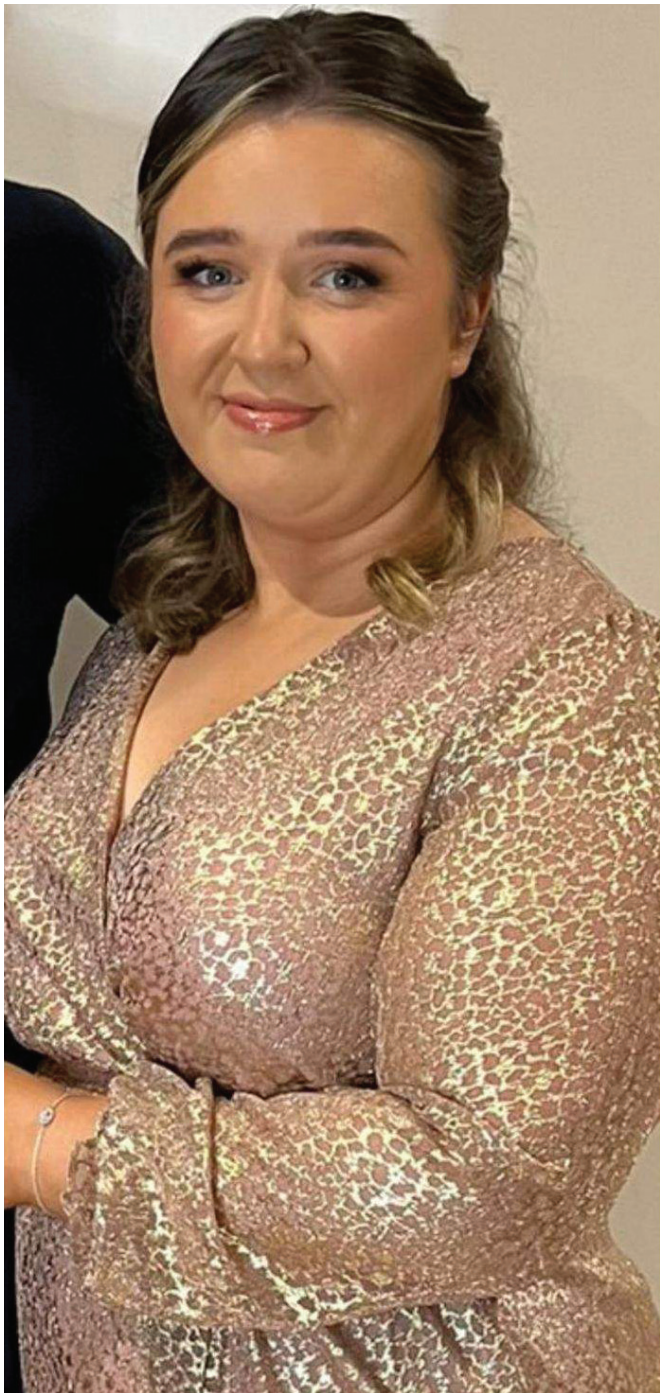
Dystonia Advocate

Dystonia is a neurological movement disorder, which may affect anyone at any age

Dystonia Ireland
33 Larkfield Grove,
Harold's Cross,
Dublin 6W
Tel: 00 353 1 492 2514
Fax: 00 353 1 492 2565
Email: info@dystonia.ie
Web: www.dystonia.ie

Edition: Spring 2025

Remembering Niamh



The Late Niamh Stafford

>>

Remembering Niamh Stafford, from Wexford with ‘a heart of gold’ aged 25.

It is with profound sadness, I regret to have to write of the sudden passing of our dear friend Niamh Stafford on Thursday, 30th May 2024, following a medical emergency at her home.

The heart - wrenching news reverberated through Wexford town, prompting the community to rally around Niamh’s heartbroken family: her fiancé William, her parents Josie and Jimmy, and her sister Roisin.

Niamh and William Murphy had celebrated their engagement the previous Christmas and were excitedly planning their wedding and future together when life took a devastating turn.

A beloved and familiar figure in the Maudlinstown community, Niamh was known for her warm, approachable nature. Working at Hanrahan’s Londis, for 10 years she brightened many people’s days with her “happy-go-lucky” demeanour, greeting customers with a smile, a chat, and an infectious laugh that could light up any room. The supermarket closed its doors on Thursday as a mark of respect, allowing friends and colleagues time to process the news.

Niamh, was also deeply involved in her local community. She was passionately committed to St. Mary’s of Maudlinstown, where her father Jimmy serves as the secretary. The club posted a tribute online, stating: “Niamh was a big part of the Mary’s family, who played for both the girls’ and boys’ teams underage. She was also a regular volunteer for our club and was always there to lend a hand. She will be sadly missed by all at St. Mary’s of Maudlinstown.”

Local Labour councillor George Lawlor expressed the profound impact of Niamh’s passing on Wexford’s south end, describing it as “a tragedy of unspeakable proportions.” He added, “Niamh comes from a family steeped in community service and involvement. The trauma being felt by her family and the community she was embedded in is unimaginable. Her passing has sent shockwaves through the Maudlinstown area.”

Niamh developed Cervical Dystonia 12 years ago, a neurological movement disorder, at a young age. She overcame numerous challenges and barriers and became a person of extraordinary generosity, making immense contributions. Niamh participated in a Dystonia Europe video project to promote awareness about living with Cervical Dystonia.

In Dystonia Ireland, we have been deeply moved by the tremendous support from Niamh’s fiancé, her family, colleagues, sponsors, friends, and the community in Maudlinstown and Wexford town. Their efforts in raising funds in her memory are significantly contributing to our ongoing Dystonia Ireland Medical Research Projects.

Left to Right Stephen Fenlon, Maria Hickey, Dystonia Ireland, Sharon Cullen, Katie Holmes, Pauline Moore, Michael Hanrahan, Owner, Philly Cullen, Manager

Last October, I received a telephone call from Philly Cullen, the manager of Hanrahan’s Londis The Faythe, where Niamh worked for 10 years. They graciously offered to host a fundraising Halloween Fancy Dress Event in Wexford on Saturday, 2nd November 2024, in her honour and to support our charity, Dystonia Ireland.

The event was a remarkable success, with an outstanding turnout from the entire community of Maudlinstown and Wexford town.Together, they raised an astonishing €7,474. When we saw the final total, it was a wonderful surprise, and it was clear that a lot of hard work and organising went into making this event such a success. In fact, they are hoping to hold another event this year in memory of Niamh.

It is people like you all that are such an inspiration to us in Dystonia Ireland.

Thank you, Pauline and Caroline

A heartfelt thank you to Pauline Murphy and her sister Caroline for taking on the Camino Walk Way in memory of Niamh Stafford. As the mother of Niamh's cherished fiancé, William, Pauline and Caroline embraced this challenge to honour Niamh, proudly wearing their Dystonia Ireland t-shirts throughout the journey.

"Ask The Experts" Meeting 2025

We are planning to hold a Dystonia Patients Conference this year. "Ask The Experts" As soon as the information and details become available, we will promptly inform you so that you can mark in your diary.

CHRISTMAS CARDS

A heartfelt thank you to all our members, friends, and family for purchasing our Christmas Cards, which continue to be incredibly popular. As you know, this is one of our main fundraising methods and a crucial way to raise awareness about dystonia. For those of you who take large quantities of cards each year to distribute across your regions or workplaces, we deeply appreciate the tremendous effort you put in. Your dedication always exceeds expectations!

Thanks to your kindness and relentless support for this fundraiser, we can continue to fund some of our Dystonia Ireland Medical Research Projects and keep Dystonia Ireland running smoothly.

DONATIONS

Sincere thanks once again to EI Electronics, Shannon, Co. Clare for your company's incredibly generous donation of €5,000 towards Dystonia Ireland. Your support means a great deal to us.

Thank You For Your Great Support

Dystonia Ireland would like to extend our heartfelt gratitude to each and every one of you for your unwavering kindness, generosity, and support. Your donations, participation, and encouragement of our annual fundraising events mean the world to us.

We would also like to take this opportunity to thank our Patron, Mr. Brian Kerr, our Medical Patrons, the Neurology Medical Teams, and the Irish Dystonia Research Group. We deeply appreciate all that you do for us!

Dysphonia International: Supporting People with Voice Disorders

About Spasmodic Dysphonia

Spasmodic dysphonia (SD), a form of laryngeal dystonia, is a neurological voice disorder that affects the muscles of the larynx, causing involuntary spasms that disrupt normal speech. These spasms lead to voice breaks, strained or strangled-sounding speech, and difficulty maintaining a steady voice. The condition is a form of focal dystonia, meaning it specifically affects one area of the body—in this case, the vocal cords.

About Dysphonia International

Dysphonia International, formerly known as the National Spasmodic Dysphonia Association (NSDA), is a global charity organization committed to improving the lives of individuals affected by spasmodic dysphonia (laryngeal dystonia) and related voice conditions through research, education and awareness, and support. Our community includes people with voice conditions, care partners, researchers, and healthcare professionals.

Building a Community that Cares about You and Your Voice

A cornerstone of Dysphonia International's work is fostering connections among people affected by voice conditions. We facilitate both in-person and virtual support groups, providing a place for individuals to share experiences, gain insights, and share their journeys with others who understand their challenges. Dysphonia International's Area Contact Leaders around the world, like Yvonne Elliott Coyne in Ireland, play a crucial role in facilitating these networks, ensuring that you, regardless of your location, can access the help you need. By encouraging dialogue, we try to help individuals overcome feelings of isolation and promotes a sense of belonging. Our local support groups serve as hosts for our virtual meetings, but all are welcome to attend. The meetings are listed on our website.

Educational Outreach and Raising Awareness

Education about voice disorders among the public, medical communities, and policymakers builds understanding by highlighting the challenges posed by voice issues. Dysphonia International has a wealth of information available through webinars, workshops, symposiums, and published materials, helping people living with a voice condition and medical professionals stay informed about the latest developments. Raising public awareness about voice disorders is another key focus of ours. We try to dismantle misconceptions about voice conditions through podcasts, speaking opportunities, and events like World Voice Day (April 16) and Walk for Talk (October 16-19, 2025). You are welcome to participate in any of our awareness events.

