

PSYCHOLOGICAL AND GENETIC CONTRIBUTIONS TO THE  
DEVELOPMENT OF SOCIAL COGNITION IN CHILDREN

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This study examined the development of social cognition in children with and without autism spectrum disorder (ASD), as well as the influence of behavioral and molecular genetics on these higher-order cognitive abilities. Specifically, it was hypothesized that children with ASD would perform more poorly on all social cognitive tasks compared with typically developing peers. In addition, it was hypothesized that typically developing children who performed better on a simpler social cognitive task at ages 3 or 4 would perform better at follow-up (i.e., one time between the ages of 6-10). Lastly, it was hypothesized that children who had at least one risk allele in both the DRD4 and the 5-HTTLPR polymorphisms would perform worse than those who had at least one risk allele in either polymorphism, who, in turn, would perform worse than children without any risk alleles. The twin sample included 62 families of multiples (twins, triplets, or quadruplets) who were recruited through the Southern Illinois Twins and Siblings Study (SITSS), and the ASD sample included 25 children who were recruited from the Center for Autism Spectrum Disorders at SIU. Significant group differences were found for children's performance on all of the social cognitive tasks. Furthermore, results showed that some areas of social cognition (theory of mind and the understanding of non-literal language) are more influenced by genetic factors than are other cognitive skills. Lastly, results from the molecular genetic analyses suggest that basic social cognitive skills (e.g., theory of mind) may be influenced by underlying biological factors in the serotonergic and dopaminergic pathways. The

present study provided useful information on how psychological and genetic factors influence the development of social cognitive abilities in children with and without ASD.

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## CHAPTER 1

### INTRODUCTION

Social cognition involves a complex interplay of different abilities, drawing from social, cognitive, emotional, and behavioral domains. Understanding social cues, being able to take another's perspective, interpreting others' emotional displays, learning from prior social interactions, and applying that learning to future interactions are the necessary steps to a successful social exchange (Baron-Cohen, 2000; Crick & Dodge, 1994; Lemerise & Arsenio, 2000). Theory of mind (ToM) tasks, which measure the ability to infer others' mental states, are commonly used to study social cognition in children (Wellman, Cross, & Watson, 2001). Recently, Peterson, Wellman, and Slaughter (2012) validated the addition of a new step to the well-established preschool ToM Scale created by Wellman and Liu (2004). This new step extends developmental milestones to include an understanding of nonliteral language such as sarcasm in slightly older children (i.e., school-age). Limited research using this newly validated ToM Scale exists; therefore, this study added to the literature in this area. In addition, the Social Responsiveness Scale, 2<sup>nd</sup> Edition (SRS-2; Constantino & Gruber, 2012), a newly released edition that includes a social cognition treatment subscale, was used in this study. The study used both a longitudinal and a cross-sectional design and included typically developing twins, as well as children diagnosed with autism spectrum disorder (ASD).

The first goal of this study was to better understand the development of social cognition in typically developing children. This was done by using data previously collected from twin pairs who participated in the Southern Illinois Twins and Siblings Study (SITSS; DiLalla, 2002a). To assess longitudinal progression in the acquisition of ToM skills, 6- to 10-year-old twins were administered five ToM measures in the follow-up portion of this study. Data

collected from the ToM tasks at ages three and four was used to predict current performance on the ToM tasks in this study.

The second goal of this study was to compare the social cognitive abilities of typically developing twins to those of children with autism spectrum disorder (ASD), who historically have performed poorly on measures of ToM (Baron-Cohen, Leslie, & Frith, 1985). Therefore, children between the ages of 6 and 10 who have been diagnosed with ASD were administered the same five measures of ToM as the ones administered to the twins. Given that group differences emerged when comparing the performance of children with ASD to typically developing children, this provides further support for the idea that children with ASD have impairments in social cognitive abilities.

The third goal of this study attempted to shed light on the relation between genetic influence and the development of social behaviors in young children with and without social impairment. Several recent studies have shown that social cognition may be related to genetic variations in the dopamine and/or serotonin systems (Lackner, Bowman, & Sabbagh, 2010; Lackner, Sabbagh, Hallinan, Liu, & Holden, 2012; Skuse & Gallagher, 2011). Thus, the neurotransmitters dopamine and serotonin were selected for this study because of their implication in the development of social cognitive abilities (Rogers, 2011; Skuse & Gallagher, 2011). More specifically, the rewards associated with experiencing social interactions are related to the dopamine system, whereas the emotional experience of social interaction is associated with serotonin. Therefore, this study examined how dopaminergic and serotonergic risk alleles (i.e., DRD4 and 5-HTTLPR, respectively) were related to social behaviors such as ToM.

