# Presentation of Myotonic Dystrophy type 1 and 2 within a five-generation family

Published: 15-10-2013 Last updated: 26-04-2024

K2. objective

**Ethical review** Approved WMO

**Status** Pending

**Health condition type** Other condition

**Study type** Observational invasive

## **Summary**

#### ID

NL-OMON36875

#### **Source**

ToetsingOnline

#### **Brief title**

Presentation of MD1 and 2 within a five-generation family

## **Condition**

- Other condition
- Chromosomal abnormalities, gene alterations and gene variants

## **Synonym**

PROMM, Steinert's disease // Ricker's disease

#### **Health condition**

multisysteemaandoeningen

## Research involving

Human

## **Sponsors and support**

**Primary sponsor:** Universitair Medisch Centrum Sint Radboud

**Source(s) of monetary or material Support:** onderzoeksbudget afdeling neuromusculaire

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#### ziekten

#### Intervention

**Keyword:** myotonic dystrophy type 1 (Steinert), myotonic dystrophy type 2 (PROMM), pedigree, phenotype

## **Outcome measures**

## **Primary outcome**

K2. primary study parameters

## **Secondary outcome**

K2. secundary study parameters

# **Study description**

## **Background summary**

K2. background

## Study objective

K2. objective

## Study design

K2. design

## Study burden and risks

K2. burden and risks

## **Contacts**

#### **Public**

Universitair Medisch Centrum Sint Radboud

Geert GrootepleinZuid 10

2 - Presentation of Myotonic Dystrophy type 1 and 2 within a five-generation family 13-05-2025

Nijmegen 6525 GA

NL

## **Scientific**

Universitair Medisch Centrum Sint Radboud

Geert GrootepleinZuid 10 Nijmegen 6525 GA NL

## **Trial sites**

## **Listed location countries**

**Netherlands** 

# **Eligibility criteria**

## Age

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

## Inclusion criteria

- member of the 5-generation family
- age >- 18 years

## **Exclusion criteria**

age < 18 years

# Study design

## **Design**

Study type: Observational invasive

Masking: Open (masking not used)

Control: Uncontrolled

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Primary purpose: Diagnostic

## Recruitment

NL

Recruitment status: Pending

Start date (anticipated): 01-09-2012

Enrollment: 15

Type: Anticipated

## **Ethics review**

Approved WMO

Date: 15-10-2013

Application type: First submission

Review commission: CMO regio Arnhem-Nijmegen (Nijmegen)

## **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 NL41136.091.12