

# Assessing underlying skills of individuals with Rett syndrome

**Dr. Gillian S. Townend**

Researcher, Rett Expertise Centre Netherlands-GKC, Maastricht University Medical Center, The Netherlands

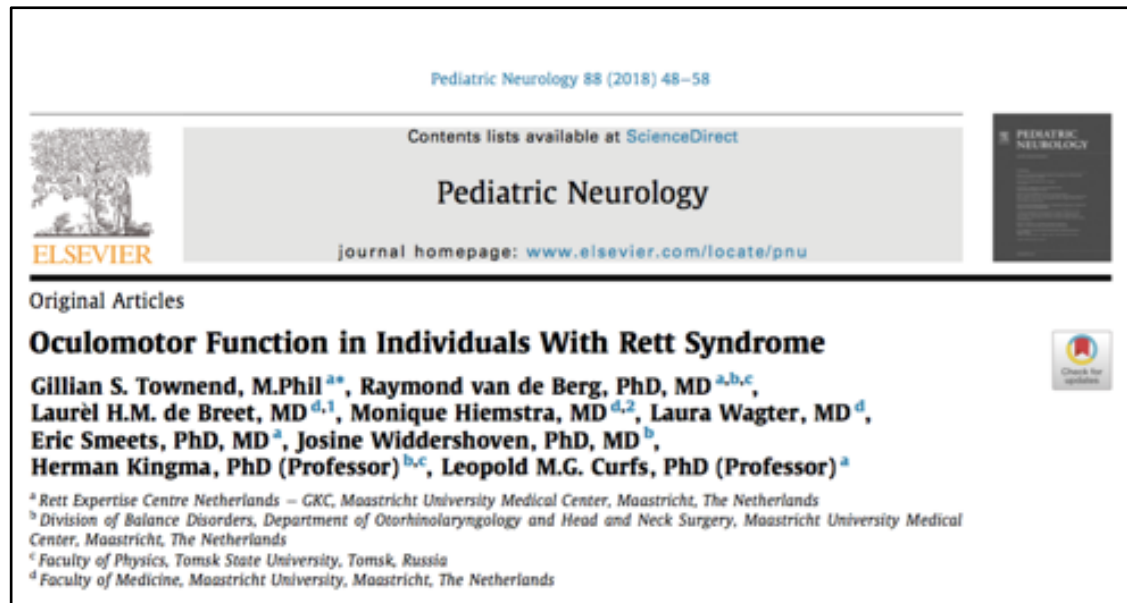
and

Joint Project Lead, Communication and Education Support, Rett UK

6<sup>th</sup> European Rett Syndrome Conference, Tampere, Finland

28<sup>th</sup> September 2019

# Part One: Assessing eye movement and oculomotor function



Gillian S Townend, Raymond van de Berg, Laurèl HM de Breet, Monique Hiemstra, Laura Wagter, Eric Smeets, Josine Widdershoven, Herman Kingma, Leopold MG Curfs

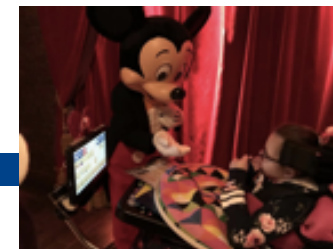
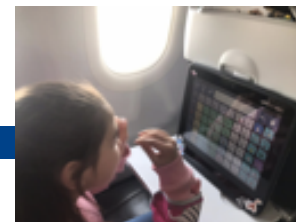
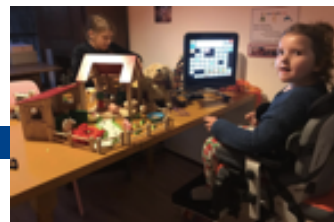
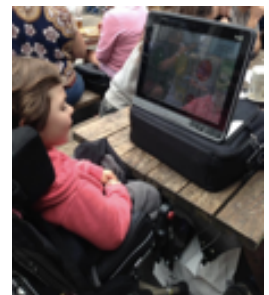
# Relevance of eye gaze research:



- 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



Neul JL et al. (2010). Rett syndrome: Revised diagnostic criteria and nomenclature. *Annals of Neurology*, **68**(6), 944-950.



## 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)

Sargent J et al. (2013). Use of eye-pointing by children with cerebral palsy: what are we looking at? *International Journal of Language & Communication Disorders*, **48**(5), 477-85.

