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Contributing to the early detection of Rett syndrome: The potential role of auditory Gestalt perception

Peter B. Marschik [a], Christa Einspieler [a,*], Jeff Sigafoos [b]

[a] Institute of Physiology (IN:spired, Developmental Physiology and Developmental Neuroscience), Center for Physiological Medicine, Medical University of Graz, Austria

[b] Victoria University of Wellington, New Zealand

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ABSTRACT

To assess whether there are qualitatively deviant characteristics in the early vocalizations of children with Rett syndrome, we had 400 native Austrian–German speakers listen to audio recordings of vocalizations from typically developing girls and girls with Rett syndrome. The audio recordings were rated as (a) inconspicuous, (b) conspicuous or (c) not able to decide between (a) and (b). The results showed that participants were accurate in differentiating the vocalizations of typically developing children compared to children with Rett syndrome. However, the accuracy for rating verbal behaviors was dependent on the type of vocalization with greater accuracy for canonical babbling compared to cooing vocalizations. The results suggest a potential role for the use of rating child vocalizations for early detection of Rett syndrome. This is important because clinical criteria related to speech and language development remain important for early identification of Rett syndrome.

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1. Introduction

Rett syndrome, first described by the Austrian neuropaediatrician Andreas Rett (1966), is a profoundly disabling neurodevelopmental disorder that is almost entirely confined to females (Hagberg, Aicardi, Dias, & Ramos, 1983; Neul et al., 2010). It is considered to be an important etiological factor for severe/profound intellectual disability in females with a prevalence of 1:5000 to 1:10,000 live female births (Laurvick et al., 2006). Mutations in the X-linked gene *MECP2* were identified as the main cause for Rett syndrome (Amir et al., 1999) affecting a wide range of neurodevelopmental functions such as cognitive processes, purposeful hand use, and communicative abilities (Cass et al., 2003; Kerr, Archer, Evans, & Gibbon, 2006; Matson, Fodstad, & Boisjoli, 2008; Neul et al., 2010; Sigafoos et al., 2011). There are also patients with Rett syndrome originating from mutations in other genes (e.g. *FOXG1*, *CDKL5*) and individuals with *MECP2* mutations who show no clinical signs. Therefore, the clinical criteria for diagnosis of this disorder are important for early identification and differential diagnosis.

Apart from the classic Rett syndrome, three main variant forms have been described: the preserved speech variant (PSV, Zappella Variant), the early seizure variant (Hanefeld Variant) and the congenital variant (Rolando Variant; Neul et al., 2010). All forms of Rett syndrome, the classic phenotype and its variants, typically show a four-stage course of development: the

* Corresponding author at: Institute of Physiology, Center for Physiological Medicine, Medical University of Graz, Harrachgasse 21/5, 8010 Graz, Austria. Tel.: +43 316 380 4266; fax: +43 316 380 9630.
E-mail address: christa.einspieler@medunigraz.at (C. Einspieler).

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pre-regression period is followed by an onset of stagnation or regression (at the age of 6–18 months); during the rapid destructive stage (between 1 and 3 years), speech and purposeful hand use are lost and characteristic hand stereotypies become more prominent; the pseudo-stationary stage between pre-school and school age is characterised by cardinal features like breathing irregularities, seizures, autistic-like symptoms, social impairments, unsteady gait, apraxia, and intellectual disability; during the late deterioration stage (at age 15–30 or later), reduced mobility, dystonia and scoliosis are some of the prominent features (Cass et al., 2003; Charman et al., 2002; Dunn & MacLeod, 2001; Hagberg et al., 1983; Kaufmann et al., 2011; Kerr, 2001; Neul et al., 2010; Percy et al., 2010; Percy, 2011; Rajaei et al., 2011).

