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# Associations between ASD Symptomatology and Cognitive Functioning in Siblings

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UNIVERSITY OF MIAMI

ASSOCIATIONS BETWEEN ASD SYMPTOMATOLOGY AND COGNITIVE  
FUNCTIONING IN SIBLINGS

By

Susan I. Acosta

A DISSERTATION

Submitted to the Faculty  
of the University of Miami  
in partial fulfillment of the requirements for  
the degree of Doctor of Philosophy

Coral Gables, Florida

December 2009

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FUNCTIONING IN SIBLINGS

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Siblings of children with autism (ASD-sibs) often exhibit deficits in social reciprocity and cognitive deficits similar to those of their affected siblings. The purpose of this study was to examine the associations between degree of autistic symptomatology and degree of cognitive functioning in sibling pairs seen as part of a longitudinal ASD-focused sibling study. Both cognitive functioning and autistic symptomatology were assessed using continuous measures in sibling pairs. Three sets of bivariate correlations were conducted to examine the relationships between autistic symptomatology and cognitive ability. One ANCOVA and 6 ANOVAS were also conducted to identify possible group differences between younger siblings of children with diagnoses of ASDs (ASD-sibs) and younger siblings of children without diagnoses of ASDs (COMP-sibs). When associations were examined in the entire sample, all correlations examined were significant,  $p < .05$ . However, when examined by group, no associations between younger and older siblings were significant. Negative correlations were found between ASD symptomatology and cognitive functioning within the younger sibling, and between ASD symptomatology and cognitive functioning within the older sibling. Thus, within the ASD group, level of autistic symptomatology was negatively associated with level of cognitive functioning with individuals. Results indicate that intellectual disability (i.e.,

impaired cognitive functioning) runs in concert with symptomatology among children with ASDs and among their younger siblings. Additionally, by three years of age, ASD-sibs were receiving lower scores than COMP-siblings in the areas of receptive language, expressive language, and in visual reception. Clinically, the identification of specific limitations in ASD-sibs has important implications for intervention programs which could help to prevent or ameliorate poor outcomes.

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## Chapter 1: Introduction

### Associations between ASD symptomatology and cognitive functioning in siblings

Autism spectrum disorders (ASDs) are characterized by impairment in communication skills and social interaction, and the presence of restricted, repetitive and stereotyped patterns of behavior (American Psychiatric Association [APA], 1994). Children with ASDs vary in their degree of impairment in these domains. Deficits in a variety of cognitive domains also are frequently associated with these disorders (Ozonoff, McMahon, Coon, and Lainhart, 2002; Fombonne, Bolton, Prior, Jordan, & Rutter, 1997, etc.). Siblings of individuals with autism (ASD-sibs) often exhibit deficits in social reciprocity (Constantino et al, 2006; Landa & Garrett Mayer, 2006; Zwaigenbaum et al., 2005), as well as cognitive deficits similar to those of their affected siblings (Yirmiya et al., 2006; Ozonoff, McMahon, Coon and Lainhart, 2002; Plumet, Goldblum, & Leboyer, 1995).

The purpose of this study was to examine the associations between degree or level of autistic symptomatology and degree or level of cognitive functioning in sibling pairs seen as part of a longitudinal ASD-focused sibling study. A secondary goal was to determine what differences, if any, exist between siblings of ASD children (ASD-sibs) and comparison siblings (COMP-sibs) in the level of ASD symptomatology, level of cognitive functioning, and related deficits. In the present study, both cognitive functioning and autistic symptomatology were measured in sibling pairs. However, unlike in previous studies, both cognitive functioning and autistic symptomatology were assessed using continuous measures in sibling pairs.

*Genetic component of autism and at-risk ASD-sibs.* ASDs have a strong genetic basis. In identical twin pairs, the concordance rate for autism is 60% and the concordance rate for a disorder on the autism spectrum is more than 90% (Veenstra, Vanderweele & Cook, 2003). Also, among twin pairs, Bailey et al. (1995) found that 92% of monozygotic twins and 10% of dizygotic twins were concordant for cognitive and/or social deficits.

The recurrence risk for ASD in younger siblings of children with ASD has been estimated to be 6-8% (Piven et al., 1997), and the prevalence of the broader autism phenotype in siblings has been reported to be as high as 20% (Bolton, Macdonald, Pickles, & Rios, 1994). However, recently published prospective studies of younger siblings have revealed substantially higher rates of ASD, ranging from 29% (19/65; Zwaigenbaum et al, 2005) to 37% (22/60; Landa & Garrett-Mayer, 2006). Though ASD-sibs have an increased risk of meeting criteria for an ASD, even those who do not meet criteria may manifest impairments involving subtle ASD-linked deficits, such as language delays, difficulties with sensory integration, and potential difficulties with emotion regulation and communication (Yirmiya et al., 2006, Landa & Garrett-Mayer, 2006; Zwaigenbaum et al., 2005). Recent studies have examined social and communication difficulties in ASD-sibs, as deficits in these areas are among the best predictors for a diagnosis of autism (Yirmiya et al., 2006). ASD-sibs are more likely than their peers to display subthreshold levels of autistic social impairment (Constantino et al, 2006; Yirmiya et al, 2006 ). Twenty percent of full siblings of autistic individuals who do not receive an ASD diagnosis present with language delay or behavioral inflexibility and/or inhibition by 24 months of age (Zwaigenbaum et al, 2005).

Subsequent research focusing on sibling concordance for level of cognitive functioning in autism has been revealing. Szatmari et al. (1996) used Intraclass Correlation Coefficients to compare measures of intelligence, social and communication skills, and autism characteristics among 23 multiplex families. They found high correlations for all domains measured, ranging from 0.57 to 0.62. Additionally, MacLean et al. (1999) found that, in sibling pairs in which both had an ASD diagnosis, nonverbal IQ and adaptive behaviors in socialization and communication were all significant at  $p < .01$  and in the moderate range (0.40-0.50).

The current study assessed autistic symptomatology and cognitive functioning along a continuum of severity in a group of older diagnosed ASD-children and their younger siblings. Previous studies have examined associations between level of ASD symptomatology within twin pairs (MacLean et al, 1999) or in multiplex families in which all members of sibling groups had ASD diagnoses (Szatmari et al, 1996). No study previously has examined associations in ASD symptomatology and cognitive functioning among younger siblings of affected children.

*ASDs and Cognitive Functioning.* The long-term prognosis for any given child with an ASD is largely based on the joint impact of the severity of expression of ASD symptomatology and his or her level of general intelligence (Coplan, 2003). Individuals with ASDs exhibit varying levels of functioning within the cognitive domain. Though the majority of individuals with autism have traditionally fallen below a standard score of 70 (Fombonne, 1999; Rutter, Bailey, Bolton, & Le Couter, 1994), they have also demonstrated a wide range in their IQ profiles (Ghaziuddin & Mountain-Kimchi, 2004). Fein, Pennington, Markowitz, Braverman, and Waterhouse (1986) found that while the

majority of individuals with autism assessed scored lower in verbal than performance tasks, 20% showed no discrepancy between the two areas. With respect to the verbal domain of cognitive development, approximately half of young children with autism fail to acquire speech as their primary mode of communication (Prizant, 1996). Despite the fluctuation in cognitive profiles, young autistic children, even if verbal, almost universally have comprehension deficits, in particular deficits in understanding higher order complex questions (Klin et al., 2004). The current study examined the cognitive functioning of older diagnosed ASD-children, additionally assessing whether performance on subtests of Verbal and Nonverbal functioning was associated with level of ASD symptomatology. It was hypothesized that a given child's level of autistic symptomatology would be associated negatively with that child's level of cognitive functioning. It was also hypothesized that level of ASD symptomatology would be specifically associated with reduced performance on the Verbal and Nonverbal subtests of the cognitive measures.

*Cognitive Functioning in Siblings.* The intelligence scores of older and younger typically-developing siblings are consistently associated, with an average correlation of 0.49 (Elbedour, Bouchard, & Hur, 1997; McCall, 1970). Previous studies of cognitive functioning in ASD populations have reported associations of verbal and nonverbal IQ in children with ASDs and their affected siblings. Specifically, when examining multiplex families in which two or more siblings have an ASD, Szatmari et al. (1996) found an Intraclass Correlation Coefficient among siblings of .37 for verbal IQ and an ICC of .62 for nonverbal IQ. The ICC describes how strongly units in the same group resemble each other – in this case, the degree to which siblings resemble each other in terms of IQ.

MacLean et al. (1999) reported an ICC of .42 for nonverbal IQ among affected siblings. Szatmari et al. (1996) additionally examined relationships between IQs of children with ASDs and their unaffected siblings and reported a non-significant Pearson correlation of .23.

Relatedly, ASD-sibs exhibit delays when compared to younger siblings of children who do not have a diagnosed ASD (i.e., COMP-sibs) in certain aspects of cognitive functioning, such as spatial abilities and verbal skills (Yirmiya et al., 2006; Plumet, Goldblum, & Leboyer, 1995). ASD-sibs exhibited significantly lower IQ scores and poorer reading and spelling performances than COMP-sibs (Fombonne, Bolton, Prior, Jordan and Rutter, 1997). Yirmiya et al. (2006) found that, at 14 months of age, ASD-sibs achieved lower language scores on the Bayley Scales of Infant Development than COMP-sibs. Moreover, they made fewer nonverbal requesting gestures than COMP-sibs, precursors for later language development (Yirmiya et al., 2006).