# 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



## 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:

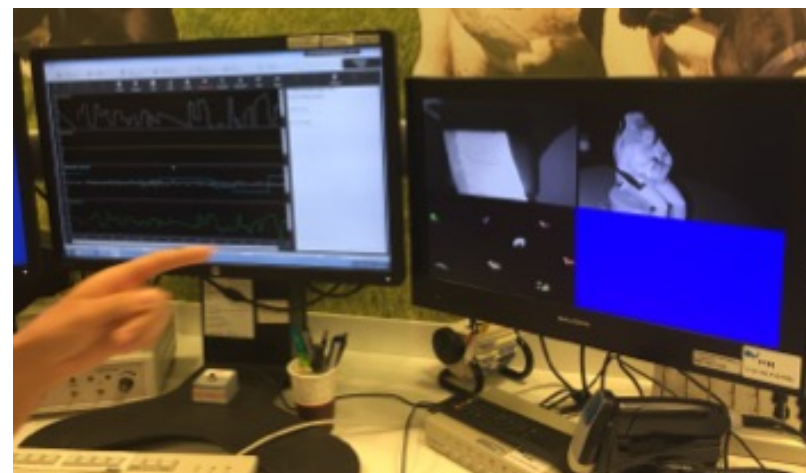
- Traditional electronystagmography (ENG)
- Two groups of children/young people
  - Rett syndrome
  - typically-developing

## Participants:

Group	Mean age (yrs:mths)	Range (yrs:mths)	SD
<b>Rett syndrome N=18</b>	9:7	2:7 – 25:11	6:5
<b>Controls N=16</b>	7:8	3:6 – 18:7	4:0

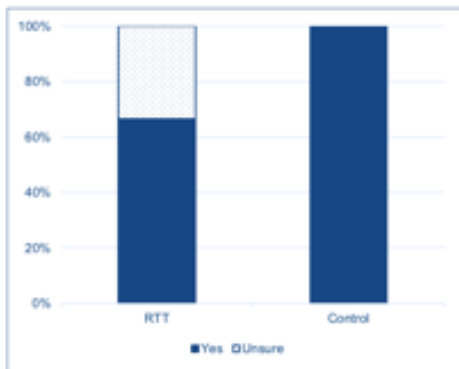


## ENG test materials & conditions:

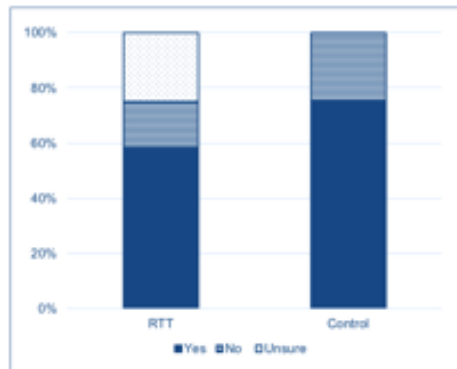


# Results:

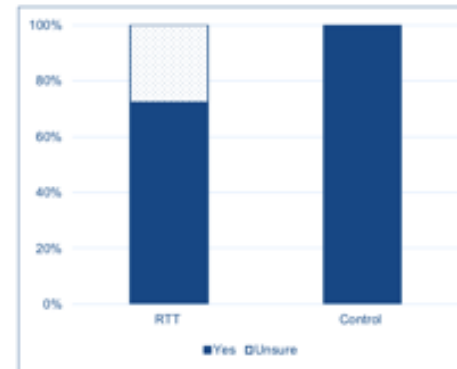
SP present



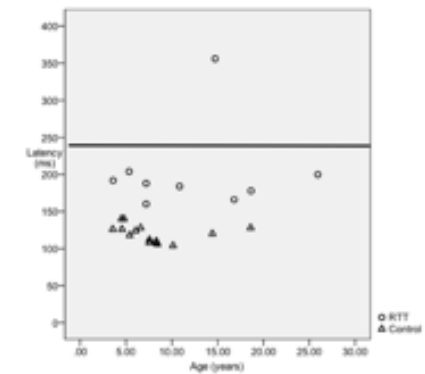
SP completely smooth



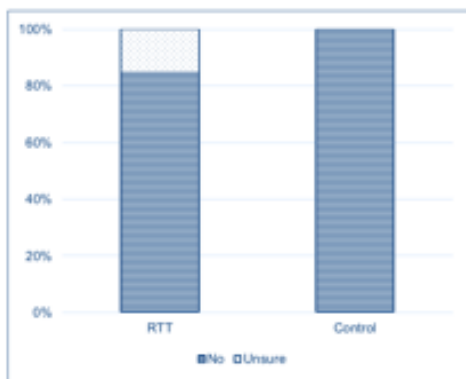
Saccades present (to R & L)



