Psychopathology and cognition in CNV disorders

Published: 23-10-2019
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The objective of this study is to describe cognitive profile and dimensions of psychopathology in subjects with CNV disorders.

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

Summary

ID

NL-OMON52635

Source
ToetsingOnline

Brief title
CNV disorders

Condition

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

Synonym
chromosomal abnormalities, Copy number variation

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: CNV, Cognition, psychopathology

Outcome measures

Primary outcome

Dimensions of psychopathology measured with different questionnaires/interviews

and cognitive functioning measured with a neuropsychological test battery.

Secondary outcome

NA

Study description

Background summary

Rare recurrent copy number variants (CNV*s), including chromosomal variations at loci 22q11.2 and 16p11.2, are among the most common genomic disorders and are associated with increased risk for neuropsychiatric disorders and cognitive dysfunction across the lifespan. However, clinical representations are heterogeneous and include symptoms of depression, anxiety, ADHD and psychosis.
Treatment advances of psychiatric disorders have been limited by lack of mechanistic understanding of the pathophysiology of the disorders. Increasing our understanding and development of treatment of mental illness requires integration of basic and clinical research with cutting-edge approaches in a developmental context. For early detection and novel therapeutics it is essential to elucidate the trajectory of neurodevelopmental processes and identify biomarkers. Genetic disorders such as 22q11 and 16p11 CNV disorders are unique human models to study the development of neuropsychiatric profiles and to fill the gaps in our knowledge. Research on psychopathology and cognitive function in genetic disorders offers an unique possibility to track development of psychiatric symptoms and cognitive functioning in order to identify genetic and environmental risk factors. Therefore, we wish to study psychopathology, cognition and genetic markers in people with CNV disorders.

Study objective

The objective of this study is to describe cognitive profile and dimensions of

psychopathology in subjects with CNV disorders.

Study design

This study is a cohort describing cognitive and psychopathological profiles in CNV disorders.

Study burden and risks

The risks and burden associated with this study are minimal and consist of one venipuncture. In accordance with treatment guidelines, adults with a CNV disorder, e.g. 22q11 and 16p11 CNVs, undergo a venipuncture at least once a year to check various somatic parameters. An annual check of psychological well-being and cognitive functioning is also recommended for these patients. Participation in this study therefore hardly enhances the burden for patients. CNV disorders are relatively rare but have the potential to generate necessary insights into the genetic and environmental mechanisms that underlie psychopathology. It is therefore important to study these unique disease models.

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)
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 CNV disorder confirmed by FISH, micro-array or MLPA analysis.
- Mentally competent (ability to give informed consent) and aged 12-16 years. In this case consent will also be given by the parents or legally authorized representative of the subject.
-Mentally competent (ability to give informed consent) and aged 16 years and older.
-Mentally incompetent aged 12 years and older. In this case consent will be given by the legal representative of the participant.

Exclusion criteria

A potential subject who meets any of the following criteria will be excluded from participation in this study:
-Medical or neurological disorders that may affect brain function (e.g. head trauma, CNS tumor)

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):	18-12-2019
Enrollment:	400
Type:	Actual

Ethics review

Approved WMO Date:	23-10-2019
Application type:	First submission
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)
Approved WMO Date:	11-03-2020
Application type:	Amendment
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)
Approved WMO Date:	27-06-2022
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: 25035
Source: NTR
Title:

In other registers

Register	ID
CCMO	NL70681.068.19