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A comparison of formal and informal methods for assessing language and cognition in children with Rett syndrome

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ABSTRACT

Background: Opinions about the cognitive and receptive language skills of people with Rett syndrome (RTT) range from severe intellectual impairment to near-normal development. Assessment is challenging because most are non-verbal, with no purposeful hand use. Clarkson et al. (2017) adapted the Mullen Scales of Early Learning for use with eye gaze technology (MSEL-A/ET) for people with RTT.
Aims: To investigate and compare the performance of children with RTT on formal and newly-designed informal assessments of language and cognition using eye gaze/tracking technology.
Methods and procedures: Ten children with RTT aged 4:0–6:8 were assessed on the MSEL-A/ET for Visual Reception (VR) and Receptive Language (RL), and standard MSEL for Expressive Language (EL). Informal assessments of the same skills were embedded in activities such as reading and cake-decorating.
Outcomes and results: Standard scores on MSEL-A/ET VR and RL subtests ranged from 'very low' to 'above average'. All children scored 'very low' on standard EL assessment. Informal assessments added information about EL, with children producing 1–3 word utterances and a range of communicative functions through an eye gaze device.
Conclusions and implications: Combining low-tech augmentative and alternative communication, eye gaze technology, informal activities and formal assessment, yields greater insight into children's abilities. This is important in informing suitable support and education for the individual.

What this paper adds

Traditional ways of assessing language and cognition rely upon motoric and/or verbal responses. Due to their loss of oral speech and purposeful hand use, and wider disturbances in motor movement and planning, people with Rett syndrome find themselves unable to respond to command; in many cases this may be (erroneously) interpreted as a sign of cognitive impairment or lack of understanding. Eye gaze, and eye tracking technology in particular, offer an access method that can facilitate delivery of both formal and informal assessment tasks, as well as opening up opportunities to engage in learning and communication. This study demonstrates how assessments can be adapted for eye gaze access, and how assessments delivered using eye gaze and eye tracking technology can reveal a

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broad range of cognitive ability within a small test sample. It also shows how combining formal assessments with informal tasks can yield higher quality information than employing formal assessments alone. Some individuals respond better on informal assessments, others on formal, and taken together a more rounded picture of an individual is created. This study, although small-scale, confirms the imperative for assessments to be adapted to meet an individual's physical and verbal constraints so that hidden cognitive abilities can be uncovered and an individual can be supported in the most appropriate and effective way to maximise their learning and communication.

1. Introduction

Rett syndrome (RTT) is a rare neurodevelopmental disorder typified by near normal development in the first 6–18 months of life followed by a loss of acquired skills. Following regression, most individuals with RTT lose mobility, purposeful hand use and oral speech. Disturbances in motor movement and planning, such as dystonia and dyspraxia (impaired control of voluntary movement, exacerbated by anxiety), affect all individuals with RTT, impacting communication and making accurate assessment challenging (Byiers & Symons, 2013; Djukic & McDermott, 2012). These difficulties have led to generalisations about intellectual and language abilities, with many (still current) scientific publications describing RTT as a leading cause of severe cognitive impairment (Berger-Sweeney, 2011; Chapleau et al., 2013; Gold, Krishnarajy, Ellaway, & Christodoulou, 2018; Parisi, Di Filippo, & Roccella, 2016). Inconsistencies in performance from day to day or moment to moment within individuals have further contributed to this view, with occasional evidence of higher level performance regarded as a 'fluke'. Such conclusions are at odds with anecdotal reports and personal stories shared on social media and websites of Rett associations across the world[2] and there is increasing evidence that cognitive ability has been under-estimated, largely due to the physical limitations that hamper assessment. The need for increased awareness of the (potential) range of cognitive ability and the impact of concomitant features of Rett syndrome on communication abilities, as well as the need to approach assessment of skills in a different way, was highlighted in the international Rett Syndrome Communication Guidelines that were published in 2020 (Townend, Bartolotta, Urbanowicz, Wandin, & Curfs, 2020; Townend, Bartolotta, Urbanowicz, Wandin, & Curfs, 2020). The need to develop more sensitive and appropriate assessment methodologies has also been recognised by other authors (Byiers & Symons, 2012; Demeter, 2000; Loffler & Gordon, 2018; Townend et al., 2017).

