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The impact of familial autism diagnoses on autism symptomatology in infants and toddlers

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ABSTRACT

Debate regarding the etiology of Autism Spectrum Disorders (ASD) is on the rise with numerous theories being put forth. Currently, the theory with the most empirical support is the interaction of multiple genes. Many studies have provided evidence that as the incidence of ASD increases so do genetic similarities. However, very little research has focused on the presentation of ASD symptomatology in those individuals with or without ASD diagnoses who have biological relatives with or without ASD diagnoses. The aim of the current study was to first examine the percentage of toddlers with and without ASD who had biological relatives with ASD. Next, the impact familial ASD had on ASD symptomatology within infants and toddlers with and without diagnoses of ASD was investigated. In the first study, 438 toddlers with an ASD diagnosis and 1,071 who were atypically developing without an ASD diagnosis were examined. A greater percentage of toddlers with ASD were noted to have a biological relative with an identified ASD in comparison to atypically developing toddlers. In the second study, no significant differences emerged between groups dependent on familial ASD of symptoms of autism as measured by the *Baby and Infant Screen for Children with aUtIsm Traits* (*BISCUIT*). As such, despite previous research indicating a strong genetic link to ASD, this link is undoubtedly complex and not necessarily related to ASD symptomatology. Suggestions for further research are provided.

1. The impact of familial autism diagnoses on autism symptomatology in infants and toddlers

Autism Spectrum Disorders (ASD) share overlapping behavioral characteristics including deficits in socialization and communication, and excesses in repetitive behaviors and restricted interests (Cederlund, Hagberg, & Gillberg, 2010; Gillis, Callahan, & Romanczyk, 2011; Kopp, Beckung, & Gillberg, 2010; Lacroix, Guidetti, Rogé, & Reilly, 2009; Matson, Dempsey, & Fodstad, 2009a; Matson, Boisjoli, Hess, & Wilkins, 2010a; Matson & Wilkins, 2007, 2009; Oeseburg, Groothoff, Dijkstra, Reijneveld, & Jansen, 2010; Sevlever & Gillis, 2010; Smith & Matson, 2010c; Wing, Gould, & Gillberg, 2011). Although an extensive amount of research has been conducted on ASD, the etiologies of the disorders comprising the spectrum are relatively unknown (Matson, Dempsey, & Fodstad, 2009; Smith & Matson, 2010a, 2010b). Only about 10% of ASD cases are nomothetic, with the remaining percentage resulting from an unknown origin or cause (Limprasert, 2008). Many different etiologies have been linked to ASD, but lack empirical support or have been discounted (e.g., Measles, Mumps, and Rubella vaccination [MMR]; Nebel-Schwalm & Matson, 2008). To date, the most consistently supported link to ASD is the interaction

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of multiple genes (Cook, 1998; Mazefsky, Goin-Kochel, Riley, & Maes, 2008; Muhle, Trentacoste, & Rapin, 2004; Nebel-Schwalm & Matson, 2008).

Twin and sibling studies have provided support for the genetic causes of autism. In regards to twin studies, monozygotic (MZ) twins show a much higher concordance rate for ASD when compared to dizygotic (DZ) twins. More specifically, concordance rates for MZ and DZ twins range from 36% to 91% and 0% to 10%, respectively (Bailey et al., 1995; Folstein & Rutter, 1977; Muhle, Trentacoste, & Rapin, 2004; Steffenburg et al., 1989). When investigating the broader ASD phenotype and not actual ASD diagnoses, these percentages increase for both MZ and DZ twins (Bailey et al., 1995). As a result of these findings, it is hypothesized that the cause of ASD is one of multiple, not single, genetic interactions (Muhle et al., 2004). If a single gene could be linked to the cause of ASD, then the concordance rate of ASD between DZ and MZ twins would be more similar.

Sibling studies have also yielded concordance rates for ASD. For instance, after conducting a review and aggregating the results of family studies together, the risk of a sibling having ASD was reported at 2.2% (Szatmari, Jones, Zwaigenbaum, & MacLean, 1998). Other researchers, however, have reported higher rates ranging from 4 to 8% (Chakrabarti & Fombonne, 2001; Muhle et al., 2004). Additionally, Szatmari and colleagues' (1998) review revealed that the rate of concordance in family members decreases when the family members were second- and third-degree relatives, as opposed to only first-degree relatives.

In addition to the genetic implications of autism, studies on autism symptoms and birth order have also been conducted. For example, Reichenberg, Smith, Schmeidler, & Silverman, 2007 conducted an investigation aimed at examining symptom profiles of autism between first and second born children within multi-incidence families. Children born second within multi-incidence families had significantly worse useful phrase speech. However, first born children had significantly worse repetitive behaviors. Thus, even within a family, heterogeneity of symptoms and severity differences appear to exist.

In sum, twin and family studies have demonstrated genetic links to ASD. Given that the broader phenotype of ASD has higher concordance rates than concordance rates of actual ASD diagnoses, would symptom severity be greater for those within multi-incidence families? Research on the differences in severity of ASD symptomatology between individuals with and without family members (i.e., first- or second-degree) diagnosed on the autism spectrum has not been investigated, and therefore was the aim of the current study. Additionally, prior to exploring ASD symptomatology between individuals with and without family members diagnosed with an ASD, a preliminary examination was completed to determine the percentage of toddlers with and without an ASD who had a biological family member diagnosed with an ASD.

2. Study 1

2.1. Method

2.1.1. Participants

A total of 1,509 toddlers, ages 17 through 37 months, were selected for inclusion in this study from a total sample of 2,222 toddlers. All of the toddlers received services through EarlySteps, which is Louisiana's Early Intervention System under the Individuals with Disabilities Education Act, Part C, which provides services to infants and toddlers and their families from birth to 36 months. Children qualify for EarlySteps if they have a developmental delay or a medical condition likely to result in a developmental delay. Those selected for inclusion in the study had provided information regarding whether or not a biological family member had been diagnosed with an ASD. All 1,509 toddlers were assigned diagnoses of ASD or non-ASD based on currently used methodologies including reference to the *DSM-IV-TR* criteria (American Psychiatric Association [APA], 2000), *Modified Checklist for Autism in Toddlers* scores (Robins, Fein, Barton, & Green, 2001), and developmental scores on the *Battelle Developmental Inventory, Second Edition* (*BDI-2*; Newborg, 2005). Interrater reliability of diagnoses was obtained by a second doctoral level psychologist for a subset of participants within the entire sample ($n = 195$), who had several years of experience assessing and treating children with developmental disabilities. Identical diagnostic methodologies were used. Interrater reliability for diagnoses was excellent with a kappa value of .94, $p < .001$. Within the current sample, 438 participants were diagnosed as having an ASD while the remaining 1,071 were classified as atypically developing (i.e., non-ASD). Demographic characteristics for all participants are presented in Table 1.

2.2. Measure

2.2.1. Baby and infant screen for children with autism traits (BISCUIT) demographic form

The *BISCUIT* Demographic Form is administered by an interviewer to the parent/caregiver of the toddler. Interviewers record the parent/caregiver's responses with respect to the infant's demographic information: name, date of birth, gender, ethnicity, birth weight, current weight, and height. The demographic form also inquires about information related to the child's developmental history and current status: parental concerns regarding their child's development, child's age when parents' first concern regarding development was noted, age at which various developmental milestones were attained, current medical diagnoses, and current medications. Additionally, informants are asked to indicate if any of the child's family