

Published in final edited form as:

*Dev Neurorehabil.* 2014 February ; 17(1): 34–38. doi:10.3109/17518423.2013.837537.

## Three different profiles: Early socio-communicative capacities in typical Rett syndrome, the preserved speech variant and normal development

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### Abstract

**Background and aims**—This is the first study aiming to compare pre-diagnostic socio-communicative development of a female with typical Rett syndrome (RTT), a female with the preserved speech variant of RTT (PSV), and a control toddler.

**Methods**—We analysed 1275 minutes of family videos at the participants' age between 9 and 24 months and used the Inventory of Potential Communicative Acts (IPCA) to delineate their repertoires of communicative forms and functions.

**Results**—The results revealed different profiles for the three different conditions. The repertoire of communicative gestures and (pre)linguistic vocalizations was most comprehensive in the control toddler, followed by the female with PSV and the female with RTT.

**Conclusion**—These findings contribute to the growing knowledge about early developmental abnormalities in RTT. In order to define distinctive profiles for typical and atypical RTT and evaluate their specificity a larger body of evidence is needed.

### Keywords

Communication impairment; Family videos; Preserved speech variant; Rett syndrome; Socio-communicative development; Video analysis

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#### *Declaration of interest*

The authors report no declarations of interest. The authors alone are responsible for the content and writing of this article.

## 1 Introduction

According to the staging model of the neurodevelopmental disorder Rett syndrome (RTT), early development has been considered apparently normal and followed by a profound regression of skills [1–3]. There is, however, accumulating evidence of abnormality in various developmental domains before the onset of regression [4–7]. As RTT is usually diagnosed during the toddler years or even later, most of the findings on early pre-diagnostic development were obtained either from parental interviews or from retrospective video analyses. Our own studies on individuals with typical RTT or the relatively milder preserved speech variant (PSV) revealed speech-language and socio-communicative impairments already during the first two years of life and before the onset of regression [6, 8–12]. These results were benchmarked against what is commonly known about early development in these domains but there are no studies directly comparing socio-communicative forms (i.e. behaviours that the child uses to express oneself, e.g. body movements, vocalizations) or functions (i.e. the use of various forms to e.g. request an object, comment on something) of individuals with typical RTT or PSV with normally developing children.

In order to provide a first delineation of socio-communicative development and to document intra-individual profiles as well as inter-individual differences in this domain, we collected and analysed a comprehensive audio-video footage of one female with typical RTT, one with PSV and one typically developing toddler to answer the following questions: (i) Which communicative forms can be observed between 9 and 24 months of age? Are there inter-individual differences? (ii) Which communicative functions are present in the participants' repertoires? How do they change over time? (iii) Are there inter- and intra-individual differences in the development of non-linguistic and (pre)linguistic verbal behaviours?

## 2 Methods

### 2.1 Participants

In this study we included three female participants: participant A with a clinical diagnosis of typical RTT; participant B diagnosed with PSV; and participant C, a typically developing toddler. All participants were singletons; pregnancies and deliveries were uneventful; birth weight, birth lengths, occipitofrontal circumferences, and Apgar scores were within the normal ranges; the participants, all first borns, were from German speaking families of the same socio-economic status. Genetic testing revealed the following pathogenic *MECP2* mutations: p.Arg168\* R168X for participant A; c.378-43\_964delinsG for participant B [13]. Participant A now, at the age of 12 years, communicates primarily through non-verbal behaviours, whereas participant B achieved comprehensive linguistic skills (current age of 15 years); these are reported elsewhere [14]. The study was approved by the local research ethics committees. All parents gave their informed consent to this research and to the publication of the results.

### 2.2 Procedure

The procedure of this study was similar to previous studies conducted on individuals with PSV [10] and typical RTT [8]. Besides comparing two forms of RTT with a typically

developing toddler we here focused on an extended time period from 9 to 24 months of age. Our analysis was based on family video recordings made by the participants' parents who were not aware at that time about their daughters' medical condition. The available footage to analyse socio-communicative forms and functions (total recording time: 1275 minutes; A: 977 minutes; B: 126 minutes; C: 172 minutes) allowed for a splitting into three predefined age intervals across the subjects: 9-12 months (342 minutes); 13-18 months (623 minutes); and 19-24 months of age (310 minutes).

