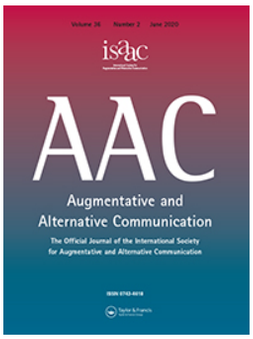
Development of consensus-based guidelines for managing communication of individuals with Rett syndrome



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RESEARCH ARTICLE

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Development of consensus-based guidelines for managing communication of individuals with Rett syndrome

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ABSTRACT

Difficulties with communication have a profound impact on the lives of individuals with Rett syndrome and their caregivers. Globally, many families report difficulty accessing appropriate and timely information and services from professionals with expertise in augmentative and alternative communication (AAC) as it pertains to Rett syndrome. To address this need, international consensus-based guidelines for managing the communication of individuals with Rett syndrome were developed by combining available evidence and lived experience with expert opinion. A two-phase Delphi survey was built on statements and recommendations extracted from a review of over 300 pieces of literature combined with survey responses from communication professionals and caregivers. All statements that reached a pre-determined threshold of ≥70% agreement were incorporated into guidelines that consist of 268 statements and recommendations relating to (a) rights of the individual; (b) beliefs and attitudes of communication partners; (c) professional knowledge and team work; (d) strategies to optimize engagement; (e) assessment; and (f) intervention (targets and goals, techniques), including the use of AAC. To date, this project is the largest of its kind, with 650 participants from 43 countries contributing to development of consensus-based guidelines for Rett syndrome.

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KEYWORDS
Augmentative and alternative communication; clinical practice guidelines; consensus-based guidelines; Delphi survey; expert consensus; Rett syndrome

Rett syndrome is a neurological disorder known to occur in approximately 1 in 10,000 females (Fehr et al., 2011; Laurvick et al., 2006) and more rarely in males (Reichow, George-Puskar, Lutz, Smith, & Volkmar, 2015). Core diagnostic criteria include the loss of spoken language and functional hand use (Neul et al., 2010). Individuals with Rett syndrome experience life-long difficulties with communication; most are non-speaking and many use some form of augmentative and alternative communication (AAC) that is often accessed by eye gaze (Bartolotta, Zipp, Simpkins, & Glazewski, 2011; Townend et al., 2017; Urbanowicz, Leonard, Girdler, Ciccone, & Downs, 2016). There is wide variation within and among countries in relation to knowledge and experience of Rett syndrome as well as health, social care, and education policies, practices, and funding. This has led to discrepancies and inequity in communication services and supports offered (Townend et al., 2015). Many families struggle to find speech-language pathologists (SLPs) and other communication professionals who are experienced in Rett syndrome and to access the advice, equipment, and technological support required to develop their child's communication (Townend et al., 2016; Wandin, Lindberg, & Sonnander, 2015). For the purposes of this paper, a *communication professional* is defined as any professional with a key role in the development and support of communication skills. In addition to SLPs, professionals may include occupational therapists (who often specialize in assistive technology and/or AAC), music therapists, and education or special education staff. Many of these professionals are challenged to find the information and support they require to build their expertise in this area (Townend et al., 2016; Wandin et al., 2015). This paper describes a project that seeks to meet some of those needs.

Rett syndrome is diagnosed according to a set of clinical criteria (Neul et al., 2010); this diagnosis may be strengthened by identification of a mutation in the methyl-CPG-binding protein 2 (*MECP2*) gene (Amir et al., 1999). The first clinical signs are commonly reported as emerging between 6 and 18 months of age, when parents begin to notice a slowing down in attainment of developmental milestones, followed by a loss or regression in previously acquired skills. Some individuals may be able to speak a small core of single

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Supplemental data for this article can be accessed here.

words or phrases prior to regression and retain or add to them post-regression, while others may never develop spoken language (Renieri et al., 2009; Urbanowicz, Downs, Girdler, Ciccone, & Leonard, 2015). Nonetheless, communication skills can continue to develop throughout life (Schönewolf-Greulich, Stahlhut, Larsen, Syhler, & Bisgaard, 2017). Studies that describe communication skills across the lifespan of individuals with Rett syndrome report the use of multiple modalities to express a range of communicative functions with apparent intent (Bartolotta et al., 2011; Didden et al., 2010; Urbanowicz, Leonard, et al., 2016). Eye gaze is the most commonly reported modality, followed by body movements and the use of AAC systems such as non-electronic communication boards (Bartolotta et al., 2011; Didden et al., 2010; Neul et al., 2014; Urbanowicz, Downs, Girdler, Ciccone, & Leonard, 2016). Motor movement and planning difficulties affect the use of gestures as well as the timing and consistency of communication and the accurate assessment of cognition (Byiers & Symons, 2012, 2013; Djukic & McDermott, 2012). Communication is further impacted by co-existing features such as seizures, breathing irregularities, heightened anxiety, fatigue, sleep disturbances, difficulties with sensory regulation, gastro-intestinal problems, and scoliosis (Neul et al., 2010).