Assessments of language and cognition in RTT may be direct or indirect (Sigafoos, Kagohara, & van der Meer, 2011). Indirect measures utilise observational checklists and parent/carer reports. Some are used widely, with diverse populations across the world, such as the Vineland Adaptive Behavior Scales (VABS) (Dahlgren Sandberg, Ehlers, Hagberg, & Gillberg, 2000; Kaufmann, Tierney, & Rohde, 2012; Vignoli, Fabio, & La Briola, 2010) for assessing broad cognitive skills or the Inventory of Potential Communicative Acts for linguistic/communicative behaviours (Didden, Korzilius, & Smeets, 2010; Hetzroni & Rubin, 2006; Sigafoos, Woodyatt, & Keen, 2000). Others are unique, where research teams (and clinicians) devise their own questionnaires and interview schedules (Bartolotta, Zipp, Simpkins, & Glazewski, 2011; Cass, Reilly, & Owen, 2003; Neul, Lane, & Lee, 2014; Urbanowicz, Leonard, Girdler, Ciccone, & Downs, 2016). Where direct assessment of an individual's skills is required, the Rett Syndrome Communication Guidelines recommend that tests should be adapted for eye gaze (with or without eye tracking technology) in order to compensate for the verbal and motoric limitations experienced by people with RTT (Townend et al., 2020a). However, the Guidelines also recognise that formal (standardised) assessments are unlikely to fully reflect the underlying skills of someone with RTT as the direct questioning nature of the tasks is likely to increase pressure, potentially triggering or intensifying the anxiety and motor planning issues (dyspraxia) that are features of RTT, thereby diminishing accuracy of responses. In contrast, the 'non-confrontational' nature of soft-questioning that can be built into more informal style assessments is likely to yield more accurate results. Thus, a combination of assessment approaches – direct and indirect, formal and informal – is recommended by the Guidelines in order to gain a holistic picture of an individual's underlying skills and potential.

Some studies using eye gaze-based assessments have directly examined visual perception and/or language-based concepts like size and colour, while others have explored cognitive aspects such as attention and visual or social preferences (Baptista, Mercadante, Macedo, & Schwartzman, 2006; de Lima Velloso, de Araújo, & Schwartzman, 2009; Djukic, Valicenti McDermott, Mavrommatis, & Martins, 2012; Rose et al., 2013; Rose, Djukic, Jankowski, Feldman, & Rimlerd, 2016; Rose, Wass, Jankowski, Feldman, & Djukic, 2017; Vignoli et al., 2010; von Tetzchner et al., 1996). Two recent studies that have adapted formal or standardised assessments for use with children with RTT are those conducted by Ahonniska-Assa, Polack, and Saraf (2018) and Clarkson, LeBlanc, and DeGregorio (2017). In the former, the Peabody Picture Vocabulary Test (PPVT-4) was adapted for eye tracking to assess the receptive vocabulary of children with RTT aged 3:4–12:2. This included short 'practice sessions' to help participants become more familiar with using the technology. Results ranged from moderate-to-severe cognitive impairment (11/17), through mild cognitive impairment (4/17), to low average/within normal range (2/17). Clarkson et al. (2017) explored cognitive and motor development more broadly, using the Mullen Scales of Early Learning (MSEL) and VABS to assess visual reception (VR), receptive language (RL), expressive language (EL) and fine motor skills (FM) in children with RTT. The VR and RL parts of the MSEL were adapted to allow for eye gaze/eye pointing and body movement in response to object and picture materials (MSEL-A) and for eye tracking on an eye gaze-controlled computer (MSEL-ET). The EL and FM parts of the MSEL were not adapted. Participants ranged from 1:10–10:11; some were assessed on the MSEL-A and some on both the MSEL-A and MSEL-ET. Clarkson et al. found a much broader range of ability in the VR and RL domains of

[2] See for example: https://www.rettuk.org/wp-content/uploads/woocommerce_uploads/2018/08/An-Introduction-to-AAC-for-People-with-Rett-Syndrome-and-other-Complex-Communication-Needs-Email.pdf https://www.rettuk.org/resources/resources-for-families/resources-news/ https://rett.nl/de-vereniging/prettpraat/.

the MSEL than on the VABS, with the VABS showing "global deficits" (Clarkson et al., 2017, p.424) while some participants scored above average for their age on the MSEL-A/ET. Although results did not differ between the MSEL-ET and the MSEL-A, the test was completed in a shorter time on the MSEL-ET, suggesting that eye tracking technology may be a more time efficient method of delivering assessments.

1.1. Aims

Elements of both the Ahonniska-Assa and Clarkson studies were incorporated into the present study, and then extended, with the main aim being to investigate and compare the performance of children with RTT on formal and informal assessments of VR, RL and EL. Specific questions of interest included:

- What do the results of the MSEL-A/ET reveal about the cognitive and language abilities of individuals with RTT?
- Are there differences between results on the formal MSEL-A/ET and the informal assessment tasks?
- Do the results of the informal assessments add information not otherwise seen on the formal assessment?

2. Material and methods

2.1. Participants

An invitation letter and consent form were circulated by Rett UK[3] to all families whose child met the following eligibility criteria:

- Clinical diagnosis of RTT (which may/may not be confirmed genetically);
- Aged 4:0–6:11.

Once the consent form was returned, a member of the research team made contact with each family to discuss the requirements of the study further. When the family agreed to proceed, a 30-minute pre-assessment interview was conducted by telephone. This was based on the adapted MSEL-protocol. A questionnaire was also sent to families to complete in advance and return to the assessors on the day of the assessment. The interview and questionnaire together were used to elicit background information on the child, their physical and communicative skills, and their device familiarity. The information also prepared the assessors for reading the child's communicative behaviours during the assessment, for example, alerting them to the child's idiosyncratic yes/no responses and the wait time that may be required before expecting a response. The parent questionnaire that was developed for the study can be seen in Appendix A; the MSEL-protocol is available from Clarkson et al. (2017).

