Psychopathology and Cognition in CNV disorders

Gepubliceerd: 01-12-2019 Laatst bijgewerkt: 15-05-2024

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.

Ethische beoordeling	Positief advies
Status	Werving gestart
Type aandoening	-
Onderzoekstype	Observationeel onderzoek, zonder invasieve metingen

Samenvatting

ID

NL-OMON25035

Bron
Nationaal Trial Register

Verkorte titel
CNV-disorders

Aandoening

Copy number variant (CNV) disorders

Ondersteuning

Primaire sponsor: azM
Overige ondersteuning: NIH

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

• Dimensions of psychopathology measured with questionnaires/interviews.
• Cognitive functioning measured with a neuropsychological tests.

• Collection of blood samples for genetic analysis and cellular phenotyping.

Toelichting onderzoek

Achtergrond van het onderzoek

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.

Doel van het onderzoek

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.

Onderzoeksopzet

One

Onderzoeksproduct en/of interventie

None

Contactpersonen

Publiek

Maastricht University
claudia vingerhoets

0647988146

Wetenschappelijk

Maastricht University
claudia vingerhoets

0647988146

Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

- 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.

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

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

Onderzoeksopzet

Opzet

Type:	Observationeel onderzoek, zonder invasieve metingen
Onderzoeksmodel:	Parallel
Toewijzing:	N.v.t. / één studie arm
Blindering:	Open / niet geblindeerd

Controle:	N.v.t. / onbekend

Deelname

Nederland

Status:	Werving gestart
(Verwachte) startdatum:	01-11-2019
Aantal proefpersonen:	250
Type:	Verwachte startdatum

Voornemen beschikbaar stellen Individuele Patiënten Data (IPD)

Wordt de data na het onderzoek gedeeld: Nog niet bepaald

Ethische beoordeling

Positief advies

Datum:	01-12-2019
Soort:	Eerste indiening

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

ID: 52635
Bron: ToetsingOnline
Titel:

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

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

Resultaten