Finding Answers through Research

Dystonia is thought to be a network or circuit disorder involving abnormal interactions in regions of the brain that coordinate and control movement. The brain communicates with other parts of the body through the transmission of signals along specialized neuropathways. When these connections are disrupted, as seen in SD, the effects ripple outward, significantly impacting the voice and complicating daily life for those affected.

Dysphonia International plays a crucial role in finding answers for these voice disorders aimed at improving diagnosis, management, treatment, and, ultimately, a cure. Some of the ways the organization does this is through funding seed grants to support breakthrough research and expand scientific understanding, sponsoring research travel awards to multiple professional voice conferences to increase engagement, providing opportunities for collaboration by connecting researchers, and supporting resources, including a brain bank and a global patient registry.

Advances in imaging techniques like functional MRI are starting to shine a light and provide valuable insights into these pathways, which may lead to a deeper understanding of the disorder's mechanisms. In addition, this may pave the way for targeted therapies to address the root causes rather than just managing symptoms. By restoring communication within the brain's motor circuits, researchers may be able to reverse unfavorable neuroplasticity and recalibrate disrupted pathways. Through continued exploration and targeted research, we move closer to restoring voices and enhancing our broader understanding of brain function and its role in regulating communication and movement.

Kim Kuman, Executive Director, Dysphonia International

Innovations in Treatment Approaches

Neuromodulation refers to the use of targeted therapies that alter or regulate the activity of specific neural pathways or brain circuits. For spasmodic dysphonia, neuromodulation aims to normalize the misfiring of signals in the brain and restore smoother communication between the brain and the laryngeal muscles. It targets the root cause of the disorder—disrupted neural circuits—rather than just alleviating symptoms. Research is ongoing to determine the most effective neuromodulation techniques for SD, optimize delivery methods, and ensure long-lasting effects without significant side effects.

Dysphonia International is funding research on external and internal neuromodulation therapies along with grants on imaging. Internal approaches include deep brain surgery, which is currently used for other forms of dystonia. External approaches include transcranial direct current stimulation (tDCS), transcranial magnetic stimulation (TMS), and vibrotactile stimulation (VTS). This is an exciting new area of research to help advance the options for treatment for spasmodic dysphonia.

Collaboration and Advancement

Dysphonia International is pleased to partner with organizations like Dystonia Ireland to advance the field and deepen understanding of dystonia. Committed to supporting individuals with these conditions, our organization continues to expand its outreach, education, and research efforts. By bringing together those affected, researchers, and healthcare professionals, we aim to improve the quality of life and drive innovation in voice disorder treatment. Learn more at dysphonia.org and connect with Ireland Area Contact Leader Yvonne Elliott Coyne at yvonneelliottcoyne@gmail.com.

World Voice Day

World Voice Day is an annual event dedicated to recognising the significance of the human voice and raising awareness about various voice-related issues.

To mark this occasion, Dystonia Ireland and Dysphonia International are hosting their first joint Zoom meeting on Thursday, April 10th, at 8 pm GMT. This event will feature a medical expert and is open to all individuals with voice-related concerns. We also warmly welcome speech therapists and medical doctors to join us. This is a fantastic opportunity to make new friends, learn more about spasmodic dysphonia, and spread awareness. Awareness is crucial for early diagnosis, best treatment practices, funding for research, and, ultimately, finding cures.

To join the meeting, please click on the link or scan the QR Code.

https://us02web.zoom.us/meeting/register/EQh3NeGJTmGYAaHD_4w38Q

SD JOURNEY

By Marie Drumgoole

My name is Marie Drumgoole. In 2014, I was a busy full-time general practitioner in a mixed urban/ rural practice in Donegal town. I loved my job and although it was challenging work, I got enormous job satisfaction. I had 3 children who were nearly educated and certainly at age 53, I was right in the middle of a busy career with no intentions of retiring until 65.

I initially noticed a catch in my voice. I ignored it for a while, but it gradually began to interfere with my work and everyday activities. I started to struggle in noisy environments, on the phone, and when under pressure. When I opened my mouth to speak it was cracked, strangulated, low volume and very broken. It was intermittent at first, worse after a long day, if tired, if ill, and if under pressure or needing to speak on the phone. Gradually, it worsened until I was conscious of it all the time - like some nasty little gremlin lurking in my vocal cords, waiting to grab them and lock them together when I needed to speak.

It seemed ironic that I had always felt communication was one of my stronger personality traits. I did a lot of debating and public speaking, and enjoyed telling a good joke in good company. I just LOVED talking!

But, life is what happens when you are busy making other plans.

As a doctor, the tone of the voice is an essential tool to delivering proper care to the patients and their families. It is used to reassure and comfort, it's essential in emergencies to sound competent and in control, it's needed to relay information and give clear instructions to patients regarding treatments and medications and it is essential when giving bad news, to relay empathy and genuine feeling. All of this, I found, was becoming increasingly difficult. My consultations with patients were suddenly becoming about me and not the patient; "what's wrong with your voice Doctor?", "That's a terrible dose", "'have you tried honey and lemon?" The deaf elderly could not hear me. On one occasion, I was calling for an ambulance for a patient needing to be transferred to hospital. The person in ambulance control seemed more inclined to send the ambulance for me instead. I felt that as a primary care provider, my communication skills were gradually being eroded and leaving me feeling ineffectual and far from top of my game.

I started doing some research and managed to make my own provisional diagnosis. I called Dystonia Ireland on one of my busy days out on calls, when I was becoming increasingly frustrated at the struggle to talk and Maria Hickey was extremely helpful. She suggested some ENT and Neurology consultations.

Having received the official diagnosis of 'Adductor Spasmodic Dysphonia' from Mr. Niall Costigan (neurologist) in the Mater and Mr. Mark Rafferty (ENT) in St Vincent's, I was told the condition would worsen initially and then stabilise after 3 years. It was permanent. I went on a mission to try and fix it. After all, as a doctor, surely I could heal myself? I was most fortunate to get a quick diagnosis and I am aware that others are not so fortunate. In 2017 I made the difficult decision to come out of work, and so on January 14th I walked away, bag in hand, with the intention of taking 6 months, and returning when sorted.

I attended the Royal Victoria Hospital and received a trial of botox injections under Prof. Timon and his wonderful team. I went for speech therapy, attended a neuro-physiotherapist, tried kinesiology and homeopathy and even went to a healer, unfortunately with no major result.

In all my medical training I had never heard of Spasmodic Dysphonia and neither had any of my colleagues. Saying that, we would have been vaguely aware of the 'dystonias' although our medical knowledge would have been very limited.

My head was wrecked with the guilt of developing such a rare and little known condition that was inevitably going to end a very rewarding career and yet I was conscious of many others who continued to work with more serious and life limiting conditions. I was also conscious of the unfortunate people who suffer from the painful and debilitating dystonias, a much more difficult condition to cope with. I felt a little shame at even contemplating abandoning my post, my wonderful colleagues and lovely patients, but ultimately that is what I did.

Everybody's journey with SD is different. I was fortunate to have good illness cover and the background knowledge to move me forward. I also had wonderful support from family and friends. It can be a very scary and protracted journey for many and although they say it is a rare condition, I suspect the incidence might be rising.

I adjusted to the condition. I learnt to become a better listener, I shortened my sentences when speaking, and used facial expression, humour and hand signals when not understood. Initially I forced volume into my voice out of pure frustration, which certainly aggravated the condition. Now I am more chill and although I do not relish phone calls or crowded noisy environments, I try not to avoid them and pick my company carefully. I get tired when speaking for long periods and I don't tell jokes anymore.

Saying that, I have relished my free time and explored hobbies and activities that I never thought I would get a chance to enjoy. It took two years before I could honestly say that I had come to terms with SD and my premature retirement. After that transition, I started concentrating on activities that didn't necessarily need voice.

I have become a beekeeper, have helped publish and illustrate a book for children with anxiety, have become a competitive member of a dragon boat team, got a motor boat license, started sea swimming, walked the camino and play trad in our local.

So for all those struggling, **THERE IS LIFE WITH SD**. It does not need to define you. I have accepted the presence of my SD Gremlin. We will never be friends, but I have learnt that it is possible to have a relatively peaceful co-existence.

Marie Drumgoole

My SD Journey

by Yvonne Elliott Coyne

"Ah, have you a sore throat?"

I remember it vividly; the first time I was asked this question and became aware that I actually didn't have a sore throat! To this day I'm not sure why I answered yes – I knew I didn't have a sore throat or had i? I wasn't sure, my voice was breaking. Maybe it was nerves……

That was over 25 years ago. I have been asked that same question countless times over those years. My answer had always remained the same. I felt embarrassed. I wasn't sure what was happening to my voice. My voice breaking on a regular basis, certain words I suddenly couldn't 'get out'. I was always a bit shy in crowds or in the presence of strangers, so I convinced myself it was a nervous thing

Throughout those 25 years I managed my 'nervous problem' as best as I could. Excuses became the norm – I had a cold, a sore throat, a cough…. I discovered when I laughed it subsided – or at least wasn't as noticeable so I would converse whilst laughing as much as I could – within reason! Telephone calls were a nightmare to me – I struggled so much to answer a call and say 'Hello'.

A simple thing like saying 'Hello'. What was wrong with me? Was I that nervous? I became really anxious about being nervous. I began to avoid certain situations where I would need to talk.

Being a Holistic Therapist, I have studied stress and the effect of stress on the body, so I began working on my stress levels. Massage, Aromatherapy, Meditation, Breathing Exercises to name a few... All of them so relaxing and beneficial at the time but as soon as I would begin to speak, my voice continued to break.

I felt like a different version of myself, a quieter version... I felt trapped in my own head. I had so much to say but couldn't get the words out. I didn't like this 'New' me. I was always a yapper! In the comfort of my own circles, I would have talked for Ireland. I had worked dealing with the public all through the years, in a shop, a hair salon, as a receptionist in a big London company.

I had always been able to converse with people and answer 60 extensions at a reception desk.

What had happened?