Results of family studies clearly indicate a significant familial clustering of cognitive disabilities in ASD-sibs, which mirror cognitive impairments seen in individuals with ASDs. However, it is unclear from previous studies whether an association in level of cognitive function exists between older children with an ASD and their younger siblings. As such, the current study examined this association and also closely examined symptom severity and cognitive functioning within older diagnosed ASD-children and within their younger siblings. Another aim of the current study was to determine whether level of cognitive functioning was associated with level of ASD symptomatology. It was hypothesized that level of cognitive functioning in the older sibling would be associated with level of cognitive functioning in the younger sibling,

both for siblings in general and within groups. Also, as ASD-sibs have been found to display deficits which mirror those cognitive impairments seen in individuals with ASDs, it was hypothesized that level of ASD symptomatology in an individual would be associated negatively with his or her sibling's level of cognitive functioning.

The current study further investigated the presence of autistic symptomatology and related impairments in sibling pairs. By assessing ASD symptomatology along a continuum of severity, the associations between level of ASD symptomatology in older and younger siblings were able to be examined. ASD-sibs are more likely than their peers to display subthreshold levels of autistic social impairment (Constantino et al, 2006) and deficits in social and communicative functioning (Landa & Garrett Mayer, 2006; Zwaigenbaum et al., 2005; Bolton, Macdonald, Pickles, & Rios, 1994; Pickles et al., 2000; Rutter, 2000). As such, it was hypothesized that level of ASD symptomatology in older siblings would be associated with level of ASD symptomatology in their younger siblings.

## Chapter 2: Method

*Participants.* Infants in the present study were recruited from a larger sample in which the social, emotional, and cognitive development of ASD-sibs and COMP-sibs are investigated from two months of age to 36 months of age. For the purposes of this study, data collected during the 30 and 36 month visits, as well as a one-time older sibling visit, were used. ASD-sibs were recruited by referrals from the University of Miami/Nova Southeastern University Center for Autism and Related Disabilities (UM-NSU CARD), the Autism Spectrum Assessment Clinic (ASAC), and the University of Miami Psychological Services Center. A brochure was also distributed at autism-related events and at other functions to parents of infants. COMP-sibs were recruited by mail. Brochures were mailed to parents of infants whose addresses and names were obtained from Miami-Dade County birth records. Both groups of infants were also recruited by word of mouth. Infant siblings were excluded from this study if they were born at gestational age less than 37 weeks or weighed less than 2500 grams.

The cognitive functioning and level of ASD symptomatology of twenty-nine younger siblings and their older siblings was assessed (see Table 1). Group assignment was based on the presence or absence of a diagnosed ASD in the older sibling. Community diagnoses of an affected sibling(s), when present, were confirmed by an independent clinician using clinical best estimate based on a review of the older sibling's performance on multiple measures and questionnaires collected during a research visit. Previous report(s) were also used when available.

Two infants were placed in an Unresolved category due to the uncertainty of the older sibling diagnosis. The older sibling of one of these infants had a prior ASD



diagnosis, which was later rescinded through a follow-up community diagnosis. In addition, this study's independent clinician did not find evidence to substantiate the earlier community ASD diagnosis. The older sibling of the other infant did not have a prior ASD diagnosis but had research evidence of elevated ASD symptomatology (i.e., elevated score on at least one of two measures of ASD symptomatology: Social Communication Questionnaire [SCQ; Berument, Rutter, Lord, Pickles, & Bailey, 1999] or Autism Diagnostic Observation Schedule [ADOS; Lord et al., 2000], both discussed in detail below). For the purposes of this study, a score of 9 or above on the SCQ was considered elevated. On the ADOS, cutoff scores signaling an autism spectrum classification were used (i.e., Module 1 = 7, Module 2 = 8, Module 3 = 8) [Lord, Rutter, DiLavore, and Risi, 1999].

The Unresolved participants were included in correlations using the entire sample but not in those analyses that focused on the ASD-sibs, COMP-sibs, or differences between these groups.

Of the remaining 27 sibling dyads, 11 infants were placed in the COMP-sibs group as their older sibling(s) had not been diagnosed with, nor showed any research evidence, of an ASD-related disorder. The 16 remaining infants were placed in the ASD-sibs group after parents reported that at least one older sibling had been previously diagnosed with an ASD. Two younger ASD-sibs, and their older siblings, were twins. To maintain the assumption of independent sampling in the younger and older siblings, the average of the younger twins' scores on all measures were used in all younger sibling analyses, and the average of the older twins' scores on all measures were used in all older sibling analyses. Thus, the ASD-sibs group sample size was 15.

Two pairs (one from the comparison group and one from the ASD group) were excluded from analyses involving younger sibling symptomatology due to missing data. The younger sibling in one of these pairs also missed the 36 month visit, thus the Mullen was not collected, precluding their inclusion in any analyses involving younger sibling cognitive functioning.

*Younger Sibling Measures.* Cognitive functioning in younger siblings was assessed with the Mullen Scales of Early Learning (Mullen, 1995). Level of autistic symptomatology was assessed with the ADOS (Lord et al., 2000), the Autism Diagnostic Interview – Revised (ADI-R; Lord, Rutter, & Le Couteur, 1994), and either the Social Responsiveness Scale-Preschool (SRS-P; *in press*) or Social Responsiveness Scale (SRS; Constantino, 2005).

At 30 months of age, participants were administered the ADOS (Lord et al., 2000) by a trained clinician (see Table 2). The ADOS consists of a series of “presses” in which a situation is created to generate observations of the spontaneous behaviors of the individual, as well as to observe social interaction, communication, and play. Items presented differ based on the language level of the child. Module 1 is used for children who have little to no language. Module 2 is used when language is present, but speech is not fluent or spontaneous. Module 3 is administered to children who have fluent phrase speech. A Module 1 was used for 15 younger siblings and a Module 2 was used for 10 younger siblings. Two participants who missed their 30 month visit later returned to complete the ADOS at four years of age; both were administered a Module 3. For the

purposes of this study, only scores from the algorithm items in the social and communication domains were used for all participants.

At 36 months of age, a trained administrator completed the ADI-R (Lord, Rutter, & Le Couteur, 1994) with the parent. The ADI-R follows a semistructured format of interview with the parent and includes a list of items related to onset patterns, communication, social development and play, and restricted patterns of interest and behaviors. The ADI-R provides a diagnostic algorithm that is keyed to the DSM-IV (American Psychiatric Association, 1994) criteria for autism; this algorithm was utilized in the present study. Participants were also administered the Mullen (Mullen, 1995) by a trained administrator. The Mullen is a multidomain assessment scale that emphasizes the measurement of distinct abilities. It contains five domains: Visual Reception, Expressive Language, Receptive Language, Fine Motor, and Gross Motor. The last of these (i.e., Gross Motor) was not administered at this visit because children were older than 30 months of age, which is beyond the norms of this subtest. The Mullen yields standard T scores in all five domains and an Early Learning Composite score based on the first four domains. The Mullen is considered to be optimal for the assessment of young children with autism due to its separation of visual perceptual abilities from expressive and receptive language, as well as the separation of fine and gross motor skills (Klin, Saulnier, Tsatsanis, and Volkmar, 2005).

Parents also completed the SRS-P (*in press*) or SRS (Constantino, 2005), based on the age of the child at time of completion; both are parent-completed questionnaires that measure differences in reciprocal social interactions on a continuum. The SRS-P is used for children 3 years of age, while the SRS is used for children 4 years of age and

older. They each consist of 65 items covering dimensions of communication, social interactions, and repetitive and stereotyped behaviors and interests associated with ASDs. Each item rates the frequency, not the intensity, of a behavior on a scale from zero (not true) to three (almost always true). The item scores are totaled and result in a severity score. The SRS-P was collected for all but two younger siblings at the 36 month visit. For those two children, the SRS was collected as they were 4 years of age when the questionnaires were completed.

*Older Sibling Measures.* The older siblings of the infant participants were brought in by their parents for one visit when they were three years of age or older. The mean age of the older COMP-sibs at the time of their visit was 6.5 years ( $SD = 0.75$ ), while the mean age of the older ASD-sibs at the time of their visit was 6.8 years ( $SD = 2.45$ ). The age at the time of the visit was not significantly different for the two groups,  $F(1,24) = 0.08, p > .05$ . All older siblings were administered a test of cognitive functioning (discussed below). Additionally, parents of all older siblings completed questionnaires to assess level of autistic symptomatology (i.e., SRS, SCQ). Finally, all older siblings of children in the ASD group were administered the ADOS. The ADOS was also administered to the older sibling of one child originally in the comparison group because he surpassed a pre-set minimum score of a nine on the SCQ. He was placed in the Unresolved group (as discussed above).

