Investigating oculomotor and vestibular function in individuals with Rett syndrome

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* To investigate oculomotor and vestibular activity in individuals with RTT. * To compare oculomotor and vestibular activity with more functional eye gaze and communication skills. * To compare oculomotor and vestibular activity with severity of...

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Neurological disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON47151

Source ToetsingOnline

Brief title

Oculomotor and vestibular function in individuals with Rett syndrome

Condition

• Neurological disorders congenital

Synonym MECP2 Disorder; Rett syndrome

Research involving Human

Sponsors and support

Primary sponsor: Medisch Universitair Ziekenhuis Maastricht **Source(s) of monetary or material Support:** Ministerie van OC&W

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Intervention

Keyword: eye tracking, oculomotor, Rett syndrome, vestibular

Outcome measures

Primary outcome

1. Insight into the oculomotor and vestibular systems of individuals with RTT

(which may include comparison with a control/normative reference group or other

comparative group).

2. Comparison between the objective measures for each individual and their

functional use of eye gaze for communication.

3. Indication of any possible correlation between vestibular dysfunction and

development of scoliosis in individuals with RTT.

Secondary outcome

Comparison between testing via the "standard" ENG procedure and with virtual

reality goggles and/or video-tracking glasses.

Study description

Background summary

Rett syndrome (RTT) is a severe neurodevelopmental disorder predominantly affecting females. It is characterised by the progressive loss of motor and communication skills after seemingly near-normal development in the first year of life, accompanied by stereotypic hand clapping behaviours and additional features including abnormal breathing, scoliosis and epilepsy. Apraxia is one of the significant features of the syndrome, affecting (amongst others) those movements that are necessary to communicate in traditional ways such as through speech, gesture and / or manual signs. Many individuals with RTT are thought to understand more language than they are able to express. They are reported to retain good eye pointing skills, which opens up a way to communicate through eye gaze and/or eye tracking technology. However, not all individuals with RTT succeed in their attempts to use eye gaze systems for communication and it is unclear where points of breakdown lie.

It is hypothesised that neural and motor movements associated with eye gaze are (relatively) less affected by apraxia than other pathways. To date, however, the neural pathways shared by the oculomotor and vestibular systems have been little researched in RTT. Investigation of these basic pathways prior to introducing an eye gaze system for communication could indicate any underlying problems which might hinder future success and could contribute towards localisation of the problem for anyone already struggling to use such a system. These same pathways may also be significant for the development of scoliosis in adolescence in RTT. A link has been identified between oculomotor and vestibular system dysfunction and amplitude of spine deformity in cases of idiopathic adolescent scoliosis; such a relationship has not yet been explored in RTT. Evidence of a link could have implications for therapy and non-surgical, or at least pre-surgical, intervention which is significant in a medically-vulnerable population.

The oculomotor and vestibular assessments routinely conducted by the Department of Otorhinolaryngology (Division of Balance Disorders) at MUMC+ offer potentially deeper insights into (1) neural pathways and motor activity underpinning observable eye gaze/eye tracking behaviours in RTT, and (2) a possible association between visuo-ocular and vestibulo-ocular responsivity and scoliosis in RTT. These tests are already incorporated into the routine package of clinical assessment offered to individuals with RTT within the context of three-day inpatient brainstem registration. The aim now is to offer these same oculomotor and vestibular assessments to individuals with RTT attending the multidisciplinary outpatient clinic and to combine the results from both sets of subjects for research as well as clinical purposes. Once established with these groups a further aim is to invite the wider RTT community (via the Dutch Rett Syndrome Association, the Dutch Rett Syndrome Foundation or speech and language therapists) to participate in the assessments.

Collecting objective measures associated with neural pathways has a two-fold benefit: firstly, at a clinical (individual) level, the assessments contribute to the delineation of a more detailed profile of an individual*s skills and needs which can inform planning of their interventions and therapy, improving their personal quality of life; secondly, at a research (collective) level, insight into the neural pathways can be collated anonymously to contribute to the growing body of knowledge about RTT which can inform global recommendations around intervention planning and therapeutic management, improving quality of life for all individuals with RTT. These aims fit with the wishes expressed by parents.

Study objective

* To investigate oculomotor and vestibular activity in individuals with RTT.

* To compare oculomotor and vestibular activity with more functional eye gaze and communication skills.

* To compare oculomotor and vestibular activity with severity of scoliosis.

Study design

Observational study

Study burden and risks

1. Individuals who come as an inpatient onto the PICU for three-day brainstem registration:

The oculomotor, vestibular and functional communication assessments are already established as part of the clinical routine during the three-day brainstem registration. There is no added burden to subjects and families except that the parents will be asked to take a little time (10-15 minutes) to complete a questionnaire at home prior to the session, and the results from the assessments will be used for research purposes in addition to providing individual clinical data.

2. Individuals who come to the multidisciplinary outpatient Rett clinic for a one-day consultation:

The oculomotor, vestibular and functional communication assessments will be added to the investigations undertaken routinely during the multidisciplinary clinic. The added burden will amount to one-two hours of added time on the day of the clinic visit, and completion of a questionnaire at home prior to the session (10-15 minutes).

3. Individuals who would not otherwise be coming to the hospital: The time burden from participation in the study will amount to one-two hours* attendance at the hospital in addition to travel time on the day of assessment and completion of the questionnaire at home prior to the session (10-15 minutes).

In all cases, there will be no added psychological burden from completion of the functional communication questionnaire or any additional communication assessments. In order to protect the subjects and their families, the parents/guardians will be asked to give informed consent for the use of their data for research purposes and the data will be anonymised, the subject*s data for research purposes and the data will be anonymised.

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

- 1. Clinical diagnosis of RTT.
- 2. Age 2 years and above.
- 3. Parents/guardians give informed consent

Exclusion criteria

1. Unable to sit on the chair in the BalanceLab, either alone or on a parent*s knee.

2. Physical/medical status too vulnerable. For example, the subject needs constant assistance from a supportive system such as an oxygen tank.

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)

Primary purpose: Other

Recruitment

NI

Recruitment status:	Recruiting
Start date (anticipated):	08-02-2017
Enrollment:	100
Туре:	Actual

Ethics review

Approved WMO	
Date:	05-10-2016
Application type:	First submission
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)
Approved WMO	
Date:	16-01-2019
Application type:	Amendment
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht. METC azM/UM (Maastricht)

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

ССМО

ID NL57673.068.16