Psychopathology and Cognition in 22q11 copy number variation disorders

No registrations found.

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

Summary

ID

NL-OMON27226

Source
Nationaal Trial Register

Brief title
22q11 CNV disorders

Health condition

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

Sponsors and support

Primary sponsor: University Maastricht / Academic Hospital Maastricht, the Netherlands (AZM)
Source(s) of monetary or material Support: University Maastricht / Academic Hospital Maastricht, the Netherlands (AZM)

Intervention

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

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.

Study objective

Psychopathology and Cognition in 22q11 CNV disorders

Study design

one

Intervention

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

Contacts

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Eligibility criteria

Inclusion criteria

-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

Exclusion criteria

-Other chromosomal abnormalities

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other

Control: N/A , unknown

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	24-03-2015
Enrollment:	80
Type:	Actual

IPD sharing statement

Plan to share IPD: Yes

Ethics review

Positive opinion

Date:	12-06-2015
Application type:	First submission

Study registrations

Followed up by the following (possibly more current) registration

ID: 47626
Bron: ToetsingOnline
Titel:

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

No registrations found.

In other registers

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

Study results