During the older sibling's visit, a cognitive test was administered; the particular test chosen was based on child's age (see Table 2). The Mullen was administered to the six children who were at least three and less than five years of age. The Wechsler Preschool and Primary Scale of Intelligence- Third Edition (WPPSI-III; Wechsler, 2002)

was administered to the eleven children who were at least five and less than seven years of age. The Wechsler Intelligence Scales for Children-Fourth Edition (WISC-IV; Wechsler, 2003) was administered to the twelve children seven years of age or older. The merits of the Mullen have been noted previously. The Wechsler scales are considered the standards for the testing of intelligence, and their division of the various tasks into factor scores (Kaufman, 1994) can be particularly helpful in the interpretation of profiles of children with an ASD given the typical performance scatter (i.e., lower verbal, higher performance) often found in these children's profiles (McDonald, Mundy, Kasari, & Sigman, 1989). This author could not find any information on the correlation between the Mullen and Wechsler tests. However, a number of studies have shown that preschool psychological estimates of cognition have some predictive value. For example, Yang et al (2003) found that nonverbal IQ is stable over a mean interval of 22 months. Few intelligence test batteries span development from preschool to adolescence, so that different instruments are likely to be used at different ages, especially in handicapped children (Rapin, 2003).

Parents also completed the SRS (Constantino, 2005) and the SCQ (Berument, Rutter, Lord, Pickles, & Bailey, 1999) for the older sibling. The SRS was previously discussed. The SCQ is a 40-item parent report questionnaire designed as a screener for ASDs, and was administered as a parent interview in the present study. Items come from the ADI-R algorithm and evaluate reciprocal social interaction, language and communication, and repetitive and stereotyped behaviors. Behaviors are rated as either present or absent. The Current version was used in the present study for children less than five years of age, while the Lifetime version was used for children five and older.

The module of the ADOS administered was based on the language level of the child. Four older siblings were administered a Module 1, five were administered a Module 2, and nine were administered a Module 3.

*Variable Creation.* The ADI-R and ADOS are the gold standard for diagnosis, and consequently are used by many clinicians and researchers in their formulation of a diagnosis. For the purposes of diagnosis, these measures require categorization of individuals based on designated cutoff scores. Thus, a child who satisfies less than sufficient criteria for a particular domain may not be considered as meeting the requirements for a diagnosis of an ASD. However, the focus of this study was not on diagnosis, but rather the level of ASD symptomatology present. Thus, for each measure, the scores on all algorithm items were summed to represent a severity score.

For the ADI-R, and less so on the ADOS, summing the administered items may appear to artificially inflate the degree of autistic symptomatology in a more verbal child, or under-represent the symptomatology in a non-verbal child as fewer items are administered to non-verbal children. Adding the scores in essence adds the number of ways in which the child's language is unusual, and thus individuals with more complex language score as more abnormal than children who cannot speak since language items are not administered to nonverbal children (Rutter et al., 2003). Two previous studies have used an alternate method in which nonverbal children receive the highest score for language items of the ADI-R, which would otherwise not be administered (Lord, Rutter, & LeCouteur, 1994; Tadevosyan-Leyfer et al., 2003). However, using this strategy results in the allocation of extreme scores to very low-functioning children because they are non-verbal. This was not the intention of the ADOS or ADI-R developers (C. Lord, personal

communication, September 9, 2007), because when this is done in factor analyses, these measures correlate highly with nonverbal IQ. Instead, for this study, a code of 3 was converted to 2, and 8 was converted to 0 (as is specified in the manuals: ADOS [Lord et al., 2000] and ADI-R [Lord, Rutter, & Le Couteur, 1994]) prior to the summation of all algorithm item scores. Modules 1 and 2 of the ADOS have the same number of algorithm items (i.e., 5 for communication, and 7 for social). However, Module 3 has one fewer Communication algorithm item (i.e., 4 for communication, and 7 for social). Thus, in order to ensure that each set of scores carried the same weight, all Module 3 Communication item scores were multiplied by 1.25.

*Younger Sibling Variable Creation.* In order to quantify the level of ASD symptomatology present in each child, scores on all measures administered were standardized. The z-scores were constructed in two ways based on the subsequent analyses. For all analyses that included the whole sample, z-scores were calculated for the entire younger sibling sample, and for the entire older sibling sample. For the analyses that were run separately by group, the z-scores were calculated for each participant within group. In other words, z-scores for each younger ASD-sib were calculated using only the data from other younger ASD-siblings and z-scores for each younger COMP-sib were calculated using only the data from other younger COMP-siblings, and likewise for older siblings. A severity variable was created to represent level of ASD symptomatology and consisted of the average z-score on the ADOS, ADI-R, and SRS-P.

With respect to cognitive functioning, the score on the 36 month Mullen (i.e., Early Learning Composite) was used to represent Cognitive Functioning. On the Mullen,

the index score on Visual Reception was used to account for Performance Ability.

Receptive and Expressive Language index scores were averaged to account for Verbal Ability.

*Older Sibling Variable Creation.* In order to quantify the level of ASD symptomatology present in each child, scores on all measures administered were standardized (see above for details on how z-scores were created). A severity variable was created to represent level of ASD symptomatology and consisted of the average z-score on the measures of symptomatology. For children in the ASD and Unresolved groups, the mean consisted of scores on the ADOS, SCQ and SRS. For all other children, only scores from the SCQ and SRS were used.

A variable was created to represent Cognitive Functioning and consisted of either the Early Learning Composite on the Mullen or the Full Scale IQ on the WPPSI-III or WISC-IV. A separate variable was created for Performance Ability. On the Mullen, the index score on Visual Reception was used to account for Performance Ability. On the WPPSI-III, a Performance index score was generated from subtest scores on Block Design, Matrix Reasoning, Picture Concepts, and Object Assembly. On the WISC-IV, a Perceptual Reasoning index score was generated from subtest scores on Block Design, Picture Concepts, and Matrix Reasoning and used to estimate Performance Ability. A separate variable was made for Verbal Ability. On the Mullen, Receptive and Expressive Language index scores were averaged to account for Verbal Ability. On the WPPSI-III, a Verbal index score was generated from subtest scores on Information, Vocabulary, Word



Reasoning, and Similarities. On the WISC-IV, a Verbal Comprehension index score was generated from subtest scores on Similarities, Vocabulary, Comprehension, and Word Reasoning.

### Chapter 3: Results

The primary purpose of this study was to examine the associations between level of autistic symptomatology and level of cognitive functioning within sibling pairs. Three sets of bivariate correlations were conducted to examine the relationships between autistic symptomatology and cognitive ability. Each set of correlations was run first with the entire sample, then separately for COMP-sibs and ASD-sibs. Bivariate correlations were run between standardized measures of autistic symptomatology and between standardized measures of cognitive functioning and allowed for testing of all hypotheses. Post-hoc analyses included an ANCOVA and 6 ANOVAS which were conducted to identify possible group differences between younger siblings of children with diagnoses of ASDs (ASD-sibs) and younger siblings of children without diagnoses of ASDs (COMP-sibs). The ANCOVA was run to assess group differences on overall cognitive functioning and level of autistic symptomatology. Univariate analyses of variance were conducted as follow-up tests to analyze group differences on individual measures of cognitive functioning and level of autistic symptomatology (i.e., SCQ, SRS, ADOS, and ADI-R). A summary of the collected data is presented in Table 2. Scores on the collected measures are presented in Table 6.

*ASD Symptomatology and Cognitive Functioning Within Individuals.* Younger siblings' level of autistic symptomatology was negatively correlated with their cognitive functioning,  $r(27) = -.53, p < .01$ . When examined within each status group, no associations were found within the comparison group,  $r(10) = -.01, p > .05$ . However, within the ASD group, a younger sibling's level of autistic symptomatology was associated with his or her level of cognitive functioning,  $r(15) = -.54, p < .05$ . Thus,

within the ASD group, level of autistic symptomatology was associated negatively with level of cognitive functioning (see above for results of ANCOVAs displaying group differences on these variables).

Older siblings' level of autistic symptomatology was significantly negatively correlated with their level of cognitive functioning,  $r(29) = -.79, p < .01$ . When examined within each status group, no associations were found within the comparison group,  $r(11) = -.19, p > .05$ . However, within the ASD group, an older sibling's level of autistic symptomatology was negatively associated with his or her level of cognitive functioning,  $r(16) = -.66, p < .01$ . Thus, within the ASD group, level of autistic symptomatology was associated negatively with level of cognitive functioning.

*ASD Symptomatology and Cognitive Functioning by Subtest.* For younger ASD-siblings, level of autistic symptomatology was associated negatively with scores on Language,  $r(14) = -.56, p < .05$  and on the Visual Reception subtest score of the Mullen,  $r(14) = -.63, p < .05$ . For older ASD-siblings, an association was found between level of autistic symptomatology and performance on the Visual Reception subtest of the Mullen,  $r(5) = -.94, p < .05$ , but not on the Language subtests of the Mullen, nor on measures of Performance or Verbal Comprehension on the WPPSI-III or WISC-IV.

*Cognitive Functioning.* For the sample as a whole, level of cognitive functioning was associated between siblings,  $r(27) = .62, p < .01$ . When examined within each status group, level of cognitive functioning was not associated between siblings in the comparison group,  $r(10) = .56, p > .05$ , or between siblings in the ASD group,  $r(15) = .49, p > .05$ . In order for these findings to be significant at  $p < .05$ , with an effect size of 0.56, the sample would need to include five more COMP-sibs ( $N=15$ ) and four more

ASD-sibs (N=20). Results of an ANCOVA indicated that there was a group difference in cognitive functioning for younger siblings,  $F(1,23) = 5.88, p < .05$ , and older siblings,  $F(1,23) = 14.00, p < .01$ . Both younger and older siblings in the comparison group had higher cognitive scores than the participants in the ASD group (see Figure 2 for distribution of cognitive scores by group).