We analysed the footage for the occurrence of (a) socio-communicative forms, such as body movements (e.g. reaching), gestures (e.g. waving), facial expressions/eye movements (e.g. eye contact), non-linguistic vocalizations (e.g. pleasure vocalizations), and (pre)linguistic vocalizations (e.g. canonical babbling) and (b) their respective functions according to the Inventory of Potential Communicative Acts (IPCA) [15]. Communicative forms were coded by the first two authors using the Noldus Observer-XT and assigned to ten functional categories according to IPCA (i.e. social convention, attention to self, reject/protest, requesting an object, requesting an action, requesting information, comment, choice making, answer, imitation) [15]. Sequences with disagreement (7%) were discussed within the team until consensus was achieved.

### 3 Results

#### 3.1 Communicative forms

We observed 27 different communicative forms and classified them as follows: body movements; facial expressions/eye movements; gestures; non-linguistic vocalizations; and (pre)linguistic vocalizations. Developmental trends are provided in Table 1.

#### 3.2 Communicative functions

We observed three different profiles for the participants' fulfilled communicative functions throughout the three age periods: 7/9/9 observed communicative functions for participant C; 3/6/8 for B; and 6/8/6 for A. All participants exhibited some communicative behaviours that represented the three functional clusters 'social convention', 'attention to self' and 'answer' at all age periods. Except for participant B between 9 and 12 months of age, 'commenting' was present in all participants at all age periods. Whereas all individuals showed some 'imitation' behaviour, participant C was the only one to imitate (pre)linguistic vocalizations. A similar result was observed for 'requesting an object' where participants A and B showed relevant behaviours at both age periods in the second year of life whereas C already requested objects in the first year of life. 'Requesting an action' revealed a somewhat different result; participants B and C asked their caregivers to do something during the second year of life whereas A did so only once between 13 and 18 months of age. 'Reject/protest' occurred at all three age periods for participants A and C, but was not observed in B before 19 months. Participant C was the only one to 'request information' and did so in the second year of life. 'Choice making' was not observed in any of the participants throughout the observation period.

### 3.3 Non-linguistic and (pre)linguistic vocalizations

All participants used non-linguistic vocalizations for communicative purposes during all age periods. Over time, the functional use of non-linguistic vocalizations showed different patterns: they decreased in participant C, increased in B and showed an increase-decrease pattern in A (Figure 1a). (Pre)linguistic vocalizations were only observed in participants B and C (Figure 1b).

## 4 Discussion

The Inventory of Potential Communicative Acts appears to be a useful tool for delineating socio-communicative forms and functions in individuals with RTT [8, 10, 16, 17]. Our own studies [8, 10] so far benchmarked results of retrospective video analyses against what is known about normative socio-communicative development facing one limitation of this methodology, the absence of a control group. As a first step to overcome this limitation here we compared socio-communicative profiles of one female with typical RTT (participant A) and one female with PSV (participant B) with a typically developing toddler (participant C). In addition, the possibility to observe the participants over an extended period of time enabled us to document the development of various communicative forms and functions in the context of the different stages of RTT.

As hypothesized, the control toddler had the most comprehensive repertoire of communicative forms whereas the female with RTT had the smallest one at the end of the second year of life. It was interesting to observe, however, that the female with RTT showed more overall communicative forms compared to the female with PSV earlier in development (Table 1). If we only consider gestures and (pre)linguistic vocalizations, the developmental profile is as follows: C>B>A. This reflects previous reports about restricted gestural repertoires as well as the lack of attaining early speech-language milestones in RTT and PSV [6, 7, 10, 12, 18]. It is interesting to note that the female with RTT did not use her index finger to point, a communicative form that is considered to be essential for further speech-language and communicative development that typically emerges at the end of the first year of life [19]. Indeed, she neither pointed nor used (pre)linguistic vocalizations throughout the observation period. Participants B and C on the other hand used both forms to express wants and needs at all times.