For 20 odd years the 'old' me lived in my head… I would hold conversations in my head that were never said out loud. I would prepare every conversation in advance, answering the shortest way possible, choosing my sentences carefully, avoiding words that I knew I would struggle with, pausing in between sentences. I now had to stay away from crowded areas and situations if there was background noise as I struggled to project my voice. I emailed and texted rather than make a phone call. I had stopped attending courses and get-togethers with friends.

My voice was affecting my daily life! My anxiety was through the roof! This wasn't me!

I needed help!

5 years ago, I attended my local doctor for my anxiety. Life had become so difficult, the daily fears and frustrations – of talking!

My doctor sent me to ENT in Sligo which I thought to be very strange because I didn't have a sore throat!

A "Flexible Fibreoptic Laryngoscopy" exam whilst rhyming off vowels and repeating certain words revealed an involuntary spasm of the vocal cords. "Spasmodic Dysphonia"

I was diagnosed with "Adductor Spasmodic Dysphonia". I had a diagnosis. I had an actual physical condition. The tears flowed… I wasn't going mad, it wasn't all in my head, I wasn't bringing on this strangled / break in my voice, it wasn't nerves… It had a name; it was an actual thing!

I had never heard of it, it is a neurological disorder, very rare and at present there is no cure.

Botox into the vocal cords is an option to temporarily reduce spasms and improve voice quality.

I personally have not opted to take that route – as of yet. I feel if I can control my anxiety around my condition, I will be better able to manage and deal with my condition.

I searched for support groups. Surely, I couldn't be the only person with spasmodic dysphonia.

Being so rare, there is so little out there about this condition. Speech and Language Therapy helped with my breathing techniques and learning to relax more but my voice remained the same.

I have had some mixed emotions since my diagnosis, relief that this is not something I have brought on myself, fear in knowing I will have this for the rest of my life, frustration that I cannot 'just talk' like I used to and like everyone else can without having to think about it, and sadness that ill never be the 'old' me again – but I am learning to love the 'new' me. 'Old' me, 'New' me, I'm still 'Me'!

I contacted Dysphonia International just over a year ago and I can honestly say it has been the changing point for me. The support and resources they have available are second to none. Information, reading materials, online support groups and zoom calls with others who have this condition. I no longer felt 'alone'.

Dystonia Ireland have also kindly put me in contact with others who share my condition. It makes such a difference speaking with others who 'understand' the fears of talking.

Acceptance has been difficult, but I am so lucky to have such great support from my husband, my family and my friends, and reaching out to Dysphonia International and Dystonia Ireland has helped tremendously.

I am proud to say I am now an "Area Contact Leader" for Dysphonia International to act as a support and friend to others who have this condition. I have already contacted a handful of fellow SD sufferers here in Ireland and have set up a WhatsApp Support Group between us – a safe place where we can share our daily fears and frustrations and offer advice based on our own personal experiences.

A few of us got together and took part in the Dysphonia International "Walk For Talk" for awareness in October '24. A first time for Ireland taking part.

"Ah, have you a sore throat?"

No, I have "Spasmodic Dysphonia", an involuntary spasm of the vocal cords that affects my speech, but thank you for asking!

FUNDRAISING

VHI Women's Mini Marathon 2025

Please join Dystonia Ireland and thousands of amazing women on Bank Holiday Sunday 1st June 2025, for the VHI Women's Mini Marathon!

Whether you choose to walk, run, jog, or just sponsor your fundraising efforts will help us keep our organisation in operation and continue to fund our ongoing Dystonia Ireland Medical Research Projects. Rally your friends, family, or colleagues and be part of the largest one-day charity event in the country.

To participate in the VHI Women's Mini Marathon, you must register on the official VHI Women's Mini Marathon website.

General registration opens on Tuesday, March 4th, 2025. Make sure to register there to receive your race number and all relevant event information and materials.

Once registered, please contact Maria at 01 4922514, Email info@dystonia.ie Website www.dystonia.ie We will provide a Dystonia Ireland T-shirt for you to wear, post-race refreshments, sponsorship cards if needed, and assistance with setting up your online sponsorship page.

Pictured: Bernie Drumgoole, Marie Drumgoole, Caitriona Williams,Clodagh Mooney Duggan, Lauren Flanagan

NEWS

NAI Members Meet in Leinster House on the 5th March 2025

Dystonia Ireland is a member of the NAI and was represented at this meeting. The NAI organised a meeting of its member organisations and lived experience representatives from across the country today in Leinster House, calling for renewed commitment in delivering on the targets outlined in the 2019 Neurorehabilitation Strategy. NAI launched their call for the Government to invest in four community neurorehabilitation teams, promised since 2019 and committed to within the current Programme for Government.

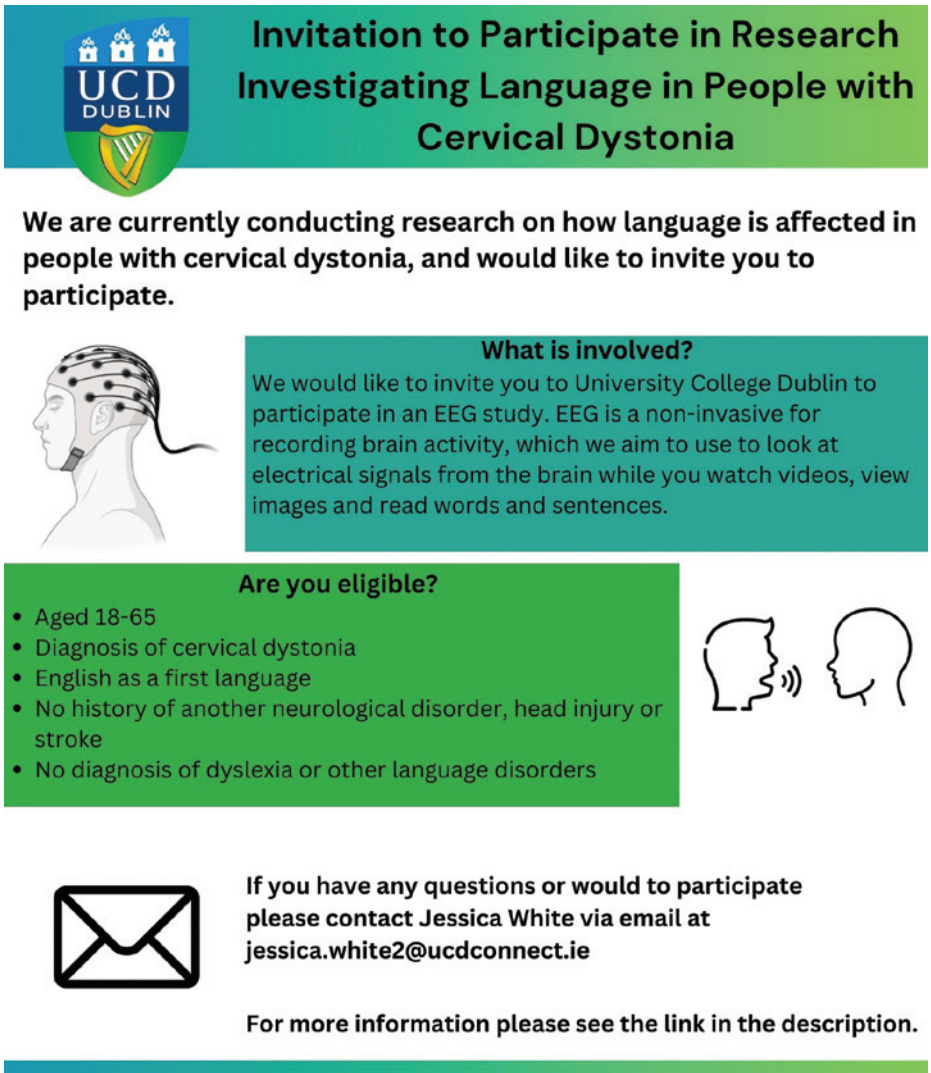
Over the coming months, NAI will organise a series of meetings highlighting the need for neurorehabilitation services across the country.

Invitation to Participate in Research Investigating Language in People with Cervical Dystonia also known as Spasmodic Torticollis.

A research team in UCD is currently conducting a study on how language is affected in people with cervical dystonia (CD) and would like to invite you to participate in the research. The study aims to use EEG (a non-invasive method for recording brain activity) to examine if there are differences in brain activity in people with CD while they view videos, images, words and sentences. You can read more about the study in the information leaflet below.

If you would like to participate or hear more about the study, please contact Jessica White (jessica.white2@ucdconnect.ie).

"Please see the following detailed information regarding the study and your participation in this research."



Participant Information Sheet

TITLE OF THE STUDY

Exploring the role of the motor system in language.

Researchers

Jessica White, UCD School of Psychology
Dr Patricia Gough, UCD School of Psychology
Dr Fiadhnait O'Keeffe, UCD School of Psychology
Prof Klaus Kessler, UCD School of Psychology

We would like to invite you to take part in a research study. The study is being conducted by Jessica White, Dr Patricia Gough, Dr Fiadhnait O'Keeffe and Prof Klaus Kessler of the UCD School of Psychology as part of a PhD research project. The purpose of this study is to understand if and how language is affected in people with cervical dystonia or persistent developmental stuttering.

What is the research about?

The present study aims to investigate how the motor system (area of the brain responsible for movement of your body) contributes to the perception of language. Particularly, we are interested in how language is affected in people who have a dysfunctional motor system such as people with cervical dystonia or persistent developmental stuttering.

Cervical dystonia (CD) is a chronic neurological condition characterised by involuntary intermittent/sustained contractions of the neck muscles. Persistent developmental stuttering (PDS) is a speech condition characterised by disruption in fluency, rhythm and timing of speech. Both conditions have their origin within

the motor system, but have symptoms beyond their typical movement and speech symptoms.

Specifically, we seek to use electroencephalography (EEG) while participants view a series of videos, images, words and sentences. EEG is a non-invasive method for recording brain activity. Please see the EEG information sheet provided. We are particularly interested in two aspects of language known as semantics and grammar. Semantics refers to our ability to understand the meaning of words and sentences; while grammar refers to our ability to understand and use structure in our language.