*ASD Symptomatology and Cognitive Functioning Between Individuals.* In the sample as a whole, older siblings with a greater level of autistic symptomatology had younger siblings with lower scores on measures of cognitive functioning,  $r(27) = -.46, p < .05$ . When examined within each status group, there was not an association between younger sibling cognitive functioning and older sibling level of autistic symptomatology in either the comparison group,  $r(10) = -.15, p > .05$ , or in the ASD group,  $r(16) = -.23, p > .05$ .

Younger siblings with a greater level of autistic symptomatology had older siblings with lower scores on measures of cognitive functioning,  $r(27) = -.49, p < .01$ . When examined within each status group, there was no longer an association between older sibling cognitive functioning and younger sibling level of autistic symptomatology in the comparison group,  $r(10) = .37, p > .05$ , or in the ASD group,  $r(15) = -.34, p > .05$ .

*ASD Symptomatology.* In the present study, ASD symptomatology was assessed and viewed along a continuum of severity. For the sample as a whole, level of autistic symptomatology in the older siblings was positively associated with level of autistic symptomatology in the younger siblings,  $r(27) = .39, p < .05$ . Level of autistic symptomatology was not associated between siblings within the comparison group,  $r(10) = .11, p > .05$ , or within the ASD group,  $r(15) = .02, p > .05$ .

*Post-Hoc Analyses.* An ANCOVA conducted using the entire sample indicated that there was a difference between composite level of autistic symptomatology for younger siblings,  $F(1,23) = 7.22, p < .05$ , and for older siblings,  $F(1,23) = 45.97, p < .01$  (see Figure 1 for distribution of symptomatology scores by group). These results remained significant after controlling for younger sibling gender. Follow-up univariate ANOVAS were conducted to determine the relevance of using parent-report versus observational measures (i.e., ADI-R and SCQ versus ADOS). These analyses indicated group differences on individual measures of autistic symptomatology. Younger siblings' scores differed on two of the three components of the ASD severity score such that younger siblings in the comparison group exhibited lower levels of autistic symptomatology than younger siblings in the ASD group. Specifically, scores differed by status group on the ADOS and ADI-R, but not on the SRS. Older siblings' scores also differed such that older siblings in the ASD group showed a greater level of autistic symptomatology than those in the comparison group on the SCQ and SRS. Tables 7 and 8 contain the means and the standard deviations of all protocols and questionnaires, as well as results of the univariate analyses of variance.

In order to assure that the inclusion of the older ASD-sibs' ADOS scores into the severity variable did not impact the results, ADOS scores were eliminated from the average z-score calculation. Thus, the ASD symptomatology variable for both older COMP-sibs and ASD-sibs was comprised solely of the standardized score on the SCQ and SRS. Findings reported above were replicated. Specifically, when associations were examined in the entire sample, all correlations examined were significant. As above, when examined by group, the only associations which remained significant were those

that were previously significant, specifically between ASD symptomatology and cognitive functioning in the younger sibling, and between ASD symptomatology and cognitive functioning in the older sibling.

As reported above, an overall association existed between level of ASD symptomatology and level of cognitive functioning for the entire sample, and for the ASD-sibs. To further investigate, analyses were conducted using specific subtests for each measure of cognitive functioning representing the two principal domains of intelligence, namely Verbal and Nonverbal Learning. ASD-sibs, on average, had lower cognitive scores than COMP-sibs. ASD-sibs also had significantly lower scores in receptive language, expressive language, and visual reception than COMP-sibs. Older ASD-sibs had significantly lower scores on the Verbal Comprehension Index of the WISC-IV than COMP-sibs (see Table 10 for all subtest mean scores and results of ANOVAs). Finally, although an ANOVA could not be conducted to compare ASD-sibs and COMP-sibs scores on the Mullen scales due to there only being one COMP-sib, the scores of ASD-sibs in the areas of receptive language, expressive language, and visual reception were found to be in the Below Average range (see Table 10).

## Chapter 4: Discussion

The purpose of this study was to examine the relationship between the severity of autistic symptomatology and cognitive functioning within and between siblings. The study was comprised of younger siblings of children with a confirmed ASD diagnosis (ASD-sibs), and a comparison group whose older siblings did not have an ASD diagnosis (COMP-sibs). For the entire sample, level of ASD symptomatology in the older sibling was associated positively with level of ASD symptomatology in the younger sibling. Also, level of cognitive functioning in the older sibling was associated positively with level of cognitive functioning in the younger sibling. Moreover, level of ASD symptomatology was associated negatively with level of cognitive functioning within siblings and between each member of a sibling pair. However, when the groups were examined separately (i.e., ASD-sibs and COMP-sibs), only some of these associations were found within the ASD-sib group, and no associations were found within the COMP-sib group (see Table 5). It appears that the significant associations that existed for the whole sample may primarily reflect mean differences in both variables between the groups.

Overall, there was a difference between the level of autistic symptomatology in younger siblings and older siblings. ASD-sibs exhibited a greater level of autistic symptomatology than COMP-sibs as measured by the ADOS and ADI-R, but not on the SRS. Older siblings in the ASD group had a greater level of autistic symptomatology than those in the comparison group on the SCQ and SRS. Additionally, both younger and older siblings in the ASD group had lower cognitive scores than siblings in the comparison group. In essence, these findings suggest that, for both children with ASDs

and their younger siblings, ASD symptomatology and cognitive functioning are associated. Also, both children with ASDs and their younger siblings display a greater level of symptomatology and lower cognitive functioning than COMP-sibs.

*The Relationship of ASD Symptomatology Between Siblings.* ASD-sibs not only have an increased risk of meeting criteria for an ASD, but many also manifest impairments that fall under the broader autism phenotype (Bailey et al., 1998). In the present study, level of ASD symptomatology in the older sibling was hypothesized to be associated with level of ASD symptomatology in the younger sibling. As expected, level of autistic symptomatology in the older siblings was positively associated with level of autistic symptomatology in the younger siblings. However, when examined within each group, symptomatology was no longer associated between siblings. It appeared that the overall association in level of autistic symptomatology between siblings was due to the tendency of ASD children and their younger siblings to have higher levels of autistic symptomatology than the comparison siblings.

These findings should be understood in the context of the current literature. Constantino et al. (2006) used the Social Responsiveness Scale (SRS) in an effort to quantify the degree to which subsyndromal autistic impairments are present among siblings of probands. The authors found that the greatest level of impairment (i.e., highest elevations on social responsiveness scale scores) was seen among ASD-sibs from multiplex families, followed by siblings of probands with any ASD. However, the authors limited their study to the brothers of ASD individuals. This was due to the lower phenotypic expression of genetic susceptibility in female subjects observed in studies of autistic traits in the general population (Constantino & Todd, 2003). In the current study,



given that nearly half of the younger ASD-sibs were female, it is important to consider that the manifestation of disrupted social responsivity may be different in girls than boys. Additionally, the SRS taps into one domain: reciprocal social behavior, which is a defining feature of ASDs. In the current study, the autistic symptomatology score was also comprised of scores on the ADOS and ADI-R, commonly referred to as the gold standards for diagnosis of ASDs. These measures use a diagnostic algorithm keyed to the DSM-IV (American Psychiatric Association, 1994) criteria for autism and take into account not only social interaction, but also communication and play. However, when SRS scores alone were compared, ASD-sibs' SRS scores were not significantly higher than COMP-sibs' SRS scores. This is likely due to the fact that the preschool version of the SRS (i.e., SRS-P) was used with the younger siblings due to the age at which the measure was collected. The SRS-P may not be able to parse apart social differences seen at three years of age as the social skills perceived to be deficient in ASDs either are too subtle in nature at such a young age or are not included in questionnaire.

*The Relationship of Level of Cognitive Functioning Between Siblings.* Studies have consistently found relationships between intelligence scores of typically-developing siblings, with an average correlation of 0.49 (Elbedour, Bouchard, & Hur, 1997; McCall, 1970). Studies of cognitive functioning in ASD populations have been primarily limited to children with ASDs and their affected siblings and have relied entirely on nonverbal measures of intelligence (Szatmari et al., 1996; MacLean et al., 1999). Szatmari et al. additionally examined relationships between the IQs of children with ASDs and their unaffected siblings and reported a non-significant Pearson correlation of .23. Specifically, ASD probands were administered the Leiter Performance Scales (Levine, 1986), which is

a nonverbal IQ test, and unaffected siblings were given the Stanford-Binet (revised edition) IQ test (Thorndike et al., 1985). In the present study of ASD siblings and their younger siblings, level of cognitive functioning in the older sibling was hypothesized to be associated with level of cognitive functioning in the younger sibling. Although an association was found between older and younger sibling cognitive functioning, this was only true when the whole sample was included. This suggests that the overall association in level of cognitive functioning between siblings was due to a tendency of comparison children and their siblings to have higher scores on measures of cognitive functioning than the ASD siblings. These associations were not significant using the small sample sizes of the current study. However, the magnitude of the correlations (COMP-sibs [ $r = .56$ ] and ASD-sibs [ $r = .49$ ] mirror those in the literature.