The final sample comprised 10 children aged 4:0–6:8, living in geographically diverse locations across the UK. The range of MECP2 mutations and selected characteristics of the participants are shown in Table 1. Table 2 presents comparative information on their communication skills and experiences of eye gaze technology prior to the study. The information in these tables was extracted from the parent questionnaire and interview.

2.2. Assessment materials

For the formal assessment, the MSEL-A/ET as adapted by Clarkson et al. (2017) was utilised. The objects used for the MSEL-A were those provided as standard with the MSEL tool kit while the picture images were copied and enlarged to facilitate eye pointing, in accordance with MSEL-A guidelines. For the MSEL-ET, images from the assessment kit were photographed and embedded into Grid 3 software from Smartbox Assistive Technology (https://thinksmartbox.com/), on a 12" I-Series eye gaze device with eye tracker (IS-4) from Tobii Dynavox (https://www.tobiidynavox.com/). Gaze Viewer™ software (Tobii Dynavox) was used to record and track the child's on-screen eye movements, with heat maps preserved for later viewing, and video cameras were positioned in front of and behind the child.

For the informal EL assessments, a range of materials was used, as listed in Section 2.3, together with the child's own eye gaze device and vocabulary system if they had one; if not, the I-Series assessment device was made available to the child and a vocabulary system selected to match any paper-based symbol system they were already using. If the child was not familiar with any symbol set, selection of a vocabulary system for the informal assessments was based on their responses during the practice session at the start of the day.

2.3. Procedure

The assessments were conducted by two members of the research team (a teacher and a speech and language therapist). They took place across a single day, in the child's own home, with one or both parents present. Breaks in assessment and/or changes in activity and positioning were allowed as often as each child required. At the start of the day, each child completed a five-point calibration and

[3] Rett UK is a national charity dedicated to supporting and empowering people with Rett syndrome and their families.

Table 1
Participant characteristics, Rett syndrome-related (as reported through the parent questionnaire).

Participant no.	Age (months)	Mutation type	Seizures	Mobility	Hand function	Spoken language	Anxiety
1	53	R270X (c.808C > T)	no	2	3	3	1
2	68	R255X (c.763C > T)	yes	4	4	3	2
3	80	T158 M (c.473C > T)	yes	4	4	4	0
4	59	S49X (c.146C > G)	yes	4	4	3	2
5	76	T158 M (c.473C > T)	no	0	4	3	3
6	48	P152R (c.455C > G)	yes	3	4	3	2
7	69	R255X (c.763C > T)	yes	4	3	2	2
8	52	SE204X (c.611_614del)	no	4	3	3	2
9	62	R306H (c.917G > A)	yes	0	1	2	3
10	49	Unknown	yes	4	3	3	1

Key Mobility: 0 = very mobile, 1 = sometimes mobile, 2 = mobile with limited support, 3 = mobile with a high level of support, 4 = not mobile at all. Hand function: 0 = always functional hand use, 1 = often functional hand use, 2 = sometimes functional hand use, 3 = occasional functional hand use, 4 = no functional hand use.
Spoken language: 0 = verbalises full sentences that make sense and are usually used appropriately, 1 = copies words and phrases heard before but not always used appropriately, 2 = verbalises a limited range of words that are sometimes used appropriately, 3 = vocalises some sounds but words are not clear, 4 = does not vocalise at all.
Anxiety: 0 = never anxious, 1= occasionally anxious, 2 = sometimes anxious, 3 = often anxious, 4 = always anxious.

Table 2
Participant characteristics, communication-related (as reported through the parent questionnaire).

Participant no.	Communicates yes/no	Makes choices (access method, no. options)	Has own eye gaze device	Length of time with own device (months)	Device availability	Modelling of language system on device	Device use	Page-set & grid size
1	2	EP from field of 2	No	N/A	N/A	N/A	N/A	N/A
2	1	PAS up to 5 choices	Yes	26[a]	1	1	1	Snap Core First 7 × 7
3	0	EP from field of 3–4	Yes	45	2	2	1	Snap Core First 6 × 6
4	1	EP from field of 3–4	Yes	26[b]	1	1	1	Snap Core First 7 × 7
5	1	PAS up to 3 choices	Yes	34	2	1	1	Super Core 50
6	2	EP from field of 2	No	N/A	N/A	N/A	N/A	N/A
7	1	PAS up to 6 choices	Yes	41	3	3	3	Symbol Talker A/B
8	1	EP from field of 3	No	N/A	N/A	N/A	N/A	N/A
9	1	EP from field of 3	Yes	4	2	1	1	Snap Core First 6 × 6
10	3	EP from field of 2	Yes	7[c]	1	2	3	Snap Core First 5 × 5