The quantitative distribution of acquired communicative functions revealed the following profiles: an increasing number of functions over time for participants B and C (even though B expressed fewer functions compared to A during the first two time periods) and an increase-decrease pattern for participant A, the female with RTT. Similarly, as reported in Didden et al. [16], even though this 2010 study involved individuals who were at different ages than the present participants, ‘social convention’, ‘commenting’, ‘answering’ and ‘requesting’ were the most prevalent communicative functions observed. In contrast to Didden et al.’s study [16], ‘attention to self’ was also present at all ages; ‘requesting information’ was only observed in the control toddler and ‘choice making’ did not occur at all.

For non-linguistic vocalizations developmental profiles were as follows: Again, for the female with PSV (B) the number of categories observed increased; for the female with RTT (A) we observed an increase-decrease pattern; the control toddler (C) showed a decrease in the use of non-linguistic vocalizations and a simultaneous increase in (pre)linguistic vocalizations used for communicative purposes (Figure 1a and 1b). The latter might be interpreted as reflecting growing speech-language capacities. In contrast, functional categories achieved through non-linguistic vocalizations and (pre)linguistic vocalizations both increased in the female with PSV.

Our study documents intra-individual developmental changes and highlights inter-individual differences in socio-communicative development in RTT and PSV. Especially when it comes to delineate communicative gestures and (pre)linguistic vocalizations used as communicative forms, the differences between RTT and PSV appeared to be more salient. Interestingly, the pattern C>B>A seems to be robust for gestures and (pre)linguistic vocalizations even though we had the by far longest recording for participant A (6 or 8 times longer compared to C or B). She did not show index finger pointing or any (pre)linguistic vocalizations in the given data set and she had the same increase-decrease developmental profile for (a) communicative forms, (b) communicative functions and (c) non-linguistic vocalizations. Her profile likely reflects the onset of regression [3, 5]; this is in contrast to the profile of the female with PSV whose regression was reported to begin after her second birthday [14].

Even though promising, our findings have to be interpreted in light of methodological limitations of retrospective video analysis [20, 21]. Still, this is the first study to directly compare RTT profiles with a typical one, but these findings are preliminary and do not allow for generalization. Also, we have to interpret the data with some caution as the available footage only allowed us to analyse the data in age blocks, consisted of recordings of different lengths and took place in different communicative settings [20, 21]. Also, the IPCA itself needs some adaptations when used together with retrospective video analysis in terms of categorizing age-specific phenomena as well as addressing the issue whether certain behaviours can properly be assessed through family videos; e.g. ‘choice making’ could not be seen in the whole data set.

In conclusion, our study provides an important contribution to the growing knowledge about early developmental abnormalities in RTT. The preliminary evidence of different and potentially specific socio-communicative profiles for typical RTT and PSV is conceptually appealing but needs further detailed studies and empirical evidence.

