Neurocognitive endophenotyping: on the intersection of genetic disorders and psychiatry

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Cognitive testing in a structured and standardized way at a defined group of patients with cognitive dysfunction, with a shared molecular etiology .The intended objectives are:- Detailed human neurocognitive profile defining for each genetic defect...

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

Summary

ID

NL-OMON38921

Source
ToetsingOnline

Brief title
Neurocognitive endophenotyping

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Developmental disorders NEC

Synonym
Cognitive Disorders, Developmental disorders

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Sint Radboud
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Endophenotype, Genetic disorders, Neurocognitive, Psychiatry

Outcome measures

Primary outcome

neurocognitive profiles

Secondary outcome

-Identification of neurocognitive markers as endophenotypes in different

neuropsychiatric diseases caused by EHMT1 defects (ID, schizophrenia and

autism).

- Identification of cross species neurocognitive markers in EHMT1 defects

Study description

Background summary

Genetic factors contribute significantly to the aetiology of neuropsychiatric disorders. These factors are multifactorial, which results in any clear relationship between the genetic defects and the range of neuropsychiatric outcomes.
In recent years, next generation sequencing has revolutionized the identification of causative genetic mutations. These result in a wide and intense range of neuropsychiatric outcomes. In animal research this has been carefully elaborated. To translate these results into human results, it is necessary to carry out tests in humans.
By precisely identifying the pathophysiological mechanisms of this specific genetic defects, we're able to identify neurocognitive profiles. This may offer clues for future research.

See also the application form in the appendix.

Study objective

Cognitive testing in a structured and standardized way at a defined group of

patients with cognitive dysfunction, with a shared molecular etiology .
The intended objectives are:
- Detailed human neurocognitive profile defining for each genetic defect.
- Identification of translational neurocognitive endophenotypes between species.
- Gain insight in cognitive endophenotypes of neuropsychiatric syndromes with a shared etiology.

See also the application form in the appendix.

Study design

Population: n = at least 60 subjects.

Measures:
- Cognitive tests:
1. parts of the CANTAB: CAmbridge Neuropsychological Test Automated Battery (CANTAB (Owen, Downes, Sahakian, Polkey, & Robbins, 1990). This is a computer tablet based cognitive assessment battery, which investigates attention, memory and reaction time.
2. parts of the ADOS: Autism Diagnostic Observation Schedule (Lord et al., 1989) is a structured and standardized psychiatric observation and examination for assessing characteristics of autism.

- Phenotypic testing:
1.clinical interviews
a. The Vineland Adaptive Behavior Scale (VABS) (Sparrow, 1984) will be used to determine the adaptive functioning of people with an intellectual disability.
b. The mini Psychiatric Assessment Schedules for Adults with Developmental Disabilities (mini PAS-ADD) (Moss S., 1997) to determine behavioral problems and psychiatric disease in subjects with an intellectual disability by interviewing the proxy.
2.child psychiatric examination: the participant is asked to sing, draw, play with a doll and tea set, puzzle together with the investigator to assess the level of functioning, motor skills, language skills and social emotional skills.
3.questionnaires, which are completed by proxy of the participants:
a. CBCL: questions about problem behavior.
B.SCQ: questions about autistic features
c.temperament questionnaire
d. Questionnaire about socio-economic features.
e. Development questionnaire

See also the application form in the appendix.

Study burden and risks

There aren't any risks associated with participation.
The toys used for playing are approved for children and don't contain any small parts, which can be dangerous.

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)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

-Presence of one of the following specific genetic variants
- EHMT1 mutation (Kleefstra Syndrome)
- KANSL1 mutation (Koolen-deVries syndrome)
- ABJRD11 mutation (KBG syndrome)

- 15q13 mutation (Prader-Willi syndrome)
- Trisomie 21 (syndroom van Down).;-biological age above 3 years

Exclusion criteria

Multiple genetic defects within one subject

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	27-12-2013
Enrollment:	60
Type:	Actual

Ethics review

Approved WMO Date:	15-08-2013
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	26-05-2015
Application type:	Amendment

Review commission: CMO regio Arnhem-Nijmegen (Nijmegen)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

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

No registrations found.

In other registers

Register	ID
CCMO	NL43187.091.13