Molecular basis of Body Integrity Identity Disorder

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1. To make a phenomenological profile of BIID individuals using a standardized questionnaire.2. To perform whole exome analyses in four phenomenological identical BIID individuals.3. To perform molecular analyses in a series of other BIID...

Ethical review	Approved WMO
Status	Pending
Health condition type	Psychiatric disorders NEC
Study type	Observational non invasive

Summary

ID

NL-OMON36710

Source
ToetsingOnline

Brief title
Molecular basis of BIID

Condition

- Psychiatric disorders NEC

Synonym
"people who'd like to be amputated", Apotemnophilia

Research involving
Human

Sponsors and support

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

Intervention

Keyword: Body Identity Integrity Disorder, Genetics

Outcome measures

Primary outcome

Gene(s) involved in BIID

Secondary outcome

Phenomenological Profile of BIID individuals

Study description

Background summary

Body Identity Integrity Disorder (BIID) is a person*s intense and longstanding desire to have one of several amputations of their limb(s). The main motivation for amputation is not feeling complete with four limbs. BIID is an infrequently described condition, is not included in DSM-IV-TR, and often not known to surgeons, neurologist and psychiatrists. The etiology of the BIID remains unclear, no genetic research to BIID has been done so far.

Study objective

1. To make a phenomenological profile of BIID individuals using a standardized questionnaire.
2. To perform whole exome analyses in four phenomenological identical BIID individuals.
3. To perform molecular analyses in a series of other BIID individuals in case a causative gene can be found by the total exome sequencing.

Study design

Questionnaire
After enrolment in the study the participants will be send a questionnaire in order to gather all relevant information on their phenotype (Appendix 3). Topics of the questions will be general demographics, medical history, family history, BIID features and DSM-axis I screening. Questionnaires can be returned either by ordinary mail or via a secured internet connection. Results of the questionnaire will be computerized and statistically analyzed.

Blood sampling
Participants that have indicated to be willing to participate in the molecular study will be sent a packet with materials and an instruction letter for blood drawing. The blood can be drawn locally by their general physician or local hospital. The instruction letter will not mention the term BIID. Materials needed to return the samples by mail to Amsterdam for free will be added.

Molecular studies
Total exome sequencing using the 454 or Solid Genome Analyzer will be performed in 4 BIID individuals. The choice of the participants will be made based on the data from the questionnaire in order to ensure the highest possible homogeneity within the group. The sequence capture technology from Ninblegen/Roche will be used for the exome sequencing. DNA from the participants will be sheared, hybridized to a set of oligonucleotides covering all exons of the human genome. Eluted DNA will be sequenced on a massive parallel sequencer (454/Solid). Bio-informatic analyses of the sequence information will be matched to the human reference genome. The variations (sequence and structural) will be compared with databases of known variation and prediction tools (SIFT, Polyphen, splice site predictors) will be used to estimate the impact of the variants. Variants will be ranked on predicted impact. Mutations in the same gene in two or more participants will get the highest priority. Further total exome sequencing in one or more additional participants will be performed if no single gene can be detected. If the total exome sequencing leads to discovery of a gene or of genes that are likely to be causative for BIID, Sanger sequencing of these gene(s) will be repeated first in the four participants in whom total exome sequencing was performed, and, if findings are confirmed, in all other participants of whom DNA samples have become available for this study.

Study burden and risks

The associated risks are restricted to blood draw in healthy individuals (apart from their BIID). Because indivduals with BIID are highly secretive concerning their BIID, the main burden of participation in this study could be the disclosure of their BIID to the researchers.

With respect to the secretivity of the BIID individuals, the design of the research has been made to protect the participants' identity as much as possible. Communication and recruitment of the individuals will mainly be part take part through the internet, which will give the participants enough time to consider their participation.

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

-Diagnosed with BIID
-Participants have to be able to read and understand the written information
-18 years of age or older

Exclusion criteria

-History of any severe psychiatric disorder other than BIID
-History of neurological disease that affects the CNS

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-12-2010
Enrollment:	10
Type:	Anticipated

Ethics review

Approved WMO

Application type:	First submission
Review commission:	METC Amsterdam UMC

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	NL34740.018.10