*ASD Symptomatology and Cognitive Functioning Within Individuals.* Research on individuals with autism (e.g., Ghaziuddin & Mount-Kimchi, 2004; Barnhill, Hagiwara, Myles, & Simpson, 2000) indicates a cognitive profile typified by higher performance on visual and abstract subtests than on verbal subtests, particularly for low-functioning individuals. However, level of intellectual function can range from profound mental retardation to the superior range on conventional IQ tests (Filipek et al., 1999). As such, when the focus is on the entire autism spectrum, cognitive impairments may not be as pronounced (Bailey et al., 1998). This is particularly true of individuals with a diagnosis of high-functioning autism [HFA] (i.e.,  $IQ > 70$ ) and Asperger's syndrome, which some believe collectively account for 50% of all children with ASDs (Kielinen, Linna, & Moilanen, 2000). In the present study, the association between level of autistic symptomatology and level of cognitive functioning was examined, first within

individuals to determine whether having an increased level of autistic symptomatology was associated with level of cognitive functioning. Among the entire sample of younger siblings, there was an association between ASD symptomatology and cognitive functioning. This was also true of older siblings. However, when examined within each group, this association was only found for ASD-sibs. Specifically, level of ASD symptomatology was associated negatively with cognitive functioning. Level of autistic symptomatology in the younger sibling of an autistic child was associated with his/her level of cognitive functioning. Also, for the older ASD siblings, level of ASD symptomatology was associated with level of cognitive functioning. These results suggest that intellectual disability (i.e., impaired cognitive functioning) runs in concert with symptomatology among children with ASDs. Importantly, it indicates that impaired cognitive functioning and ASD symptomatology covaried for younger siblings at risk for ASDs. To our knowledge, this latter association has never before been documented. This finding may be due to the presence of two groups: younger siblings who were observed to display and reported to have high level of symptoms and low cognitive functioning versus younger siblings with better outcomes who had low symptomatology and higher cognitive functioning. It may also be due to the participation of siblings who were on their way to developing autism versus those who had a more typical trajectory.

*Associations of ASD Symptomatology and Cognitive Functioning Between Individuals.* Results of family studies indicate that a relationship exists between cognitive performance in siblings of autistic individuals (Yirmiya et al., 2007; Plumet, Goldblum, & Leboyer, 1995). In the present study, level of ASD symptomatology in an individual was hypothesized to be associated with his or her sibling's level of cognitive functioning.

The existence of a relationship between ASD symptomatology and cognitive functioning between siblings was evident when examining the overall sample, but not when looking at each group separately. When the entire sample was included, having an older sibling with a greater level of autistic symptomatology was negatively associated with the younger sibling's level of cognitive functioning. Additionally, younger siblings with a greater level of autistic symptomatology had older siblings with lower scores on measures of cognitive functioning. However, when examined within each group, there were no significant associations between older and younger sibling symptomatology and cognitive functioning. Consequently, the overall association between level of autistic symptomatology and level of cognitive functioning between siblings appeared to be due to the difference in the scores between the two groups.

Level of ASD symptomatology was hypothesized to be associated with performance on the Verbal and Nonverbal subtests of the cognitive measures. ASD-sibs, on average, had lower total cognitive scores than COMP-sibs. Younger ASD-sibs also had significantly lower scores in receptive language, expressive language, and visual reception than COMP-sibs, similar to a study being conducted by Ozonoff (2007) in which early findings indicate that ASD-sibs perform significantly worse than the siblings of typically developing children and siblings of children with other developmental delays on the Receptive Language and Expressive Language subscales of the Mullen at 12, 18, and 24 months of age. Yirmiya et al. found that, at 24 months, significantly more ASD-sibs demonstrated language scores one or two standard deviations below the mean compared to COMP-sibs. At 36 months, the groups differed significantly in receptive language, and more ASD-sibs displayed receptive and expressive difficulties compared to

COMP-sibs. Additionally, Ozonoff, Rogers, Farnham, and Pennington (1993) reported that ASD-sibs performed significantly less well than did siblings of individuals with learning disabilities on the verbal comprehension and perceptual organization factors of the Wechsler Intelligence Scale for Children—Revised (WISC-R) and on the Wechsler Adult Intelligence Scale (WAIS-R). Level of autistic symptomatology in ASD-sibs was negatively associated with scores on Language and the Visual Reception subtest score on the Mullen. Older ASD-sibs had significantly lower scores on the Verbal Comprehension Index of the WISC-IV than older COMP-sibs. The scores of ASD-sibs in the areas of receptive language, expressive language, and visual reception were noted to be Below Average. For older siblings, an association was found between level of autistic symptomatology and performance on the Visual Reception subtest of the Mullen. These findings seem to indicate that, by three years of age, ASD-sibs are already receiving lower scores than COMP-siblings in the areas of receptive language, expressive language, and visual reception and supports previous findings in this area.

The current study's limitations, in addition to sample size, include the measures implemented. For example, three different cognitive tests were used for the older siblings. Though they were chosen based on the test parameters, future studies should consider the use of one cognitive test that can be used across a range of ages and ability levels. Additionally, given that intelligence is not a stable construct until a child reaches maturity (Garlick, 2002), it is possible that associations between cognitive functioning as measured in the present study and ASD symptomatology change throughout one's development. This may be of particular interest for sibling studies currently being conducted as current ASD-siblings mature and continue to be studied beyond three years

of age. Finally, it is possible that the tests did not adequately represent the true abilities of those children who presented with no or delayed language. Language delays commonly translate into impaired performance on verbally mediated tests (Barnhill, Hagiwara, Myles, & Simpson, 2000; Ghaziuddin & Mountain Kimchi, 2004; Mayes & Calhoun, 2003).

Another possibility that exists given the findings of these results is that there is not a true relationship between severity of ASD symptomatology and level of cognitive functioning. Coplan (2003) believes that ASDs of any degree of severity can be seen in association with any degree of general intelligence.

Despite its limitations, this study contributes to the emerging literature conceptualizing ASDs, not as discrete categories, but along a continuum of severity. The application of this study design to a larger sample could reveal associations between ASD symptomatology and cognitive functioning between siblings not revealed in the present study due to its limited sample size. The use of a larger sample would not only increase power, but also ensure a more diverse range of ability levels across all domains assessed in both members of the sibling pairs. Importantly, the current study's results indicate that intellectual disability (i.e., impaired cognitive functioning) runs in concert with symptomatology among children with ASDs and among ASD-sibs. Additionally, by three years of age, ASD-sibs were already receiving lower scores than COMP-siblings in the areas of receptive language, expressive language, and, for the first time, in visual reception. Follow-up of these siblings, which is underway, will help determine whether these siblings' current cognitive profiles are stable over time and are associated with

other delays. From a clinical viewpoint, the identification of specific and stable limitations in ASD-sibs has important implications for intervention programs which could help to prevent or ameliorate poor outcomes for ASD-sibs.

## References

- American Psychiatric Association (1994). *Diagnostic and statistical manual of mental disorders* (4th ed.). Washington, DC: American Psychiatric Press, Inc.
- Bailey, A., Palferman, S., Heavey, L., & Le Couteur, A. (1998). Autism: The phenotype in relatives. *Journal of Autism and Developmental Disorders*, 28(5), 369-392.
- Barnhill, G., Hagiwara, T., Myles, B. S., & Simpson, R. L. (2000). Asperger syndrome: A study of the cognitive profiles of 37 children and adolescents. *Focus on Autism and Other Developmental Disabilities*, 15(3), 146-153.
- Berument, S. K., Rutter, M., Lord, C., Pickles, A., & Bailey, A. (1999). Autism screening questionnaire: Diagnostic validity. *British Journal of Psychiatry*, 175 Nov, 444-451.
- Bolton, P., Macdonald, H., Pickles, A., & Rios, P. (1994). A case-control family history study of autism. *Journal of Child Psychology and Psychiatry*, 35(5), 877-900.
- Constantino, J. N. (2005). *The Social Responsiveness Scale*. Los Angeles: Western Psychological Services.
- Constantino, J.N., Lajonchere, C., Lutz, M., Gray, T. et al. (2006). Autistic social impairment in the siblings of children with pervasive developmental disorders. *The American Journal of Psychiatry*, 163(2), 294-296.
- Coplan J. (2003). Atypicality, intelligence, and age: a conceptual model of autistic spectrum disorders. *Developmental Medicine and Child Neurology*, 45, 712 –716.
- Ehlers, S., Nyden, A., Gillberg, C., & Dahlgren Sandberg, A. (1997). Asperger syndrome, autism and attention disorders: A comparative study of the cognitive profiles of 120 children. *Journal of Child Psychology and Psychiatry*, 38(2), 207-217.
- Elbedour, S., Bouchard, T. J. J., & Hur, Y. M. (1997). Similarity in general mental ability in Bedouin full and half-siblings. *Intelligence*, 25(2), 71-82.
- Filipek, P. A., Accardo, P. J., Baranek, G. T., Cook, E. H. J., Dawson, G., Gordon, B., et al. (1999). The screening and diagnosis of autistic spectrum disorders. *Journal of Autism and Developmental Disorders*, 29(6), 439-484.
- Fombonne, E., Bolton, P., Prior, J., Jordan, H., & Rutter, M. (1997). A family study of autism: Cognitive patterns and levels in parents and siblings. *Journal of Child Psychology and Psychiatry*, 38(6), 667-683.
- Garlick, D. (2002). Understanding the nature of the general factor of intelligence: The role of individual differences in neural plasticity as an explanatory mechanism. *Psychological Review*, 109(1), 116-136.



