Detailed Speech & Language Phenotyping in SATB2-Associated syndrome

Published: 12-09-2018
Last updated: 10-04-2024

With this study we aim to define a detailed oral motor, speech and language profile for patients with SAS.

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

Summary

ID

NL-OMON46826

Source
ToetsingOnline

Brief title
SPELA-SAS

Condition

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

Synonym
intellectual disability, SATB2-associated syndrome

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum
Source(s) of monetary or material Support: NWO Zwaartekracht subsidie (24.001.006) aan het Language in Interaction consortium

Intervention

Keyword: Language, Phenotyping, SATB2, Speech

Outcome measures

Primary outcome

A common speech and language phenotype (*deep phenotype*) for patients with SAS, with a detailed description on three different domains: oral motor skills, speech and language. In addition to that, a non-verbal and/or performance IQ score.

Secondary outcome

NA

Study description

Background summary

SATB2 is among the most frequently disrupted genes in neurodevelopmental disorders. Patients with SATB2-associated syndrome (SAS) have general developmental delays and intellectual disability, but speech/language development is relatively more severely affected. Patients with SAS generally have limited or absent speech, while language comprehension seems to be more preserved. So far, not much is known on the details of this disturbed speech and language development.
Better knowledge on the communication skills and deficits of SAS patients can guide treatment and interventions, and help in better understanding of pathogenesis of disorder.

Study objective

With this study we aim to define a detailed oral motor, speech and language profile for patients with SAS.

Study design

This study is an observational study, with a descriptive study design. Subjects with SAS will undergo speech/language testing, together with baseline

neuropsychological testing.

Study burden and risks

There are no risks associated with participation. The burden for patients will be small, and the study can only be done using this patient group. As there is currently a clear lack of knowledge about the speech/language phenotype for SAS, results from this study could help parents/caregivers and therapists in better recognizing and understanding the speech/language problems and skills. This could also guide interventions in a speech therapy setting.

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

- A molecular diagnosis of SATB2-associated syndrome (SAS)
- At least two years old at the time of testing
- Raised in Dutch-speaking family

Exclusion criteria

- the presence of another gene disruption/molecular diagnosis that is likely to contribute to the neurodevelopmental phenotype

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	11-04-2019
Enrollment:	25
Type:	Actual

Ethics review

Approved WMO Date:	12-09-2018
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	19-12-2018

Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	01-08-2019
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	NL64562.091.18