



Rett syndrome: A preliminary analysis of stereotypy, stress, and negative affect



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ABSTRACT

Rett syndrome (RTT) is a neurodevelopmental disorder primarily affecting females. It is characterized by apparently normative development of motor and communicative abilities followed by deterioration in these domains. Stereotypic hand movements are one of the core diagnostic criteria for RTT. There is some anecdotal but limited scientific evidence that changes in hand stereotypy may be a sign of increased anxiety or arousal (i.e., a 'stress response') in RTT. Understanding stress responsivity is difficult in RTT because almost all individuals are nonverbal or otherwise severely communicatively impaired. This study used direct behavioral observation to quantify and compare the frequency of hand stereotypy and signs of negative affect during presumed periods of high and low stress associated with functional analysis conditions (negative reinforcement ['escape'] and control ['free play'], respectively) for 5 females with RTT (mean age = 17.8; range 4–47). Negative affect was more likely to occur during negative reinforcement ('stress') conditions for each participant whereas hand stereotypies did not differ across conditions for any of the participants. Although preliminary, the results suggest that hand stereotypy may not be a valid behavioral 'stress-response' indicator in females with RTT. Alternatively, the approach we used may have been limited and not sufficient to evoke a stress response. Either way, more work with direct relevance to improving our understanding of hand stereotypy and anxiety in RTT in relation to social context appears warranted.

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1. Introduction

Rett syndrome (RTT) is a neurodevelopmental disorder primarily affecting females. The syndrome is typically characterized by a mutation in the methyl-CpG binding protein 2 (*MECP2*) gene with a prevalence rate of 1:10–15,000 female live births. From birth to 6–18 months, girls develop apparently normally, reaching age-appropriate milestones, such as the use of motor skills and some language. With the onset of the disorder, however, there is a loss of much of their adaptive functioning, such as purposeful hand movement, communicative abilities, and in many cases, locomotion. Subsequently, the behavioral characteristics upon which the diagnosis is based emerge including stereotypic hand movements, growth retardation, breathing abnormalities, seizures, gait abnormalities, scoliosis, and thermal dysregulation (Lotan & Ben-Zeev, 2006; Neul et al., 2011).

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There is reason to believe that anxiety and related issues are also common among individuals with RTT. MECP2 deficient mice, which have been developed to recapitulate the core features of RTT, show significantly higher levels of anxiety-like behaviors (Adachi, Autry, Covington III, & Monteggia, 2009; Fyffe et al., 2008; Gemelli et al., 2006; McGill et al., 2006), and provide evidence of pathologically altered stress-response systems. It appears that MeCP2 modulates or otherwise regulates the expression of the corticotrophin-releasing hormone (*Crh*) gene, which is a part of a well-established pathway underlying anxiety (Fyffe et al., 2008; LeDoux, 2000; McGill et al., 2006). Although there has not been comparable work specific to anxiety among individuals with RTT, parents and other caregivers commonly report anxiety and stress-related issues. For example, in one study, 74% of caregivers reported that their daughters experienced fear or anxiety in unfamiliar situations (Mount, Charman, Hastings, Reilly, & Cass, 2002). In another, it was reported that 76% of individuals with RTT exhibited brief episodes of anxiety-like behavior, and these behaviors were reported to be precipitated by external events (Sansom, Krishnan, Corbett, & Kerr, 1993).

Although it is widely assumed that individuals with RTT have problems with anxiety (Lotan & Ben-Zeev, 2006), identifying if and when anxiety is being experienced is difficult, because of severe communicative impairments and therefore limited access to an emotional state (i.e., self-report is compromised; Didden et al., 2010). Most of the available information regarding emotional states among individuals with RTT is based on proxy reports such as rating scales or questionnaires, which is similar to, but not identical to, measuring symptoms. Another approach to examining anxiety among individual with RTT may be to use direct observation – which would be similar to measuring signs. To our knowledge, only two studies have utilized direct observation to assess behavioral or emotional states among individuals with RTT (Bergstrom-Isacsson, Lagerkvist, Holck, & Gold, 2013; Woodyatt, Marinac, Darnell, Sigafos, & Halle, 2004). The results of these studies indicate that, although it is possible for trained observers to identify facial expressions and other indications of emotional/behavioral regulation in this population, agreement regarding the meaning or intention of these behaviors is generally poor, and individualized operational definitions of behavioral indications are necessary.

Considering the current state of knowledge, it is possible that caregivers of individuals with RTT may be over- or under-identifying possible sign of anxiety. Additional research is clearly needed to identify the specific behavioral indications of stress or anxiety among individuals with RTT. One possible indicator is stereotyped hand movements. Although this type of movement is one of the defining characteristics of RTT, it has been suggested that hand stereotypies may increase or intensify in response to stress or arousal (Temudo et al., 2008). In fact, one study reported differences in the percent of time that individuals with RTT engaged in hand stereotypies across social and non-social activities, although the specific patterns differed across participants (Sigafos, Woodyatt, Tucker, Roberts-Pennell, & Pittendreigh, 2000). Similarly, Hetzroni and Rubin (2006) reported that females with RTT exhibited increased hand stereotypy in response to interruptions of familiar activities like watching television or listening to music, and Stasolla and Caffo (2013) reported decreases in hand stereotypy, along with increases in indices of happiness, associated with structured activities and environmental stimulation. In all of these cases, the observed differences may have been due to changes in anxiety or arousal. Another possibility, however, is that hand stereotypies decrease when the individual is engaged with or oriented to an ongoing activity (Fabio, Giannatiempo, Antonietti, & Budden, 2009). Despite the clinical implications of the assumption that hand stereotypies are indicative of a state of anxiety among individuals with RTT, the relationship between stereotyped hand movements and stress has not been systematically studied in this population.

The purpose of this exploratory study was to build on the existing literature by testing the hypothesis that individualized behavioral indicators of negative affect (such as whining, and negative facial expressions) and the percent of time engaged in hand stereotypy would differ across controlled environmental conditions that were expected to be relatively stressful or non-stressful in a small consecutively enrolled clinical sample of females with classic RTT.

2. Methods

2.1. Participants

Five females (mean age = 17.8; range 4–47) with a clinical diagnosis of classical Rett Syndrome who met the core criteria defined by Neul et al. (2011), participated in the study. Participants were recruited through the Minnesota Rett Syndrome Research Association. The University of Minnesota Institutional Review Board approved the study and parents or legal guardians provided informed consent. Participants of all ages and levels of physical functioning were included in the study except those with a recent onset of seizure disorder. All of the participants were nonverbal. SG was the only participant to use an augmentative communication device (Tobii C12 eye tracker). SG's parents reported her using the device appropriately during activities such as mealtimes, but no formal assessments of language abilities were conducted. None of the other participants had any formal means of communication. The participant's motor skills varied greatly: Three participants (ZC, SG, and JS) were ambulatory, whereas two (ML and KP) could not walk independently. Three (ML, KP, and JS) had little to no functional hand use, and two (ZC and SG) retained some skills, including the ability to pick up toys and feed themselves. Three of the participants (ML, KP, and JS), had histories of seizure disorders. One participant (ZC) lived in a group home, while the other four participants lived at home with their parents or legal guardians.

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