- Ghaziuddin, M., & Mountain Kimchi, K. (2004). Defining the intellectual profile of Asperger syndrome: Comparison with high-functioning autism. *Journal of Autism and Developmental Disorders*, 34(3), 279-284.
- Kaufman, A. S. (1994). *Intelligent testing with the WISC-III*. Oxford, England: John Wiley and Sons.
- Kielinen, M., Linna, S. L., & Moilanen, I. (2000). Autism in northern Finland. *European Child and Adolescent Psychiatry*, 9(3), 162-167.
- Klin, A., Saulnier, C., Tsatsanis, K., & Volkmar, F.R. (2005). Clinical evaluation in autism spectrum disorders: Psychological assessment within a transdisciplinary framework. In F.R. Volkmar, R. Paul, A. Klin, D. Cohen (Eds). *Handbook of autism and pervasive developmental disorders*, Vol. 2: Assessment, interventions, and policy (3rd ed.). (pp. 772-798). Hoboken, NJ: John Wiley & Sons, Inc.
- Landa, R., & Garrett Mayer, E. (2006). Development in infants with autism spectrum disorders: A prospective study. *Journal of Child Psychology and Psychiatry*, 47(6), 629-638.
- Lord, C., Risi, S., Lambrecht, L., Cook, E. H. J., Leventhal, B. L., DiLavore, P. C., et al. (2000). The Autism Diagnostic Observation Schedule--Generic: A standard measure of social and communication deficits associated with the spectrum of autism. *Journal of Autism and Developmental Disorders*, 30(3), 205-223.
- Lord, C., Rutter, M. & Couteur, A. L. (1994). Autism Diagnostic Interview-Revised: A revised version of the diagnostic interview for caregivers of individuals with possible pervasive developmental disorders. *Journal of Autism and Developmental Disorders*, 24, 659-685.
- Mayes, S. D., & Calhoun, S. L. (2003). Ability profiles in children with autism: Influence of age and IQ. *Autism*, 7(1), 65-80.
- McCall, R. B. (1970). Intelligence quotient pattern over age: Comparisons among siblings and parent-child pairs. *Science*, 170(3958), 644-648.
- McDonald, M. A., Mundy, P., Kasari, C., & Sigman, M. (1989). Psychometric scatter in retarded, autistic preschoolers as measured by the Cattell. *Journal of Child Psychology and Psychiatry*, 30(4), 599-604.
- Mullen, E. (1995). *Infant Mullen Scales of Early Learning*. Circle Pines, MN: American Guidance Service.
- Ozonoff, S. (2007, January). *Autism in infancy: Can we detect it earlier?* Poster presented at the Annual CARD Conference, Fort Lauderdale, FL.

- Ozonoff, S., South, M., & Miller, J. N. (2000). DSM-IV-defined Asperger syndrome: Cognitive, behavioral and early history differentiation from high-functioning autism. *Autism, 4*(1), 29-46.
- Pickles, A., Starr, E., Kazak, S., Bolton, P., Papanikolaou, K., Bailey, A., et al. (2000). Variable expression of the autism broader phenotype: Findings from extended pedigrees. *Journal of Child Psychology and Psychiatry, 41*(4), 491-502.
- Plumet, M. H., Goldblum, M. C., & Leboyer, M. (1995). Verbal skills in relatives of autistic females. *Cortex, 31*(4), 723-733.
- Rutter, M. (2000). Genetic studies of autism: From the 1970s into the millennium. *Journal of Abnormal Child Psychology, 28*(1), 3-14.
- Rutter, M., Bailey, A., Bolton, P., & Le Couter, A. (1994). Autism and known medical conditions: Myth and substance. *Journal of Child Psychology and Psychiatry, 35*(2), 311-322.
- Veenstra, Vanderweele, J., & Cook, E. H. J. (2003). Genetics of childhood disorders: XLVI. autism, part 5: Genetics of autism. *Journal of the American Academy of Child and Adolescent Psychiatry, 42*(1), 116-118.
- Wechsler, D. (2002). The Wechsler Preschool and Primary Scale of Intelligence-Third Edition. San Antonio, TX: The Psychological Corporation.
- Wechsler, D. (2003). Wechsler Intelligence Scale for Children-Fourth Edition: Technical and interpretive manual. San Antonio, TX: Psychological Corporation.
- Yirmiya, N., Gamliel, I., Pilowsky, T., Feldman, R., Baron Cohen, S., & Sigman, M. (2006). The development of siblings of children with autism at 4 and 14 months: Social engagement, communication, and cognition. *Journal of Child Psychology and Psychiatry, 47*(5), 511-523.
- Yirmiya, N., Gamliel, I., Shaked, M., & Sigman, M. (2007). Cognitive and verbal abilities of 24- to 36-month-old siblings of children with Autism. *Journal of Autism and Developmental Disorders, 37*(2), 218-229.
- Zwaigenbaum, L., Bryson, S., Rogers, T., Roberts, W., Brian, J., & Szatmari, P. (2005). Behavioral manifestations of autism in the first year of life. *International Journal of Developmental Neuroscience, 23*(2-3), 143-152.
- Zwaigenbaum, L., Thurm, A., Stone, W., Baranek, G., Bryson, S., Iverson, J., et al. (2007). Studying the emergence of autism spectrum disorders in high-risk infants: Methodological and practical issues. *Journal of Autism and Developmental Disorders, 37*(3), 466-480.