Why are we doing this research?

Although the primary role of the motor system is to prepare and execute movements, research has also shown that this system may play a role in other functions such as language perception and production. As a result, we are interested in researching how language may be affected by conditions which involve dysfunction of the motor system; ultimately aiming to explore if motor system dysfunction can lead to higher-order consequences beyond typical consequences for movement and speech.

Why have you been invited to take part?

People with a diagnosis of cervical dystonia or persistent developmental stuttering are invited to take part. However, there are exclusion criteria which apply to each group.

Participants with cervical dystonia or persistent developmental stuttering:

- Should have english as their first language
- Should be aged 18-65
- Should not have another neurological condition or history of head injury or stroke.
- Should not have a diagnosis of dyslexia.
- Should not have a diagnosed psychiatric condition which can affect language (i.e., schizophrenia)
- Should not have a diagnosis of autism (research suggests differences on the EEG markers we are measuring)

STUDY PROCEDURE: What will happen if I choose to participate?

If you choose to participate, you will be invited to attend University College Dublin to take part in the study.

1. You will be asked to provide some information about yourself such as your age, gender and level of education.
2. You will be then asked to read a short extract and talk about a topic of your choice, while your voice is being recorded. This is so that we assess your fluency of speech, that is the number of pauses, hesitations or repetitions made while speaking. This will be calculated immediately after you leave the session and the audio recording will be immediately deleted.
3. You will be asked to complete a short pen and paper task which measures executive function. Executive function refers to a set of skills required to control, monitor and adjust behaviour to achieve a desired goal. You are asked to do this, as there is some research to suggest that executive function is closely related to grammatical ability.
4. Finally, you will be connected to the EEG machine and sit in front of a computer monitor to view a number of videos, images, words and sentences. You can read more about what is involved in EEG in the EEG information section at the end of this document.

a. In this task, we aim to examine how the electrical activity in the brain changes in response to your viewing of these videos, images, words and sentences.

This study is part of a larger PhD project and there will be further studies you may be interested in participating in. On the day we will ask you if you are interested in hearing about future studies. If so, we will ask for your contact details. This is completely optional. If you consent to being contacted, you are under no obligation to participate in future studies, you will just be contacted about them; at which point you can decide to refrain from participating.

We will also ask you to consent to your data from the current study, to be used in future studies if you should decide to participate. This is because the data you provide in this may be related to data we collect at a future date. Additionally, we may collect some of the same information in future studies.

PERSONAL DATA COLLECTION, USAGE, STORAGE AND DISCLOSURE

How will your data be used?

Data collected will be analysed and reported in a PhD thesis; and will be included in scientific publications and presentations. Data used for these purposes will be anonymised (not identifiable as you).

OPEN DATA SHARING

To promote open science, your anonymised data will be shared with the scientific community via an open access database on the internet. The data shared on this database cannot be traced back to you. Data sharing allows researchers to replicate findings or reuse data to answer another research question.

How will your privacy be protected?

Your data is strictly confidential. Your data will be stored linked only to you by your participant code on a password protected device.

Your data will be stored in pseudonymised form (name replaced with a code) on a password protected device. Your name will be linked to this code in a separate password protected document on the device.

Your participant code is linked to your name, so that if you decide to participate in any future studies, we can link your data sets.

Your name is also linked to the

code so that we can identify your data in the event you wish to access or delete your data. You will have 6 months to request the full deletion of your data. After which, anonymised results will have been shared in scientific publications and uploaded to an online database. At this point, we cannot trace your data back to you so it cannot be deleted. Local copies linked to you by your participant code will be stored for 3 years until completion of the PhD. After 3 years, your data will be stored in a fully anonymised format. Anonymised data uploaded to an online repository will be available indefinitely.

During data analysis, your data will be pseudo-anonymised (name replaced with a code) and stored on a password protected device. Only authorised researchers will have access to the data.

Are there any benefits to taking part?

The research performed with your participation may help us to better understand the contribution of the motor system to language and how this relationship is affected in people with CD and PDS.

Are there any risks or discomforts associated with the research?

EEG is non-invasive and very safe with no known biological risks. Saline gel will be inserted into the electrodes in the EEG cap during the EEG session and will remain in your hair. However, hair washing facilities will be provided at the end of the session. There is a low risk of irritation on the point of contact between the scalp and the saline gel. Please tell the researcher if you feel any discomfort during the session. The gel will be applied with a blunt syringe with NO needle.

You may experience some discomfort or fatigue as you spend a prolonged period sitting still (Approximately 35 minutes) and concentrating on a computer screen. You will be given regular breaks to minimise this discomfort.

All necessary measures will be taken to minimise the spread of COVID-19.

Can you change your mind at any point and withdraw from the study?

Participation in the study is completely voluntary. You do not have to participate and withdraw anytime without justification or consequence. Should you initially choose to participate but later decided to withdraw from the study, your data can be destroyed and withheld from analysis. Please note that once a paper with anonymised results has been published it cannot be retracted. Anonymised data shared on online repositories cannot be deleted, as we can no longer link it back to you. However, we will delete any local copies of your data. If you wish for your data to be deleted before anonymised data is published or uploaded to online repositories, please contact the researchers within 6 months of your participation.

Will I receive the results?

If you wish to find out about the results of the study, you can contact the researchers listed on this information sheet and we will be happy to provide a summary of results. Please note that individual results will not be available.

How is this study being funded?

This study is funded by the Irish Research Council under the Government of Ireland Postgraduate Scholarship.

ETHICAL APPROVAL

Ethical approval for this study was granted by the UCD Human Research Ethics Committee (Project ID: HS-24-35-White-Gough-Kessler).

If you have any further questions, please contact
Jessica White - jessica.white2@ucdconnect.ie
Dr. Patrica Gough - patricia.gough@ucd.ie
Dr. Fiadhnait O'Keeffe – fokeeffe@ucc.ie
Prof Klaus Kessler – Klaus.kessler1@ucd.ie

Information and support:

Stammering Ireland Website:
https://www.stammeringireland.ie/

Dystonia Ireland Website:
https://www.dystonia.ie/support/

Dystonia Ireland Helpline:
(01) 492 2514

Empowering Connections Social Event

27TH March 6pm to 8.30, Trinity Comprehensive Secondary School, Ballymun main hall

You are Strong
You are Powerful
You are Determined
Despite any obstacles that you may have

You CHOOSE to keep going and THRIVING...

We are inviting you to our Empowering Connections Social Event.

What is the event about??

It's about bringing ALL ABILITIES TOGETHER, to grow individually and collectively.

To EMBRACE OUR OWN UNQUIE STRENGTHS AND CAPABILITIES, Share stories, laughter and creativity

I really feel it's so IMPORTANT TO be around people who want to EMPOWER themselves and WANT more from life

To not feel isolated but inclusive and YOU TRULY DESERVE TO BEEN SEEN AND HEARD

A space in grow in Confidence build new friendships with good connections

Feel VALUE when you Show up right

Give yourself something to look forward too every few weeks and have fun

When is it happening?

Date 27TH March 6pm to 8.30
Venue Trinity Comprehensive Secondary School, Ballymun main hall

Who is it for?

The Empowering Connections Social Event is for ALL ABILITIES TO SHINE

Tracey McCann

REMEMBER YOU ARE EQUAL.

This event is ideal for you. Come along and listen to our inspiring speakers and musician, mingle, chat and make new connections. This is a safe space to be YOURSELF DEFINITELY and EXPAND

SO Have you got your ticket yet?

WE'D LOVE TO SEE you there

https://www.eventbrite.com/e/empowering-connections-tickets-1117609623469

The Dystonia UK Awards – Nominations

Who is your Super Star or Masked Hero?

Dystonia Ireland are pleased to announce that they have been invited for the first time to take part in the following two categories of The Dystonia UK Awards. The awards are four years old this year. These awards are your chance to nominate the incredible individuals you want to celebrate in the Republic of Ireland.

You have until Monday 24th March 2025 to submit your nominations.

Link for people to make their nominations: **https://www.dystonia.org.uk/Pages/Category/dystonia-uk-awards-nominations**

For anyone wishing to come to awards this is the link to book their place: https://www.tickettailor.com/events/dystoniauk/1426400

Scan the QR Code to vote

Dystonia Superstar

The Dystonia Superstar Award is your chance to celebrate an extraordinary individual from the dystonia community who has made a meaningful difference. Whether they have dystonia themselves or not, you have the power to decide what makes them a true Dystonia Superstar.

We're excited to see lots of brilliant individuals from England, Scotland, Wales, Northern Ireland and the Republic of Ireland.

Masked Hero

The Masked Hero Award recognises a medical professional who has had a wonderful impact on your life, from doctors and nurses, to physiotherapists and other professionals who are dedicated to helping the dystonia community. We want to celebrate those special people in the UK and Republic of Ireland whose care, attention and commitment have gone above and beyond.

For those of you who don't have access to a computer we have included these articles in our newsletter. The latest Dystonia Europe Newsletter can be viewed at **www.dystonia-europe.org**

RESEARCH

Analysis of Movement Disorders in Cervical Dystonia Through Three Clinical Studies

Jean-Pierre Bleton, Doctor in Human Movement Sciences, Researcher, physiotherapist, Adolphe de Rothschild Foundation Hospital (Paris)

1. Biomechanical analysis of cervical rotation movement

Publication of the study funded by the Breughel grant (AMADYS)

Clinical Biomechanics 107 (2023) 106037

Contents lists available at ScienceDirect

Clinical Biomechanics

journal homepage: www.elsevier.com/locate/clinbiomech

Assessment of axial rotation movement in cervical dystonia using cone-beam computed tomography

Jean-Pierre Bleton [a,b,*], Raphaël Portero [b], Kévin Zuber [b], Sophie Sangla [a], Jean-Philippe Brandel [a], Marie Vidailhet [c,d,g], Serge Mesure [e], Marc Williams [f], Julien Savatovsky [f]

[a] Neurology Department, Hôpital Fondation Adolphe de Rothschild, Paris, France
[b] Clinical Research Department, Hôpital Fondation Adolphe de Rothschild, Paris, France
[c] Sorbonne Université, F-75005 Paris, France
[d] Assistance Publique-Hôpitaux de Paris, Hôpital Pitié-Salpêtrière, Paris, France, Department of Neurology, Groupe Hospitalier Pitié-Salpêtrière, 47 boulevard de l'Hôpital, F-75013 Paris, France
[e] Aix-Marseille University, CNRS, ISM UMR 7287, F-13288 Marseille, France
[f] Department of Radiology, Hôpital Fondation Adolphe de Rothschild, Paris, France
[g] Inserm U1127, CNRS UMR 7225, UM 75, ICM, F-75013 Paris, France

The study on "*Assessment of axial rotation movement in cervical dystonia using cone-beam computed tomography*" involved researchers from the A. de Rothschild Foundation Hospital (Jean-Pierre Bleton PT, PhD, Kévin Zuber MSc, Sophie Sangla, MD, Jean-Philippe Brandel MD, Marc Williams MD, Julien Savatovsky MD), Paris-Est Créteil University (Raphaël Portero PhD), Salpêtrière Hospital and Brain Institute (Marie Vidailhet MD, PU-PH) and Aix-Marseille University (Serge Mesure PT, PhD).