Overall, this study aimed to advance knowledge of the psychological and genetic contributions to the development of social cognition in both typically and atypically developing

samples of children. Research such as this is needed to better understand the complexities of social interactions and the ability to take another person's perspective, especially when someone else's beliefs are different from one's own. This study has potential to be useful clinically in terms of better understanding a potential source of difficulty that school-aged children may be having in peer relationships (i.e., inability to take another's perspective). Lastly, the inclusion of a clinical comparison group allowed for the investigation of group differences in ToM task performance, which could inform development of social cognitive curricula used in school- and community-based interventions.

## CHAPTER 2

### LITERATURE REVIEW

#### **Social Cognition**

The development of social cognition in children has been a rapidly expanding area of interest over the past few decades. The understanding of this higher-order ability warrants attention given the fact that humans spend a significant amount of time in social relationships with others (Astington, 1993). Despite the popularity of the topic, researchers disagree on fundamental aspects of its definition such as when this ability emerges in children. A growing body of literature supports the idea that children as young as infants have the ability to navigate social situations by using social cues (Rakoczy, 2012; Yott & Poulin-Dubois, 2012). For example, an infant who is startled by a barking dog for the first time may look to her parent to see if the dog is “safe” by referencing her parent’s facial expression (e.g., a smile or a fearful face). Assessing social cognitive abilities is difficult in young children given their limited verbal abilities. Much of the previous research in this area has focused on precursors, or building blocks, of social cognition in younger children, which include engaging in joint attention, understanding the “intentionality” of actions, recognizing that others have different perspectives, and using imagination in play (Baron-Cohen, 1991; Miller, 2006).

Social cognition goes beyond perspective taking, however, because it is a heterogeneous term that encompasses literature from social psychology (e.g., schemas, attributions, stereotypes) and cognitive psychology (e.g., reasoning, attention, memory; Fiske & Taylor, 1991).

Perspective taking, on the other hand, is defined as a construct that can be assessed only by behavioral means, but it does not account for the contributions from cognitive and emotional states. Furthermore, social cognition can be defined as the ability to understand others’ attitudes, beliefs, values, desires, and social knowledge (Astington, 1993). It also can refer to aspects of

higher cognitive functioning that allow for the understanding of “one’s own and others’ minds” (Baron-Cohen, 2000). Although most children develop the skills needed for navigating the social world as they progress through childhood and adolescence, some children and adolescents have more difficulty than others. For example, children and adolescents with externalizing disorders such as conduct disorder, especially those with comorbid aggressive features, are often impaired in social cognitive abilities (Lochman & Dodge, 1994). These children are believed to have difficulty processing social information when interacting with others. In addition, children with autism spectrum disorder (ASD) are also impaired in social cognitive functioning. In fact, Baron-Cohen (2000) argued that all individuals with ASD have deficits in social cognition, suggesting that it may be a hallmark characteristic of the disorder. Individuals who have difficulty navigating social situations such as children with conduct disorder or ASD have a wide variety of social impairments, ranging from mild to severe. It is believed that these individuals are impaired in their ability to follow the necessary steps to reach the appropriate behavioral outcome when processing social cues or other social information.

### **Social Information Processing Theory**

Social Information Processing Theory (Crick & Dodge, 1994) is a well-known model that was modified from an original model by Dodge (1986). This theory states that people enter into social relationships with prior knowledge of how to navigate new social situations based on their previous experiences. According to Crick and Dodge, people typically rely on past memories, schemas, and social scripts in order to dictate their future behavior. Furthermore, individuals placed in novel social situations are believed to process social information in a systematic way through a series of processing steps. These processing steps include: 1) encoding of external and internal cues; 2) interpretation of those cues; 3) selection of goals; 4) mental access to possible

behavioral responses; 5) selection of a particular response; and 6) behavioral enactment of selected response. Crick and Dodge suggested that social information processing through the use of these six steps is not linear. Furthermore, they believe each step can interact with the others, and the social information processing is subject to feedback at any step along the way.