In recent years, intervention goals for individuals with Rett syndrome have expanded from establishing basic communication skills (simple choice-making and requesting) to facilitating a wider range of communicative behaviors and developing reading and writing skills (Byiers, Dimian, & Symons, 2014; Sigafoos et al., 2009; Simacek, Reichle, & McComas, 2016; Stasolla et al., 2014, 2015; Stasolla & Caffò, 2013; Wandin et al., 2015).

Caregivers and professionals require training and support to communicate effectively with individuals with Rett syndrome (Bartolotta & Remshifski, 2013; Townend et al., 2016; Wandin et al., 2015). For the purposes of this paper, *caregiver* is an umbrella term that includes parents and others who may have a parental or legal guardian role, including grandparents, adult siblings and paid caregivers (e.g., in a residential care setting). A lack of clarity about best practices has been evident in the sharing of information during country update sessions at international Rett syndrome conferences. During the 3rd European Rett Syndrome Conference held in Maastricht in 2013, for example, representatives from national Rett syndrome associations called for more evidence-based knowledge to guide clinical practice and the provision of more equitable services (Townend et al., 2015).

Clinical practice guidelines can help establish a universal baseline of awareness and knowledge, improve quality and effectiveness of care, decrease variation in clinical practices, and promote consistency of management independent of location or level of expertise (Kredo et al., 2016; Mei, Anderson, Waugh, Cahill, & Morgan, 2018; Woolf, Grol, Hutchinson, Eccles, & Grimshaw, 1999). This is especially important for rare diseases where expertise is scarce (Sejersen et al., 2014). Clinical practice guidelines are "systematically developed statements to assist practitioner and patient decisions about appropriate health care for specific clinical circumstances" (Institute of Medicine Committee to Advise the Public Health Service on Clinical Practice Guidelines, 1990, p. 38). Traditionally, an evidence-based approach is utilized to develop guidelines (Guyatt et al., 2008; Scottish Intercollegiate Guidelines Network, 2015; Woolf, Schunemann, Eccles, Grimshaw, & Shekelle, 2012). With a rare disease like Rett syndrome, however, meta-analyses, randomized clinical trials, and rigorous systematic reviews that would normally underpin such an approach are lacking (Alonso-Coello et al., 2010). Identifying evidence-based assessment and intervention strategies is challenging, given the limitations in available literature and the low incidence of the disorder.

In the absence of high-quality evidence, consensus-based guidelines may fill the void. In this approach, recommendations are based on "current best practice and available evidence" (Mei et al., 2018, p. 329) that can be obtained via use of a Delphi survey (of at least two rounds), completed by a panel of experts in the field (Waggoner, Carline, & Durning, 2016). During each round, panel members rate statements or recommendations and provide comments that are analyzed and integrated into the next round of questions. The aim is to reach a consensus on the responses. The Delphi method has been used to develop consensus-based guidelines for a range of clinical conditions (e.g., Mei et al., 2018; Tuffrey-Wijne et al., 2016) as well as other aspects of Rett syndrome, including scoliosis (Downs et al., 2009), growth and nutrition (Leonard et al., 2013), and bone health (Jefferson et al., 2016).

In light of the clear need for guidance for caregivers and communication professionals wanting to support the communication development of individuals with Rett syndrome, the aim of this project was to create consensus-based guidelines for managing communication of individuals with Rett syndrome. Specific objectives were to (a) identify and promote awareness of the communication challenges and needs of this population; (b) identify and share best practices for communication assessment and intervention; (c) provide information that families can use as a starting point for discussion with professionals, regardless of their location or the age of the individuals they serve; and (d) provide information that professionals can use to inform and guide their work with this population.

Method

Research design

The project comprised three broad phases: (a) a literature review, (b) the development and use of two surveys to obtain input from caregivers and communication professionals, and (c) the development and use of a Delphi survey to determine consensus from an expert panel. Ethical approval was granted by the Ethical Review Committee Psychology and Neuroscience (ERCPN), Maastricht University, The Netherlands. Specific approval for conducting a focus group and pilot surveys in the United States was granted by the Institutional Review Board at Stockton University, New Jersey.

Phase 1: Literature review

The purpose of the literature review was two-fold: (a) to gather research-based information that could contribute to the formulation of draft statements and recommendations for the guidelines, and (b) to identify gaps in the literature to inform development of the caregiver and professional surveys in the next phase of the project.

Search methods

Database searches were conducted by the first author in April 2016 and repeated in September 2016, April and September 2017, and again in April 2018 to capture any newly-published articles. The following databases were searched without a limit in terms of publication date: Cumulative Index of Nursing and Allied Health Literatures, Educational Resources Information Clearinghouse, Medline via PubMed, PsychArticles, and PsychInfo. The basic scope of the search can be seen in Table 1; the full search strategy can be requested from the authors. Any systematic reviews were separated out and potentially relevant studies in the review papers were traced back to their original source (i.e., ancestry search). To supplement information on communication assessment and intervention in Rett syndrome yielded by the peer-reviewed literature, online internet searches were also conducted to identify gray literature such as books, book chapters, non-peer-reviewed papers, policy documents produced by professional bodies (e.g., American Speech-Language-Hearing Association, Royal College of Speech and Language Therapists, Speech Pathology Australia), and publications from Rett syndrome organizations and associations. In addition, broader AAC literature was also consulted.