Key Yes/No: 0 = can always communicate yes/no, 1 = can usually communicate yes/no, 2 = can sometimes communicate yes/no, 3 = can occasionally communicate yes/no, 4 = can never communicate yes/no.
Choices: EP = eye pointing, PAS = partner assisted scanning.
Device availability: 0 = always available, 1 = often available, 2 = sometimes available, 3 = occasionally available, 4 = never available.
Modelling: 0 = always modelled, 1 = often modelled, 2 = sometimes modelled, 3 = occasionally modelled, 4 = never modelled.
Device use: 0 = literate and uses keyboard to type sentences, 1 = combines 2–3 symbols to form a message, 2 = makes purposeful one-word selections, 3 = makes random word selections, 4 = uses the device for games only.
[a] Used only for games for first 8 months and primarily for communication thereafter.
[b] Used only for games for first 16 months and primarily for communication thereafter.
[c] Used mainly for games rather than communication.

played games on the assessment eye gaze device during a practice session. The practice activities were selected from Look to Learn™ (Smartbox), especially those from the 'sensory', 'explore', and 'target' categories. These ice-breakers served a dual purpose, to establish rapport with the assessors and to increase familiarity with the device. This was especially important where the child had no previous experience of using an eye gaze device. Assessment tasks were then administered, with the order of activities varying per child.

2.3.1. Informal tasks

The informal tasks were organised around three themes:

1 Playing games using the eye gaze device. Activities from Look to Learn™ were used to transition the session from practice into assessment mode. Activities from the 'choose' category of Look to Learn™ were introduced as well as simple communication grids

centred around activities such as singing and blowing bubbles. Observations of skills included the ability to track static and moving targets, to search for hidden items, to make choices, and to ask for 'more', 'again'. This corresponded with the VR sections of the MSEL which includes tracking of targets, as well as providing an opportunity to observe informal EL use during the activities.

2 A book discussion. Each child was presented with a choice of picture books and asked to select one to look at on screen. This was read aloud by either the assessor or the computer and paused at regular intervals to allow for discussion with the child. The child's eye movements and on-screen selections were tracked, for example, to determine whether they looked at an animal as it was being talked about. Informal written observations were made in each of the assessment areas (VR, RL and EL).

3 Cake decorating activity. Each child was encouraged to instruct/direct the assessor to decorate cupcakes. By employing 'soft' questioning, VR and RL skills were assessed. For example, as an RL task correlating with the size-finding task in the RL MSEL-A, big and small chocolate buttons were placed in front of the child and the assessor said "Mum would like the big chocolate button on her cake, I wonder if you can help me find it?". As a VR task, a range of differently-shaped decorations were placed in front of the child while the assessor said "Mum really wants a shape like this one on her cake, can you help me find it?". The items and questions were adjusted to reflect differing levels of complexity across both VR and RL domains. Participants had access to both core and topic-based pages on the eye gaze device and EL was assessed through observation, with prompted and spontaneous language production noted by the assessors.

2.3.2. Formal tasks

The formal tasks were organised according to the three selected MSEL categories:

1 MSEL-A/ET to evaluate VR.
2 MSEL-A/ET to evaluate RL.
3 Standard MSEL to evaluate EL skills.

Children were given the opportunity to use either the MSEL-A or MSEL-ET or a combination of both for the VR and RL tests, as the results of the Clarkson study showed no loss of integrity by using one or other adaptation. These were administered according to the Clarkson protocol, with the assessors keeping a record of the adaptions.

2.4. Data recording and analysis

2.4.1. Informal tasks

An observation record sheet was developed for contemporaneous note-taking during the informal assessments. This can be seen in Appendix B. Each child was required to give a correct response two or more times to be credited with a skill. For example, if a selection of tubes of food colouring was presented when making icing for cupcakes, the child had to find a minimum of two named colours to decorate cakes for different members of the family. The nature of the assessment allowed for the child's own interests to be followed e.g. reading, baking, singing, and consequently there was not the opportunity to assess all elements for every participant.

For a positive score on the informal EL assessment, the child had to demonstrate a skill more than twice, with the words used in an appropriate context.

2.4.2. Formal tasks

Participants' responses to the VR and RL tasks on the MSEL-A/ET were recorded at the time of assessment using the adapted MSEL scoring sheet and the adapted scoring procedures. To achieve a baseline value, the child had to complete three tasks correctly in a row; they were judged to have reached a ceiling when they failed, or completed incorrectly, three consecutive tasks.

For the EL section of the MSEL, the scoring sheet was completed through combined parental report and assessor observation as it was not possible to test speech skills directly.

For those aged up to 5:10 (the upper age band of the MSEL), the raw score could be used to derive an age-related score (in months), a T-score, percentile rank, and descriptive score. For older children, no T-score could be calculated and a descriptive category of 'very low' was assigned as their biological age exceeded the highest age for which the MSEL was designed.

At the end of the assessment period, qualitative analysis of results focused on the questions outlined in Section 1.1.

Table 3
Assessor agreement on raw scores when scored independently.

Participant no.	Visual Reception		Receptive Language	
	Assessor 1	Assessor 2	Assessor 1	Assessor 2
1	31	31	29	29
2	42	42	35	35
3	47	47	n/a	n/a
5	n/a	n/a	32	32
7	45	45	43	43
8	40	40	39	39

Note: occasional differences of opinion between assessors as to whether a response was equivocal or unequivocal are not reflected in the final scores.