It was published in the July 2023 issue of *Clinical biomechanics*.

Cervical dystonia is characterised by localised abnormal movements and/or postures of the head and neck. A general consensus defines four unidirectional dystonic patterns of the head and neck: torticollis (axial rotation in the transverse plane), laterocollis (lateral bending in the coronal plane), and retrocollis or antecollis (extension or flexion in the sagittal plane).

The cervical dystonia analysis grid proposed by Reichel Gerhard (*Basal Ganglia, 2009*) combines biomechanical analyses of the cervical spine with neuro-pathological knowledge of cervical dystonia. It distinguishes two subtypes: the upper cervical spine subtype (caput or CAP) and the lower cervical spine subtype (collum or COL). The CAP subtype covers the upper cervical spine (from the occipital bone to the second cervical vertebra, or axis), while the COL subtype covers the lower cervical spine (from the third to the seventh cervical vertebra).

The aim of this study was to analyse the location of bad positioning and head rotation difficulties induced by cervical dystonia, distinguishing between segments of the cervical spine (COL and/or CAP).

Analysis of neck and head postures and movements was carried out using a cone beam scanner. This X-ray imaging technique provides 2D and 3D images similar to those obtained with conventional scanners, with the advantage that they can be obtained in a seated position (i.e. in a natural posture).

Twenty volunteers took part in the study: ten suffering from cervical dystonia with rotatory predominance (study group) and ten without dystonia (control group), matched for age, sex and laterality. Their postures and cervical rotation movements were measured at each joint complex in three situations: head in natural position, right rotation and left rotation.

The results showed that:

1. Posture: In the natural position, the total cervical spine is significantly further from the neutral position (axis of symmetry) in people with cervical dystonia than in the unaffected control group.

2. Range of rotation: The range of rotation of the cervical spine is significantly reduced in participants with cervical dystonia, particularly in the upper cervical spine.

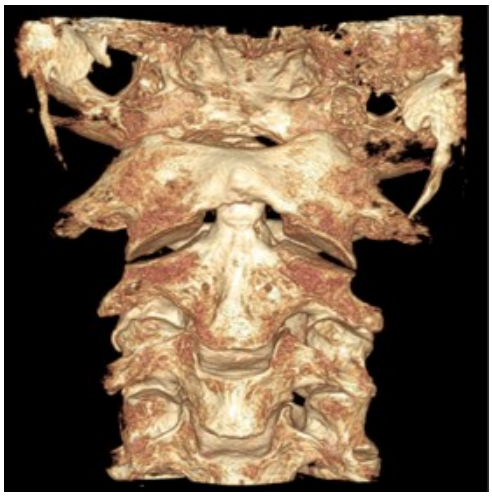
Conclusion:

These results confirm that it is essential to take into account the involvement of rotator muscles at the upper cervical level in treatments with botulinum toxin injections and when designing rehabilitation programs.

Front view of the cervical region from the base of the occipital bone to the 4th cervical vertebra (C4), showing the alignment of the cervical region in a non-dystonic person (left view) and the dystonic rotation between the atlas (C1) and axis (C2) in a person with cervical dystonia (right view).

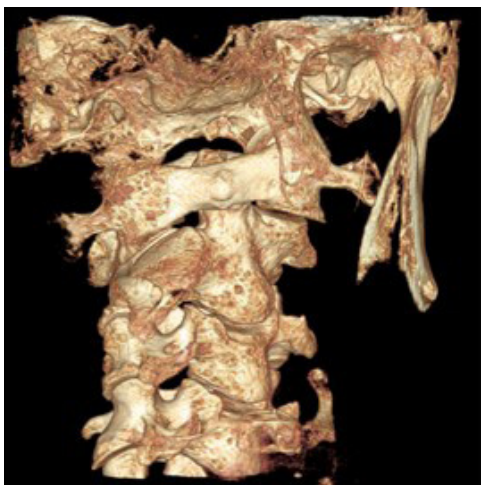
The results of this study underline the importance of analysing and understanding the role of the muscles responsible for rotation between the Atlas (C1) and Axis (C2) vertebrae, in particular that of the inferior oblique head muscle (OCI). A new study is currently underway to investigate this further (see *clinicalTrials.gov - NCT: 05327985*)

Non-dystonic person front view



Dystonic person front view

(right rotatory torticollis)



RESEARCH

2. Perception disorders of the straight-ahead position

Annals of Physical and Rehabilitation Medicine
Volume 66, Issue 8, November 2023, 101753

Letter to the editor

Repositioning errors of the head in straight-ahead position in cervical dystonia: Influence of clinical features and movement planes

Jean-Pierre Bleton [a b], Sophie Sangla [a], Raphaël Portero [b], Dominique Garric [c], Vincent Guiraud [d], Pierre Portero [c], Jean-Philippe Brandel [a], Marie Vidailhet [e f g h], Serge Mesure [i]

The study on "*Repositioning Errors of the head in straight-ahead position in cervical dystonia: Influence of clinical features and movement planes*" involved researchers from the A. de Rothschild Foundation Hospital (Jean-Pierre Bleton PT, PhD, Sophie Sangla, MD, Jean-Philippe Brandel MD), Paris-Est Créteil University (Raphaël Portero PhD, Dominique Garric PhD, Pierre Portero PhD, PU-PH), University Hospital Group Paris psychiatry and neurosciences (Vincent Guiraud), Salpêtrière Hospital and Brain Institute (Marie Vidailhet MD, PU-PH) and Aix-Marseille University (Serge Mesure PT, PhD). It was published in the journal *Annals of Physical and Rehabilitation Medicine* in November 2023.

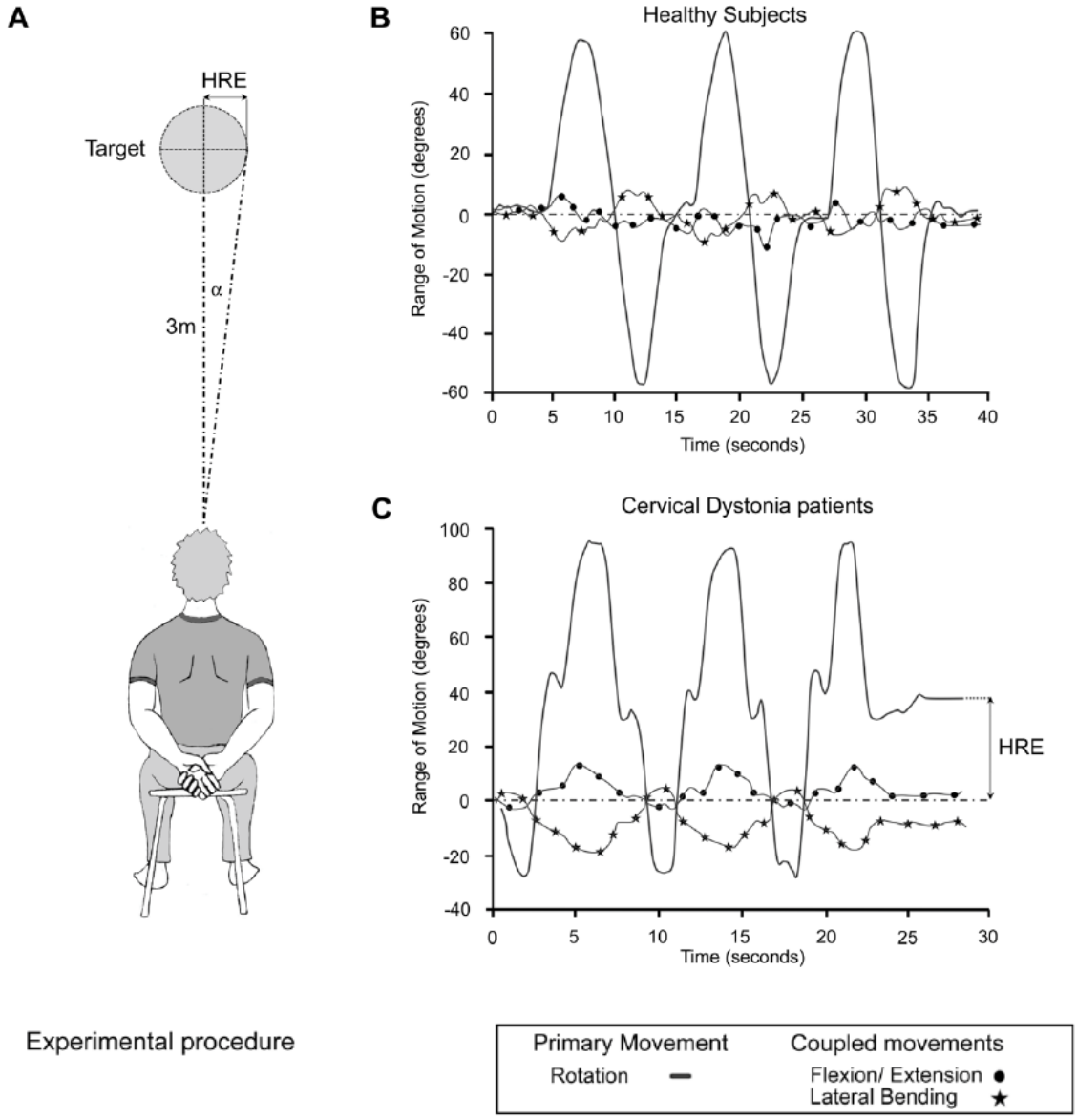
The most visible symptoms of cervical dystonia are bad head positioning and uncontrolled neck movements. But other systems can also be affected, such as the system for sensing and integrating information on body position, known as proprioception.

