Imaging genetics in Autism Spectrum Disorders

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The objective of this study is to investigate the effects of replicated and theoretically interesting autism risk genes on brain anatomy in ASD. We will use psychiatric genetics and gene expression patterns to predict neuroimaging findings in ASD.

Ethical review Approved WMO

Status Recruitment stopped

Health condition type Psychiatric and behavioural symptoms NEC

Study type Observational non invasive

Summary

ID

NL-OMON35266

Source

ToetsingOnline

Brief title ImaGenASD

Condition

Psychiatric and behavioural symptoms NEC

Synonym

autism ASD

Research involving

Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Utrecht

Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: autism, autism spectrum disorders, genetics, MRI

Outcome measures

Primary outcome

The primary outcome measures will be genotype on autism risk genes and measures of brain structure, such as volume, gray and white matter density and white matter integrity.

Secondary outcome

Secondary outcome measures will be the results form questionnaires, interview and psychological assessment.

Study description

Background summary

Autism is a neurodevelopmental disorder that is characterized by impairments in social interaction and communication and stereotypic patterns of behavior. Epidemiological studies have indicated that the risk for autism spectrum disorders (ASD) is largely genetically determined: The prevalence of autism in siblings of individuals with autism is 2-8%, and concordance in monozygotic twins is as high as 60-91%. However, results from genetic linkage and association studies are inconsistent and the path by which a genetic risk for autism can result in the development of symptoms is not yet understood. Results from neuroimaging and post-mortem studies have indicated that functional and structural abnormalities in several brain regions are related to autism and related disorders.

Study objective

The objective of this study is to investigate the effects of replicated and theoretically interesting autism risk genes on brain anatomy in ASD. We will use psychiatric genetics and gene expression patterns to predict neuroimaging findings in ASD.

Study design

All subjects will be at least 6 years of age. Subjects will be asked to participate in a psychological assessment (maximum duration 2.5 hrs, an abbreviated 1 hr version will be used whenever possible) and subjects (or their parents) will be asked to participate in a structured interview (1.5 hrs), as well as fill out some questionnaires (45 min). For school age subjects, they will also be asked to approach a teacher to fill out a questionnaire (20 min). Subjects (and their parents) will be asked to provide a DNA sample for genetic analysis, either by blood, saliva or cheekswab. Subjects will be asked to participate in an MRI scanning session (45 min). Prior to the MRI scan, all child subjects (6 - 12 yrs) will participate in a protocolized practice session using an MRI simulator to desensitize them to the scanner environment, and prevent any anxiousness or nervousness. Only after acclimating the subject to the scanner environment, so that both the subject and parent are comfortable with the procedure, will the subject be taken to the actual scanner. As much time will be taken as is needed, but actual practice sessions usually take up to 30 minutes. If the subject or the parent is uncomfortable with any aspect of the procedure the study will be cancelled. The same procedure can be used with older subjects if the researcher or subject feels this is advisable.

Study burden and risks

There are no known risks associated with MRI acquisition, or any of the proposed methodologies, and we believe the impact on subjects will be minimal. Research into the genetic basis of neurobiological deficits in ASD will improve our insight into the pathophysiology of these disorders. Studies that elucidate the neurobiology of ASD will ultimately facilitate future design of new and effective ways to treat this disorder.

Contacts

Public

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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

General Inclusion criteria

- 1) Aged at least 6 years ;Inclusion criteria for subjects with ASD
- 1) DSM-IV (APA, 1994) diagnosis in the autism spectrum; Inclusion criteria for controls
- 1) no DSM-IV (APA, 1994) diagnosis
- 2) no scores in the clinical range on the Child Behavior Checklist (CBCL) and Teacher Rating Form (TRF)
- 3) IQ > 70

Exclusion criteria

- 1) major illness of the cardiovascular, the endocrine, the pulmonal or the gastrointestinal system
- 2) presence of metal objects in or around the body (pacemaker, dental braces)
- 3) history of or present neurological disorder
- 4) for individuals over 12 years of age: legal incompetence, defined as the obvious inability to comprehend the information that is presented by the investigator and is outlined in the Information letter and on which the decision to participate in the study is to be based

Study design

Design

Study type: Observational non invasive

Intervention model: Other

Allocation: Non-randomized controlled trial

Masking: Open (masking not used)

Primary purpose: Basic science

Recruitment

NL

Recruitment status: Recruitment stopped

Start date (anticipated): 19-05-2008

Enrollment: 600

Type: Actual

Ethics review

Approved WMO

Date: 12-02-2008

Application type: First submission

Review commission: METC Universitair Medisch Centrum Utrecht (Utrecht)

Approved WMO

Date: 05-07-2010

Application type: Amendment

Review commission: METC Universitair Medisch Centrum Utrecht (Utrecht)

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

NL19167.041.07