Inclusion criteria

Peer-reviewed literature was included if it referred to any aspect of communication and/or cognition and/or literacy and individuals with Rett syndrome. Because high-quality evidence was not anticipated, literature of any level of evidence was accepted. Gray literature was included if it referred to communication and/or cognition and/or literacy in relation to individuals with Rett syndrome or to use of AAC with other developmental disability groups. AAC literature was included if it described strategies and techniques that had been identified in the gray literature as being used with at least some individuals with Rett syndrome or with individuals with limited or no spoken language.

The first and third authors independently filtered the peer-reviewed titles and abstracts according to the predefined inclusion criteria. Full-text articles were obtained for all eligible articles and divided among the research team for analysis. Inter-rater agreement on inclusion was checked and any areas of uncertainty or dispute were resolved through discussion. Overall, 222 peer-reviewed papers met the inclusion criteria in that they referred to communication and/or cognition and/or literacy in individuals with Rett Syndrome. All of the authors worked together to identify an additional 53 items of gray literature that met the broader inclusion criteria of referring to communication and/or cognition and/or literacy and Rett syndrome and/or use of AAC with other developmental disability groups. Finally, the second and fourth authors conducted the search for extra AAC literature that described in greater detail the strategies and techniques revealed by the gray literature searches as being used with at least some individuals with Rett syndrome or other individuals with limited or no spoken language. Through this process an additional 37 peer-reviewed papers were added, making a total of 312 documents for review.

Data extraction

Data were extracted from all of the documents ($n = 312$) and compiled into three spreadsheets. The first was for peer-reviewed papers that provided background information (focusing on the development of communication and/or more general descriptions of communication skills of individuals) ($n = 89$), the second for peer-reviewed papers reporting on empirical assessment and intervention studies ($n = 170$), and the third for the gray literature documents ($n = 53$). Extracted data for the peer-reviewed papers included (a) study identifiers such as keywords, title, author, location, and year, (b) aims, (c) design and conduct of study, (d) participants and cohort size, (e) findings, and (f) recommendations/main conclusions. For the gray literature, extracted data included (a) type, title, and source of document, and (b) summary of purpose and recommendations. All documents were also assigned a level of evidence rating according to a scale given as an example by the American Speech-Language-Hearing Association (ASHA, n.d.), as applicable to the types of study design and documents that were reviewed. No other quality analysis was conducted. Recommendations and conclusions extracted from all types of literature were then grouped into broad themes (e.g., features of Rett syndrome that impact communication, assessment, intervention techniques and strategies) and used as a starting point for developing the survey questions for the next phase of the project.

Table 1. Potential search terms for conversion to database-specific keywords.

Participant terms	Communication terms		Intervention terms	Other terms
Rett, Rett syndrome, Rett disorder	Communication, language, speech, vocalization, gesture, understanding, comprehension, cognition, cognitive skills	Learning, literacy, reading, behavior, attention, eye gaze, eye tracking, augmentative and alternative communication, AAC	Assessment, intervention, management, therapy, outcome	Best practice, guidelines, state-of-the-art, comparison, review

Phase 2: Input from caregivers and communication professionals

For the second phase of the project, two surveys were developed and used to obtain real-world experiences of caregivers and professionals to further inform the development of the guidelines.

Survey development

Initial data for the surveys were gathered at the Rettsyndrome.org conference in Chicago in 2016. A community consultation was conducted with conference attendees to obtain face validity on the questions to be asked in both surveys. A small paper-based pilot of those surveys was also conducted. Ten communication professionals and one individual with Rett syndrome participated in the consultation process; seven professionals and 19 caregivers completed the pilot surveys. Following the pilot, two online surveys were developed and delivered online using Qualtrics software, version 2016 (Qualtrics, Provo, UT). These surveys were used to collect data from a broad international sample of communication professionals and caregivers. The survey for communication professionals (professional survey) was only available in English and asked about each participant's professional background, Rett syndrome-related knowledge and experience, Rett syndrome-related service delivery, and eye-gaze technology and Rett syndrome. In order to complete the professional survey, each professional must have worked with at least one person (of any age) with Rett syndrome and be able to respond to the survey in English. The survey for caregivers (caregiver survey) was available in 16 languages (Chinese, Danish, Dutch, English, Finnish, French, German, Greek, Hebrew, Italian, Lithuanian, Polish, Portuguese, Russian, Spanish, and Swedish). It collected basic demographic data (e.g., country of residence, age of person with Rett syndrome, daytime activities, and residential situations), and asked about each participant's experiences of communication services and support, exposure to eye gaze technology, and access to information. The choice of survey languages was largely determined by the opportunity sample of volunteer translators who worked with the project team, many of whom were parents from national Rett syndrome parent associations. Quality control of the translations was conducted by the first author in conjunction with colleagues at Maastricht University. The caregiver survey was open to any caregiver who was able to complete it in one of the 16 languages available. For both surveys, participants were recruited through social media, specialist Rett syndrome clinics and expertise centers, and national Rett syndrome associations.

