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Early socio-communicative forms and functions in typical Rett syndrome

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A R T I C L E   I N F O

A B S T R A C T

Rett syndrome (RTT) is a severe neurological disorder characterized by a developmental regression in motor and speech-language domains. There is, however, limited research on socio-communicative development of affected children before the onset of regression. We analyzed audio–video recordings made by parents of six 9- to 12-month old girls later diagnosed with typical RTT, applying the Inventory of Potential Communicative Acts (IPCA) to identify early communicative forms and functions. Each girl used at least one communicative form (e.g., body movement, eye gaze, or vocalizations) to gain attention and answer, but none were observed to make choices or request information. Varying numbers of children were observed to perform other communicative functions according to the IPCA including social convention, rejecting or requesting an object. Non-verbal forms (e.g., reaching, moving closer, eye contact, smiling) were more common than non-linguistic verbal forms (e.g., unspecified vocalizations, pleasure vocalizations, crying). (Pre-)linguistic verbal forms (e.g., canonical or variegated babbling, proto-words) were not used for communicative purposes. These data suggest that atypical developmental patterns in the socio-communicative domain are evident prior to regression in young individuals later diagnosed with RTT.

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1. Introduction

In the consensus redefinition of the clinical criteria for the diagnosis of Rett syndrome (RTT; Neul et al., 2010), four main criteria were identified: (a) partial or complete loss of purposeful hand skills, (b) partial or complete loss of spoken language, (c) appearance of gait abnormalities (dyspraxic or absence of ability), and (d) emergence of stereotypic hand movements (e.g., hand washing-like movements, hand clapping, and hand to mouth stereotypies). The developmental trait of regression followed by recovery or stabilization and these four criteria are required for adequate diagnosis whereas supportive criteria

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(such as commonly observed breathing disturbances when awake, diminished response to pain, and growth retardation) – need not to be present (Neul et al., 2010).

Even though it has been assumed that early development of children later diagnosed with RTT was apparently normal, there is growing evidence that the pre-regression period is not asymptomatic (e.g., Burford, Kerr, & Macleod, 2003; Einspieler, Kerr, & Prechtl, 2005a; Einspieler, Kerr, & Prechtl, 2005b; Leonard & Bower, 1998; Marschik, Einspieler, Oberle, Laccone, & Prechtl, 2009; Marschik, Einspieler, & Sigafoos, 2012a; Marschik et al., 2012b, 2012c, 2012d, 2013; Marschik, Lanator, Freilinger, Prechtl, & Einspieler, 2011; Neul et al., 2010; Tams-Little & Holdgrafer, 1996; Temudo, Maciel, & Sequeiros, 2007). Indeed, studies into the early development of individuals with typical RTT, and individuals with the relatively milder preserved speech variant (PSV), have revealed atypical patterns in early speech-language development. These atypical patterns have been observed during the first two years of life and prior to the period of regression (Marschik, Einspieler, Prechtl, Oberle, & Laccone, 2010; Marschik et al., 2009, 2012b, 2012c, 2012d).

With respect to socio-communicative development of individuals with RTT, a number of studies reported the presence of various idiosyncratic behaviors like non-conventional vocalizations or eye gaze that appear to serve one or more communicative functions (Dahlgren Sandberg, Ehlers, Hagberg, & Gillberg, 2000; Marschik et al., 2012b, 2012d; Sigafoos et al., 2000a, 2011; Sigafoos, Woodyatt, Tucker, Roberts-Pennell, & Pittendreigh, 2000b). However, there has been limited study into the early socio-communicative forms (e.g., body movements, vocalizations, proto-words) and functions (e.g., request an object, comment, choice making, imitation) of young girls who are later diagnosed with RTT. Studies of this population might identify developmental deficits with respect to early communicative development in the pre-regression period of RTT. Identification of any such deficits might in turn be useful for early detection of RTT. To this end, we conducted a retrospective analysis of home video-recordings of early communicative forms and functions in six girls from 9 to 12 months of age who were later diagnosed with typical RTT. The analysis focused on the following questions: (a) Which, if any, potential communicative acts can be observed during the pre-regression period? (b) What, if any, communicative functions are present in the participants' communicative repertoires? (c) Are there differences with respect to the use of verbal versus nonverbal communicative behaviors?

