Specifying cognition, social interaction and autism-like traits in Fragile X

Published: 18-04-2018
Last updated: 13-04-2024

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

Summary

ID

NL-OMON46517

Source
ToetsingOnline

Brief title
Special X

Condition

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

Synonym
Fragile X Syndrome, FXS

Research involving
Human

Sponsors and support

Primary sponsor: Erasmus MC, Universitair Medisch Centrum Rotterdam
Source(s) of monetary or material Support: Sophia Stichting Wetenschappelijk Onderzoek (SSWO)

Intervention

Keyword: Autism Spectrum Disorder, Cognition, Developmental Delay, Fragile X Syndrome

Outcome measures

Primary outcome

For the three domains, the main study parameters are:

- ASD severity: ASD severity scores from structured behavioral observations.

- cognitive abilities: IQ, measured using an age-appropriate IQ test.

- (neuro)biological characteristics: brain activation patterns measured using

fNIRS.

Secondary outcome

Our secondary parameters will be: Biological measures (i.e. eye-movements and

FMRP levels; see section 8.3 for an extensive description of all assessments

and outcomes).

Study description

Background summary

Fragile X syndrome (FXS) is the most common single genetic cause of intellectual disability (ID) and autism spectrum disorder (ASD). Despite the high prevalence of ASD, relatively little is known about the exact profile of these symptoms in FXS, and the role of ID remains unclear. Further, little is known regarding the common or distinct neural mechanisms underlying ASD symptomatology in FXS and non-syndromic ASD. Due to the mild to severe ID and behavioral problems in FXS, studies investigating task-related brain activity in young children with FXS are lacking. Finally, the relation with other biological measures, such as expression levels of the fragile x mental retardation 1 (FMR1) gene protein product (FMRP) remains to be elucidated.

Study objective

Our primary objective is to compare children with FXS with children with

non-syndromic ASD, a developmental delay (DD) and typically developing (TD) children on three domains: ASD severity, cognitive abilities and (neuro)biological characteristics.
Furthermore, we aim to study ASD severity and (neuro)biological characteristics in mothers of children with FXS (permutation carriers), compared to their children (full mutation carriers) as well as a control group of mothers of typically developing children, in order to study the effects of carriership.

Study design

Cross-sectional observational study

Study burden and risks

This is a non-therapeutic study. The burden of the study for parents and children will mainly consist of the time and effort spent participating in the study visits and filling out questionnaires (parents only). A mobile lab will be used to visit participants at a location of their choosing to reduce the burden of participation. There are no known risks in the administration of neuropsychological tasks, filling out questionnaires, eye-tracking, fNIRS or hair plucking for FMRP. fNIRS is a non-invasive method for quantifying brain activity using only a headcap and allowing patients to freely move around. FMRP levels will be assessed from hair roots, a non-invasive procedure specifically suitable for young and intellectually disabled populations. With regard to group relatedness; because we aim to generate knowledge regarding the ASD profile and its underlying etiology in patients with FXS, no other study groups can be studied instead. Similarly, since knowledge in children with the disorder is especially lacking, we will study a pediatric population. To be able to parse out the effect of ID on the ASD profile, and to study the specificity of ASD symptoms and severity for FXS, a comparison with developmentally delayed controls will have to be made.
As an indirect benefit this study could contribute to more focused diagnosis, improved assessment of treatment efficacy and the development of personalized treatment for children with FXS or ASD as well as help unravel the etiology of ASD.

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

Children aged 6-18 years, (corrected to) normal vision, Dutch as first language. Four groups of 55 participants are included.
Two groups of mothers (n<=40 per group) will be included, mothers of children in the FXS and in the TD group.
Group specific in- and exclusion criteria are provided in section 4.2 and 4.3 of the research protocol.

Exclusion criteria

Four groups of participants are included. Group specific in- and exclusion criteria are provided in section 4.2 and 4.3 of the research protocol.

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):	06-02-2019
Enrollment:	300
Type:	Actual

Ethics review

Approved WMO

Date:	18-04-2018
Application type:	First submission
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)

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	NL62172.078.17