Data analysis

The professional survey yielded 120 responses from 19 countries while the caregiver survey received 490 responses across all 16 languages, from individuals living in 39 countries. Data from the surveys were downloaded from Qualtrics into an Excel file and coded using a content analysis framework (Sandelowski, 2000) with NVivo 11 qualitative data analysis software[1] (QSR International, 2015). The initial codes were developed by the first author and checked for consistency by the third author; any disagreements were resolved through discussion within the research team. According to the content analysis process, broad themes or categories were first identified; each category was then further divided into sub-categories, into which the information extracted from the surveys and the literature review was organized as statements and recommendations. For the purposes of the project, *statements* were defined as descriptions or explanations of background status (e.g., "For any individual with Rett syndrome, their level of receptive language [understanding] is usually better than their ability to express themselves") while *recommendations* were defined as obligations or directives to be followed or acted upon (e.g., "AAC should be made available to every individual with Rett syndrome").

Phase 3: Delphi survey

For the third phase of the project, a two-round Delphi survey was used to gain consensus agreement on the statements and recommendations that had been extracted from the literature and surveys. Participation was by invitation only. Subject-matter experts with significant lived experience of Rett syndrome, in either a caregiver or professional capacity, were identified from the respondents to the online surveys in Phase 2 and from their reputation as experts in the field. Caregivers were required to have a child (of any age) with Rett syndrome and diverse experience of the wider Rett community (e.g., through serving on the board of a national parent association or working for a Rett syndrome organization). Professionals needed to be experienced in the delivery of communication assessment and intervention, either directly or through managing others working in a communication professional role; this allowed for the inclusion of medical practitioners working in a management capacity within a Rett expertise center or Rett specialist clinic. Additionally, the professionals should have worked with at least eight individuals with Rett syndrome for a period of at least one year. In this way, the composition of the Delphi panel was intended to reflect a range of stakeholders (Eccles, Grimshaw, Shekelle, Schunemann, & Woolf, 2012).

In total, 68 caregivers and professionals were invited to participate in the Delphi survey and 36 accepted. All 36 participants completed Round 1 of the Delphi survey and 35 of the 36 completed Round 2. Of the 36, 21 lived in one of 10 countries within Europe, 11 were from the United States, three were from Australia, and one was from Israel. Participants varied, with the highest representation being SLPs (29%) followed by parents (22%). A more detailed breakdown of participants can be seen in Table 2.

[1]NVivo is a product of QSR International, https://qsrinternational.com/nvivo/nvivo-products/.

Table 2. Delphi survey participants by role.

Role	*n* (%)[a]
Speech-language pathologist[b]	13 (29)
Parent	10 (22)
Other therapist (occupational, music)[b]	6 (13)
Rett clinic director/coordinator	5 (11)
Education staff (teacher, educational psychologist, social education worker)[b]	4 (9)
Researcher (University professor, lecturer)	4 (9)
Medical doctor	3 (7)

[a]$N = 36$, but several participants reported more than one role.
[b]These categories included individuals who also identified as assistive technology and/or augmentative and alternative communication specialists.

Table 3. Delphi survey: question types and numbers.

	Statements and recommendations[a]				
Round	Agreement[b]	Importance[c]	All/most/some/no-one questions[d]	Open-ended questions	Case scenarios
1	195	84	41	32	8
2	125	5	3	0	0

[a] Five-point Likert rating scales were used to rate statements and recommendations for agreement and importance.
[b] Agreement rating, 1 (strongly agree), 2 (agree), 3 (neither agree nor disagree), 4 (strongly disagree), 5 (disagree).
[c]Importance rating, 1 (extremely important), 2 (very important), 3 (moderately important), 4 (slightly important), 5 (not at all important).
[d]Participants were asked to grade intervention goals according to whether they were suitable for all individuals, most individuals, some individuals, or no-one with Rett syndrome.

Table 4. Delphi survey results: levels of consensus.

		Statements and recommendations reaching consensus									
		Agreement[a]					Importance[b]				
Round	Overall level of consensus n/N (%)	1	2	3	4	5	1	2	3	4	5
1	267/279 (96)	124	47	6	8	1	48	32	1	0	0
2	124/130 (95)	74	38	9	0	0	1	2	0	0	0

[a]1 (strongly agree), 2 (agree), 3 (neither agree nor disagree), 4 (strongly disagree), 5 (disagree).
[b]1 (extremely important), 2 (very important), 3 (moderately important), 4 (slightly important), 5 (not at all important).