2. Method

2.1. Participants

Six girls with typical RTT were included in this study. All of them had been recorded by their parents (see Section 2.2) when they were between 9 and 12 months of age. Three of them came from German-speaking families and three from English-speaking families. All were singletons from uneventful pregnancies and deliveries. Birth weight, birth lengths, occipitofrontal circumferences, and Apgar scores were within the normal ranges. All were later confirmed to be MECP2 positive (mutations in the gene encoding Methyl-CpG-binding protein 2 are present in 95–97% of individuals with typical RTT; Neul et al., 2008) and classified as having typical RTT. The study was approved by the local research ethics committees and parents gave their informed consent for the research and publication of the results.

2.2. Procedure

The procedures were similar to those used in our previous study on individuals with PSV (Marschik et al., 2012b). Analyses were based on coding of extensive video footage recorded by the participants' parents when the children were from 9 to 12 months of age, the last months before the onset of regression. At this time, the parents were not aware that their daughters had RTT. The audio–video recordings were made during typical family routines (e.g., play situations, bathing, feeding) and special events (e.g., family gatherings). A research assistant naïve to the purpose of the study checked the recordings for sufficient length and quality standards, copied the relevant recordings, and prepared them for coding. The footage of all six participants comprised a total of 459 min.

The audio–video recordings were coded for the occurrence of potential communicative acts, such as body movements (e.g., turning to or moving toward a person, reaching and touching), vocalizations (e.g., pleasure bursts, crying, babbling, proto-words) and facial expressions. This coding was based on the Inventory of Potential Communicative Acts (IPCA; Sigafoos, Arthur-Kelly, & Butterfield, 2006; Sigafoos et al., 2000a). The IPCA is an assessment tool that has been used to gather descriptive information on the potential communicative forms and functions of individuals with developmental disabilities and severe communication impairments, including individuals with RTT (Didden et al., 2010; Marschik et al., 2012b; Sigafoos et al., 2000a, 2000b, 2006).

All potential communicative behaviors (i.e., all behaviors of the participants coded as an attempt to communicate) observed in the audio–video recordings were transcribed by the first author and rechecked by a second transcriber against the recordings to ensure accuracy and consistency. Sequences with disagreements (13%) were discussed within the team until consensus was achieved. Based on these transcriptions, the verbal and nonverbal communicative forms that were observed were assigned a communicative function based on the classification system of the IPCA (Sigafoos et al., 2000a, 2006). These functions were: (a) social convention (e.g., greeting, indicating farewell, responding to name), (b) attention to self (e.g., getting attention, seeking comfort, showing off), (c) reject/protest (e.g., rejecting non-preferred objects/activities), (d) request object (e.g., requesting a preferred toy or snack), (e) request action (e.g., help with dressing, someone to come/be

Table 1
Communicative forms observed in six females between 9 and 12 months of age who were later diagnosed with typical RTT.

Body movements	Facial expression/eye movements	Vocalizations	Gestures
Reaching	Eye contact	Unspecified vocalization	Waving (indicating greeting)
Moving closer	Smiling	Fussing	
Moving/turning away		Crying	
Touching/tweaking persons		Pleasure vocalization	
Patting		Laughing	
Retaining objects			
Imitation of manual routine			

Table 2
Communicative functions observed (●) or not observed (□) for each child between 9 and 12 months of age and later diagnosed with typical RTT. Categories according to the Inventory of Potential Communicative Acts (IPCA; Sigafoos et al., 2000a, 2006).

	Social convention	Attention to self	Reject/ protest	Request object	Request action	Request information	Comment	Choice making	Answer	Imitate	Total
Child 1	□	●	□	□	□	□	●	□	●	□	3
Child 2	□	●	●	●	●	□	●	□	●	●	7
Child 3	●	●	●	□	□	□	●	□	●	●	6
Child 4	□	●	●	●	□	□	□	□	●	□	4
Child 5	●	●	□	□	□	□	●	□	●	●	5
Child 6	●	●	●	●	□	□	●	□	●	□	6
Total	3	6	4	3	1	0	5	0	6	3	

near), (f) request information (e.g., clarification, information about something), (g) comment (e.g., happy, frightened, pain), (h) choice making (e.g., making a choice between two or more objects, indicating when to start or stop an activity), (i) answer (e.g., reacts when someone talks to her, indicates yes/no in response to a question), and (j) imitation (e.g., imitating another person's speech or gestures). Behaviors assigned to one of these functional categories were further classified as verbal or nonverbal (e.g., body movements). Verbal behaviors were further classified into one of two subcategories, that is non-linguistic vocalizations (e.g., vegetative sounds, fussing) and (pre-)linguistic vocalizations (e.g., canonical or variegated babbling, proto-words). The coding was carried out using the Noldus Observer-XT (www.noldus.com; version: 11.0).