Figure 1

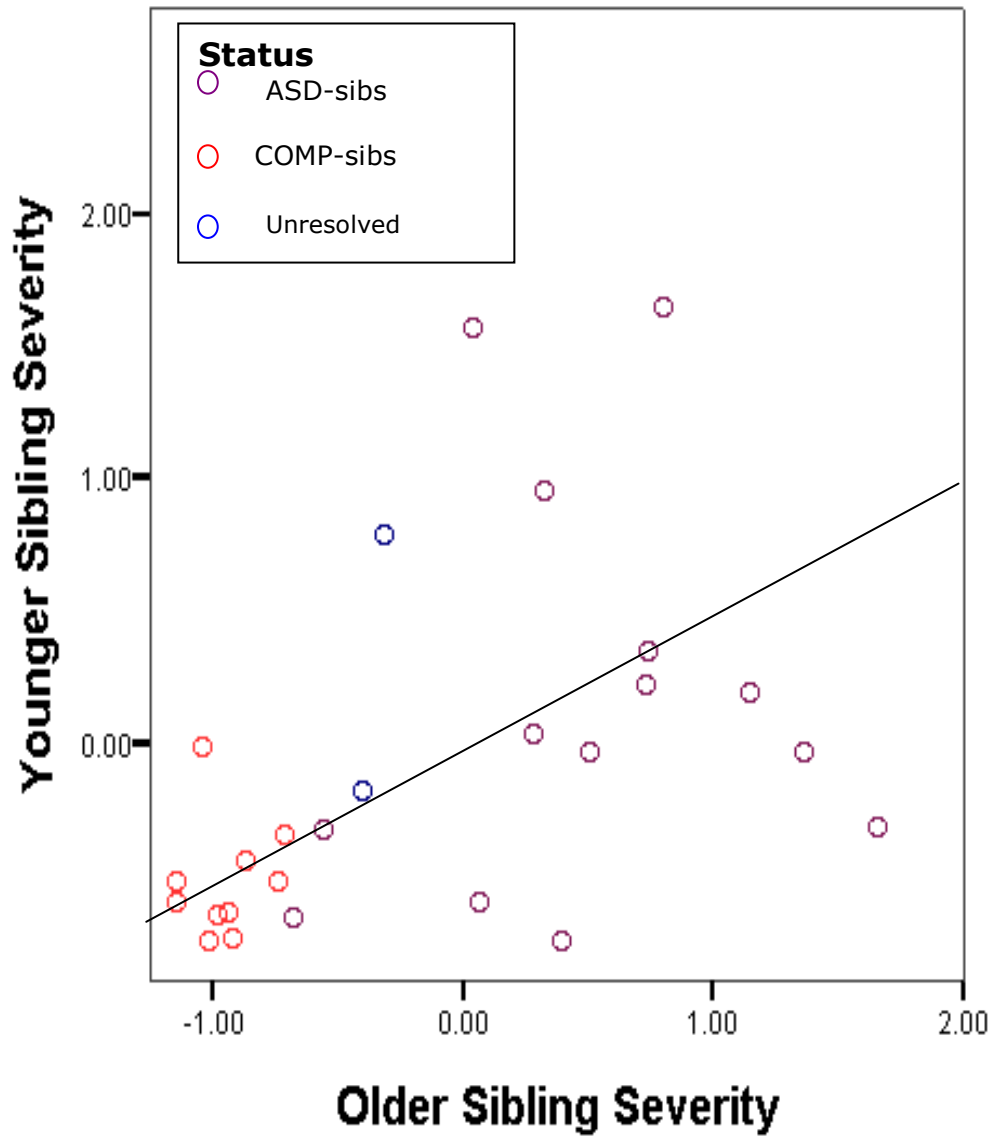


Figure 2

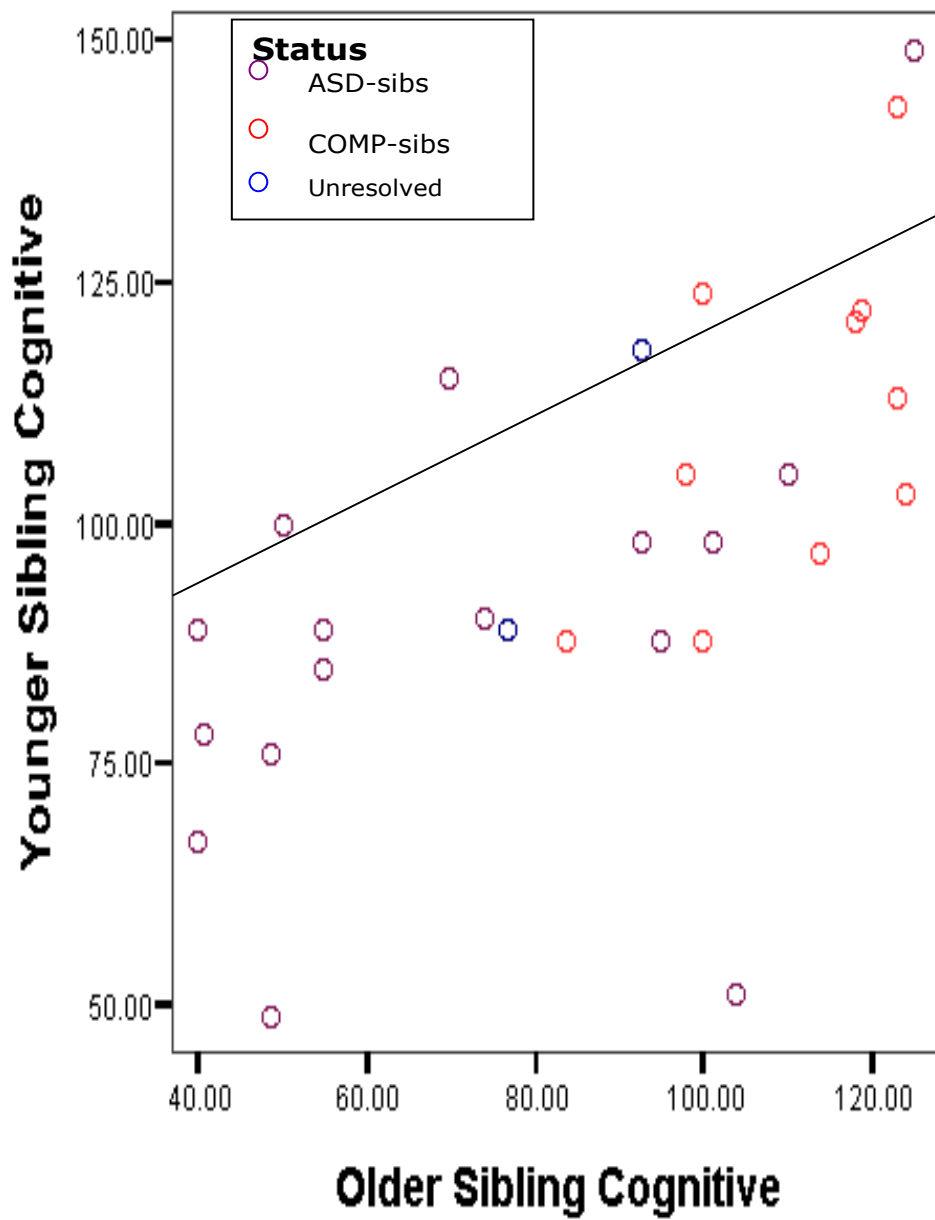


Table 1 Participant Demographics

| <i>Demographics</i>                     | <i>ASD-sibs (n =16)</i> | <i>COMP-sibs (n=11)</i> | <i>Unresolved(n=2)</i> |
|---|-------------------------|-------------------------|------------------------|
| <i>Younger Sibling Gender [% / (n)]</i> |                         |                         |                        |
| Male                                    | 56.3 / (9)              | 45.5 / (5)              | 50.0 / (1)             |
| Female                                  | 43.7 / (7)              | 54.5 / (6)              | 50.0 / (1)             |
| <i>Older Sibling Gender [% / (n)]</i>   |                         |                         |                        |
| Male                                    | 100 / (16)              | 18.2 / (2)              | 100 / (2)              |
| Female                                  | 0 / (0)                 | 81.8 / (9)              | 0 / (0)                |
| <i>Ethnicity [% / (n)]</i>              |                         |                         |                        |
| White/Non-Hispanic                      | 56.3 / (9)              | 36.4 / (4)              | 0 / (0)                |
| White/Hispanic                          | 37.5 / (6)              | 45.5 / (5)              | 100 / (2)              |
| Biracial                                | 6.2 / (1)               | 9.1 / (1)               | 0 / (0)                |
| Asian                                   | 0 / (0)                 | 9.1 / (1)               | 0 / (0)                |
| <i>Maternal Education [% / (n)]</i>     |                         |                         |                        |
| High School                             | 0 / (0)                 | 0 / (0)                 | 50.0 / (1)             |
| Some College                            | 12.5 / (2)              | 0 / (0)                 | 0 / (0)                |
| 2-year College                          | 6.3 / (1)               | 36.4 / (4)              | 0 / (0)                |
| 4-year College                          | 31.3 / (5)              | 36.4 / (4)              | 0 / (0)                |
| Advanced Professional                   | 50.0 / (8)              | 27.3 / (3)              | 50.0 / (1)             |
| <i>Paternal Education [% / (n)]</i>     |                         |                         |                        |
| High School                             | 18.8 / (3)              | 9.1 / (1)               | 0 / (0)                |
| Some College                            | 18.8 / (3)              | 9.1 / (1)               | 0 / (0)                |
| 2-year College                          | 37.5 / (6)              | 45.5 / (5)              | 0 / (0)                |
| 4-year College                          | 25.0 / (4)              | 27.3 / (3)              | 50.0 / (1)             |
| <i>Degree</i>                           |                         |                         |                        |

Table 2

*Completed Protocols and Questionnaires, by status group (YS = younger sibling, OS = older sibling)*

| Group      | YS Mullen | YS ADOS                                      | YS ADI | YS SRS               | OS Cognitive                               | OS SCQ | OS SRS       | OS ADOS                                      |
|------------|-----------|--|--------|----------------------|--|--------|--------------|--|
| Comparison | 10        | 7 (Module 1)<br>4 (Module 2)                 | 10     | 8 (SRS-P)<br>2 (SRS) | 1 (Mullen)<br>5 (WPPSI-III)<br>5 (WISC-IV) | 11     | 11           | NA   |
| ASD        | 16        | 9 (Module 1)<br>6 (Module 2)<br>1 (Module 3) | 16     | 16                   | 5 (Mullen)<br>4 (WPPSI-III)<br>7 (WISC-IV) | 16     | 16           | 5 (Module 1)<br>5 (Module 2)<br>6 (Module 3) |
| Unresolved | 2         | 1 (Module 1)<br>1 (Module 3)                 | 2      | 2                    | 2 (WPPSI-III)2                             | 2      | 1 (Module 2) | 1 (Module 3)                                 |

Note: NA= Not Administered. OS ADOS not administered in comparison group unless older sibling participant evidenced elevated ASD symptomatology on a screener.

Table 3

*Correlations between younger sibling (YS) autistic symptomatology and cognitive functioning and older sibling (OS) autistic symptomatology and cognitive functioning within sample as a whole*

|                   | YS cognitive | YS symptomatology | OS cognitive | OS symptomatology |
|-------------------|--------------|-------------------|--------------|-------------------|
| YS cognitive      | --           | -.53**            | .62**        | -.46*             |
| YS symptomatology | --           | --                | -.49**       | .39*              |
| OS cognitive      | --           | --                | --           | -.79**            |
| OS symptomatology | --           | --                | --           | --                |

\*\*p<.01

\*p<.05

Table 4

*Correlations between younger sibling (YS) autistic symptomatology and cognitive functioning and older sibling (OS) autistic symptomatology and cognitive functioning for comparison group*

|                   | YS cognitive | YS symptomatology | OS cognitive | OS symptomatology |
|-------------------|--------------|-------------------|--------------|-------------------|
| YS cognitive      | --           | -.01              | .56          | -.15              |
| YS symptomatology | --           | --                | .37          | .11               |
| OS cognitive      | --           | --                | --           | -.19              |
| OS symptomatology | --           | --                | --           | --                |

\*\*p<.01

\*p<.05



Table 5

*Correlations between younger sibling (YS) autistic symptomatology and cognitive functioning and older sibling (OS) autistic symptomatology and cognitive functioning for ASD group*

|                   | YS cognitive | YS symptomatology | OS cognitive | OS symptomatology |
|-------------------|--------------|-------------------|--------------|-------------------|
| YS cognitive      | --           | -.54*             | .49          | -.23              |
| YS symptomatology | --           | --                | -.34         | .02               |
| OS cognitive      | --           | --                | --           | -.66**            |
| OS symptomatology | --           | --                | --           | --                |

\*\*p<.01

\*p<.05

Table 6

Scores on Completed Protocols and Questionnaires, by status group (YS = younger sibling, OS = older sibling)

**COMP-SIBS**

| Mullen | ADOS-Y | ADI | SRS-Y | Cognitive (SS) | SCQ | SRS-O |
|--------|--------|-----|-------|----------------|-----|-------|
| 88     | 4      | 4   | 12    | 84             | 6   | 6     |
| 105    | 2      | 3   | 29    | 98             | 3   | 15    |
| 97     | 5      | 10  | 38    | 114            | 3   | 10    |
| 88     | 2      | 3   | 40    | 100            | 5   | 30    |
| 124    | 1      | 5   | 22    | 100            | 1   | 23    |
| 143    | 1      | 6   | 40    | 123            | 3   | 28    |
| 121    | 0      | 12  | 32    | 118            | 3   | 40    |
| 113    | 6      | 1   | 16    | 123            | 3   | 19    |
| 122    | 2      | 2   | 41    | 119            | 2   | 6     |
| 103    | 4      | 4   | 19    | 124            | 1   | 12    |
| ---    | 7      | --- | ---   | 116            | 5   | 32    |