Delphi procedure and data analysis

The Delphi survey was conducted online using the Qualtrics software. In both rounds participants were asked to rate statements and recommendations according to five-point Likert scales of agreement (1 *strongly agree*, 2 *agree*, 3 *neither agree nor disagree*, 4 *disagree*, 5 *strongly disagree*) or importance (1 *extremely important, 2 very important*, *3 moderately important*, *4 slightly important, 5 not at all important*). They were also asked to provide responses to a range of open-ended questions, several multiple-choice or yes/no questions, questions asking participants to grade intervention goals according to whether they were suitable for *all individuals, most, some, no-one with Rett syndrome*, and a number of case scenarios. After each round, the responses were analyzed by the third author using IBM SPSS Statistics, version 24 (IBM Corporation, New York) to identify those statements and recommendations reaching consensus. As in previous guidelines developed for the management of scoliosis (Downs et al., 2009), nutrition and growth (Leonard et al., 2013), and bone health (Jefferson et al., 2016) in individuals with Rett syndrome, the consensus level was set at a minimum of 70% of responses within one category of the median. In addition, the qualitative responses to Round 1 were coded by the fourth author to facilitate extraction of new information for Round 2.

Following analysis of Round 1, the statements and recommendations included in Round 2 were those that (a) had not reached a consensus of 70% in Round 1, (b) had reached consensus but were rephrased or combined to reduce repetition or increase clarity, (c) were completely new following information supplied by participants in Round 1, or (d) were based on intervention goals from Round 1 (rated according to *all individuals*, *most, some, no-one with Rett syndrome*) and rewritten for the agreement rating scale in Round 2. Following participant feedback, a sixth option (*not my area of expertise*) was also added to the agreement and importance scales in Round 2.

The number of statements and recommendations, and the number and type of questions included in Rounds 1 and 2 can be seen in Table 3, while the consensus ratings achieved in both rounds can be seen in Table 4.

Copies of the caregiver and professional surveys and the Delphi Round 1 and 2 surveys are available from the authors on request.

Results

Final guidelines

Across both rounds of the Delphi survey a total of 268 statements and recommendations reached the predetermined consensus level of 70% and above. In addition, they all achieved positive ratings (*strongly agree, agree, extremely important, very important*). These statements and recommendations thus qualified as the final, consensus-based guidelines for managing the communication of individuals with

Rett syndrome. The guidelines were arranged in eight sections and made available in two formats: the raw statements and recommendations (accessible online as Supplemental Material), and a Handbook in which the statements and recommendations are supplemented with explanatory texts, case stories, and links to useful resources. An overview of each of the eight Sections is given below; however, the Supplemental Files should be consulted for full information on the final guidelines. Both the raw statements and recommendations (the Supplemental Material) and the Handbook can be accessed at: https://cris.maastrichtuniversity.nl/en/publications/rett-syndrome-communication-guidelines-a-handbook-for-therapists-.

Section 1: Guiding principles

This first section of the guidelines includes 20 statements and recommendations that set out the principles upon which the rest of the guidelines are based. This section describes the rights to communication that individuals with Rett syndrome should be able to expect (e.g., the right to an appropriate communication system and communication goals; the right to advice, support and services that start early and continue throughout life; the right to communication partners who are trained in appropriate communication strategies and techniques), based on the United Nations Convention on the Rights of Persons with Disabilities, and the Communication Bill of Rights developed by the United States National Joint Committee for the Communication Needs of Persons with Severe Disabilities (Brady et al., 2016). Beliefs and attitudes are also included in this section (e.g., communication partners should have an open mind to the communication potential of the individual with Rett syndrome; communication partners should believe that, given the opportunity, individuals with Rett syndrome should be able to communicate using AAC).

Section 2: Professional practice

This section has 29 statements and recommendations and establishes principles of teamwork and the need for collaboration between parents and professionals (e.g., every individual with Rett syndrome should be supported by a multidisciplinary team; the team should share a common vision and work collaboratively to define and agree communication goals and support plans). It explores the obligations incumbent on professionals and their employers to develop knowledge and expertise, in relation to AAC in general and Rett syndrome in particular (e.g., professionals who are inexperienced in working with individuals with Rett syndrome should seek training in relevant topics, and seek advice and support from colleagues with more specialized knowledge and expertise in the area). The role of Rett expertise centers and Rett specialist clinics is also explored in Section 2 (e.g., the individual and family may be referred to a Rett specialist clinic or expertise center in order to get a diagnosis, or for specialized assessment and advice or a second opinion at any point following diagnosis; communication professionals attached to a Rett specialist clinic or expertise center should be available to offer advice and support to, and answer questions from, locally-treating therapists, and available to respond to questions from parents and caregivers and individuals with Rett syndrome).

Section 3: Features of Rett syndrome that impact communication

This section of the guidelines has 19 statements. These present an overview of the core and supportive clinical criteria for Rett syndrome as well as additional conditions that are frequently reported to co-exist and that the Delphi panel agreed impact communication (e.g., movement disorders; breathing/respiratory irregularities; impaired sleep pattern; scoliosis; seizures; fatigue and reduced alertness; difficulties with sensory regulation, mood, and anxiety; gastro-intestinal issues; and, less frequently reported, hearing or vision-related conditions).

Section 4: Strategies to optimize engagement

This section is comprised of 26 statements and recommendations. These describe general strategies that communication partners should embed in daily life (e.g., address and talk directly to the individual rather than talking about them in their presence; be responsive, acknowledging and reinforcing all communication attempts; give feedback and attribute meaning; use multimodal communication) and more specific strategies that can be utilized in response to the key features of Rett syndrome described in Section 3. Strategies to address issues of fatigue, alertness, sensory regulation, stress, and anxiety are offered as well as tips for determining sufficient wait time.