2.5. Reliability

For all informal assessment tasks, the assessors completed the observation record sheets independently. They also scored performance on the MSEL-A/ET RL and VR tasks independently for six of the 10 children, with the remainder scored jointly. Video playback was used after the event to confirm any responses that the assessors found to be unclear at the time of assessment. However, occasional differences of opinion as to whether a response was equivocal or unequivocal were not reflected when the final scores were compared between assessors. According to the adapted MSEL protocol two equivocal responses were judged to equate to one unequivocal; in all cases, this led to the same final score being awarded by the assessors when the points assigned to the individual tasks were totalled. There were no instances of wider disagreement between the assessors. This is reflected in Table 3, which shows a 100 % inter-rater agreement rate for the assessors' scores for the participants who were rated independently on the MSEL-A/ET RL and VR.

2.6. Ethical approval

Ethical approval for the study was granted by the City, University of London School of Health Sciences Research Ethics Committee.

3. Results

As seen in Tables 1 and 2, the participants exemplified a range of MECP2 mutations and levels of severity in respect of the core characteristics of RTT and concomitant features that impact communication, that is, in their levels of spoken language, hand use, mobility, seizures and anxiety. Their experience of using eye gaze technology for communication prior to the assessment session was also very mixed. Seven participants had their own device, three did not. Two or three had mainly used their devices for games, at least in the early months, before starting to use it as a communication tool; others had access to their devices for communication from the moment of provision. Devices were made available for varying amounts of the day and across a range of settings, including home, school, and 'out and about'. There were varying vocabulary systems, with a range of grid sizes and page types, including core and fringe vocabulary pages, and topic and phrase-based pages. Two participants also had a robust low-tech paper-based communication system.

3.1. Performance on MSEL VR, RL and EL subscales

Table 4 presents the data associated with each child's performance on the VR and RL subscales of the adapted MSEL. This shows a broad range of results. Not all children were tested on every variable, depending on their levels of fatigue, willingness to engage and time taken to complete the tasks. Considering the subscales separately, three children were 'very low', two 'below average' and two 'average' on the VR subscale, while on the RL subscale, four were rated 'very low', one 'below average', two 'average' and two 'above average' when compared with the neurotypical population. Only one child of the seven tested on the VR tasks reached a ceiling; four became too tired or opted out of completing this section before they reached a ceiling and two reached the end of the test before they reached a ceiling. None of the nine tested on RL reached a ceiling, three because they reached the end of the test first, the remainder due to tiredness/opting out. One child, for example, used their communication device to say "I never want to see this again".

Table 5 presents the EL scores according to the standard MSEL ratings. This shows that all children received a rating of 'very low', scoring as able to laugh, smile and make happy vocalisations. Five out of 10 could produce 2–7 words verbally (still classifying as 'very low') but there were no skills recorded on the formal assessment beyond this. Communication via an eye gaze device could not be scored as part of this formal assessment as there was no provision for including the use of any augmentative and alternative communication (AAC) system (e.g. the use of paper-based symbol communication boards or an eye gaze device) in either the standard or adapted MSEL.

3.2. Performance on informal VR, RL and EL assessment tasks

Table 6 illustrates each of the skills observed during the informal VR and RL assessment tasks and Table 7 shows the children's EL skills as recorded on informal assessment. During the informal VR-related tasks, all of the children were able to calibrate the eye gaze device, track stationary and moving targets, and focus on shapes to reveal hidden targets. Three had the opportunity to find matching shapes from a choice of two or four and achieved this, while all of those who were presented with informal RL tasks were able to identify objects, colours and sizes, and five out of the six presented with the task were able to identify objects by use.

All participants were able to use a 20-button (or greater) grid size to demonstrate a range of communicative functions, to create one-word utterances in context e.g. 'more', 'stop' 'good', as well as to make choices and communicate spontaneously (not just on demand). Nine out of 10 were able to create two-word utterances with meaning while three children used three-word utterances in context. Five needed support to navigate between pages to find relevant vocabulary for a situation and five were able to navigate independently to find the vocabulary required across a range of pages. All children were also able to communicate 'yes' and 'no' through a range of methods, both on and off the device.

3.3. Performance across formal and informal assessments

In order to evaluate across assessment methods, analogous tasks were compared to determine whether the informal presentation of

Table 4
Performance of each child on adapted MSEL Visual Reception and Receptive Language tasks.