During a novel social interaction, a child is processing the first two steps: encoding and interpreting cues. The memory of past experiences is very important to these initial steps because a child often remembers previous social interactions that may not have gone well. A child's own internal cues that he is the target of bullying yet again will send feedback to his social processing system. For example, a child who is frequently teased by other peers is likely to view a more neutral situation as negative and hostile rather than accidental. This attribution will have a direct effect on the third step, or the generation of possible goals. Given that the child may be feeling defensive from being bullied, his first choice for a goal may be to get back at the other child who teased him. The fourth step, response access, describes an individual's ability to think back through their responses to similar past experiences for ways to deal with the current situation. If a child is most likely to retaliate in an aggressive manner when provoked, then aggressive options are most likely to come to mind in the new social situation. During the fifth step, response decision, a child selects his/her response of choice from among the possible options. Crick and Dodge noted that concepts related to social psychology such as self-efficacy, expectations, and morals are taken into consideration at this stage. For example, if a child is more confident that a particular solution is going to work, then she might be more likely to choose that option in this step. In addition, a child is more likely to select an option not only if he believes it is going to solve the problem, but also if the solution aligns with his moral views (e.g., believing it is okay to steal from another person). Lastly, the sixth step, behavioral

enactment, is the act of going through with the selected choice from the previous step. Crick and Dodge emphasize in the social information processing model that multiple factors are taken into account when choosing the final decision.

More recently, Lemerise and Arsenio (2000) have outlined some additions to the theory initially proposed by Crick and Dodge (1994). The most notable contribution was the addition of emotional processing as a factor in each of the steps outlined by Crick and Dodge. Lemerise and Arsenio argued that each person brings a unique emotional response style, arousal level, and mood to social interactions. When interacting with others in a social manner, both individuals' emotional states were important factors in determining the eventual quality of the interaction. If one person was overly emotional, this behavior was likely to influence the goal selected and subsequent actions of the other.

In summary, individuals who have difficulty navigating social interactions with others, such as those who diagnosed with conduct disorder or autism spectrum disorder (ASD), are believed to have social information processing deficits. Crick and Dodge (1994) outlined six steps necessary for processing social cues (i.e., cues taken from others and one's own internal experiences) that lead to an actual or predictable behavior. Lemerise and Arsenio (2000) modified Crick and Dodge's model by adding an emotional component to each step outlined in the Social Information Processing Theory. Both sets of researchers describe the interaction of the steps within the models and the importance of feedback at each stage. Thus, successful social interactions require the integration of social, cognitive, emotional, and behavioral aspects, learning from prior social experiences, and the ability to take another's perspective, also known as Theory of Mind, which will be discussed next.

## **Theory of mind (ToM)**

Social cognition is a broad construct that is comprised of social and cognitive components. The development of theory of mind (ToM) is an example of one such social-cognitive skill necessary for attaining social competence. Having a true understanding of ToM implies that an individual recognizes that one can never fully understand another's mind; however, one can assume that others also think and reason because they themselves are able to do the same. Wellman and Liu (2004) used the term "mental subjectivity" to describe the ability to understand another's mental state as a response to objective events or observable behaviors. Furthermore, ToM is defined as the ability to understand how human behavior is dictated by mental states of actual and false beliefs, the intentions of self and others, memory of past experiences, and desire to perform specific behavioral responses (Peterson et al., 2012). This social knowledge allows individuals to explain and predict the behavior of those with whom they are interacting (Wellman & Estes, 1986).

Given that children, especially pre-verbal infants, do not have the capacity to understand another's perspective at an adult-like level, research has focused on the developmental changes of ToM in young children. Many researchers have shown that the preschool years between three and five years of age are when most children rapidly develop the ability to complete ToM tasks (de Villiers & Pyers, 2002; Flavell, Everett, Croft, & Flavell, 1981; Wellman et al., 2001; Youngblade & Dunn, 1995). Some researchers have argued that children as young as two years old may be able to begin to understand what others want, see, and feel, albeit at a simple level (Bartsch & Wellman, 1995; Wellman, Phillips, & Rodriguez, 2000). By the age of three, young children are able to tell the difference between what others are thinking and what others are doing and from that age on can distinguish between objectivity and subjectivity (Flavell, Flavell,



Green, & Moses, 1990; Watson, Gelman, & Wellman, 1998; Wellman & Estes, 1986). One study by Flavell, Green, and Flavell (1995) showed that children who are between three and five years of age begin to differentiate between thinking (i.e., mental states) and doing (i.e., behavior). Children begin to comprehend that by understanding another's mental state, it is easier to justify why someone chooses to act in a certain way (Wellman & Lagattuta, 2000).

