

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

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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.

|                              |  |
|------------------------------|--|
| <b>Ethical review</b>        | Approved WMO                           |
| <b>Status</b>                | Recruitment stopped                    |
| <b>Health condition type</b> | Movement disorders (incl parkinsonism) |
| <b>Study type</b>            | 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 disorder. 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 |