Sections 5: Assessment

In this section, there are 42 statements and recommendations. These establish general principles of communication assessment, present the World Health Organization's (2001) International Classification of Functioning, Disability and Health as an appropriate model around which to structure holistic assessment of the individual, and explore assessment procedures and settings. Included within Section 5, the pros and cons of employing standardized or formal assessment tools versus informal methods of assessment are considered (e.g., standardized assessments of language and cognition are likely to indicate that individuals with Rett syndrome have an intellectual disability; standardized assessments may not accurately reflect an individual's underlying ability nor their communicative and learning potentials but they may be adapted to obtain information on certain specific skills), and dynamic assessment (Haywood & Lidz, 2007) is endorsed as an appropriate model for communication assessment in individuals with Rett syndrome (e.g., assessment should not take place at a single moment in time, it should be ongoing and dynamic).

Section 6: AAC assessment

This section has 23 statements and recommendations that focus specifically on issues pertaining to AAC assessment, with the starting point being that "no prerequisite skills must be demonstrated before aided AAC is considered." Components of AAC assessment are described and models of best practice for AAC assessment are presented (e.g., the six-step process (Dietz, Quach, Lund, & McKelvey, 2012), the participation model (Beukelman & Mirenda, 2013), and the model of communicative competence (Light & McNaughton, 2014)). There is also a focus on the assessment of readiness for eye-gaze technology (e.g., assessments of readiness to use eye gaze technology are best conducted through informal activities).

Section 7: Assessment of AAC system/device

Within the penultimate section of the guidelines there are 33 statements and recommendations. These address the need for extended trials and trials of more than one system or device (e.g., suitability of an AAC system or device cannot be judged adequately from a single session or a single point in time; trial periods should be for a minimum of 8 weeks; individuals should be able to trial more than one or multiple AAC systems and devices) as well as consideration of the device-specific features that must be assessed and the levels of support required (e.g., during trial periods the individual with Rett syndrome and their primary/key communication partners should be well-supported by knowledgeable and experienced professionals who are familiar with/trained to use the system/device on trial).

Section 8: Intervention

The final section of the guidelines has 76 statements and recommendations that cover general principles of intervention (e.g., communication intervention and management should start early and be lifelong) and the setting of goals and targets (e.g., goals for intervention should include development of nonverbal, low-tech and high-tech strategies; developing a yes/no response is important for all individuals with Rett syndrome), including the expansion of communication functions and ways of communicating. Strategies and techniques to support the development of communication skills are also presented (e.g., modeling; partner assisted scanning).

Evidence tables have also been compiled to complement the consensus-based guidelines, with one table corresponding to each statement or recommendation. The tables bring together in one place the supporting information collected from each of the phases of the project. They provide an overview of the supporting literature, quotes from online survey responses, and Delphi results upon which each statement or recommendation was built. The levels of evidence ratings that were assigned (ASHA, n.d.) are also included alongside the literature. An example can be seen in Table 5. The full set of Evidence tables can be requested from the authors or accessed alongside the Supplemental Material and Handbook at: https://cris.maastrichtuniversity.nl/en/publications/rett-syndrome-communication-guidelines-a-handbook-for-therapists-.

Discussion

This project followed a consensus-based approach (Mei et al., 2018) to develop guidelines for managing the communication of individuals with Rett syndrome. In the absence of high-quality evidence, current best practice was combined with available evidence from literature and surveys, and the resulting statements and recommendations were verified by expert consensus in the form of a two-round Delphi survey.

Table 5. Evidence table example: intervention techniques

Recommendation 8.75	Prompts or cues such as gestures, demonstrations, touch, and signals can be used to increase the likelihood that individuals will make correct responses.
Level of evidence	IIb
Consensus rating	Agree 27/31 (87.1%)
Supporting evidence	
Published literature	Prompts have been demonstrated to increase the frequency of correct responses in individuals with RTT; however, in certain cases the prompts could not be faded (withdrawn) without a subsequent loss of skill. (Byiers, Dimian, & Symons, 2014; Lancioni et al., 2014; Simacek, Reichle, & McComas, 2016; Stasolla et al., 2015) (Level IIb)
Gray literature	- nil -
Focus group and professional survey	"Describe the most appropriate prompts." "Pause to prompt - give the girls LOADS of time to respond and use indirect prompts."
Caregiver survey	"When needed, offer prompt."
Delphi survey	"Describing the most appropriate prompts." "Reinforcement, stimulus prompting and fading, and discrimination teaching "I would use discrimination teaching and reinforcement (and prompting as needed) to chain these open-ended choices following the "yes/no" choice."
References	Byiers, B. J., Dimian, A., & Symons, F. J. (2014). Functional communication training in Rett syndrome: A preliminary study. *American Journal on Intellectual and Developmental Disability, 119*, 340-350. doi:10.1352/1944-7558-119.4.340 Lancioni, G. E., Singh, N. N., O'Reilly, M. F., Sigafoos, J., Boccasini, A., La Martire, M. L., . . . Sacco, V. (2014). Microswitch-aided programs for a woman with Rett syndrome and a boy with extensive neuro-motor and intellectual disabilities. *Journal of Developmental and Physical Disabilities, 26*, 135-143. doi:10.1007/s10882-013-9349-x Simacek, J., Reichle, J., & McComas, J. (2016). Communication intervention to teach requesting through aided AAC for two learners with Rett syndrome. *Journal of Developmental and Physical Disabilities*, *28,* 59-81. doi:10.1007/s10882-015-9423-7 Stasolla, F., Perilli, V., Di Leone, A., Damiani, R., Albano, V., Stella, A., & Damato, C. (2015). Technological aids to support choice strategies by three girls with Rett syndrome. *Research in Developmental Disabilities, 36*, 36-44. doi:10.1016/j.ridd.2014.09.017