The focus of most research on the development of ToM in children between the ages of three and five has concerned false beliefs. False belief tasks are designed such that a child is asked to explain or predict a situation from another's perspective regarding a belief that does not match reality. For example, a child is shown a container and asked what is inside without having seen the contents. When shown that the contents do not match the container in which the items were kept, the child is then asked what another person who has not seen the inside would say is in the container. Perner and Wimmer (1985) outlined two different types of false belief attributions: first-order and second-order. First-order belief attributions involve Person A making a judgment about Person B's belief related to an event that occurred or an object that was shown to Person A but not to Person B. For example, if Person A is asked what is behind a cupboard door without having seen inside, Person A likely will give a practical response such as "dishes." Person A is shown a shoe in the cupboard (i.e., an unexpected response), and then asked what his friend might say is in the cupboard. A child who understands first-order beliefs would say that Person B thinks there are dishes in the cupboard when Person A now knows that there is a shoe in the cupboard. A second-order belief attribution involves attributing a belief to one person about another person's beliefs. An example of this is a child saying, "Jimmy thinks that Mary thinks that..." This more advanced second-order attribution ability is believed to be present by the age of seven in typically developing children (Perner & Wimmer, 1985). Many

children who are three years of age fail false belief tasks; however, by four to five years of age, most typically developing children are able to pass false belief tasks (Flavell, Everett, Croft, & Flavell, 1981; Wellman et al., 2001).

### **Implicit/indirect versus explicit/direct tasks**

Research on ToM tasks can be divided into two categories: implicit/indirect tasks and explicit/direct tasks (Apperly & Butterfill, 2009; Low & Perner, 2012). Studies differentiating between indirect and direct tasks attempt to answer the question of whether a young child under the age of three is unable to complete ToM tasks due to developmental constraints or if the method used to measure ToM is hindering their ability to answer correctly. As a way to eliminate the language demands of ToM tasks, several studies using infants have used implicit/indirect tasks such as an anticipatory looking task to measure the development of ToM. During one type of anticipatory looking task first used by Clements and Perner (1994), infants were trained to watch a hand move in a predictable pattern to grab one of two objects on opposite sides of a small platform. After the infants were habituated to this behavior, the two objects were switched and the hand either reached for the same object in the opposite location or a new object in the old location. The infants tended to look longer at the new object along the same path as before than at the new path with the old object in its new location. This suggests that infants are surprised about the new event that occurred, but this does not necessarily support the fact that infants understand others' mental states. Evidence for an earlier understanding of ToM has been shown in infants as young as 18 months old (Neumann, Thoermer, & Sodian, 2008) and 25 months old (Southgate, Senju, & Csibra, 2007); however, Sodian (2011) argued that these studies may not be measuring the same cognitive ability as the ToM tasks administered to verbal children through the use of explicit/direct tasks. Implicit/indirect tasks have the benefit of being

a more efficient and developmentally appropriate way to measure ToM in infants compared with explicit/direct tasks; however, the former are less cognitively demanding and more inflexible (Apperly & Butterfill, 2009).

The second type of task, explicit/direct tasks, is more cognitively demanding but more flexible. False belief tasks are direct measures of ToM because they elicit verbal responses from the child who is performing the task. Rather than implying the belief indirectly through a nonverbal anticipatory looking task with infants, explicit/direct tasks can be answered directly using open-ended questions. An example of this type of task is the cupboard example that was previously described. Explicit/direct tasks were the type of task used in this study because the children with and without autism spectrum disorder had the language ability necessary to respond verbally to ToM items.

Recent research by Wellman & Liu (2004) has suggested that ToM may not be an isolated skill, but rather children may progress through a series of skills related to ToM. Support for this progression comes from a preliminary meta-analysis by Wellman and Liu (2004) and prior work by other researchers (Astington, 2001; Flavell & Miller, 1998; Repacholi & Gopnik, 1997; Wellman & Woolley, 1990). Results from the meta-analysis showed that children are able to accurately judge another person's desires (i.e., whether people want a particular object or not) prior to being able to accurately judge another person's beliefs (i.e., people's subjective thoughts about an objective event). Furthermore, Wellman & Liu (2004) reported that children were able to successfully perform belief tasks that do not involve deception before being able to pass false belief tasks. Interestingly, children are even able to indicate that someone else might not know what is in a container before reporting that another person falsely believes a particular object is in the container. Although the authors indicated that the meta-analysis results were preliminary,

there was significant evidence to support the claim that ToM develops in a progressive manner, which is consistent with social cognitive theory (Bandura, 1989). Follow-up studies from the same researchers have also supported this progression (Peterson et al., 2012; Wellman, Fang, & Peterson, 2011).

