Prevalence of FXI gene mutations in women with menorrhagia.

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to investigate the presence and frequency of FXI gene mutations in patients with menorrhagia and healthy volunteers.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Coagulopathies and bleeding diatheses (excl thrombocytopenic)
Study type	Observational non invasive

Summary

ID

NL-OMON35784

Source
ToetsingOnline

Brief title
FXI gene mutations in women with menorrhagia

Condition

- Coagulopathies and bleeding diatheses (excl thrombocytopenic)
- Menstrual cycle and uterine bleeding disorders

Synonym
FXI gen mutations

Research involving
Human

Sponsors and support

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

Intervention

Keyword: bleeding disorders, FXI gene mutations, menorrhagia

Outcome measures

Primary outcome

The prevalence and frequency of FXI gene mutations in patients with menorrhagia

and healthy volunteers.

Secondary outcome

not applicable

Study description

Background summary

Menorrhagia is a common problem among women in the reproductive age. Menorrhagia or heavy menstrual bleeding is objectively defined as greater than or equal to 80 ml blood loss per menstrual cycle.3 At least 5 -10% of women in reproductive age will seek medical attention for menorrhagia.4 The World Health Organization estimates that 18 million women worldwide are affected.5 Menorrhagia is a common cause of iron deficiency anemia3;6 and may affect a woman*s quality of life, her study or work and family and social interactions.7 Menorrhagia can be caused by a wide range of disorders.8;9
We performed an observational study about the prevalence of underlying bleeding disorders in women with menorrhagia (METc 2009/217). Preliminary results of 91 women and 30 healthy volunteers showed that women with menorrhagia had significantly lower levels of factor XI (FXI) compared to controls (100 vs 125 IU/dL; p=0.000). Of all the patients four had a lower FXI level (<70%). FXI is a coagulation protein essential to normal haemostasis and acts by cleaving coagulation factor IX. FXI deficiency, also known as haemophilia C, is an autosomal bleeding disorder characterized by reduced levels of FXI in plasma with a high prevalence (about 9%) in the Ashkenazi Jewish population.10 Women with low levels FXI (<70%) are prone to excessive bleeding during menstruation. Bleeding manifestations are not well correlated with the plasma levels of FXI activity and bleeding episodes can vary widely among patients with similar FXI levels.10;11 Recent studies suggested that the incidence of mild FXI deficiency in Caucasians may be higher than expected.1;2 The factor XI (FXI) gene is located on the long arm of chromosome 4 (4q35) with 23 kb in length and consists of 15 exons and 14 introns. Until now, more than 90 FXI gene

mutations, associated with a FXI deficiency have been reported. Most of these mutations are found in the Jewish population. To our knowledge, the prevalence of FXI gene mutations in women with menorrhagia have not been reported. However, the presence of mutations in FXI gene in women with menorrhagia could probably lead to lower levels of FXI.
We want to perform an explorative study out of our previous study to investigate the prevalence of FXI gene mutations in women with menorrhagia and a control group of healthy female volunteers matched by age with normal menstrual blood loss without hormonal treatment.

Study objective

to investigate the presence and frequency of FXI gene mutations in patients with menorrhagia and healthy volunteers.

Study design

Explorative study to investigate the prevalence and frequency of FXI gene mutations in patients with menorrhagia and healthy volunteers from our previous study.

Study burden and risks

Burden and risks: 20 mL blood will be taken from the patients and the controls by venapunction. There are no benefits for the patients and the controls.

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

For patients
• Patients with regular heavy menstrual bleeding (=menorrhagia).
• Age over 18 years.
• Written informed consent.;For healthy volunteers
• women with normal menstrual blood loss.
• Age over 18 years.
• Written informed consent.

Exclusion criteria

For patients:
• Patients with postmenopausal, irregular, postcoital and intermenstrual bleeding.
• Patients with an intra-uterine device or hormonal treatment.
• Patients with anticoagulant, antithrombotic therapy or use of non-steroidal anti-inflammatory drugs (NSAIDs). ;Exclusion criteria for healthy volunteers :
• women with postmenopausal, irregular, postcoital and intermenstrual bleeding.
• women with an intra-uterine device or hormonal treatment.
• women with anticoagulant, antithrombotic therapy or use of non-steroidal anti-inflammatory drugs (NSAIDs).

Study design

Design

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

Primary purpose: Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	02-12-2011
Enrollment:	90
Type:	Actual

Ethics review

Approved WMO

Date:	26-10-2011
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Groningen (Groningen)

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	NL35433.042.11