EXT 1 and 2 Gene mutations in a Dutch population with HME/MO

Published: 06-04-2012
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How is the genetic expression of EXT1 and EXT2 in our HME/MO population and how does this related to the severity of the disease.

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON35147

Source
ToetsingOnline

Brief title
Ext 1-2 mutations in HME/MO

Condition

- Chromosomal abnormalities, gene alterations and gene variants

Synonym
Hereditary Multiple Exostosis, Multiple Osteochondromas

Research involving
Human

Sponsors and support

Primary sponsor: Onze Lieve Vrouwe Gasthuis
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Ext 1, Ext 2, HME/MO, mutation

Outcome measures

Primary outcome

Ext 1 of Ext 2 genmuation, or no mutation found.

Secondary outcome

Etnic Origin, Height, Age of onset HME/MO, Number of osteochondromas at a

certain age. Site of osteochondromas. Development of osteochondromas. Family

history

Study description

Background summary

Hereditaire Multiple Exostosis (HME)/Multiple Osteochondromas (MO) is a hereditairy skeletal disease. It is rare with a calculated prevalence of 1 in 50.000 people. HME/MO has a autosomal dominant patern. Two genes have been isolated; EXT 1 en EXT2. About 62% of the patients have a positive family history. The other cases are spontanous mutations. Patients with HME/MO develope osteochondromas at the growth plates at a young age. These osteochondromas are mainly located in the long bones but also in the pelvis, shoulder, hand and feet. The osteochondromas cause painsymptoms, limb deformities and functinal impairment in the growing child.
The Onze Lieve Vrouwe Gasthuis in Amsterdam is the refferal centre in the Nederlands for the treatment of patients with HME/MO. We have treated more then 300 patiënten with this disease. A unique cohort. We cooperate with the HME/MO patients support organisation.
There is little known about the genetic characteristics in the Dutch population. The medical genetic department of the Universitity of Antwerp has world wide experience in this field. In cooperation with them we want to investigate the incidence of these gene mutations. This is further explained in the research protocol.

Study objective

How is the genetic expression of EXT1 and EXT2 in our HME/MO population and how does this related to the severity of the disease.

Study design

Observational study in a Dutch HME/MO cohort.

Study burden and risks

Hematoma from blood test

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

Patient with HME/MO

Exclusion criteria

Previous genetic research done that showed a Ext 1 or 2 mutation.

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	06-06-2012
Enrollment:	100
Type:	Actual

Ethics review

Approved WMO Date:	06-04-2012
Application type:	First submission
Review commission:	MEC-U: Medical Research Ethics Committees United (Nieuwegein)
Approved WMO Date:	27-01-2015
Application type:	Amendment
Review commission:	MEC-U: Medical Research Ethics Committees United (Nieuwegein)

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
Other	dit volgt nog.
CCMO	NL37625.100.11