Assessing underlying skills of individuals with Rett syndrome

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Part One:
Assessing eye movement and oculomotor function

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Relevance of eye gaze research:

- Individuals with Rett syndrome demonstrate an ability to communicate with their eyes in the context of:
  - loss of speech
  - loss of purposeful hand movement
- “..intense eye communication – eye pointing” (Neul et al, 2010)
- Increasing use of eye gaze/eye tracking technology for communication

Definitions of eye pointing:

- “...the context-relevant, controlled and intentional use of gaze in order to direct one or more partner’s visual attention to any item or object for a deliberate communicative purpose” (Sargent et al, 2013)

- “...the integrated outcome of a combination of visual, social, cognitive and motor skills” (Sargent et al, 2013)

Successful communication using eye pointing/eye gaze and/or eye tracking technology requires:

- Arousal and alertness
- Visual attention & memory
- Visual perception & acuity
- Intact oculomotor system
- Motivation & desire to communicate
- Awareness & understanding of cause-effect & reciprocity
- Awareness & understanding of world
- Understanding of spoken and written language
Basic horizontal gaze pathway:

Oculomotor movements:

- Visual-ocular movements
  - Saccades
  - Smooth Pursuit (SP)
  - Optokinetic Nystagmus (OKN)
  - Vergence
- Vestibular-ocular reflex (VOR)
- Gaze-shifting – Saccades
- Gaze-holding – SP, OKN, Vergence and VOR
Aim of study:

• To explore whether individuals with Rett syndrome show the same speed & range of eye movements as their neuro-typical peers (have intact oculomotor function)

Method:

• Traditional electronystagmography (ENG)
• Two groups of children/young people
  – Rett syndrome
  – typically-developing
Participants:

<table>
<thead>
<tr>
<th>Group</th>
<th>Mean age (yrs:mths)</th>
<th>Range (yrs:mths)</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td>Controls N=16</td>
<td>7:8</td>
<td>3:6 – 18:7</td>
<td>4:0</td>
</tr>
</tbody>
</table>
ENG test materials & conditions:
Results:

SP present  

SP completely smooth  

Saccades present (to R & L)  

Saccades latency

Dysmetria present  

VOR present  

OKN present  

OKN correct
Overall findings:

• Individuals with Rett syndrome demonstrated a similar range of eye movements to their neuro-typical peers
  – all responses that could be measured showed SP, saccades, OKN, VOR

• However, challenges in assessing individuals with Rett syndrome led to limitations in quantitative analysis
  – e.g. reduced motivation/attention to test materials, low quality electrode signals

• Two areas of potential difference were saccades latency and VOR
  – individuals with Rett syndrome showed slower responses
In summary:

- The starting point for assessing the complex matrix of requirements for functional use of eye gaze/eye pointing for communication is whether an individual can maintain and shift fixation of gaze.

- This study suggests that individuals with RTT have a ‘typically-functioning’ range of eye movements (intact oculomotor function).

- However, possible differences in speed (saccades latency) should be explored further.

- Modifications should be made to traditional ENG assessment due to challenges in testing e.g. trial use of virtual reality goggles.

- Testing should be extended – to include use of eye gaze to assess cognitive and language skills.
References:


Editorial

Part Two: Assessing language and cognition

• Small-scale pilot study undertaken by Callie Ward and Gill Townend with support from Shula Chiat (City, London University)

• Based on studies published by
  - Clarkson et al. (2017) = adapted Mullen Scales of Early Development (MSEL) for eye gaze access
  - Ahonniska-Assa et al. (2018) = adapted Peabody Picture Vocabulary Test (PPVT-4) for eye gaze access
Background:

• Standard tests of cognition and language usually require motoric and/or verbal responses
• This makes assessment challenging for people with Rett syndrome
• The new communication guidelines do not endorse any specific formal (standardised) assessments, though it is recognised they may be adapted and used sometimes to test some skills
• Historically all individuals with Rett syndrome have been considered to be severely cognitively impaired
• Anecdotal evidence and recent research suggests there is a wider range of ability
• “Eye-tracking and gaze-controlled communication may be the key to unlocking the potential of people with Rett syndrome.” (Loffler & Gordon, 2018)

