Vestibular function in patients with 22q11.2 deletion syndrome

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Primary Objective: - To determine the rate of functional vestibular abnormalities in 22q11DSSecondary Objective: - To determine the rate of anatomical malformations of the inner ear in 22q11DS- To study the relation between vestibular functioning...

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-OMON45597

Source
ToetsingOnline

Brief title
Vestibular function in 22q11DS

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Congenital ear disorders (excl deafness)

Synonym
22q11.2 deletion syndrome, velocardiofacial sydnrome

Research involving
Human

Sponsors and support

Primary sponsor: Keel-, Neus- en Oorheelkunde
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: 22q11.2 deletion syndrome, motor development, semicircular canal, vestibular function

Outcome measures

Primary outcome

The main study parameters of the study is the vestibular function in patients

with 22q11.2 deletion syndrome.

We will use elektronsytagmography (rotatory chair testing and calorisation) and

oVEMP to study this.

Secondary outcome

The secondary study parameters are:

- The presence or absence of anatomical malformations of the inner ear

- The relation between anatomical malformations of the inner ear and

vestibular function in patients with 22q11.2 deletion syndrome.

This will be studied the MRI images of the temporal bones (and the vestibular

tests).

- The relation between vestibular function and motor development in patients

with 22q11.2 deletion syndrome.

For this parameter we will use data extracted from the Van Wiechenschema, where

the age of patients at which motor developmental milestones are reached, are

noted.

The last study parameters is:

- The relation between anatomical inner ear malformations and hearing

loss in patients with 22q11.2 deletion syndrome.

For this parameter the pure tone audiogram is performed (and the MRI scan of

the temporal bones).

Study description

Background summary

The 22q11.2 deletion syndrome (22q11DS) is a heterogenic syndrome, over 180 possible features are described. It is caused by a microdeletion on the long arm of chromosome 22. Recently we studied hearing and otologic features of this syndrome. During this study we examined nine available Computed Tomography (CT) scans and one Magnetic Resonance Imaging scan of the temporal bones and found that six of the 10 patients had a malformed vestibular system. The lateral semicircular canal and vestibule were fused to one single cavity in seven ears (35%) in four patients and the lateral semicircular canal was malformed with a small bony island in four ears (20%) in two patients. These results are also reported by Loos et al. who analyzed the CT scans of 11 patients with 22q11DS. They found in four ears a lateral semicircular canal and vestibule fused to one single cavity, and in 14 ears a wide vestibule, which in two ears consisted in combination with a wide lateral semicircular canal [5]. This raised the question if these anatomical anomalies lead to a loss of vestibular function. In our study, we could not find a history of balance problems described in the medical files. However, balance problems are reported in 22q11DS, but this is considered related to neuromotor deficits, including motor delay and hypotonia. In addition, balance is important for motor development, and motor delay is frequently present in 22q11DS. We hypothesize that anatomical malformations of the vestibular system lead to a loss of vestibular function, which could contribute to motor delay, rather than to dizziness in daily life in 22q11DS.

Study objective

Primary Objective:
- To determine the rate of functional vestibular abnormalities in 22q11DS

Secondary Objective:
- To determine the rate of anatomical malformations of the inner ear in 22q11DS

- To study the relation between vestibular functioning and anatomical malformations of the vestibular system in 22q11DS
- To study the relation between vestibular function and motor development in 22q11DS

Other Objectives:
- To study the relation between hearing loss and malformations of the inner ear in 22q11DS

Study design

The design of the study will be a cross-sectional study. The duration of the study will depend on the speed of inclusion, but we expect it to take no more than one year. The study will be conducted in the University Medical Center Utrecht (UMCU). The patients will be recruited from a psychiatric study (METC protocol number 08-345/K), a random sample of patients (the first 30 patients who meet our inclusion criteria) will be asked for informed consent. Hereafter the patient will undergo a pure tone audiogram, tympanogram, an electronytagmography, ocular vestibular evoked myogenic potentials (oVEMP) and an MRI scan of the temporal bones. The electronystagmography, oVEMP and audiometric tests will take place in the otorhinolaryngology department and the MRI scan will be performed in the radiology department. Next, we will retrospectively collect data on motor development by contacting the Child Health Care Center (Gemeentelijke Gezondheidsdienst). Here we will collect the Van Wiechenscheme where the age of the patient at which motor milestones are reached is noted.

Study burden and risks

This study can only be performed in this patient population, because we want to study this syndrome specifically. Furthermore, we want to study the effect of vestibular abnormalities on motor development. Since these data are collected in all patients at a young age and kept for 15 years, we only want to include children aged 12-17 years. Other than knowledge on his or her anatomy and function of the vestibular system the patient will not benefit from this study. During and shortly after rotatory chair and caloric testing the patient will experience dizziness and/or nausea, which will disappear within minutes. There are no risks or other discomforts involved in this study. The patient will be asked to visit the hospital once or twice. The vestibular tests and hearing tests can be easily combined in one visit and will take about two hours. The MRI scan of the temporal bones will be combined with an MRI of the brain, performed in a psychiatric study (METC protocol number 08-345/K). Our MRI scan will add an extra 8 minutes to the total scan time.

Contacts

Public
Selecteer

Heidelberglaan 100
Utrecht 3584 CX
NL
Scientific
Selecteer

Heidelberglaan 100
Utrecht 3584 CX
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)

Inclusion criteria

- Signed informed consent
- 12-17 years old
- Diagnosed with 22q11.2 deletion syndrome with fluorescence in situ hybridization or multiplex ligation-dependent probe amplification confirming the presence of a deletion.

Exclusion criteria

- Unable to undergo electornystagmography, due to a tympanic membrane perforation or tympanic membrane tube, or due fear or other similar reasons.
- Unable to undergo MRI scanning, due to claustrophobia, unable to potentially lay still, presence of metal objects in the body that cannot be taken of, possibility of pregnancy.

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	13-12-2017
Enrollment:	30
Type:	Actual

Ethics review

Approved WMO

Date:	26-07-2017
Application type:	First submission
Review commission:	METC NedMec

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	NL60737.041.17