Although an apparently normal early development had initially been regarded as one of the criteria for classic Rett syndrome, various scientists now considered the disorder to be a developmental disorder that manifests shortly after birth (e.g. Burford, Kerr, & Macleod, 2003; Einspieler, Kerr, & Prechtl, 2005a; Einspieler, Kerr, & Prechtl, 2005b; Marschik, Einspieler, Oberle, Laccone, & Prechtl, 2009; Marschik, Lanator, Freilinger, Prechtl, & Einspieler, 2011a; Temudo, Maciel, & Sequeiros, 2007). A detailed tracking down of parents' suspicions about early maldevelopment was derived from retrospective home movie analysis (Marschik & Einspieler, 2011). Apart from abnormal early motor development in infants with Rett disorder (Einspieler et al., 2005a, 2005b; Marschik et al., 2009) our studies have also contributed to the delineation of early abnormalities in another key clinical feature of Rett syndrome, a deficit in the developing linguo-cognitive domain (Marschik, Einspieler, Prechtl, Oberle, & Laccone, 2010; Marschik et al., 2011a, 2011b). In particular, we observed an intermittent character of typical and atypical vocalizations from the first months of life onwards. One of the most salient features was an abnormal vocalization type of inspiratory character, i.e. proto-vowel or proto-consonant alternations produced on ingressive airstream and breathy voice characteristics (Marschik et al., 2010, 2011b). Based on these observations and the fact that the typical sound repertoire of the infant develops systematically, we decided to focus on the earliest stages of sound productions to evaluate their potential role for the early detection of RTT.

The most intensively investigated early vocalization type is crying, which has been discussed – if it does not decline with growing age – as a marker for later deviations such as aggressive behavior, eating and sleeping difficulties, hyperactivity or even severe developmental disorders such as autism spectrum disorders or Rett syndrome (Bahi-Buisson et al., 2010; Esposito & Venuti, 2010; Forsyth & Canny, 1991; Papousek & Von Hofacker, 1998; Papousek, Wurmser, & von Hofacker, 2001; Von Kries, Kalies, & Papousek, 2006; Wolke, Rizzo, & Woods, 2002). Apart from crying, characteristics of canonical babbling that should emerge by no later than 10 months of age have been discussed as indicators for developmental disabilities (Nathani, Oller, & Neal, 2007; Oller, 1995; Oller, Eilers, Neal, & Schwartz, 1999): children with Rett syndrome, Down syndrome, Williams syndrome, cerebral palsy or profound hearing impairment were reported to enter the babbling-stage with a significant delay (Levin, 1999; Lynch et al., 1995; Marschik et al., 2011b; Masataka, 2001; Nathani et al., 2007; Tams-Little & Holdgrafer, 1998). Our own studies stressed that it is not only the developmental delay, but also rather a qualitatively deviant appearance of early vocalizations which could be essential for the early detection of Rett syndrome (Marschik et al., 2010, 2011b). Based on these findings we were curious as to whether professional- and/or naive-listeners to audio recordings were capable of distinguishing normal from abnormal early vocalizations.

The aims of the study were to address the following issues and questions: (a) Are listeners able to differentiate between qualitatively normal and abnormal early vocal behavior? (b) Is there a difference in the judgements of age-specific vocalizations during the precanonical stage and canonical stage? (c) Is there a difference in the accuracy of judgements between professional-listeners and naive-listeners; between different age groups of listeners; between female and male listeners; and between parents and non-parents?

2. Methods

2.1. Participants

In total 400 native Austrian–German speakers (267 females and 163 males) were enrolled in an audio experiment on the "assessment of early vocalizations" (RTT-audio experiment from there on). The majority of participants was between 19 and 30 years old; $n = 361$; 24 participants were between 31 and 40 years and 15 were over 40 years of age. The majority of participants ($n = 377$) did not yet have any children of their own.

We classified participants into two groups based on their reported professions: First, professional-listeners ($n = 61$) consisted of speech-language therapists ($n = 47$), clinical linguists ($n = 5$), phoniatricians ($n = 4$), and developmental psychologists ($n = 5$). However, none of these professionals had expertise in early vocal development of children with genetic disorders. The second group consisted of naive-listeners ($n = 339$), i.e. medical students ($n = 261$) and students of general linguistics ($n = 78$). None of these individuals had training in speech-language development.

2.2. Procedure

From an extensive collection of video footage, audio files were created of either normal vocalizations (cooing and babbling) from typically developing girls (recordings B, D, E) or the vocalizations of girls with Rett syndrome (recordings A, C, F). The sound files of girls with Rett syndrome corresponded to previously reported abnormal inspiratory vocalizations and sequences with intermittent and rapidly changing character of normal and abnormal vocalization patterns (Table 1; Marschik et al., 2010, 2011b). Furthermore, we selected one vocalization sequence of a girl with Rett syndrome, which had

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