Peculiarities in the gestural repertoire: An early marker for Rett syndrome?

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ABSTRACT

We studied the gestures used by children with classic Rett syndrome (RTT) to provide evidence as to how this essential aspect of communicative functions develops. Seven participants with RTT were longitudinally observed between 9 and 18 months of life. The gestures used by these participants were transcribed and coded from a retrospective analysis of a video footage. Gestures were classified as deictic gestures, play schemes, and representational gestures. Results of the analysis showed that the majority of gestures observed were of deictic character. There were no gestures that could be classified as play schemes and only two (e.g., head nodding and waving bye bye) that were coded as representational or symbolic gestures. The overall repertoire of gestures, even though not necessarily delayed in its onset, was characterized by little variability and a restricted pragmatic functionality. We conclude that the gestural abilities in girls with RTT appear to remain limited and do not constitute a compensatory mechanism for the verbal language modality.

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1. Introduction

Rett syndrome (RTT) is a severe neurodevelopmental disorder that generally affects females and is mainly caused by mutations in the X-linked MECP2 gene (Amir et al., 1999; Neul et al., 2010). It is associated with severe intellectual disability, autistic-like behavior, communicative restrictions and difficulties in hand use coinciding with specific stereotyped movements such as hand-wringing or washing-like movements (Carter et al., 2010; Cass et al., 2003; Hagberg, Aicardi, Dias, & Ramos, 1983; Kaufmann et al., 2012; Kerr, Archer, Evans, & Gibson, 2006; Matson, Fodstad, & Bojsjoli, 2008; Neul et al., 2010). One of the necessary criteria for classic RTT is a recognizable regression that is followed by a period of recovery or stabilization (Neul et al., 2010). Regression is defined by a loss of previously acquired skills, specifically spoken language and purposeful hand use. The general development before regression was initially believed to be asymptomatic, but recent
research has provided mounting evidence of subtle abnormalities in the motor, speech-language and communicative repertoires during the first year of life (e.g. Burford, 2005; Einspieler, Kerr, & Prechtl, 2005a,b; Kerr et al., 2006; Leonard & Bower, 1998; Marschik, Einspieler, Oberle, Laccone, & Prechtl, 2009; Marschik, Einspieler, & Sigafoos, 2012; Marschik, Kaufmann, et al., in press; Marschik, Pini, et al., 2012; Tams-Little & Holdgräfer, 1996).

The recent description of early peculiarities in the speech-language and motor domain have contributed to a better understanding of how MECP2 deficiencies influence early (functional) brain development, and consistent additions to this body of knowledge might lead to a more timely diagnosis. As diagnosis to date is usually made at a mean age of 3 years and usually after the onset of regression (Fehr et al., 2011; Laurivick et al., 2006), there are limited possibilities to study the pre-regressional development (Marschik & Einspieler, 2011). Retrospective video analysis and retrospective questionnaires (parental interviews) are currently the methods of choice to track down early developmental peculiarities in speech-language and communicative development during this period. Questionnaires have proven to be a valuable source of documenting various concurrent behaviors including speech-language functions in children with developmental disabilities (Charman, Drew, Baird, & Baird, 2003; Luyster, Lopez, & Lord, 2007). However, retrospective assessments of communicative functions must be interpreted more cautiously when considering the following factors affecting reliability: (a) long time lag between the interview/questionnaire and period of interest; (b) memory bias of parents with affected children; and (c) the lack of parental training in the observation of linguistic or cognitive skills (Einspieler, Widder, Holzer, & Kenner, 1988; Marschik & Einspieler, 2011; Marschik, Einspieler, Garzaroli, & Prechtl, 2007). Consequently, the best available method for obtaining a detailed description of various developmental domains in children with late-clinical-onset disorders is the retrospective video analysis of non-standardized home movies (Einspieler et al., 2005a; Maestro et al., 2001; Marschik & Einspieler, 2011; Ozonoff et al., 2011; Palomo, Belinchón, & Ozonoff, 2006; Saint-Georges et al., 2010).