Proprioception is the ability to perceive and regulate the position of different parts of the body without using sight. In the case of cervical dystonia, perception of the position and movement of the head in space may be impaired.

The present study sought to understand the impact of cervical dystonia on proprioception. To this end, researchers assessed the accuracy with which participants could reposition their heads to a neutral position (straight ahead) with their eyes closed. 29 people with cervical dystonia and 29 without participated in the study. Participants were seated in a rigid chair, with their backs resting against the backrest. Each participant's 3D head position was measured using a small electronic sensor placed on the forehead, capable of measuring head orientation and movement. To obtain the initial neutral head position, participants had to look at a target 3 meters away at eye level. Then, with their eyes closed, they had to move their head and try to return to the initial position.

The results showed that people with cervical dystonia made greater repositioning errors than those without, particularly in the rotational plane for those with torticollis and in the sagittal plane for those with antecollis. These results confirm that rotation is a major feature of cervical dystonia.

They also show the importance of analysing this condition in its three-dimensional aspects, and of proposing proprioception rehabilitation programs to complement botulinum toxin injections and motor rehabilitation.



A- Experimental position

B- Rotational displacement and replacement in neutral position with eyes closed of a control subject

C- Same test performed by a person suffering from cervical dystonia, showing a replacement error (HRE - Head Repositioning Error)

RESEARCH

3. Cerebellum and neuroplasticity: a path to be explored to acquire the right movement

Front Neurol. 2024; 15: 1381390.
Published online 2024 Apr 30. doi: 10.3389/fneur.2024.1381390
PMCID: PMC11091337
PMID: 38746658

Combination of anodal tDCS of the cerebellum with a goal-oriented motor training to treat cervical dystonia: a pilot case series

Jean-Pierre Bleton, [1] Charlotte Cossé, [2] Tiphanie Caloc'h, [2] Alcira Suarez Moreno, [2] Elisabeth Diverres, [2] Pascal Derkinderen, [3] Julien Nizard, [4,5] Jean-Pascal Lefaucheur,[✉5,6,*] and Jean-Paul Nguyen [2]

The study on "*Combination of anodal tDCS of the cerebellum with a goal-oriented motor training to treat cervical dystonia: a pilot cases series*" involved researchers from the Parkinson's Unit/Neurology Department of the A. de Rothschild Foundation Hospital (Jean-Pierre Bleton PT, PhD), Transcranial Stimulation Unit, Bretéché Clinc, Elsan Group, Nantes (Jean-Paul Nguyen MD, PU-PH, Charlotte Cossé, Tiphanie Caloc'h, Alcira Suarez Moreno MD and Elisabeth Diverres PT), Neurology Department, Nantes University Hospital Center (Pascal Derkinderen MD, PhD, PU-PH), Pain, Palliative and Support Care Department and UIC22, Hôpital Laennec Hospital, Nantes University Hospital Center (Julien Nizard MD, PhD, PU-PH), Clinical Neurophysiology Unit, Henri Mondor Hospital, Créteil (Jean-Pascal Lefaucheur MD, PhD, PU-PH). It was published in the journal *Frontiers in Neurology* in April 2024.

Cerebral plasticity, or neuroplasticity, is the brain's ability to remodel itself in response to experience or learning. The cerebellum plays a crucial role in brain plasticity and motor learning.

Dystonia is associated with poorly adaptive plasticity.

Transcranial Direct Current Stimulation (tDCS), which applies a small, constant electrical current to the scalp to modulate neuronal excitability, could improve symptoms of dystonia by altering the excitability of neurons in the cerebellum and basal ganglia. This could help restore more normal plasticity.

In this pilot study, tDCS of the cerebellum was combined with specific motor rehabilitation programs (motor training) in people suffering from cervical dystonia poorly controlled by botulinum toxin injections.

The first phase involved administration of the tDCS protocol alone (three or five daily 20-minute sessions over one week). The second phase combined administration of the tDCS protocol (also three or five daily sessions over one week) with motor training. The 20-minute motor training protocol was adapted to the clinical characteristics of each patient and carried out during the tDCS sessions.

Analysis of the study results revealed that a personalised motor rehabilitation program combined with tDCS of the cerebellum resulted in significant and longer-lasting improvements than stimulation alone.

This study suggests that combining tDCS with a personalised motor rehabilitation program could promote more adaptive brain plasticity, thus contributing to the reduction of motor symptoms. However, further studies involving larger numbers of subjects are needed to confirm these results.

The researchers would like to express their sincere thanks to the people with cervical dystonia who agreed to take part in these studies. Their contribution is invaluable. We would also like to thank the AMADYS Association for its unwavering support for research, without which these advances would not have been possible.

AMADYS and DYSTONIA EUROPE would like to thank Jean-Pierre Bleton for providing patients with a comprehensible summary of these scientific articles.

Translated from French to English by E. Ponseel and G. Ainsley.

Project Cervical Dystonia Patient Journey Update

The work on the project Cervical Dystonia Patient Journey continues. It all started four years ago when we developed a patient journey/patient experience map for people living with cervical dystonia in Europe.

It is being translated into other languages and these are all available on the ERN-RND website.

https://www.ern-rnd.eu/disease-knowledge-hub/dystonia/

There you find the cervical dystonia patient journey in: English, Czech, Dutch, Finnish, French, German, Hungarian, Italian, Norwegian, Polish, Portuguese and Spanish. The goal is to have it translated to most European languages.

Last year at the EAN Congress in Budapest an actress performed the Cervical Dystonia Patient Journey in the Scientific Theatre. The ERN-RND team thought this was so good that they decided to record it in a more professional way.
At our annual meeting in Timisoara, we were delighted to show for the very first time "Lilly's Journey" – a 25-minute long video with actress Carmel Stephens from Ireland.

She is doing a wonderful interpretation of the many stages of the patient journey such as: what it is like to discover the first strange symptoms of dystonia, the struggle to find out what it is, the search for the best treatment and how to learn to live with it.

You find the video here:
https://www.youtube.com/watch?v=PxJmQA1ewi0

We hope this video will help to raise awareness and spread knowledge about dystonia.

Monika Benson
Executive Director
Dystonia Europe

MY DYSTONIA STORY

Meet Mikko from Finland

My name is Mikko Juutinen. I am 43 years old, and I live in Muhos, Finland. I have a wife Riikka and we have two children. I am a dentist, but I haven't done any clinical work for almost two and half years. I am specialising in healthcare. I work as a specialist dentist. I am responsible unit manager for wellbeing in the county of North Bothnia. I also work as a part time university teacher in the University of Oulu. I teach dentistry students in my specialised field. Also, I teach first- and second-year students about what it is like to work as a dentist and how to connect with patients.

My first symptoms of Dystonia were with my left thumb in 2017. It started during patient work. First it was a few times a day but in a few weeks the symptoms were worse and I had to go to an occupational health doctor. First, she sent me to a hand surgeon and after that my hand was getting MRI-examination. There wasn't anything that caused the symptoms so next I went to a clinical neurophysiologist, and she examined me and also found nothing which caused my symptoms. She said it could be a neurological condition. My occupational health doctor didn't know what was wrong with me either and at the same time symptoms started with my right hand thumb. I was in a seminar where we were having dinner and there was a neurologist on the same table. We were having coffee with cognac, and I asked about my symptoms and that clinical neurophysiologist also said it could be a neurological condition. Right away I was told my symptoms were neurological and that it is task-specific dystonia which can be treated with botulinum toxin injections. Next week I visited my occupational health doctor, and she said to me that she doesn't know at all what is causing the symptoms. I told her about the talk with a neurologist who said that it is task-specific dystonia. After that she called the university hospital clinic of neurology and sent me there.

It took almost a year before my first injections. They wanted to do head and neck MRI-examinations and take a lot of blood tests. During that year both of my big toes started to have the same symptoms as my thumbs. In 2019 when I first got my injections, my neurologist and I were looking in an anatomy book to determine the correct muscles to be injected to allow me to continue my work as a dentist.

I have been injected four times a year since then. My dystonia symptoms have also spread, my both calves and 2nd-finger and upper back are affected and now on my injection list. Now a total of 12 muscles are injected. My cousin also has dystonia. He has cervical dystonia and spasmodic dysphonia. We live in different areas, but we can support each other.

I struggled with my work for three years, and it started to get harder and harder. Sometimes I thought that I might be more of a danger to the patient than a help. But it was such an important thing for me to work as a dentist and I liked that a lot.

From left: Mikko Juutinen from Finland, Vidar Bjørkli from Norway and Jukka Sillanpää from Finland

During those years I had to re-think my occupation and I decided to apply for specialised studies. Now I don't have to do any patient work but still have a lot of knowledge from the years I worked with patients and I can use that experience in my work today.

My hobbies have also changed with dystonia. Before I was a runner, but now I can't take two running steps. I have learned to make knives and sometimes I think I should try that again. I am a big fan of football and Arsenal is my club. Scouting has been my hobby for a decade, and it is still my most important hobby. Also, organisation work is my hobby. When I got diagnosed with Dystonia I became a member of the Finnish dystonia association. I was a member for a few years when they asked me to become a member of the board. I was a member of the board for three years when I was asked to become the next president of the Finnish dystonia association. I have been a delegate on Dystonia Europe's Ddays in Copenhagen, Dublin and Timișoara. It has been a good time every time and it has been a privilege to get to know people around Europe with dystonia. At the beginning of 2024 I started a new role as President of the Finnish Dystonia Association and it has been a busy year! Also, I have done my specialised studies during this year. Luckily there is only one year left with this study.

