Psychopathology and Cognition in CNV disorders

No registrations found.

Ethical review	Positive opinion
Status	Recruiting
Health condition type	-
Study type	Observational non invasive

Summary

ID

NL-OMON25035

Source
Nationaal Trial Register

Brief title
CNV-disorders

Health condition

Copy number variant (CNV) disorders

Sponsors and support

Primary sponsor: azM
Source(s) of monetary or material Support: NIH

Intervention

Outcome measures

Primary outcome

• Dimensions of psychopathology measured with questionnaires/interviews.
• Cognitive functioning measured with a neuropsychological tests.
• Collection of blood samples for genetic analysis and cellular phenotyping.

Secondary outcome

Not applicable

Study description

Background summary

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.
Recurrent copy number variants (CNV's), including chromosomal variations at loci 22q11.2, 16p11.2, 1q21 and 15q11.2, are among the most common genomic disorders and are associated with increased risk for neuropsychiatric disorders and cognitive dysfunction across the lifespan. Clinical presentations are heterogeneous and include symptoms of depression, anxiety, ADHD and psychosis.
Genetic variants with high penetrance, such as the CNVs at 22q11, 16p11, 1q21 and 15q11.2, 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 a unique possibility to track development of psychiatric symptoms and cog-nitive functioning in order to identify genetic and environmental risk factors.
Therefore, we aim to study psychopathology, cognition, genetic markers, physical conditions and biological mechanisms 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 and collect biomaterials for genetic and mechanistic investigation.

Study design

One

Intervention

None

Contacts

Public
Maastricht University
claudia vingerhoets

0647988146
Scientific
Maastricht University
claudia vingerhoets

0647988146

Eligibility criteria

Inclusion criteria

• A genetically confirmed pathogenic CNV.
• Mentally competent (ability to give informed consent) and aged 16 years and older or
• Mentally incompetent aged 16 years and older.

Exclusion criteria

• Non-CNV related acquired brain trauma (e.g. trauma after an accident)
• Present use of illicit drugs

Study design

Design

Study type:	Observational non invasive
Intervention model:	Parallel
Allocation:	Non controlled trial
Masking:	Open (masking not used)
Control:	N/A , unknown

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-11-2019
Enrollment:	250
Type:	Anticipated

IPD sharing statement

Plan to share IPD: Undecided

Ethics review

Positive opinion

Date:	01-12-2019
Application type:	First submission

Study registrations

Followed up by the following (possibly more current) registration

ID: 52635
Bron: ToetsingOnline
Titel:

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

No registrations found.

In other registers

Register	ID
NTR-new	NL8198
CCMO	NL70681.068.19
OMON	NL-OMON52635

Study results