Given the methodological restrictions of assessing developmental profiles during the pre-regression period in RTT, there are only a few studies that report on early speech-language and communicative functions. The scaffolding function of early language capacities that precede and predict later ones is the reason why it is of high relevance to focus on this critical period of development in girls with RTT.

Increased language comprehension abilities appear to be a route to productive language abilities, and communicative gestures appear to facilitate this transition (Capone & McGregor, 2004; Iverson & Goldin-Meadow, 2005; Volterra & Eting, 1990). For example, in the first year of life, typically developing children begin to communicate through vocalizations, eye gaze, and gestures to express their needs and desires (e.g. Bates, Camaioni, & Volterra, 1975; Karmiloff & Karmiloff-Smith, 2001; Stone, Ousley, Yoder, Hogan, & Hepburn, 1997; Trevarthen & Hubley, 1978). Gestural development usually begins with the use of deictic gestures or pre-linguistic gestures (or performatives; e.g. showing, giving, pointing, reaching out) around 10–12 months of age. Performatives are usually observed a month before the first words (Bates, Benigni, Bretherton, Camaioni, & Volterra, 1979; Bates et al., 1975; Goodwyn & Acredolo, 1993) and followed by play schemes where children depict an object in terms of its functions. Representational (sometimes referred to as symbolic) gestures usually emerge before the 25-word milestone (Acredolo & Goodwyn, 1988; Capone & McGregor, 2004), and can be differentiated from play schemes in that the referent is not manipulated (e.g., using the hand to symbolize a flying airplane) and they do not change with context. Representational gestures are often complementary to spoken words (Iverson, Capirici, & Caselli, 1994), and as gestures and vocalizations simultaneously appear they increase the saliency of communicative acts that facilitate interpretations by the caregiver. This in turn increases parental responsiveness to communication (Karmiloff & Karmiloff-Smith, 2001; Vallotton, 2009; Yoder, Warren, Kim, & Gazdag, 1994).

Only a few studies have so far dedicated their focus of interest to the development of gestures in RTT. Lavás, Slotte, Jochym-Nygren, Van Doorn, and Witt-Engerström (2006) reported that, among a cohort of 125 girls with RTT, 50% were able to use eye-pointing, index finger pointing, or other gestures, without further specifying the latter. Pre-regressional gesture use was first described by Tams-Little and Holdgräfer (1996) by means of parent-completed questionnaire. They also focused on forms and functions of gestures and speculated about having discovered an early marker for RTT. Our own studies on females with the preserved speech variant (PSV) of RTT, a milder variant with relatively better speech-language abilities, revealed restricted repertoires of their socio-pragmatic functions and communicative gestures during the second year of life after a period with abnormal inspiratory vocalizations (i.e. proto-vowel or proto-consonant alternations produced on ingressive airstream: Marschik, Einspieler, et al., 2012; Marschik, Kaufmann, et al., in press; Marschik, Pini, et al., 2012).

Because our previous studies of individuals with PSV revealed a restricted repertoire of intentional gestures (Marschik et al., 2009; Marschik, Kaufmann, et al., in press) we were curious as to how this essential aspect of communicative functions develops in individuals with classic RTT. In order to obtain a better understanding of the development of communicative gestures in females with RTT, we designed the present study to address the following questions: (1) At what age do intentional gestures first occur? (2) What gestures can be observed during the first 18 months of life in individuals with RTT? (3) How complex is the gestural repertoire during this age period? and (4) How can gestures be categorized (deictic/proto-symbolic/representational)?

2. Methods

2.1. Participants

The present study focused on the acquisition and composition of the gestural repertoire of seven females with RTT who were longitudinally observed between 9 and 18 months of life. Four were from English speaking families (Cases 1, 2, 3, and 6)
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