## Acknowledgement

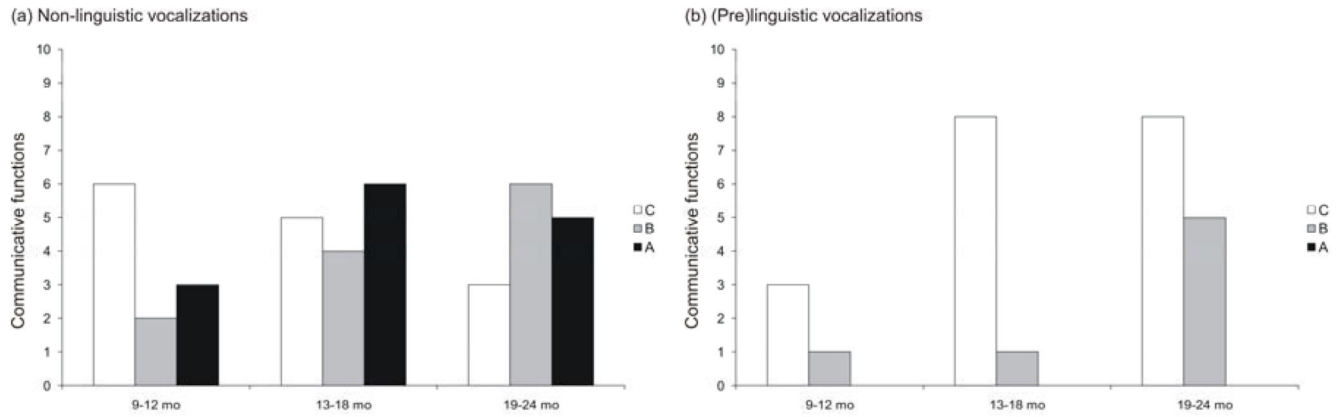
We would like to thank all parents for providing their videos. The study was supported by the Austrian Science Fund (FWF; P25241), COST Action BM1004, Country of Styria, and the Lanyar Foundation (P325, P337, P374).

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**Figure 1.**

(a). Number of communicative functions according to the Inventory of Potential Communicative Acts (IPCA) [15] fulfilled through non-linguistic vocalizations by a typically developing female (C), a female who was later diagnosed with the preserved speech variant (B) and a female later diagnosed with typical Rett syndrome (A) between 9 and 24 months of age; (b). Number of communicative functions according to the IPCA [15] fulfilled through (pre)linguistic vocalizations.



Table 1

Observed communicative forms of a typically developing female (C), a female who was later diagnosed with the preserved speech variant (B) and a female later diagnosed with typical Rett syndrome (A) between 9 and 24 months of age; ● indicates the presence of a communicative form; □ indicates that a communicative form could not be observed.

Communicative forms	C			B			A		
	9-12 mo	13-18 mo	19-24 mo	9-12 mo	13-18 mo	19-24 mo	9-12 mo	13-18 mo	19-24 mo
Body movements									
Moving/turning away	□	●	□	□	□	●	●	□	●
Moving closer	□	●	●	□	●	●	●	●	□
Taking person somewhere (by hand)	□	□	●	□	□	□	□	□	□
Touching (person)	□	●	●	●	□	●	□	□	●
Reaching	●	●	●	□	●	●	●	●	●
Moving object away	●	□	●	□	□	□	□	□	□
Retaining object	●	□	□	□	□	□	□	□	□
(Imitation of) manual routine	●	●	●	□	□	●	●	●	□
Facial expressions/Eye movements									
Eye contact	●	●	●	●	●	●	●	●	●
Smiling	●	●	●	□	●	●	●	●	●
Index finger pointing	●	●	●	●	●	●	□	□	□
Waving indicating hello/bye bye	●	●	●	□	●	□	●	●	□
Extending arms seeking comfort	□	●	●	□	□	●	□	●	□
Demonstrating an object	□	●	□	□	●	□	□	□	□
Shaking the head indicating no	□	□	●	□	□	●	□	□	□
Passing an object	□	●	□	□	□	□	□	●	□
Please/I want	●	●	●	□	□	□	□	□	□
Sending kisses	□	□	□	□	□	●	□	□	□
Non-linguistic vocalizations									
Fussing	●	●	●	□	□	●	□	□	●
Crying	●	●	□	□	□	□	□	□	●
Pleasure vocalizations	●	●	●	□	●	□	□	□	●
Laughing	●	●	●	□	□	●	□	□	●

		Communicative forms			C			B			A		
	Unspecified vocalizations	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
(Pre)linguistic vocalizations	Babbling (canonical and variegated)	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	Onomatopoeics	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	(Proto-)words	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	Word combinations	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
	Total: Communicative forms per age period	16	19	20	6	10	15	11	13	10	15		
	Total: Different communicative forms	26			18			15					