3. Results

3.1. Communicative forms

The children engaged in 15 different behaviors that were classified as communicative forms. The specific forms observed in the audio–video recordings are listed in Table 1. Overall, we observed seven different body movements, a facial expression, eye gaze/movement, five different types of vocalizations, and one gesture.

3.2. Communicative functions

For each child, we coded use of the 10 different communicative functions from the IPCA. As shown in Table 2, the total number of different communicative functions observed per child ranged from three (Child 1) to seven (Child 2). Every child was observed to (a) direct attention to herself and (b) answer. However, none of the participants showed any instances of (a) requesting information or (b) making choices. The remaining communicative functions (e.g., social convention, reject/ protest, request object, request action, comment, and imitate) were observed in from one to five children.

3.3. Verbal versus nonverbal communicative forms

Table 3 shows the extent to which communicative functions were expressed with non-verbal behavior versus non-linguistic vocalizations or (pre-)linguistic vocalizations. Non-verbal behavior dominated over non-linguistic vocalizations in

Table 3
Number of children using non-verbal behavior versus non-linguistic or (pre-)linguistic vocalizations for each of 10 communicative functions according to the Inventory of Potential Communicative Acts (IPCA; Sigafoos et al., 2000a, 2006).

	Social convention	Attention to self	Reject/ protest	Request object	Request action	Request information	Comment	Choice making	Answer	Imitate
Non-verbal behavior	3	6	2	3	1	0	5	0	6	3
Non-linguistic vocalizations	1	4	4	2	0	0	3	0	3	0
(Pre-)linguistic vocalizations	0	0	0	0	0	0	0	0	0	0

seven of the eight functional categories that were observed. Only for the reject/protest function did more children use non-linguistic vocalizations. None of the children were observed to use (pre-)linguistic utterances (such as canonical or variegated babbling or proto-words) for communicative purposes.

4. Discussion

A number of studies reported that cognitive processing, speech-language development, and social reciprocity are impaired in individuals with typical RTT and PSV (Cass et al., 2003; Charman et al., 2002; Kaufmann et al., 2012; Kerr, Archer, Evans, & Gibbon, 2006; Marschik et al., 2013; Matson, Dempsey, & Wilkins, 2008; Renieri et al., 2009; Sigafoos et al., 2011; Uchino, Suzuki, Hoshino, Nomura, & Segawa, 2001). Whereas the majority of studies focused on the post-regression period of development, emerging data suggest atypical patterns such as inspiratory vocalizations, reduced volubility or a limited gestural repertoire in the speech-language domain prior to the period of regression (Marschik et al., 2009, 2010, 2012a, 2012c, 2012d; Tams-Little & Holdgrafer, 1996). The present study extends this research by focusing on socio-communicative forms and functions in the pre-regression period.

With respect to communicative forms evident in the pre-regression period, we observed various body movements, one gesture, and several vocalizations that were classified as serving one or more communicative functions (e.g., to express needs or to draw attention to oneself). The body movements used included reaching, turning toward or touching a person, smiling, eye contact, and the gesture waving. These behaviors could be viewed as part of the repertoire of forms for shared intentionality that typically emerge at the end of the first year of life (Chapman, 2000; Tomasello & Carpenter, 2007; Tomasello, Carpenter, Call, Behne, & Moll, 2005). On the other hand, the verbal behaviors we recorded were exclusively of non-linguistic character (e.g., fussing, crying, or laughing). (Pre-)linguistic vocalizations such as canonical or variegated babbling or the use of proto-words were not observed. The latter observation is quite different from what one would expect in typically developing children at the end of the first year of life (e.g., Paul, 2007), but consistent with our previous findings showing that most girls with typical RTT did not achieve early speech-language milestones (Marschik et al., 2013). Overall, our present findings with respect to communicative forms are in line with other reports showing profoundly restricted communicative forms among individuals with typical RTT or PSV and other neurodevelopmental disorders, such as fragile X syndrome and Angelman syndrome (Bonati et al., 2007; Duker, van Driel, & van de Bercken, 2002; Hinton et al., 2013; Losh, Martin, Klusek, Hogan-Brown, & Sideris, 2012; Marschik et al., 2012b; Sigafoos et al., 2011; Tams-Little & Holdgrafer, 1996).