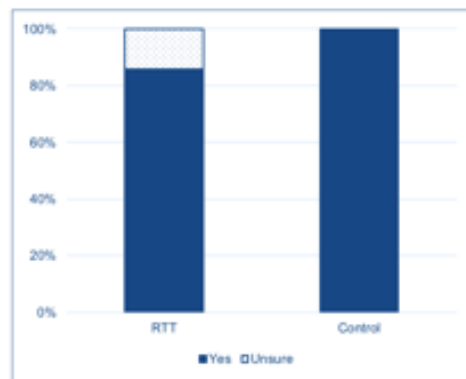
Saccades latency



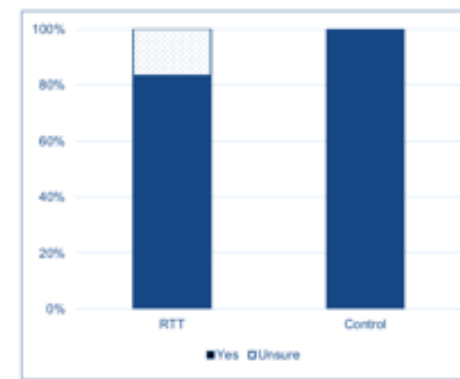
Dysmetria present



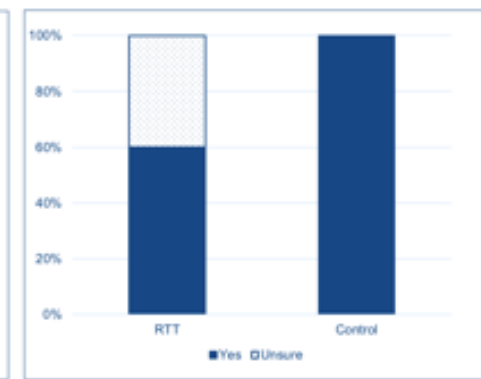
VOR present



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:

- Townend GS, van de Berg R, de Breet LHM, Hiemstra M, Wagter L, Smeets E, Widdershoven, J, Kingma H, Curfs LMG. (2018). Oculomotor function in individuals with Rett syndrome. *Pediatric Neurology*, **88**, 48-58.
- de Breet L, Townend GS, Curfs LMG, Kingma H, Smeets E, Widdershoven J, van de Berg R. (2019). Challenges in evaluating the oculomotor function in individuals with Rett syndrome using electronystagmography. *European Journal of Paediatric Neurology*, **23**(2), 262-269.

### **Editorial**

- Fabio RA. (2019). Attention measures of patients with Rett syndrome need to overcome the challenges in evaluating the oculomotor function using electronystagmography. *European Journal of Paediatric Neurology*, **23**(2), 232.

## 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)

Loffler G, Gordon GE. (2018). Cognitive function in Rett syndrome: profoundly impaired or near normal? *European Journal of Paediatric Neurology*, **22**(1), 2-3.

## 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

VR observations:

Able to calibrate device	Selects stationary targets	Selects moving targets	Reveal hidden things	Finds matching shape from range of 2	Finds matching shape for range of 4
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RL observations:

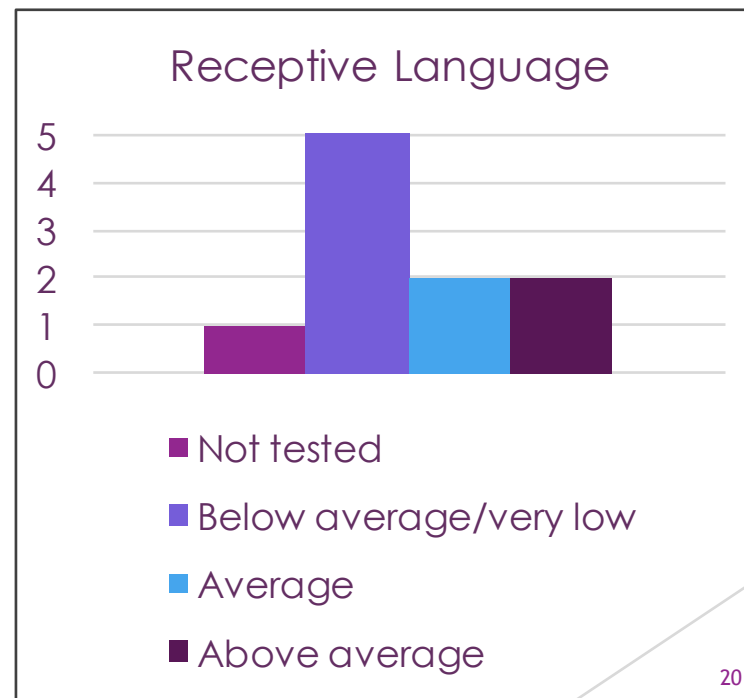
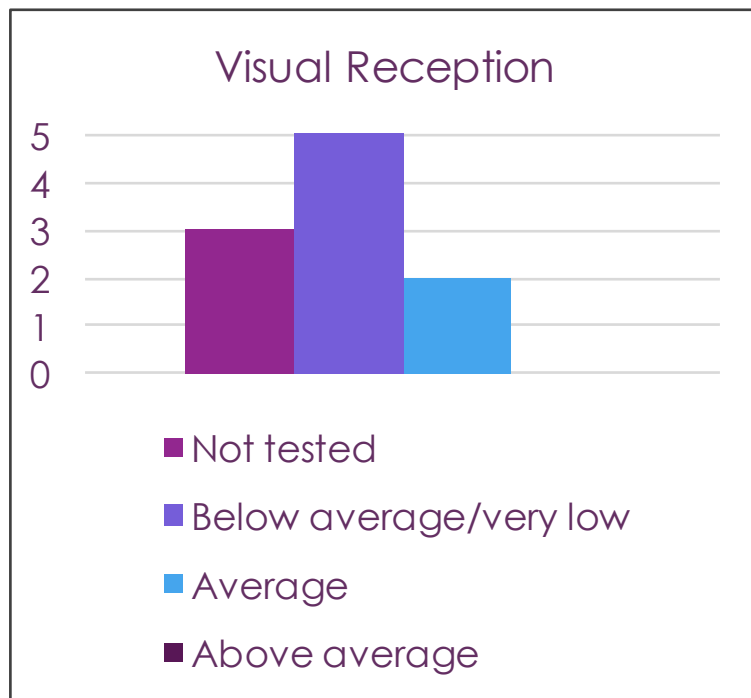
Size concepts	Identifies colours	Identifies objects	Identifies use of objects
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EL observations:

