Language impairment in the 22q11 deletion syndrome

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Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Mental impairment disorders
Study type	Observational non invasive

Summary

ID

NL-OMON54844

Source
ToetsingOnline

Brief title
Language impairment in 22q11DS

Condition

- Mental impairment disorders

Synonym
developmental language disorder, developmental language impairment

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Utrecht
Source(s) of monetary or material Support: NWO - Social Sciences and Humanities

Intervention

Keyword: 22q11DS, language development, language disorder, SLI

Outcome measures

Primary outcome

(1) Similarity/ difference in developmental language profiles of children with 22q11DS and children with SLI, in comparison to neurotypical (typically developing) controls.

(2a) Similarity / difference of the 22q11DS and SLI groups with respect to neurocognitive mechanisms hypothesized to be critical for language acquisition: short-term/working memory, executive function, and implicit learning.

(2b) The degree of association between neurocognitive performance and language outcomes in the 22q11DS and SLI groups.

Secondary outcome

NA

Study description

Background summary

Developmental language impairments have a profound impact on an individual*s life. Difficulties in acquiring a native language can result from unfavorable physical, psychological, social or educational conditions. In addition, approximately 7% of all children show severe and persistent delays in language skills without any obvious neurological, psychological or social causes. This condition is called specific language impairment (SLI). SLI is diagnosed by exclusion; there is no unambiguous clinical marker. Consequently, the SLI population is heterogeneous, and this hampers efforts to elucidate the pathway from (genetic) cause, via neurocognitive deficits to impaired language. An approach that is expected to bring new insights is to compare SLI with the 22q11.2 deletion syndrome (22q11DS). Virtually all children with 22q11DS show severe language delays in the early stages of development. This leaves room for

the hypothesis that the language delays in 22q11DS are a self-contained symptom, unrelated to any of the other known physical or psychological symptoms. If so, the 22q11DS population shares a characteristic with the SLI population. In contrast to SLI, 22q11DS has a uniform genetic etiology - a microdeletion in the long arm of chromosome 22 - which also provides for an unmistakable clinical marker, which can be detected at an early age. Our current knowledge about the development of language in 22q11DS is limited and direct comparisons with SLI have not yet been made. The present study aims to fill this void by performing an in-depth longitudinal comparison of language development in young children with 22q11DS and age-matched children with SLI. SLI has been associated with specific neurocognitive deficits, particularly with regard to short-term and working memory, executive functions, and implicit learning. If the language difficulties in 22q11DS are similar in nature to those seen in SLI, we expect to also see similar neurocognitive deficits in the 22q11DS population. For this reason we will study the neurocognitive profles of both populations and relate these to their developmental language profiles. This study will thus yield new knowledge on (disordered) development of language in children with 22q11DS and its neurocognitive underpinnings. This study will not only enhance our understanding of the 22q11DS phenotype, but can also be used to improve speech-language diagnostics and prognostics in this population, which in turn will have positive impact on counseling parents/caretakers, and managing their expectations with regard to child*s condition.

Study objective

The first objective of this study is to provide a detailed, quantitative and qualitative description of early language development (*developmental language profile*) in children with 22q11DS, comprising expressive and receptive abilities, covering all levels of linguistic structure (sounds, words, grammar), as well as the use of language in conversation and narration. This developmental language profile will be compared to that of children with SLI, so as to determine whether the two populations are phenotypically similar, as well as to the developmental language profile of healthy, typically developing children.

The second objective is to determine if children with 22q11DS and children with SLI show deficiencies in short-term and working memory, executive function, or implicit learning, and to assess the degree to which such deficiencies are (causally) related to language impairments in these populations.

Study design

Longitudinal observational case-control study, involving three groups of children.

Study burden and risks

No immediate benefits are expected for the participants in this study. The results of this study will enhance our understanding of the 22q11DS phenotype, and are also expected to be an impetus for improving speech-language diagnostics and prognostics in this population. In addition, the results may be conducive to improving advising parents/caretakers, and managing their expectations with regard to their child*s condition.
The instruments and procedures used are routinely employed by psychologists and linguists studying the development of language and its neurocognitive underpinnings. There are no known risks. The burden imposed on participants and their parents/caretakers is related to the time investment they are asked to make: the study involves 1 to 5 assessment sessions, three of which will take 1,5 hours (2 x 45 min.), one which will take 45 minutes, and one of which will take between 45 and 75 minutes spread out over a period of 4;8 year for children with 22q11DS. For children with SLI the study involves 1 to 3 assessment sessions, of which only the first will take 1,5 hours (2 x 45 min.) and the second and third will take 45 minutes, spread out over a period of max. 1 year. For controls the study involves 1 to 4 assessment sessions, of which only the first and fourth will take 1,5 hours (2 x 45 min.) and the second and third will take 45 minutes, spread out over a period of 4;8 year. As our research questions concern the development of a complex, multifaceted cognitive capacity, i.e., changes in ability as a function of age, a design that is less burdensome is not possible. To reduce the burden on participants and their parents/caretakers we have reduced the number of test procedures and test sessions as much as possible. Clearly, the age range of the participants is dictated by the phenomenon under study, primary language acquisition and development.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Children (2-11 years)

Inclusion criteria

Age: 3-6,5 years
Growing up in monolingual, Dutch-speaking families
Patient groups: a diagnosis of 22q11 deletion syndrome OR Specific Language Impairment (which is not strictly a medical condition)
Control group: healthy, typically developing children; confirmed absence of any of the above conditions.

Exclusion criteria

severe hearing loss (35 dB or worse)
growing up in a family in which one or more other languages in addition to Dutch are regularly used to communicate with the child

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:	Other

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	02-01-2019
Enrollment:	0
Type:	Actual

Ethics review

Approved WMO Date:	04-01-2018
Application type:	First submission
Review commission:	METC NedMec
Approved WMO Date:	07-11-2018
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	04-04-2019
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	05-09-2019
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	18-09-2019
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	29-01-2020
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	21-08-2020

Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	26-11-2020
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	14-03-2023
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	11-06-2024
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	NL63223.041.17