The limited repertoire of communicative forms is mirrored by the relatively limited number of communicative functions that any given child was observed to indicate as reported in Table 2. However, it is possible that the absence of a communicative function in the audio–video recordings we analyzed, does not necessarily mean that the function was absent from the child's repertoire because there might not have been an opportunity for some communicative functions during the video (Marschik & Einspieler, 2011; Palomo, Belinchón, & Ozonoff, 2006). It remains open whether some of the functions were classified as absent because they were (a) not present in the data set or (b) reflecting an atypical developmental trait; the latter includes functions that (i) are potentially acquired at a later age (delay) or (ii) remain absent throughout development (non-achievement).

Two functions (i.e., drawing attention to self and answering) were observed in all six children, but answering was mainly manifest as the child orienting to the speaker when her name was called. In addition, all but one child was observed to show some commenting behaviors (e.g., laughing or pleasure vocalizations) adequate to certain situations. What may be more telling are those situations where children were not observed to communicate in any way, such as showing no apparent reaction when they got hurt. The issue of pain-insensitivity has been noted in the RTT literature (Neul et al., 2010).

Surprisingly, none of the participants engaged in behaviors indicative of a request for information or choice making which was also observed for individuals with PSV during the second year of life (Marschik et al., 2012b). Whereas only one child (Child 2) ever requested an action at the end of the first year, the majority of females with PSV did so during the second year of life (Marschik et al., 2012b). The lack of such requesting could again be related to lack of opportunity during the times when the videos were recorded or more likely that these skills were too advanced for this age. Chapman (2000) noted that requests for information usually appear in the second year of life. Social conventions (e.g., greeting) were seen in three of the six children, but in each of these children this was in response to adult initiations, rather than being self-initiated. Three children showed an absence of orienting toward a person when called, a phenomenon discussed in the early development of individuals with autism spectrum disorders (e.g., Baranek, 1999; Fostad, Matson, Hess, & Neal, 2009; Kaufmann et al., 2012; Osterling & Dawson, 1994; Osterling, Dawson, & Munson, 2002; Werner, Dawson, Osterling, & Dinno, 2000; Zwaigenbaum et al., 2013).

Even though non-verbal behaviors and non-linguistic vocalizations were both interpreted to cover eight or six functional categories respectively, non-verbal behaviors were more commonly observed. Only for the function of rejecting/protesting non-linguistic vocalizations were predominant (Table 3). A preference of non-verbal communicative forms compared to verbal communicative forms was previously reported for girls with RTT or PSV (Lavås, Slotte, Jochym-Nygren, van Doorn, & Engerström, 2006; Marschik et al., 2012b).

The results of the present study must be interpreted with caution due to the well-known methodological limitations of retrospective video analysis (e.g., Marschik & Einspieler, 2011; Palomo et al., 2006). Further limitations are the small sample size, the absence of a control group and the lack of available norms for the IPCA protocol. We can thus only interpret the data benchmarked against what is generally known about communicative development (forms and functions) in the literature.

Nevertheless, this is the first study to examine the early communicative forms and functions in children with RTT in their pre-regression period. Our data advance the understanding of the early developmental impairments associated with *MECP2* mutations (Burford, 2005; Burford et al., 2003; Marschik et al., 2012a, 2012b, 2012c, 2012d, 2013; Tams-Little & Holdgrafer, 1996). Even though there is the need to assess early socio-communicative functions in a larger group of individuals with RTT across a wider age range and if the database is comprehensive enough also in terms of type-token-ratio (i.e., how many of each function benchmarked over time), these findings are useful as a preliminary description of the developing socio-communicative domain in typical RTT. Our data advance the clinical understanding of pre-regressional development in RTT and might prove useful with respect to the early detection of children with a range of neurodevelopmental disorders that appear to have a clinical onset after the first year of life.

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