Clinical implications

Implications for caregivers and professionals

The guidelines provide information that is valuable to both caregivers and professionals. They offer strategies that can be implemented at home by caregivers and provide a focal point around which discussions between caregivers and professionals can be structured. When used by communication professionals, the guidelines are intended to complement or supplement the requirements of mandatory bodies, for example, in relation to continuing professional development for SLPs and other therapists. The guiding principles set the context and the rest of the guidelines address what those principles mean in daily life and how they can be achieved in practice. The importance of being open minded as a communication partner (open to possibilities and potential, and to establishing the best conditions for communication) is clearly reflected throughout the guidelines, as is the need to develop partnerships between caregivers and professionals and to maintain a shared team vision when working with individuals with Rett syndrome. This is especially crucial when seeking to maximize progress and reduce device abandonment (Baxter, Enderby, Evans, & Judge, 2012; Holmqvist, Thunberg, & Peny Dahlstrand, 2018; Judge & Townend, 2013).

Rett-specialist clinics and expertise centers can play an important role in offering guidance and support to local teams, although it is recognized that availability and access to such centers will vary greatly within and among countries. Where these clinics and centers do exist, they should aim to make their services more widely available by offering alternative means of access, for example, through video link/teleconference. Where such clinics and centers are not available, clinicians and caregivers can use the guidelines (a) to help frame their own work in developing communication with the individuals they support, and (b) as leverage in discussions with local and national service planners and providers, for example, when working toward the development of specialist clinics and arguing for changes in funding of services and equipment.

Implications for assessment and intervention

The guidelines endorse informal methods of assessment rather than formal assessment tools, and recommend that assessments are ongoing and dynamic, undertaken in naturalistic settings. Trials of any AAC system should also be conducted over a prolonged period of time, scaffolded by the communication partner modeling use of the system rather than by testing of the individual. Recommendations for introducing and implementing AAC depart from a *candidacy model*, which requires individuals to demonstrate prerequisites for AAC before being introduced to any aided (low- or high-tech) AAC system. Belief in this model has been debated, and debunked, in the AAC literature for many years (Romski & Sevcik, 2005), and the results of the Delphi survey confirmed that this is no longer the prevailing view among caregivers and communication professionals working with individuals with Rett syndrome. AAC in all its forms is recognized in the guidelines as crucial for development of communication and literacy skills and, in line with widely accepted best practice in AAC (Romski, Sevcik, Barton-Hulsey, & Whitmore, 2015), it is recommended that AAC should be made available as early as possible and continued throughout life. The guidelines stress that multiple modalities should be utilized and individuals should have access to both low- and high-tech systems. Individuals are not required to achieve a minimum level of competence with low-tech AAC before high-tech options can be introduced; neither should they be expected to rely only on the high-tech. Multiple strategies and forms of communication should be encouraged, while recognizing that eye gaze is likely to be the most reliable form of access, to both low- and high-tech AAC, for most individuals with Rett syndrome (Bartolotta et al., 2011).

A wide variety of goals for intervention are described in the guidelines, beginning with goals for new/early communicators and concluding with goals for reading and writing. The ultimate aim is to enable all individuals with Rett syndrome to become more autonomous as communicators. No expectations are placed on the level each individual will reach in their quest for autonomy; each will begin their journey from a different starting point and will finish at a different end point, but the ultimate aim underpinning these consensus-based guidelines is for all individuals with Rett syndrome to be given opportunities to develop their skills and exposed to the conditions that support them to do so.

Many of the recommendations described above are clearly not limited to individuals with Rett syndrome and are applicable to a wide range of individuals with developmental disabilities and/or complex communication needs; however, there are also statements and recommendations that are specific to Rett syndrome, which is a complex condition affecting multiple body systems. As such, the guidelines include information on symptoms and features that may impact communication. In particular, they draw attention to the fact that "communication skills of individuals with Rett syndrome will fluctuate based on internal and external factors" and that communication partners should expect inconsistency. The guidelines describe the difficulties in motor movement and motor planning that affect individuals with Rett syndrome and the impact that movement disorders, such as dyspraxia/apraxia and altered muscle tone, can exert on an individual's ability to respond. Key recommendations include compensatory strategies such as reducing the motor demands of a task as the cognitive load increases, allowing for a longer response time, and deployment of eye gaze as (usually but not only) the best way to access AAC. Many of the strategies for optimizing communication that are included in the guidelines are intended to be used by all communication partners, in all settings, across the lifespan, and not confined to formal or structured therapy situations alone. This is crucial for maximizing potential, generalizing skills across contexts, and improving communication with multiple communication partners (Romski et al., 2015).

