

# Familial coritcal myoclonic tremor with epilepsy, follow-up and extension of the Dutch pedigree.

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<b>Ethical review</b>	Approved WMO
<b>Status</b>	Will not start
<b>Health condition type</b>	Movement disorders (incl parkinsonism)
<b>Study type</b>	Observational invasive

## Summary

### ID

NL-OMON38543

### Source

ToetsingOnline

### Brief title

FCMTE follow-up and extension of the pedigree

### Condition

- Movement disorders (incl parkinsonism)

### Synonym

Familial Cortical Myoclonic Tremor with Epilepsy, trembling with epilepsy

### Research involving

Human

### Sponsors and support

**Primary sponsor:** Academisch Medisch Centrum

**Source(s) of monetary or material Support:** Ministerie van OC&W

## Intervention

**Keyword:** epilepsy, FCMTE, myoclonus, tremor

## Outcome measures

### Primary outcome

Making use of clinical, clinical neurophysiological and neuropsychological examinations, we expect to observe an increase of symptoms and signs of disease, which could be influenced by the disease itself, by prescribed medication, or by comorbidity.

### Secondary outcome

Not applicable.

## Study description

### Background summary

\*Familiar cortical myoclonic tremor with epilepsy\* (FCMTE) is an autosomal dominant inherited disorder with epileptic seizures in combination with muscle jerks (myoclonus) of the limbs.

Long term follow-up of FCMTE patients is missing in the literature; as a result of which disease progression in individual FCMTE patients is not well known. Additionally, follow-up and expansion of the pedigree, allowing proper phenotyping, is essential for genetic research.

### Study objective

In recent years, we have identified a Dutch family and performed a large number of studies which have helped us gain insight into the disorder. (MEC protocols 02/223 and 03/140, Dissertation Van Rootselaar, 2006). We want to call on family members who have in the past indicated a willingness to be approached for future research, and include them in a follow-up study. In addition to that, we want to try and identify new family members. The goal of this protocol is to shed a light on follow-up studies in this pedigree.

### Study design

The follow-up portion of this study will be executed as a prospective cohort study. Comparison between clinical neurophysiological and NPO data with healthy subjects will be performed using the case-control design.

### **Study burden and risks**

Participants will undergo one site visit, during which a neurological examination will be performed, in addition to standardized clinical evaluations with the aid of clinical scales and videos. During the same visit, participants will undergo neurophysiological examination and neuropsychological assessment.

The proposed investigation bears virtually no risks and is usually well tolerated. Patients will benefit from the insight gained into progression of the disease, mainly because they can subsequently be better informed. Furthermore, insight into the effects of medication and comorbidity could lead to better management of FCMTE. Healthy controls will have no special benefits.

## **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

Patient inclusion criteria: Definitely or possible affected and age above 18. Healthy subjects undergoing neurophysiological examination: Matched for age and gender. Healthy subjects undergoing neuropsychological assessment: Matched for age and gender and for socio-economic status.

## Exclusion criteria

Exclusion criteria patients: None. Exclusion criteria healthy subjects: no known neurological disorders and medication usage which can affect task performance.

## Study design

### Design

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

### Recruitment

NL	
Recruitment status:	Will not start
Enrollment:	35
Type:	Anticipated

## Ethics review

Approved WMO

Date: 28-05-2013  
Application type: First submission  
Review commission: METC Amsterdam UMC

## 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	NL44044.018.13