Mikko Juutinen
President Finnish Dystonia Association

Dr. Stavros Tsagkaris Receives the David Marsden Award 2024

Our congratulations to Dr. Stavros Tsagkaris, from the Evelina London Children's Hospital in London, UK and the David Marsden Award 2024 for his paper: Metabolic Patterns in Brain 18F-fluorodeoxyglucose PET Relate to Aetiology in Paediatric Dystonia https://dystonia-europe.org/wp-content/uploads/2024/07/tsagkaris-et-al-brain-656e1e43a6c91253670147.pdf

On Saturday 29 June Dr. Tsagkaris was presented with the award by Dystonia Europe Executive Director Monika Benson at the Basal Ganglia Session at the 10th EAN (European Academy of Neurology) Congress in Helsinki, Finland. Dr Tsagkaris expressed his thanks to Dystonia Europe for the award and then presented the work of his research.
Monika Benson, Executive Director of Dystonia Europe says:

"We are delighted that this year's award goes to Dr. Tsagkaris for his important research on children with dystonia, since this is a field of research within dystonia that needs much more focus. We wish Dr. Tsagkaris good luck and look forward to follow his work".

About the winner:

Dr Tsagkaris is a senior Paediatrics trainee in London, UK. He is an aspiring Paediatric Neurologist, entering Neurology training.

Dr Tsagkaris completed his medical degree in National and Kapodistrian University of Athens, Greece. He completed a clinical research fellowship at Evelina London Children's Hospital Complex Motor Disorders Service under the supervision of Dr Jean-Pierre Lin, in collaboration with King's College London PET Imaging Centre, as a member of Professor Alexander Hammers' team.

His ongoing research work focuses on the field of movement disorders and particularly dystonia in children, focusing on neuroimaging, neurophysiology and neuromodulation. Dr Tsagkaris' special interest is functional neuroimaging in children with dystonia, aiming to deepen our understanding of the pathophysiology of the disease.

Dr. Stavros Tsagkaris, Evelina London Children's Hospital in London, UK and David Marsden Award Winner 2024

RESEARCH

About the research:

The exact mechanisms via which dystonia symptoms develop are not clearly understood. Brain imaging is a great way to unravel abnormalities that could be related to the disease. There has been extensive research on the structural brain abnormalities related to development of dystonia. However, there is a relative lack of imaging markers revealing the functional characteristics of different brain regions in dystonia, especially in the paediatric population. One way to assess the function of brain areas is Positron Emission Tomography (PET) scanning, where a glucose analogue is injected into the bloodstream and brain images are taken after absorption, to reflect the glucose uptake in the brain.

Dr Tsagkaris along with his colleagues examined the glucose uptake patterns in the brains of patients with different types of dystonia, including genetic as well as acquired dystonia cases. The researchers found that, on the one hand, most aetiologic subgroups shared some patterns of glucose metabolism, possibly reflecting innate characteristics of dystonia. On the other hand, the different groups also had “signature” characteristics, which distinguished them from the rest and therefore pointed towards distinct pathophysiological mechanisms.

The results offer further insights into the pathophysiology of dystonia, enhancing our understanding of the underlying biological mechanisms related to the disease. The various areas of altered brain metabolism revealed from this study relate to disease phenotype, suggesting their potential function on controlling movement and posture. They also introduce the possibility of PET scanning imaging patterns potentially being used as a biomarker for the various disease groups in the future.

Moreover, in combination with information obtained by structural neuroimaging as well as neurophysiology testing, this research work can help better inform decisions on management options, including deep brain stimulation, providing more individualized patient care.

About the award:

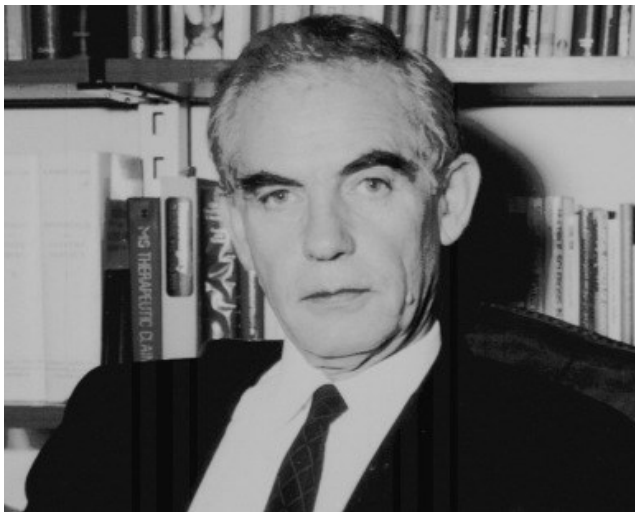
The Award was an idea of late former President of European Dystonia Federation Didi Jackson from Germany.

Late Professor David Marsden was the Dean of the Institute of Neurology in Queen Square in London and he was one of the founding members of the Movement disorder society. Many movement disorder experts across Europe and beyond first learned about dystonia from Professor Marsden. He especially encouraged young scientists to become involved in the field. When Dystonia Europe was founded in 1993 Professor Marsden gave us a lot of good advice, and took a genuine interest in the development of our organisation.

It was therefore entirely appropriate that David Marsden should be honoured by an Award given by patients in his name.

The David Marsden Award was first presented in 2003 to Professor Mark Edwards. Since then there has been a total of 12 awards. In the beginning the award was every other year but from 2020 it is an annual award. Next call for applications for the David Marsden Award of 2025 will start from 1 September.

Monika Benson
Executive Director
Dystonia Europe



Prof C David Marsden (1938 – 1998) .

Discovering a Missing Piece of the Dystonia Jigsaw: An International Collaboration on Understanding Brain Glucose Activity Patterns in Children and Young People with Dystonia

By Dr Jean-Pierre Lin, Evelina London Children's Hospital, London, UK

The award of the Dystonia Europe David Marsden Prize to Dr Stavros Tsagkaris represents a significant recognition of the importance of our work in investigating brain activity patterns in childhood and adolescent dystonias task[1], [2] representing a 20-year project to carefully unravel the mechanisms of dystonia in children and young people.

Setting up a children's dystonia diagnostic and management service:

In 2005 the embryonic Complex Motor Disorders Service (CMDS) at the Evelina London Children's Hospital set up a Multidisciplinary Team (MDT) to deliver a Deep Brain Stimulation (DBS) Service for children with dystonia and other movement disorders in collaboration with neurosurgical colleagues: Mr Richard Selway, Prof. Keyoumars Ashkan and latterly Mr Harutomo Hasegawa from the King's College Hospital Neurosurgery Department.

In 2008 Dr Jean-Pierre Lin was awarded a Guy's and St Thomas' Charity New Services and Innovation Grant G060708 for pump-priming support to build a dedicated CMDS MDT Team, leading to the current clinical-research structure.

Dystonia Brain Function:

Given the relative rarity of individual causes of dystonia in childhood the CMDS MDT set out to gather the broadest possible information that could result in a better understanding of the impact of dystonia on function, comfort, growth and development and well-being in children and young people, by collecting very detailed clinical information on each child. Together with dedicated scales to measure function, dysfunction and dystonia alongside genetic, metabolic, imaging and neurophysiological parameters.

Neurophysiology of dystonia:

Dr Verity McClelland spearheaded neurophysiological testing of the *central motor conduction time* **(CMCT)**[4] with transcranial magnetic stimulation (**TMS)** and evaluation of the sensory pathways from *somatosensory evoked potentials* **(SEP)**[4]**.** This was followed by simultaneous brain and muscle activity patterns during simple manual tasks[5].

Brain Activity Patterns in childhood dystonia:

Two imaging techniques were used to study the brain of children with dystonia: MRI brain imaging, led by *Dr Daniel Lumsden*, produces detailed structural brain images. *Fluorodeoxy- Glucose Positron Emission Tomography* brain scans fused with CT imaging **(FDG-PET-CT)**, led by *Dr Jean-Pierre Lin* and *Prof. Alexander Hammers*, allow us to study brain activation patterns in dystonia.[6,7]

How investigations and clinical evaluation are combined in assessing childhood dystonia:

Dr Stavros Tsagkaris studied the similarities and differences in regional brain glucose activity (i.e. glucose uptake and metabolism) in quietly awake children with dystonia which has shone a new light on the similarities and differences between different causes of dystonia when compared to healthy controls without dystonia.

This international multidisciplinary collaboration has enabled an understanding of direct brain activity assessment in childhood dystonias.

Future directions:

The very large comprehensive dataset collected by the CMDS team over the last 20 years will allow careful unravelling of the many factors underpinning childhood-onset dystonias by describing and following up brain activity patterns (glucose consumption). With this data we can track the impact of age, timing and aetiology (i.e. causes) of dystonia.

Dr. Stavros Tsagkaris and Dr. Jean-Pierre Lin,
Evelina London Children's Hospital in London, UK

Future applications of FDG-PET-CT brain imaging will include 'before' and 'after' dystonia intervention imaging studies to evaluate the impact of advanced therapies such as deep brain stimulation or gene therapies on childhood brain activity patterns.

We intend to build on previous data[7] to characterise individual brain energy consumption patterns to fully explore the impact of the many variables, singly and in combination, underlying the similarities and differences in dystonia, expressed in all age groups, and relative responsiveness to DBS and other advanced therapies.

Our aim is to generate new hypotheses regarding causal mechanisms of dystonia leading to new management strategies.

From left:
Executive Director Dystonia Europe Monika Benson,
DMA winner Dr. Stavros Tsagkaris and Dr. Jean-Pierre Lin

Acknowledgements:

Dr Jean-Pierre Lin supervised the clinical data collection; *Professor Alexander Hammers*, head of the King's College London PET Centre at St Thomas's Hospital, supervised the FDG-PET-CT analyses and *Dr Eric Guedj* of the Unveristé of Aix Marseille, France, kindly provided the healthy adult control FDG-PET-CT brain scan data.

