Psychopathology and Cognition in 22q11 CNV disorders

Published: 24-12-2014
Last updated: 15-05-2024

To describe cognitive profile and psychopathology in adults with 22q11 CNV disorders.

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

Summary

ID

NL-OMON47626

Source
ToetsingOnline

Brief title
22q11 CNV disorders

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Schizophrenia and other psychotic disorders

Synonym
22q111 CNV disorders, disorders of chromosome 22

Research involving
Human

Sponsors and support

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

Intervention

Keyword: 22q11 CNV, cognition, psychopathology, schizophrenia

Outcome measures

Primary outcome

Neuropsychological outcome measures (working memory, attention, social cognition,verbal memory, processing speed, visual memory, planning), a cognitive composite score bases on the total score of the CANTAB subtests (representing cognitive function), IQ, psychiatric diagnosis

Secondary outcome

n/a

Study description

Background summary

Schizophrenia (SZ) and psychotic disorders are devastating lifelong illnesses that are disabling and costly to patients, families, communities, and healthcare systems. Treatment advances have been limited by lack of mechanistic understanding of the pathophysiology of the syndrome. The NIMH mission of transforming the understanding and treatment of mental illness requires integration of basic and clinical research with cutting-edge approaches in a developmental context. As in most medical disorders, by the time SZ is diagnosed it has reached a stage where treatment is a challenge. For early identification and novel therapeutics it is essential to elucidate the trajectory of neurodevelopmental processes and identify biomarkers. Such information on specific disease models can provide a translational bridge through which this major neuropsychiatric syndrome can join the *precision medicine* revolution. 22q11 CNV disorders like 22q11 deletion syndrome (22q11DS) en 22q11 duplication syndrome (22q11Dup) are unique human models to study the development of schizophrenia and to fill the gaps in our knowledge. People with 22q11DS have a 25-30 times increased risk to develop schizophrenia, the highest known genetic risk factor. The clinical presentation between schizophrenia and 22q11DS and idiopathic schizophrenia show a lot of similarities and both are associated with cognitive decline. In contrast, people with 22q11Dup seem to be protected from schizophrenia. Research on schizophrenia and 22q11CNV disorders offer a unique possibility to follow patients from an early age onwards in order to identify molecular risk factors. Therefore, we wish to study psychopathology, cognition and genetic markers in people

with 22q11CNV disorders.

Study objective

To describe cognitive profile and psychopathology in adults with 22q11 CNV disorders.

Study design

The study design concerns an observational cross-sectional study, investigating cognitive and psychopathological profiles in 22q11CNV disorders.

Study burden and risks

The risks and burden associated with participation in this study are minimal and consists of one venapuncture and collection. This research is justified considering that according to current medical guidelines people with 22q11DS are advised to have yearly blood checks for monitoring somatic parameters and yearly monitoring of psychiatric and cognitive functioning (Bassett et al 2011). Because 22q11CNV disorders are rare disorders, and understanding the underlying mechanisms of schizophrenia is important, it is important to study this group of patients.

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

In order to be eligible to participate in this study, a subject must meet all of the following criteria:
-A deletion or duplication at chromosome 22q11 confirmed by FISH, micro-array or MLPA analysis.
-ability to give informed consent
-Written informed consent by participant
-Age 18 -65 years

Exclusion criteria

A potential subject who meets any of the following criteria will be excluded from participation in this study:
-chromosomal abnormalities other than 22q11 CNVs

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped

Start date (anticipated):	24-03-2015
Enrollment:	100
Type:	Actual

Ethics review

Approved WMO Date:	24-12-2014
Application type:	First submission
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)
Not approved Date:	03-06-2016
Application type:	Amendment
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

ID: 27226
Source: Nationaal Trial Register
Title:

In other registers

Register	ID
CCMO	NL50158.068.14
OMON	NL-OMON27226