Tonic dystonia: clinical and genetic characterisation, longitudinal changes and risk factors

Published: 27-03-2009
Last updated: 19-08-2024

The aim of this research project is to study a large a group of patients with tonic dystonia longitudinally, to accurately describe the phenotype, to investigate the potential underlying mechanisms and to identify possible causes and risk factors.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Movement disorders (incl parkinsonism)
Study type	Observational non invasive

Summary

ID

NL-OMON56936

Source
ToetsingOnline

Brief title
Tonic dystonia Cohort

Condition

- Movement disorders (incl parkinsonism)

Synonym
muscle tone regulation disorder

Research involving
Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum
Source(s) of monetary or material Support: Ministerie van Economische Zaken (BSIK03016)

Intervention

Keyword: complex regional pain syndrome, dystonia, movement disorders

Outcome measures

Primary outcome

Medical history is obtained following a semi-structured interview. The primary patient group will be evaluated with the CRPS diagnose form. 2 EDTA tubes of blood (20 ml) will be collected for genetic testing only during the first visit.

Several self-administered questionnaires will be completed: 1). Hospital anxiety and depression scale (HADS); 2). Tampa scale for Kinesiophobia; 3). Pain coping inventory (PCI); 4). Numeric rating Scale Pain (NRS-Pain); 5). McGill Pain Questionnaire (MPQ); 6). Dissociative experience scale (DES); 7); Somatoform dissociation questionnaire (SDQ20); 8). SCOPA AUT; 9). Radboud Skills Questionnaire; 10). Questionnaire on walking & rising; 11). Questions about profession and education (first visit)

The severity and progression of the dystonia, together with the impairments on daily activities, will be measured with the *DYstonia Assessment Scale (DYAS)* rating scale and the *Burke Fahn Marsden* (BFM) scale. A rapid finger movement task is performed to test
the velocity and fluency of movements (bradykinesia).

Quantative sensory testing is used to assess wind-up, pain thresholds and

perception to touch, temperature and vibration. Diffuse Noxious Inhibitory Control (DNIC), a test that interrogates the function of endogenous pain regulation, will also be determined. Surface temperature of hands and feet are determined with an infrared skin temperature device. The ability to accurately recognise the laterality of pictures of left and right extremities is performed with the Recognise© lateralization computer program.

Secondary outcome

not applicable

Study description

Background summary

Dystonia is the most common type of movement disorder that may develop after peripheral trauma, but can also arise in combination with Complex Regional Pain Syndrome (CRPS) or chronic pain. The phenotype of this dystonia differs from the primary *mobile* form and is usually *tonic*, referring to the presence of continuous muscle contractions leading to abnormal postures, from which return to the neutral position is not possible or only with great difficulty. The pathophysiology of tonic dystonia is unknown and there is still controversy about the contribution of psychological factors in the development of the disorder. Evidence from primary dystonia suggests that the condition may arise in response to trauma or pain in subjects with an increased susceptibility, whereby a (pre-existent) maladaptive plasticity and disturbances in sensimotor integration may lead to dystonia.

Study objective

The aim of this research project is to study a large a group of patients with tonic dystonia longitudinally, to accurately describe the phenotype, to investigate the potential underlying mechanisms and to identify possible causes and risk factors.

Study design

The proposed study is a combination of a prospective cohort study and a case control comparison. The primary patient group will be examined once a year at

the LUMC, for a period of 4 years, while the healthy control group will only be measured once and the patient control group at year 1 and 3.

Study burden and risks

The tests are non-invasive. Although the chances are small, it can not be excluded that patients with tonic dystonia develop more severe symptoms, patients with CRPS an exacerbation of the complaints and more pain.it can not be excluded In these situations the study will be adapted to the persons wishes, or will be ended. If necessary, the neurologist on duty will be consulted.

Tonic dystonia is an invalidating disorder with an unknown cause, however little research has been performed to learn more about the disoder. Tonic dystonia is not always recognized by doctors, and is sometimes referred as a psychogenic disorder. This study asks some effort from the patients and is not directly helping the individual, but will provide more insight in the disorder and could be beneficial for treatment and understanding patients in the future.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

patients with tonic dystonia

Exclusion criteria

- mobile dystonia
- patients with a known genetic form of dystonia, e.g. DYT1-DYT17, Wilson*s disease
- lesions or diseases of the central nervous system (e.g. as a result of head trauma)
- implantation of drug-delivery pump

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	27-03-2009
Enrollment:	425
Type:	Actual

Ethics review

Approved WMO Date:	27-03-2009
Application type:	First submission
Review commission:	METC Leids Universitair Medisch Centrum (Leiden)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL21732.058.09