Limitations and future directions

A number of limitations must be considered when interpreting the results of this project and the quality of the guidelines. First, the literature review included peer-reviewed literature across all levels of evidence without undertaking a quality appraisal of each study. Gray literature was also included that was not empirical or peer-reviewed. When taken together, these elements open up a possible risk of bias from low quality, small-scale and opinion-related data. Second, the literature searches, survey for communication professionals, and Delphi survey were restricted to the English language, thus, research, knowledge, and practices from non-English speaking individuals and countries may be poorly represented in the guidelines. Even so, 120 communication professionals from 19 countries contributed to the professional surveys, 36 experts (caregivers and professionals) from 13 countries took part in the Delphi survey, and the caregiver surveys were translated into 16 languages, receiving responses from 39 countries. In many ways this demonstrates a good spread of data and experience. Third, although the levels of consensus reached for the statements and recommendations in the Delphi survey were very high, the nature of the consensus methodology means that outliers or strong opinions that are expressed at either end of the spectrum may not be represented in the final guidelines. It could also be debated whether a 70% cutoff is high enough to determine consensus. However, this level was supported by previous research utilizing a consensus methodology to develop guidelines for Rett syndrome (Downs et al., 2009; Jefferson et al., 2016; Leonard et al., 2013). Fourth, participants were given the option to select *not my area of expertise* as a response to parts of the Delphi survey. As a consequence, some questions were completed by a small number of respondents only. The results for these questions could be considered skewed according to the background of those who chose to answer. In the field of rare disorders, however, numbers of potential participants are naturally low. Responses to the questions about formal assessments were particularly low, which is to be expected, as this is a niche area and availability of assessments will vary according to country and language. Because of the small number of participants familiar with each of the named assessment tools, no specific recommendations relating to their use are included in the guidelines. Further research is needed on the application of formal assessments to obtain accurate and reliable information on the communication skills of individuals with Rett syndrome. One final consideration in relation to the Delphi methodology is how statements or recommendations reaching a consensus of *strongly disagree* or *disagree* were analyzed and incorporated into the guidelines. This may have required a caution or warning to be issued in the guidelines when considering certain techniques or approaches. Statements and recommendations with a median response of *strongly disagree* or *disagree* in Round 1 were reworded and recirculated in Round 2 (e.g., "prerequisite skills such as understanding of cause and effect and showing communicative intent must be demonstrated before AAC should be considered" received a rating of "strongly disagree" in Round 1 and was flipped to "there are no prerequisite skills that must be demonstrated before aided AAC should be considered" in Round 2, where it received a consensus rating of "strongly agree"). As a result, no statements or recommendations achieved a consensus rating of strongly disagree or disagree at the end of Round Two. Therefore, this was not an issue in the final guidelines formulation.

Dissemination and uptake are both key to the success of the guidelines but there are no guarantees without having strategies in place to enable and support this (Kredo et al., 2016; Shekelle, Woolf, Grimshaw, Schunemann, & Eccles, 2012). A number of related efforts are already underway in several countries across the world. These include the creation of professional support and training materials, clinical networks, and translation of the guidelines into multiple languages. Future research will need to focus on implementation and evaluation of the guidelines, especially to ascertain whether they can, in practice, be adopted and adapted to suit the health, education and social care policies and practices and funding situations within and between countries. The guidelines will also need to be reviewed and updated as new evidence comes to light. Research is required to develop more appropriate methods for assessing the communication and cognitive skills of individuals with Rett syndrome, methods that adequately recognize the impacts of a multiple system rare disease on communication, cognition, physiological, and motor responses. Further research should also be conducted into how appropriate interventions can be applied to ensure communication skills are maximized throughout life. In developing these guidelines, one individual with Rett syndrome participated in the initial community consultation that took place in the United States of America. This signals a step toward including people with complex communication needs more directly in research relating to their needs and recognizing their opinions and experiences can and should be heard. This should be an important driver for future research.

Conclusion

The first international consensus-based guidelines for the management of communication in individuals with Rett syndrome have been created with a total of 650 participants from 43 different countries contributing across all stages of the project, representing a variety of roles, and living and working in a variety of contexts. Therefore, the resulting guidelines should be applicable in multiple diverse situations and settings across the world. The guidelines have the potential to improve assessment, intervention and long-term management of communication for individuals with Rett syndrome, to stimulate training and implementation of best practice world-wide, and to promote research to fill the gaps in the currently-limited evidence-base. The publishing of these guidelines is the first step in the development of more global consensus-based (and later evidence-based) practices in the assessment, intervention and management of communication in individuals with Rett syndrome.

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