Funding and support:

J-P Lin: Guy's and St Thomas' Charity New Services and Innovation Grant G060708; Dystonia Society UK Grants 01/2011 and 07/2013 and Action Medical Research GN2097; GSTT Charity Complex Motor Disorders Service Research and Education Fund A13.

A Hammers: Wellcome EPSRC Centre for Medical Engineering at King's College London (WT 203148/Z/16/Z) and also from the Department of Health via the National Institute for Health Research (NIHR) comprehensive Biomedical Research Centre award to Guy's & St Thomas' NHS Foundation Trust in partnership with King's College London and King's College Hospital NHS Foundation Trust.

V. McClelland: is currently supported by a Clinician Scientist Fellowship from the Medical Research Council UK (MRP0068681) and by a Continuation Funding grant from the Rosetrees Trust (CF-2021-2-112) and was previously supported by a postdoctoral clinical research training fellowship from the Medical Research Council UK (MRP0068681) and a project grant from the Rosetrees Trust (A1598).

1. Stavros Tsagkaris, Eric K C Yau, Verity McClelland, Apostolos Papandreou, Ata Siddiqui, Daniel E Lumsden, Margaret Kaminska, Eric Guedj, Alexander Hammers, Jean-Pierre Lin.

Metabolic patterns in brain [18] **F-fluorodeoxyglucose PET relate to aetiology in paediatric dystonia.** *Brain*, Volume 146, Issue 6, June 2023, Pages 2512–2523, https://doi.org/10.1093/brain/awac439

2. TA Szyszko, JT Dunn, MJ O'Doherty, L Reed, JP Lin.

Role of 18F-FDG PET imaging in paediatric primary dystonia and dystonia arising from neurodegeneration with brain iron accumulation

Nuclear medicine communications. 2015, 36 (5), 469-476

3. VM McClelland, A Valentin, HG Rey, DE Lumsden, MC Elze, R Selway, G Alarcon, JP Lin.

Differences in globus pallidus neuronal firing rates and patterns relate to different disease biology in children with dystonia

Journal of Neurology, Neurosurgery & Psychiatry 2016, 87 (9), 958-967

4. Verity M McClelland, Doreen Fialho, Denise Flexney-Briscoe, Graham E Holder, Markus C Elze, Hortensia Gimeno, Ata Siddiqui, Kerry Mills, Richard Selway, Jean-Pierre Lin.

Somatosensory evoked potentials and central motor conduction times in children with dystonia and their correlation with outcomes from deep brain stimulation of the globus … pallidus internus.

Clinical Neurophysiology 2018, 129 (2), 473-486

5. VM McClelland, Z Cvetkovic, JP Lin, KR Mills, P Brown

Abnormal patterns of corticomuscular and intermuscular coherence in childhood dystonia

Clinical Neurophysiology 2020, 131 (4), 967-977

6. H Gimeno, JP Lin

The International Classification of Functioning (ICF) to evaluate deep brain stimulation neuromodulation in childhood dystonia-hyperkinesia informs future clinical & research …priorities in a multidisciplinary model of care.

European Journal of Paediatric Neurology 2017, 21 (1), 147-167

7. SA Shah, P Brown, H Gimeno, JP Lin, VM McClelland

Application of machine learning using decision trees for prognosis of deep brain stimulation of globus pallidus internus for children with dystonia

Frontiers in neurology 2020, 11, 825

Dystonia Research in Finland – Brain Lesions, Brain Networks and Brain Stimulation

Turku Brainlab, a non-profit research laboratory at University of Turku in Finland, has recently ramped up dystonia research in Finland.

Turku Brainlab hosts approximately 30 researchers from numerous countries and disciplines, including neurology, psychiatry, radiology, neuroscience and psychology. The unique strength of the lab is in its multimodal approach, leveraging several cutting-edge neuroimaging and brain stimulation techniques. Turku Brainlab is led by Prof. Juho Joutsa, chief neurologist at Turku University Hospital, who has studied movement disorders for the past 15 years.

Prof. Joutsa completed his postdoctoral training at Massachusetts General Hospital and Harvard Medical school, where he and his Australian colleague Dr. Daniel Corp, PhD, led a pioneering study localising the brain network of cervical dystonia based on causal brain lesions and a novel technique called lesion network mapping (Corp & Joutsa et al., *Brain 2019*). This study prompted a years-long collaboration between Joutsa and Corp with their common cause of finding solutions to dystonia. Dr. Corp is currently working at the Turku Brainlab alongside Prof. Joutsa. This breakthrough in cervical dystonia, together with a generous donation from an individual person (who prefers to stay anonymous) upon Prof. Joutsa's return to Finland in 2018, sparked a new era of dystonia research at University of Turku.

Currently, researchers in Turku leverage several approaches to study the neurobiological mechanisms of dystonia, with the aim of localising brain networks that can be targeted by therapeutic brain stimulation . The core of this work is lesion network mapping, which they also used in their cervical dystonia study published in 2019.

This technique leverages causal lesions (i.e. cases where a focal brain damage has led to development of dystonia) and combines the lesion locations with the brain connectome (wiring diagrams of the human brain). This approach circumvents one of the big challenges in neuroimaging research by using causal sources of information, instead of only analysing correlations and group differences in brain activity, as in traditional imaging approaches. By studying these lesions, the researchers extended upon their 2019 study in cervical dystonia to show that different types of dystonia are caused by damage to different brain structures, indicating that all dystonias are not the same neurobiologically *(Corp…Joutsa, Neurology 2022*). They are now using lesion network mapping to localise the causal networks across dystonia types.

The ultimate goal in Turku Brainlab is to translate their findings to therapies. Brain stimulation is increasingly used to treat brain disorders but is currently only available for a handful of neurological and psychiatric disorders. One of the main challenges is to know exactly where in the brain to stimulate. Without the correct target, the stimulation is futile. Luckily, we already know an efficacious target for deep brain stimulation in dystonia. However, we still don't fully understand its mechanisms of action. To address this, Turku Brainlab conducted the first study in Finland combining deep brain stimulation with positron emission tomography (PET), which measures glucose metabolism in the brain (Honkanen et al., *JNNP 2024*). The researchers showed that the effects of deep brain stimulation are in fact not mediated via changes in the DBS target activity but outside this region in brain areas that are connected to it, suggesting that the target is a network rather than a single anatomical region.

While this helps to define the mechanisms and neural substrates of DBS efficacy, because DBS is invasive it is only used in severe cases where other treatments are inadequate. In addition, although the treatment is highly efficacious on average, not all types of dystonia have good response to DBS, and there may also be side-effects.

Prof Juho Joutsa, MD, PhD

Therefore, the team is currently trying to translate their findings to novel brain stimulation treatment using transcranial magnetic stimulation, which is a non-invasive form of therapy. The preliminary results are encouraging, as they have already found out that transcranial magnetic stimulation to the cervical dystonia network results in changes in brain metabolism within the network, demonstrating engagement of this target network (Kokkonen et al. *Submitted*). In parallel, Dr. Corp is overseeing a pilot study at Deakin University in Australia, showing promising clinical benefits of stimulation to this exact site over 10 days (9 patients with cervical dystonia tested so far). If successful, this would set the stage for a larger-scale randomised controlled trial, which would be necessary to conduct before the new therapeutic approach could be brought to clinical care. Moving this line of research forward is mostly dependent on the available funding, which is unfortunately relatively limited for dystonia research.

This would also allow us to expand this approach to other types of dystonia. The road from initial discovery to new treatment is always long but we believe it can be achieved with persistence and teamwork between clinicians, scientists, funders, and most importantly, individuals with dystonia, as without their support none of this work would be possible.

Juho Joutsa, MD, PhD
Professor of Neurology, University of Turku
Chief neurologist, Turku University Hospital
Turku, Finland

References:

Corp DT, Joutsa J, Darby RR, Delnooz CCS, van den Warrenburg BPC, Ren J, Batla A, Bhatia KP, Jinnah HA, Liu H, Fox MD. Network localization of cervical dystonia based on causal brain lesions. *Brain 2019;142 (6):1660-74.*

Corp DT, Greenwood C, Morrison-Ham J, Pullinen J, McDowall G, Younger E, Jinnah H, Fox MD, Joutsa J. Clinical and structural findings in patients with lesion-induced dystonia: descriptive and quantitative analysis of published cases. *Neurology 2022;99(18):1957-67.*

Honkanen EA, Rönkä J, Pekkonen E, Aaltonen J, Koivu M, Eskola O, Eldebakey H, Volkmann J, Kaasinen V, Reich MM, Joutsa J. GPi-DBS induced brain metabolic activation in cervical dystonia. *J Neurol Neurosurg Psychiatry, 2024;95(4):300-8.*

Kokkonen A, Corp DT, Aaltonen J, Hirvonen J, Kirjavainen AK, Rajander J, Joutsa J. Brain metabolic response to repetitive transcranial magnetic stimulation to lesion network in cervical dystonia. *Submitted* (preprint available at https://doi.org/10.31219/osf.io/chxkt)

Dystonia Ireland Membership

We have always tried to keep the cost of membership as low as possible. The membership per annum is €20. Your membership fees and donations will make it possible for us now to provide you with regular Newsletters and hopefully a better service for all!

Membership form

Title (Mr/Mrs/Ms/Dr)

First Name

Surname

Address

Tel. No.

Mobile

E-mail

Year of birth (optional)

Occupation (optional)

Membership

(€20) every 12 months €

Voluntary donation €

Total enclosed €

Membership type New ❑ Renewal ❑

Method of payment Cheque ❑ Postal order ❑ Cash ❑

Would you like to act on a voluntary basis for Dystonia Ireland? Yes ❑ No❑

If you wish please give details of anything which helps your dystonia (use a separate sheet if necessary). This information may be published in our newsletter or on our website.

Please tick here if you wish to remain anonymous ❑

Bankers Order

Please complete and return this form to us and we will send it on to your bank.

Bank & Branch

Sort code

Address

Please pay the sum of €_____to the account of Dystonia Ireland at AIB Bank, 9, Terenure Road East, Rathgar Dublin 6. Sort Code: 93-34-57. Current Account No. 11953031. on this date and on the corresponding date each succeeding year until further notice.

Signed Account Number

Address

Date