Participant		Visual Reception							Receptive Language						
No.	Age (months)	Raw score	Age-equivalent score (months)	T-score	Percentile rank	Descriptive category	Ceiling reached	Access method	Raw score	Age-equivalent score (months)	T-score	Percentile rank	Descriptive category	Ceiling reached	Access method
1	**53**	31	30	20	1	Very low	Y	ET EP	29	31	20	1	Very low	N	ET EP
2	**68**	42	46	20	1	Very low	N	ET EP	35	41	20	1	Very low	N	ET EP
3	**80**	47	57	–	–	Very low	E	ET EP	n/t	n/t	n/t	n/t	n/t	n/t	n/t
4	**59**	49	66	56	73	Average	E	ET EP	47	65	58	79	Above average	E	ET EP
5	**76**	n/t	n/t	n/t	n/t	n/t	n/t	n/t	32	36	–	–	Very low	N	ET EP BM
6	**48**	n/t	n/t	n/t	n/t	n/t	n/t	n/t	46	62	69	97	Above average	E	ET EP
7	**69**	45	57	31	3	Below average	N	EP	43	55	32	4	Below average	N	EP
8	**52**	40	43	35	7	Below Average	N	ET EP	39	47	43	24	Average	E	ET EP
9	**62**	n/t	n/t	n/t	n/t	n/t	n/t	ET EP BM	32	36	20	1	Very low	N	ET EP
10	**49**	41	45	41	18	Average	N	ET EP	36	42	40	16	Average	N	EP

Key ET = eye tracking; EP = eye pointing; BM = body movement; n/t = not tested; – = tested but cannot be calculated as child over 70 months of age (upper age limit of test); Y = ceiling reached of 3 consecutive incorrect responses; N = ceiling not reached as child opted out/became too fatigued/needed change of activity; E = end of test was reached before ceiling was reached.

Table 5
Performance of each child on MSEL Expressive Language tasks.

Participant no.	Age (months)	Raw score	Age-equivalent score (months)	Descriptive category
1	53	12	12	Very low
2	68	5	4	Very low
3	80	5	4	Very low
4	59	12	12	Very low
5	76	9	12	Very low
6	48	9	12	Very low
7	69	12	12	Very low
8	52	6	5	Very low
9	62	12	12	Very low
10	49	4	3	Very low

tasks offered more, less or the same information about an individual's ability as the formal assessment. Of the seven participants who completed both the formal and informal VR tasks, five showed lower results on the informal part of the assessment while two demonstrated the same skill set in both the formal and informal tasks. Of the seven participants who completed both the formal and informal RL tasks, two showed higher results on informal assessment, demonstrating a greater range of skills than on the formal assessment, two showed the same results in both assessments, and three demonstrated fewer skills in the informal assessment than in the formal assessment. For the EL assessments, all children demonstrated a greater ability to communicate expressively in the informal than formal assessments.

4. Discussion

This study of a small sample extended the work of Clarkson et al. (2017) by using the MSEL adapted for eye gaze/eye pointing and eye tracking technology, and supplementing this with informal assessments that also utilised eye gaze and eye tracking technology. Inclusion of play-based activities, both as practice on the device at the start of the day and as informal assessment tasks, was inspired by Ahonniska-Assa et al. (2018). The broad range of responses across all tasks supported the findings of both of these studies, that children with RTT do demonstrate a range of cognitive abilities. While 5/7 of the sample who were assessed for VR skills and 5/9 of those assessed for RL skills had scores that were 'below average' or 'very low', 2/7 of those assessed for VR and 4/9 of those assessed for RL were rated 'average' or 'above average' compared with their neurotypical peers, according to the MSEL scoring scale. In addition, all children were able to achieve a baseline assessment score that equated to 29 months of age or above, which indicates that even those rated as 'very low' have an ability to learn. This is a higher developmental age than the 1–18 months attributed to individuals with RTT in some of the earlier publications exploring receptive language and cognition (Byiers & Symons, 2012; Demeter, 2000). It should also be remembered that two of the children who were judged to be 'very low' were above the upper age limit for the MSEL and, as no T score could be calculated, were automatically assigned a rating of 'very low' which may not be reflective of actual ability. It is also potentially significant that only one child reached a 'true' ceiling, according to the formal MSEL scoring criteria, and that was on one set of tasks only. In all other instances, children reached the end of the assessment before reaching a ceiling or were too tired/opted out before reaching a ceiling, meaning that their 'true' upper limits were unexplored.

Comparison of performance across the informal and formal assessments yielded mixed results, with individuals demonstrating a variety of response and task preferences. Contrary to expectations, some of the children demonstrated a higher level of skill on the formal MSEL-A/ET VR and/or RL tasks, some showed equivalent skills across both sets of tasks, and some performed better on the informal tasks. For two children in particular, there was a wide discrepancy in skills shown on the formal and informal assessments. Both children declined to participate in the MSEL-A/ET VR tasks and their results on the MSEL-A/ET RL were 'very low'. For example, they were unable to demonstrate skills such as identifying colours and sizes. In contrast, however, they accurately and repeatedly demonstrated these skills when asked in the informal cake decorating activity. This conforms with Ahonniska-Assa's findings that some children "showed relatively good eye-gaze control in play sessions but little interest in the PPVT-4 tasks, resulting in arbitrary choices" (Ahonniska-Assa et al., 2018, p.42) and supports the view that, in some areas and for some individuals, informal assessment can teach us more than formal assessment. Yet, conversely, the findings of the present study also suggest that for others the opposite is true. This may be due in part to the challenge of presenting some of the more complex tasks in an informal manner, some questions just 'need to be asked' and cannot be hidden in fun activities, meaning that informal assessments may not always facilitate higher level tasks. Furthermore, it is noteworthy that some children liked 'performing' when tested formally and enjoyed 'showing off' their skills. Although not typically reported within the RTT community, the possibility that some individuals are able to perform to maximum effect on demand should also be recognised. This suggests that the testing of any skill or ability should always take into account, and be adapted to, individual needs, preferences and response styles, just as individualised approaches to testing and teaching, and recognition of individual learning styles, are to be recommended in the wider, neurotypical population.