**ASD-SIBS**

| Mullen | ADOS-Y | ADI | SRS-Y | Cognitive (SS) | SCQ | SRS-O | ADOS-O |
|--------|--------|-----|-------|----------------|-----|-------|--------|
| 100    | ---    | 3   | ---   | 50             | 5   | 59    | 22     |
| 78     | 6      | 10  | 54    | 41             | 22  | 133   | 14     |
| 89     | 5      | 11  | 47    | 55             | 19  | 93    | 19     |
| 67     | 7      | 10  | 43    | 40             | 21  | 125   | 22     |
| 149    | 5      | 0   | 36    | 125            | 3   | 53    | 7      |
| 115    | 3      | 5   | 43    | 70             | 33  | 145   | 19     |
| 97     | 11     | 8   | 17    | 93             | 23  | 49    | 14     |
| 89     | 6      | 13  | 18    | 40             | 26  | 120   | 20     |
| 90     | 1      | 8   | 21    | 74             | 15  | 92    | 8      |
| 85     | 17     | 27  | 101   | 55             | 17  | 110   | 11     |

**ASD-SIBS, cont.**

| Mullen | ADOS-Y | ADI | SRS-Y | Cognitive (SS)SCQ | SRS-O | ADOS-O |    |
|--------|--------|-----|-------|-------------------|-------|--------|----|
| 76     | 15     | 23  | 63    | 49                | 18    | 134    | 16 |
| 88     | 4      | 2   | 18    | 95                | 15    | 105    | 11 |
| 49     | 18     | 13  | 35    | 49                | 11    | 70     | 19 |
| 51     | 9      | 26  | 76    | 104               | 14    | 101    | 7  |
| 98     | 7      | 11  | 3     | 101               | 13    | 16     | 8  |
| 105    | 7      | 11  | 25    | 110               | 10    | 76     | 15 |

**Unresolved**

| Mullen | ADOS-Y | ADI | SRS-Y | Cognitive (SS)SCQ | SRS-O | ADOS-O |    |
|--------|--------|-----|-------|-------------------|-------|--------|----|
| 118    | 15     | 4   | 91    | 93                | 10    | 24     | 11 |
| 89     | 5      | 3   | 50    | 77                | 9     | 81     | 3  |

Table 7

Means and standard deviations of protocols and questionnaires, by status group (YS = younger sibling, OS = older sibling)

| <u>Group</u>         | <u>YS Mullen</u> | <u>YS ADOS</u>   | <u>YS ADI</u> | <u>YS SRS</u>   | <u>OS Cognitive</u>  | <u>OS SCQ</u> | <u>OS SRS</u> | <u>OS ADOS</u>  |
|----------------------|------------------|--|---------------|---|--|---------------|---------------|---|
| Comparison<br>(n=11) | 110.4 (17.5)     | <i>Module 1</i><br>2.9 (2.5)<br><i>Module 2</i><br>3.5 (1.9)                           | 5 (3.5)       | <i>SRS-P</i><br>28.5 (12.4)<br><i>SRS</i><br>30.5 (2.1) | <i>Mullen</i><br>118<br><i>WPPSI-III</i><br>103.8 (15.1)<br><i>WISC-IV</i><br>116.4 (9.7)        | 3.1 (1.6)     | 20.1 (11.4)   | NA  |
| ASD<br>(n=15)        | 89.2 (24.0)      | <i>Module 1</i><br>9.0 (5.6)<br><i>Module 2</i><br>7.0 (5.3)<br><i>Module 3</i><br>4.0 | 11.3 (8.0)    | <i>SRS</i><br>40.1 (25.7)                               | <i>Mullen</i><br>66.6 (32.7)<br><i>WPPSI-III</i><br>100.0 (4.6)<br><i>WISC-IV</i><br>64.8 (26.6) | 16.4 (7.7)    | 92.6 (36.2)   | <i>Module 1</i><br>16.0 (3.7)<br><i>Module 2</i><br>16.8 (6.7)<br><i>Module 3</i><br>11.0 (5.0) |
| Unresolved<br>(n=2)  | 103.5 (20.5)     | <i>Module 1</i><br>5.0<br><i>Module 3</i><br>12.0                                      | 3.5 (0.7)     | <i>SRS-P</i><br>70.5 (29.0)                             | <i>WPPSI-III</i><br>85.0 (11.3)  | 9.3 (1.1)     | 52.5 (40.3)   | <i>Module 2</i><br>3.0<br><i>Module 3</i><br>11.0   |

Note: NA= Not Administered. OS ADOS not administered in comparison group unless older sibling participant evidenced elevated ASD symptomatology on a screener.

Table 8

*Between Group Analyses of Variance for Measures of ASD symptomatology (YS = younger sibling, OS = older sibling)*

| Source   | <i>df</i> | <i>error</i> | <i>F</i> | <i>p</i> | <i>COMP-sibs mean(SD)</i> | <i>ASD-sibs mean (SD)</i> |
|----------|-----------|--------------|----------|----------|---------------------------|---------------------------|
| YS ADOS  | 1         | 24           | 8.28     | < .01    | 3.1 (2.3)                 | 8.1 (5.1)                 |
| YS ADI-R | 1         | 24           | 5.62     | < .05    | 5.0 (3.5)                 | 11.3 (8.0)                |
| YS SRS   | 1         | 23           | 1.66     | > .05    | 28.9 (11.0)               | 40.1 (25.7)               |
| OS SCQ   | 1         | 25           | 32.10    | < .01    | 3.1 (1.6)                 | 16.4 (7.7)                |
| OS SRS   | 1         | 25           | 40.81    | < .01    | 20.1 (11.4)               | 92.6 (36.2)               |

Table 9

Means and standard deviations of cognitive subtests, by status group, (YS = younger sibling, OS = older sibling)

| <u>Group</u>                  | <u>YS Mullen</u>   | <u>OS Mullen</u>   | <u>OS WPPSI-III</u>                                 | <u>OS WISC-IV</u>  |
|-------------------------------|--|--|---|--|
| Comparison<br>( <i>n</i> =11) | Receptive Language<br>53 (9.7)<br>Expressive Language<br>58.4 (13.4)<br>Visual Reception<br>60.2 (13.1)    | Language Average<br>60.5<br>Visual Reception<br>56.0               | Verbal<br>111.0 (24.3)<br>Performance<br>97.3 (7.8) | Verbal Comprehension<br>116.2 (9.1)<br>Perceptual Reasoning<br>95.2 (46.8) |
| ASD<br>( <i>n</i> =15)        | Receptive Language<br>40.5 (13.9)<br>Expressive Language<br>47.3 (12.7)<br>Visual Reception<br>44.9 (18.2) | Language Average<br>30.0 (18.7)<br>Visual Reception<br>32.8 (17.6) | Verbal<br>104.7 (4.6)<br>Performance<br>102.0 (1.7) | Verbal Comprehension<br>68.0 (24.0)<br>Perceptual Reasoning<br>74.0 (27.4) |
| Unresolved<br>( <i>n</i> =2)  | Receptive Language<br>42.0 (5.7)<br>Expressive Language<br>50.5 (2.1)<br>Visual Reception<br>52.5 (10.6)   |  | Verbal<br>103.3 (20.1)<br>Performance<br>96.6 (9.9) |  |

Table 10

*Between Group Analyses of Variance for Measures of ASD symptomatology (YS = younger sibling, OS = older sibling)*

| Source                        | <i>df</i> | <i>error</i> | <i>F</i> | <i>p</i> | <i>COMP-sibs mean(SD)</i> | <i>ASD-sibs mean (SD)</i> |
|-------------------------------|-----------|--------------|----------|----------|---------------------------|---------------------------|
| YS Mullen Expressive Language | 1         | 24           | 4.35     | < .05    | 58.4 (13.4)               | 47.3 (12.7)               |
| YS Mullen Receptive Language  | 1         | 24           | 6.13     | < .05    | 53 (9.7)                  | 40.5 (13.9)               |
| YS Mullen Visual Reception    | 1         | 24           | 5.21     | < .05    | 60.2 (13.1)               | 44.9 (18.2)               |
| OS Mullen Language Average    | 1         | 5            | 2.22     | > .05    | 60.5                      | 30.0 (18.7)               |
| OS Mullen Visual Reception    | 1         | 5            | 1.45     | > .05    | 56.0                      | 32.8 (17.6)               |
| OS WPPSI Verbal               | 1         | 7            | 0.19     | > .05    | 111.0 (24.3)              | 104.7 (4.6)               |
| OS WPPSI Performance          | 1         | 7            | 0.97     | > .05    | 97.3 (7.8)                | 102.0 (1.7)               |
| OS WISC Verbal Comprehension  | 1         | 12           | 18.07    | < .01    | 116.2 (9.1)               | 68.0 (24.0)               |
| OS WISC Perceptual Reasoning  | 1         | 12           | 1.09     | > .05    | 95.2 (46.8)               | 74.0 (27.4)               |