		On eye gaze device				Language functions				
Has a way to communicate yes & no	Grid size used	Uses single buttons in context	Combines two buttons in context	Combines three buttons in context	Navigates independently between pages	Expresses a choice from 2 or more items	Gives instructions	Asks questions	Answers questions	Communicates spontaneously

## Results: Formal assessment (MSEL)

Expressive Language = all “very low” e.g. laugh, happy vocalisations  
(MSEL does not recognise/was not adapted for AAC use)



# Results: Informal assessments – VR & RL

Child		Visual Reception						Receptive Language				
No.	Age (months)	Able to calibrate device	Selects stationary targets	Selects moving targets	Reveal hidden things	Finds matching shape from 2	Finds matching shape from 4	Size concepts	Identifies colours	Identifies objects	Identifies use of objects	
1	53	Y	Y	Y	Y	-	-	-	-	Y	Y	
2	68	Y	Y	Y	Y	Y	Y	-	-	-	-	
3	80	Y	Y	Y	Y	-	-	Y	Y	Y	N	
4	59	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	
5	76	Y	Y	Y	Y	-	-	Y	Y	Y	Y	
6	48	Y	Y	Y	Y	-	-	-	-	-	-	
7	69	Y	Y	Y	Y	-	-	Y	Y	Y	-	
8	52	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	
9	62	Y	Y	Y	Y	-	-	Y	Y	Y	Y	
10	49	Y	Y	Y	Y	-	-	Y	-	Y	-	

## Results: Informal assessments – EL

Child		On eye gaze device						Language functions				
No.	Age (months)	Has a way to communicate yes & no	Grid size used	Uses single buttons in context	Combines two buttons in context	Combines three buttons in context	Navigates independently between pages	Expresses a choice from 2 or more items	Gives instructions	Asks questions	Answers questions	Communicates spontaneously
1	53	I	20	Y	Y	N	N	Y	N	-	Y	Y
2	68	Y	56	Y	Y	N	Y	Y	Y	Y	Y	Y
3	80	Y	36	Y	N	N	N	Y	Y	N	N	Y
4	59	Y	56	Y	Y	N	N	Y	Y	Y	Y	Y
5	76	Y	54	Y	Y	Y	Y	Y	Y	N	Y	Y
6	48	Y	20	Y	Y	N	Y	Y	Y	-	Y	Y
7	69	Y	40	Y	Y	N	N	Y	Y	N	Y	Y
8	52	Y	20	Y	Y	Y	Y	Y	Y	-	Y	Y
9	62	Y	42	Y	Y	Y	Y	Y	Y	Y	Y	Y
10	49	Y	30	Y	Y	N	N	Y	Y	-	-	Y

## 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:

- Ward C, Chiat S, Townend GS. A Comparison of Formal and Informal Methods for Assessing Language and Cognition in Children with Rett Syndrome. *Submitted*.
- Clarkson T et al. (2017). Adapting the Mullen Scales of Learning for a standardised measure of development in children with Rett syndrome. *American Association on Intellectual and Developmental Disabilities*, **55**(6),419-431.
- Ahonniska-Assa J et al. (2018). Assessing cognitive functioning in females with Rett syndrome by eye tracking methodology. *European Journal of Paediatric Neurology*, **22**(1),39-45.

**Thank you for your attention!**

[g.townend@maastrichtuniversity.nl](mailto:g.townend@maastrichtuniversity.nl)

or

[gill.townend@rettuk.org](mailto:gill.townend@rettuk.org)