It should be borne in mind that there was no adequate way in which to assess and score EL on formal assessment. Self-expression through use of any form of AAC could only be recorded in informal tasks. Clarkson et al. (2017) did not adapt the EL part of the MSEL for good reason, and just how formal assessments can be adapted in this area remains a question for further investigation. Should symbol communication boards or pages on an eye gaze device be created specifically with vocabulary relevant for a particular assessment, or are words then being put into an individual's mouth? Can an assessor really conclude the word was in the individual's

Table 6
Visual Reception and Receptive Language skills observed during Informal Assessment tasks.

Participant		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	Identifies 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	–

Key Y = skill demonstrated; N = given opportunity but skill not demonstrated; – = no opportunity to demonstrate skill.

Table 7
Expressive Language skills observed during Informal Assessment tasks.

Participant		Communicates yes/no	On eye gaze device					Language functions				
No.	Age (months)		Grid size	Uses single buttons in context[a]	Combines two buttons in context[b]	Combines three buttons in context[c]	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

Key Y = skill demonstrated reliably; I = skill demonstrated with some inconsistency; N = given opportunity but skill not demonstrated; – = no opportunity to demonstrate skill.

[a] This may include one-word utterances and/or pre-set phrases on a single button.

[b] This may include combining two single words to create a novel utterance ('more cake', 'you stir') or combining a single word with a pre-set phrase ('Can I have some more?'+ 'icing').

[c] This may include combining three single words to create a novel utterance ('big' + 'white + 'star', 'you' + 'read' + 'book') or combining single words with pre-set phrases ('Can I have a turn?' + 'stir'+ 'want').

lexicon before it was offered on a page placed in front of them? Alternatively, should an individual be presented with a robust vocabulary, that can be accessed through an eye gaze device or by Partner Assisted Scanning[4], in order to see whether they can then find the relevant words from amongst a very large number that may be spread across topics and multiple pages. Is that really testing their lexicon or is it testing their familiarity with the particular page-set, and their memory and navigation skills if they do know the system in question, or are they being presented with an impossible task and set up to fail if they have never seen or used the system before? It is certainly an area of investigation fraught with difficulty. In contrast, however, informal assessments employed in the current study, adapted to the moment and to the child and using varied vocabulary-based systems, revealed a range of expressive language functions and content amongst the participants, all of whom scored ’very low’ on the formal assessment, confirming that their level of ability would have been seriously under-estimated if relying on formal assessment results alone.

5. Limitations and suggestions for future research

Although RTT is defined by a small set of core clinical criteria, it is characterised by a wide range of symptoms and comorbidities, all of which can vary in severity between individuals, resulting in a highly heterogeneous group. The small sample size could have skewed the study had it failed to provide a wide enough representation of the selected age group. Fortunately, this was not the case, as can be seen by the variability in participant characteristics. Across the sample, both more and less common MECP2 mutations were represented, and there was variation in levels of mobility, hand function, anxiety, seizures and spoken language. There was also variation between participants’ levels of exposure to and/or use of an eye gaze device prior to the study. Thus, even with a small sample size there was no apparent bias towards inclusion of participants only at the severe or conversely the mild end of the spectrum that is RTT. As a small-scale pilot, this study paves the way for future research with a larger cohort.

The upper age limit associated with the standardised scoring mechanism of the MSEL could be seen as a limitation, both of the study and of the applicability of the MSEL-A/ET. Based on the use of this specific assessment, its target age range, and the results obtained, we can only say with certainty that a range of cognitive and language abilities were shown in children with RTT under the age of seven years. Further research needs to be undertaken with older groups to ascertain whether this spread continues into adulthood. In order to do this, however, it would first be advisable to validate the use of the MSEL with older populations, both with neurotypical individuals and those with conditions other than RTT, or to select a different assessment tool already validated for older age groups.

A further limitation is that the informal activities employed in the study were not empirically validated. Nor was there a rigid protocol that set out a uniform set of tasks and activities with tightly prescribed steps for their delivery. Rather, the assessors had a range of activities to hand that were subjectively judged to be testing equivalent concepts to those covered by the MSEL. The exact selection of activities, supportive strategies, and any individual adaptations with which they were offered, was left open so that the assessors could be led by the child, their interests and motivation, levels of alertness, comfort and fatigue. This did not mean that the child was given extra clues or provided with the ‘answer’; the assessors still required the child to demonstrate the skills targeted without help (unlike dynamic assessment where the child receives progressive cuing). This also suggests an area for further research, to validate a set of tasks and activities and a protocol for delivery that allows for an acceptable degree of freedom in a more informal style of assessment.

