Haplotyping in patients with genetically proven Myotonic Dystrophy type 2

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Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Neurological disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON43063

Source
ToetsingOnline

Brief title
DM2 haplotype

Condition

- Neurological disorders congenital

Synonym
Myotonic Dystrophy type 2, Proximal Myotonic Myopathy (PROMM)

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: autoimmune dissease, Haplotype, myotonic dystrophy type 2

Outcome measures

Primary outcome

Will the genetic material in the 3q21.3 region be the same in all patients with

DM2

Secondary outcome

Do DM2 patients with an autoimmune disease have a different haplotype than that

of DM2 patients without an autoimmune disease.

Study description

Background summary

Our research CMO nr. 2007/176 showed that the frequency of autoimmune diseases (21% vs 2%) and the frequency of autoantibodies (25% vs 2%) were both significantly ($p<0.01$) higher in DM2 patients compared to DM1 patients. Data on DM1 patients were comparable with those of the general population. In a follow-up study we will investigate the possible underlying mechanism of this association. In the 3q21.3 region of the DM2 mutation, an interesting set of genes for autoimmune diseases is known, especially in the CD80/CD86 domain. Different polymorfisms in CD80 and CD86 seem to be associated with t-cel mediated autoimmune diseases or comparable clinic such as sensitivity for infections or a higher chance of acute rejection after organ transplantations, although results seem as well to be conflicting.

Study objective

In this study we want to investigate the CD80/CD86 region whether polymorfism in the 3q21 region of the DM2 population are associated with a high prevalence of autoimmune diseases. If this is indeed the case, this will help our insight in the CD80/CD86 region about the rise of autoimmune diseases.

Study design

To answer the above question we want to investigate the genetic variation in

the 3q21.3 region. We will start haplotyping in the this region. For this research, we will use the DNA of genetically diagnosed DM2 patients in the Netherlands since 2010 (n=35). In this study the material will mainly be:
- lifematerial that was collected in connection with another goal (than scientific research) and that is concerned. It comes from DNA diagnostic resulting from the diagnosis DM2. Thus no in general, additional blood punctures will have to be performed.
- In a maximum of 5 cases a new bloodpuncture will have to take place, namely because the patient chooses to have a new venaprick above sending her DNA-material from the UMCU (University Medical Center Utrecht) to the Radboudumc.

Study burden and risks

The only risk is a single venaprick.

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

genetically proven DM2, minimum age: 18 years old

Exclusion criteria

n.a.

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	02-06-2017
Enrollment:	35
Type:	Actual

Ethics review

Approved WMO

Date:	12-01-2017
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	NL58134.091.16