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Profiling early socio-communicative development in five young girls with the preserved speech variant of Rett syndrome

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ABSTRACT

Rett syndrome (RTT) is a developmental disorder characterized by regression of purposeful hand skills and spoken language, although some affected children retain some ability to speech. We assessed the communicative abilities of five young girls, who were later diagnosed with the preserved speech variant of RTT, during the pre-regression period (aged 12–24 months). Videotapes, obtained by parents during routine family situations and celebrations, were analyzed to identify communicative forms and functions used by these toddlers. Non-verbal communicative forms dominated over verbal-communicative forms for six of the eight identified communication functions. Although the girls used various non-verbal forms to make requests, for example, none of the individuals were observed to make choices or request information. Early peculiarities in the speech-language domain during the first year of life became more prominent and evident during the second year of life as general differences between typical development and atypical development become more obvious in RTT. These findings highlight the importance of assessing socio-communicative forms and functions at early age in children with RTT. The results suggest that speech-language functions did not appear to play a major role in the children's communicative attempts. We conclude that, even among children with the preserved speech variant, socio-communicative deficits are present before regression and persist after this period.

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1. Introduction

Rett syndrome (RTT) is a neurodevelopmental disorder associated with profound intellectual disability, severe communication impairment, autistic-like behavior, and stereotyped hand movements coinciding with difficulties in purposeful hand use (Carter et al., 2010; Cass et al., 2003; Hagberg, Aicardi, Dias, & Ramos, 1983; Kerr, Archer, Evans, & Gibbon, 2006; Matson, Fodstad, & Boisjoli, 2008; Neul et al., 2010). Since the identification of mutations in the X-linked

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MECP2 gene as the main etiology of RTT (Amir et al., 1999), the phenotypical features and neurobiological mechanisms underlying RTT have become increasingly better understood. Clinical phenotypes and diagnostic criteria were recently refined for classic RTT as well as for the three main variant forms: the early seizure variant (Hanefeld Variant), the congenital variant (Rolando Variant), and the preserved speech variant (PSV, Zappella Variant; Neul et al., 2010). A requirement for the diagnosis of classic RTT and its variants is the presence of a period of regression followed by recovery or stabilization. In classic RTT, these two periods are part of a four-stage course. First, the early period with subtle signs of abnormality is followed by a stage of marked and progressive deterioration that leads to dramatic loss of the ability in adaptive functioning, functional hand use, mobility, language and communicative functions. Following this regression, characteristic hand stereotypies become more prominent and cardinal features like breathing irregularities, seizures, autistic-like behavior, social impairments, unsteady gait, apraxia, and intellectual disability become more evident. During the late deterioration stage reduced mobility, dystonia and scoliosis are some of the prominent features (Cass et al., 2003; Charman et al., 2002; Hagberg et al., 1983; Kerr, 2001; Neul et al., 2010; Percy et al., 2010; Rajaei et al., 2011).

Focusing on the pre-regression period of RTT, our studies have so far contributed to the delineation of early abnormalities in both developing motor behaviors and early verbal behaviors (Einspieler, Kerr, & Prechtl, 2005a, 2005b; Marschik, Einspieler, Oberle, Laccione, & Prechtl, 2009; Marschik, Einspieler, Prechtl, Oberle, & Laccione, 2010; Marschik, Einspieler, & Sigafoos, 2012; Marschik, Lanator, Freilinger, Prechtl, & Einspieler, 2011; Marschik, Pini, et al., 2012). Of special interest is the analysis of early vocalizations in females with PSV, a mild variant of RTT associated with relatively better speech-language abilities. In previous studies of this variant, we observed an intermittent mix of typical and atypical vocalizations from the first months of life onwards. One of the most salient features was an abnormal inspiratory type of vocalization i.e., proto-vowel or proto-consonant alternations produced on ingressive airstream and breathy voice characteristics (Marschik, Pini, et al., 2012). This atypical quality of vocalization was evident to professionals and to naive listeners, thus suggesting that auditory Gestalt perception is a potential contributor for the detection of early deviations in females with RTT (Marschik, Einspieler, et al., 2012).

An extension to these findings and of high relevance to the understanding of speech-language and communicative development, is the study of pre-regressional pragmatic functions and socio-communicative capacities in RTT (e.g., request for an object, comment, choice making, imitation). It has been reported that individuals with RTT use various idiosyncratic behaviors (e.g., eye gaze, non-conventional vocalizations, facial expressions, or body movements) for communicative purposes (Dahlgren Sandberg, Ehlers, Hagberg, & Gillberg, 2000; Sigafoos et al., 2011; Sigafoos, Woodyatt, Keen, et al., 2000; Sigafoos, Woodyatt, Tucker, Roberts-Pennell, & Pittendreigh, 2000). An efficient tool to accurately document these potential communicative forms and functions of children with severe communication impairment, which has also been applied to individuals with RTT during their later stages of development, is the Inventory of Potential Communicative Acts (IPCA; Didden et al., 2010; Sigafoos, Arthur-Kelly, & Butterfield, 2006; Sigafoos, Woodyatt, Keen, et al., 2000; Sigafoos, Woodyatt, Tucker, et al., 2000). However, to the best of our knowledge, the IPCA has not yet been used to document pre-regressional socio-communicative capacities of children with the PSV of RTT. As pre-regressional behavior might be a precursor to post-regressional communicative functions in RTT, we evaluated the development of potential communicative functions of conventional and/or unconventional character during the second year of life of five young girls with PSV. This study addressed the following questions: (a) What, if any, potential communicative acts can be observed during the second year of life in young girls with the PSV of RTT? (b) What pragmatic functions are represented in the girls' communicative acts repertoires? (c) Do young girls with PSV use non-verbal communicative forms more frequently than verbal communicative forms? And (d) How complex is the gestural repertoire during this age period in PSV?

2. Methods

2.1. Participants

Participants were five young girls with PSV who were longitudinally observed from 12 to 24 months of age, before the onset of any RTT associated regression. Four of them came from Italian-speaking families and one participant was German (Case 5). All females were singletons, born as a result of uneventful pregnancies and deliveries. The mean birthweight was 3048 g (SD = 216 g) and birth lengths, occipitofrontal circumferences, and Apgar scores were in the normal range. Genetic testing revealed the following *MECP2* mutations: C468G in one, c.1163del44 in one, R133C in two, and a large intragenic deletion (c.378–43_964delinsGA) in one participant. The motor development of Case 5 and the vocalizations during infancy for all participants were described in Marschik et al. (2009) and Marschik, Pini, et al. (2012). All participants met the clinical criteria for PSV (Neul et al., 2010; Renieri et al., 2009). The study was approved by the local research ethics committees. Parents gave their informed consent to this longitudinal research and to the publication of the results.

2.2. Procedure

The data for this study was extracted from video footage made during typical family routines (e.g., play situations, bathing, feeding, etc.) and special events (e.g., Christmas or birthdays). All videos had been made by the parents, who were not aware at that time that their daughters had RTT. The footage of all five participants comprised a total of 224 min recorded in 223 clips (medians: 33 min; 26 clips). A research assistant naive to the purpose of the study checked the recordings for

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