The prevalence of Premature Ovarian Failure (POF) and Fragile X-associated Tremor Ataxia Syndrome (FXTAS) in families with FMR2 gene mutation (FRAXE).

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More knowledge about the clinical aspects of the FMR2 gene mutation can complete the couseling. Carriers with the desire to start a family can take the chance on POF in consideration. The clinical symptoms of FXTAS can be differentiated from other...

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

Summary

ID

NL-OMON30168

Source
ToetsingOnline

Brief title
FRAXE and POF, FXTAS

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Endocrine disorders of gonadal function
- Movement disorders (incl parkinsonism)

Synonym
fragile X syndrome, mild mental retardation

Research involving

Human

Sponsors and support

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

Intervention

Keyword: FRAXE, FXTAS (Fragile X-associated Tremor Ataxia Syndrome) POF (Premature Ovarian Failure)

Outcome measures

Primary outcome

Pheno-genotyping of FMR2 gene mutation.

Secondary outcome

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Study description

Background summary

In 1992 a mutation consisting of a CGG-repeat expansion in the FMR2 gene, has been discoverd (Sutherland and Baker). The FMR2 genes (FRAXE) fragile site is situated at Xq28, close to the well-known fragile X syndrome (FRAXA) site at Xq27. The underlying molecular basis for FRAXE is an unstable CGG-repeat identical to that of FRAXA. BesideS mental retardation FRAXA has been associated with premature ovarian failure (POF) (Hundscheid 2001) and fragile X-associated tremor ataxia syndrome (FXTAS) (Hagerman 2004). We ask ourselves whether the FMR2 gene mutation also enhances the risk of POF, FXTAS or other additional health risks.

Study objective

More knowledge about the clinical aspects of the FMR2 gene mutation can complete the couseling. Carriers with the desire to start a family can take the chance on POF in consideration. The clinical symptoms of FXTAS can be differentiated from other degenerative neurological diseases and benefit from therapy.

Study design

A retrospective diagnostic survey among carriers of FMR2 gene mutation and non-carrier familymembers.

Study burden and risks

Minimal risk/burden: an interview, physical and neurological examination and a blood sample.

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

Familymembers of a family with cytogenetically confirmed mutation in FMR2 gene.

Exclusion criteria

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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:	Pending
Start date (anticipated):	15-04-2006
Enrollment:	50
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	NL11949.091.06