Research in Developmental Disabilities 34 (2013) 3133–3138

Contents lists available at SciVerse ScienceDirect

Research in Developmental Disabilities

Early socio-communicative forms and functions in typical Rett syndrome

Katrin D. Bartl-Pokorny [a,1], Peter B. Marschik [a,1], Jeff Sigafoos [b,*], Helen Tager-Flusberg [c], Walter E. Kaufmann [d], Tobias Grossmann [e], Christa Einspieler [a]

[a] Institute of Physiology (Research Unit iDN-interdisciplinary Developmental Neuroscience; IN:spired), Center for Physiological Medicine, Medical University of Graz, Austria
[b] Victoria University of Wellington, Wellington, New Zealand
[c] Department of Psychology, Boston University, Boston, USA
[d] Boston Children's Hospital and Harvard Medical School, Boston, USA
[e] Max-Planck Institute for Human Cognitive and Brain Sciences, Leipzig, Germany

ARTICLE INFO

Article history:
Received 26 June 2013
Accepted 26 June 2013
Available online 24 July 2013

Keywords:
Family videos
Home videos
Infants
Retrospective analysis
Rett syndrome
Socio-communicative development
Video analysis

ABSTRACT

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.

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

* Corresponding author at: School of Educational Psychology, Victoria University of Wellington, P.O. Box 17-310, Karori 6147, Wellington, New Zealand. Tel.: +64 4463 9772.
E-mail address: jeff.sigafoos@vuw.ac.nz (J. Sigafoos).
[1] These authors contributed equally to the conduct of the study and writing of this manuscript.

0891-4222/$ – see front matter © 2013 Elsevier Ltd. All rights reserved.
http://dx.doi.org/10.1016/j.ridd.2013.06.040

(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