Psychopathology and Cognition in 22q11 copy number variation disorders

Gepubliceerd: 12-06-2015 Laatst bijgewerkt: 15-05-2024

Psychopathology and Cognition in 22q11 CNV disorders

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

Samenvatting

ID

NL-OMON27226

Bron
Nationaal Trial Register

Verkorte titel
22q11 CNV disorders

Aandoening

22q11 CNV
cognitie schizofrenie psychopathologie.
22q11 CNV cognition schizophrenia psychopathology

Ondersteuning

Primaire sponsor: University Maastricht / Academic Hospital Maastricht, the Netherlands (AZM)
Overige ondersteuning: University Maastricht / Academic Hospital Maastricht, the Netherlands (AZM)

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

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

Toelichting onderzoek

Achtergrond van het onderzoek

The 22q11.2 chromosomal region is one of the regions that has received interest from psychiatric geneticists for over 20 years. A deletion at 22q11.2 is the first and only copy number variant (CNV) unequivocally implicated in schizophrenia, and this was known long before genome-wide analysis of CNVs for schizophrenia were published. In up to 30% of people carrying the deletion at 22q11.2, a psychotic picture fulfilling the DSM criteria for schizophrenia emerges during adolescence or adulthood. More recently, duplication of the same chromosomal region has been associated with a distinct syndrome, but with several features overlapping with 22q11.2 deletion syndrome including velopharyngeal insufficiency, congenital cardiac anomalies, cognitive deficits, behavioural problems, and psychiatric disorders like autism and attention-deficit hyperactivity disorder. Interestingly, psychotic disorders associated with 22q11.2 duplication have not yet been reported in the literature, which is possible due to the fact that most cases described to date involved children. Moreover, results from a recent hallmark study suggest that duplications at 22q11.2 might protect against schizophrenia.

Doel van het onderzoek

Psychopathology and Cognition in 22q11 CNV disorders

Onderzoeksopzet

one

Onderzoeksproduct en/of interventie

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

Contactpersonen

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Wetenschappelijk

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Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

-A deletion or duplication at chromosome 22q11.2 confirmed by FISH, micro-array or MLPA analysis.

-Ability to give informed consent

-Written informed consent by participant.

-Age 18-65 years

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

-Other chromosomal abnormalities

Onderzoeksopzet

Opzet

Type:	Observationeel onderzoek, zonder invasieve metingen
Onderzoeksmodel:	Anders

Controle: N.v.t. / onbekend

Deelname

Nederland

Status:	Werving gestopt
(Verwachte) startdatum:	24-03-2015
Aantal proefpersonen:	80
Type:	Werkelijke startdatum

Voornemen beschikbaar stellen Individuele Patiënten Data (IPD)

Wordt de data na het onderzoek gedeeld: Ja

Ethische beoordeling

Positief advies

Datum:	12-06-2015
Soort:	Eerste indiening

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

ID: 47626
Bron: ToetsingOnline
Titel:

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

Register	ID
NTR-new	NL5118
NTR-old	NTR5250
CCMO	NL50158.068.14
OMON	NL-OMON47626

Resultaten