association between dystrophia myotonica type II and autoimmune disease

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researchs aims- to assess the frequency of auto-immune disease in patients with dystrophia myotonica type II compared to patients with dystrophia myotonica type I- to assess the frequency of auto-antibody formation in patients with dystrophia...

Ethical review	Approved WMO
Status	Pending
Health condition type	Autoimmune disorders
Study type	Observational invasive

Summary

ID

NL-OMON30830

Source
ToetsingOnline

Brief title
DMII and autoimmune disease

Condition

- Autoimmune disorders
- Musculoskeletal and connective tissue disorders congenital

Synonym
dystrophia myotonica type II, muscle disorder

Research involving
Human

Sponsors and support

Primary sponsor: Sint Maartenskliniek
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: autoantibodies, autoimmune disease, dystrophia myotonica type II

Outcome measures

Primary outcome

frequency of autoimmune disease according to clinical criteria or

classification criteria in both cohorts

Secondary outcome

frequency of autoantibody fromation in both cohorts

Study description

Background summary

Dystrophia myotonica (DM) is a genetical muscle disorder of wich two subtypes exist (DMI and DMII).
Recently an observation was made that the prevalence of T-cel and autoantibody mediated auto-immune disease was increased in a nationwide cohort of patients with DM I. Several causes for this association can be conceived , including genetical linkage.
The genetical cause of DM II is a mutation of the ZNF-9 gene on locus 3q21.3 en 3q13.3-q24 and in these regions some interesting genes can be found for auto-immune disease, especialy the CD80/CD86 domain. CD80/CD86 is a ligand for the costimulatory T-cel recepter CD28 and also the locus is at this moment the only candidate gene for RA susceptability outside chromosome 6 (HLA genes). Several polymorphisms of CD80/CD86 have been recognised and some seem associated with auto-immune diasease. An increased prevalence of autoimmune disaese and autoanti-body formation associated with this monogenetic disorder would provide insigth in the aetiology of auto immune disorders.

Study objective

researchs aims
- to assess the frequency of auto-immune disease in patients with dystrophia myotonica type II compared to patients with dystrophia myotonica type I
- to assess the frequency of auto-antibody formation in patients with dystrophia myotonica type II compared to patients with dystrophia myotonica

type I

Study design

Observational study comparing two cohorts

Study burden and risks

venous blood punction (hematoma)

Contacts

Public
Sint Maartenskliniek

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6522JV
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Scientific
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Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

dystrophia myotonica type II
informed consent

Exclusion criteria

none

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)

Primary purpose: Basic science

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-09-2007
Enrollment:	64
Type:	Anticipated

Ethics review

Approved WMO

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	NL18871.091.07