Finally, the fact that each assessment session took place over the course of one day can be seen as both a pro and a con of the study. On the one hand, responses to the different assessment styles may have been influenced by order of presentation, with the impact varying per child and the order effect contaminating the impact of the assessment style. This was an effect the assessors tried to minimise by randomising the order between individuals. On the other hand, however, the length of time spent in each child’s home was a luxury that allowed the assessors to be guided by the child’s needs, to take breaks as required, and afforded a more relaxed atmosphere conducive to putting the child at their ease within a familiar environment. It also meant that the results of the assessment were not influenced by variations in the child’s health, for example, as might have been evidenced had the sessions been spread over multiple sessions on different days. Future research could investigate further the impact of the assessment environment, and length and spread of sessions.

6. Conclusion

Eye gaze, and eye tracking technology in particular, offers an access method that can facilitate delivery of both formal and informal assessment tasks, as well as opening up opportunities to engage in learning and communication. Where children have heterogeneous abilities and major physical constraints, standardised assessment is necessarily and understandably problematic, particularly for establishing EL performance when children have little or no oral speech. However, different children responded to different styles of assessment, suggesting that we cannot make generalisations about optimal methods for assessing all children with RTT. Instead, researchers must employ a range of assessment methods to identify as accurately as possible each individual’s knowledge and potential, and use this information to provide opportunities for the person with RTT to learn at a level and pace that is correct for that individual, so that those who have a higher ability are not limited in their learning, and those who have learning difficulties are not set up to fail.

[4] An AAC strategy whereby a communication partner reads out a list of items for the individual to select from using a yes/no response.

Declaration of Competing Interest

Callie Ward has a family member with Rett syndrome. The authors have no other conflicts of interest to declare.

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Appendix A. Parent Questionnaire

Child's mutation type is known: ………………………………………..

Please rate each question about your child by circling the relevant box. If you do not understand a question or are not able to answer, please leave it blank and use the box below to explain why you were not able to answer it. Please use the box to write any additional notes you would like to add for any of the questions.

Language and Communication

Please rate your child's ability to communicate yes and no:

Can always communicate yes and no	Can usually communicate yes and no	Can sometimes communicate yes and no	Can occasionally communicate yes and no	Can never communicate yes and no

Any further information:

Please rate the level of modelling of your child's language system throughout the day:

Always modelled	Often modelled	Sometimes modelled	Occasionally modelled	Never modelled

Any further information:

If your child has access to a high-tech communication device, please rate the level of time it is available throughout the day:

Always available	Often available	Sometimes available	Occasionally available	Never available

Any further information:

If your child has a high-tech device, please circle what they can do on the device:

They are literate and can use the keyboard to type sentences.	They can put 2-3 world symbol selections together to form a message.	They can make purposeful one-word selections.	They make random word selections.	They use the device for games.

Any further information:

Please rate your child's verbal ability:

They verbalise full sentences that make sense and are usually used appropriately.	They copy words and phrases they have heard before but do not always use them appropriately.	They verbalise a limited range of words and sometimes use them appropriately.	They vocalise some sounds but it is not clear what words they are saying.	They do not vocalise at all.

Any further information:

<u>Background information</u>

Please rate your child's level of anxiety:

Always anxious	Often anxious	Sometimes anxious	Occasionally anxious	Never anxious

Any further information:

Please rate your child's mobility:

Very mobile	Sometimes mobile	Mobile with limited support	Mobile with a high level of support	Not mobile at all

Any further information:

Please rate your child's hand use:

Always functional hand use	Often functional hand use	Sometimes functional hand use	Occasionally functional hand use	No functional hand use

Any further information:

Please rate the level of literacy input your child has had:

Teaching to read sentences.	Teaching to read words.	Formal phonics teaching.	Exposure to some letters.	No literacy teaching.

Any further information:

Please rate your child's reading ability:

They can read books with longer words and sentences e.g. Kip went to the park in the rain.	They can read books with short word sentences e.g. Kip met a bat	They can read some simple CVC words e.g. cat, dog	They are engaged and enjoy looking at books	They cannot read

Any further information:

Appendix B. Informal Assessment, Observation Record Sheet

Note – informal observation sheets were intentionally brief, allowing space for assessors to write notes and list examples of skills observed during the session.

Visual Reception

Ability to calibrate?
Track using games? e.g. stationary targets, moving targets
Look for things using games? e.g. animals hidden behind farm doors
Matching? - Shapes, colours, sizes - most likely in baking activity
From 2?
From 4?

Receptive language

Looks to correct colours?
Looks to correct sizes?
Look to relevant objects? e.g. bowl when talking about needing it for baking
Identifies use of objects?

Expressive language

Communicates yes/no?
Makes choices?
1-word messages?
2-word messages?
3-word messages?

Answers direct questions? e.g. What do you think? How might the character in the book be feeling?
Asks questions?
Spontaneously communicates?
Independently navigates?
Gives instructions?

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