Aim of study:

- To test visual reception (VR), receptive language (RL) and expressive language (EL) skills of children with Rett syndrome using eye gaze access
- To compare performance on formal (standardised) assessments and informal assessment tasks

Method:

- Formal assessment of VR, RL, EL skills using adapted MSEL (MSEL-A and/or MSEL-ET) (Clarkson et al, 2017)
- Informal activities such as storybook reading and cake decorating to test the same skills in each child
Participants & procedures:

- 10 children with Rett syndrome
- Age range 4:0 years – 6:8 years
- Assessments conducted in each child’s home
- Across a single day for each child – with breaks as often as needed
- Assessment tasks varied (within limits) between children
- Two assessors
- Two video cameras – one in front / one behind child
- GazeViewer™ – to capture eye movements on screen
Formal assessment – MSEL VR & RL tasks

- As adapted by Clarkson et al. (2017)
  - For eye tracking on an eye gaze controlled computer (MSEL-ET)
  - For eye gaze/eye pointing and body movement in response to object and picture materials (MSEL-A)
Informal assessment tasks

- Games on eye gaze device to check ability to track stationary & moving targets
- Storybook activity
- Cake decorating activity

<table>
<thead>
<tr>
<th>VR observations:</th>
<th>Able to calibrate device</th>
<th>Selects stationary targets</th>
<th>Selects moving targets</th>
<th>Reveal hidden things</th>
<th>Finds matching shape from range of 2</th>
<th>Finds matching shape for range of 4</th>
</tr>
</thead>
<tbody>
<tr>
<td>RL observations:</td>
<td>Size concepts</td>
<td>Identifies colours</td>
<td>Identifies objects</td>
<td>Identifies use of objects</td>
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<tr>
<td>EL observations:</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>On eye gaze device</td>
<td>Language functions</td>
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<td>Has a way to communicate yes &amp; no</td>
<td>Grid size used</td>
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</table>
Results: Formal assessment (MSEL)

Expressive Language = all “very low” e.g. laugh, happy vocalisations
(MSEL does not recognise/was not adapted for AAC use)
<table>
<thead>
<tr>
<th>No.</th>
<th>Age (months)</th>
<th>Visual Reception</th>
<th>Receptive Language</th>
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<tr>
<td></td>
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<td>Able to calibrate device</td>
<td>Selects stationary targets</td>
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<tr>
<td>1</td>
<td>53</td>
<td>Y</td>
<td>Y</td>
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<td>2</td>
<td>68</td>
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### Results: Informal assessments – EL

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<th>Age (months)</th>
<th>Has a way to communicate yes &amp; no</th>
<th>Grid size used</th>
<th>Uses single buttons in context</th>
<th>Combines two buttons in context</th>
<th>Combines three buttons in context</th>
<th>Navigates independently between pages</th>
<th>Expresses a choice from 2 or more items</th>
<th>Gives instructions</th>
<th>Asks questions</th>
<th>Answers questions</th>
<th>Communicates spontaneously</th>
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<tbody>
<tr>
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<td>20</td>
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Discussion points:

• Marked discrepancy between EL skills on formal & informal assessment - informal tasks were more adaptable to/able to recognise AAC use
• All participants who took part in VR & RL tests scored at 29 months of age or above which is higher than other (earlier) studies suggest
• Many participants achieved a Baseline score only & did not reach a Ceiling, therefore, results reflect minimum achievement, not maximum
• Findings suggest that some children with Rett syndrome have age appropriate (or above) cognitive and receptive language skills
• And, despite limitations of Rett syndrome individuals are able to learn and demonstrate skills
• Both formal and informal assessments can give a better understanding of skills/abilities – but need to look for & adapt to preferences of each person
Limitations & future considerations:

• Small-scale pilot study = small sample size
• Length of assessment / time required
• Variability in assessments & subjectivity within informal assessments
• Varying effects of symptoms of Rett syndrome
• Ceiling level not reached for everyone
• Individuals above upper age limit of standardised scoring scale
• But...results are promising!
• ...and suggest a larger scale study should be pursued
References:


Thank you for your attention!

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