### **Developmental progression of theory of mind**

The second part of the study by Wellman & Liu (2004) tested the differences in abilities based on the results from the preliminary meta-analysis and results from previous studies suggesting that the skills may develop in a linear fashion. The results of the second part of the study provided psychometric support for a 5-step model of progressive ToM skills. Performance on ToM tasks in a sample of 75 children between three and five years of age were analyzed using a Guttman (1944, 1950) scale, which is a measure of how actual item responses fit an ideal pattern. The results revealed that 80% of children (i.e., 60 out of 75) had a similar progression of skill acquisition. More specifically, children were able to pass the following tasks in sequential order: diverse desire (understanding that people express differing desires for the same object), diverse belief (understanding that people have different beliefs about the same objective event), knowledge access (understanding that someone may be unaware of a certain fact), contents false belief (understanding that, despite a certain fact, someone might believe something differently), and lastly real-apparent emotion (understanding that someone can show a different emotion than the one they are feeling). One drawback to the Guttman scaling is that if two items are similar, one item is excluded because it is believed to be representing a similar, or redundant, construct. The Guttman scale in the Wellman & Liu (2004) study excluded two items: explicit false belief and belief-emotion. Although these items were excluded from the Guttman scale, that does not necessarily mean they are not important in the development of ToM abilities. Therefore, these

tasks were included in the present study, especially in light of the importance of emotion in social information processing as previously discussed (Lemerise & Arsenio, 2000). Given that all children over the age of four are presumed to be able to pass the diverse desire and diverse belief tasks, those were not administered as part of this study.

In addition to the four remaining tasks (i.e., contents false belief, explicit false belief, belief emotion, and real-apparent emotion) outlined by Wellman & Liu (2004), an additional task was added to tap the understanding of non-literal language (i.e., irony, sarcasm) in this study given that the children in this study were 6- to 10-year-olds. Peterson et al. (2012) argued for the addition of a more advanced ToM task for school-age children given that most children will have successfully mastered the understanding of false belief tasks by the age of five. In the study by Peterson et al. (2012), 184 children aged 3-12, who were typically developing, deaf, or had autism or Asperger syndrome, completed Wellman & Liu's (2004) 5-step ToM tasks with the addition of a new step tapping sarcasm. The results indicated that children with autism or Asperger syndrome were not only delayed in their ability to acquire ToM, but their pattern of abilities was different than the other groups. More specifically, these children were more likely to pass the hidden emotion (i.e., real-apparent emotion) task before the false belief task. This suggests that children with autism may not only be delayed in developing social cognitive skills in general, but they also may exhibit an atypical pattern of acquiring the ability to understand others' mental states compared to typically developing children. In contrast, the typically developing children progressed on schedule through the items in the predicted pattern previously outlined. Moreover, the deaf children were delayed in their abilities, but they had the same pattern as the typically developing children. Thus, children with autism and children who were deaf had delays compared with typically developing children on the new scale even after

controlling for age and language ability. Peterson and colleagues showed that the additional ToM step (i.e., understanding non-literal language such as sarcasm) was a reliable and valid addition to the previous ToM Scale outlined by Wellman & Liu (2004). The authors added that the inclusion of this step allows for an increase in the age range of children in future studies of the developmental progression of social cognitive abilities.

In conclusion, a substantial amount of research in the past has focused on the ability of children to successfully complete explicit/direct ToM tasks such as false belief tasks. However, a recent review by Wellman (2002) argues that ToM is comprised of several different sequential components that build on and interact with one another. A 5-step ToM Scale was developed by Wellman & Liu (2004) and was recently extended by adding a sixth step, which assesses the ability to understand non-literal language (Peterson et al., 2012). Typically developing and deaf children appear to follow a predictable pattern, with each successive step building on the previous one. Children with ASD are not only delayed in their ability to perform these tasks, they also complete the tasks in a somewhat different order (i.e., understanding hidden emotion tasks before false belief tasks). Thus, studies on the atypical development of ToM abilities should include children with autism spectrum disorder.

### **Autism Spectrum Disorder (ASD)**

Autism spectrum disorder (ASD) is a severe, pervasive, and complex neurodevelopmental disorder characterized by impairments in social-emotional functioning and communication abilities and the presence of restricted, repetitive, or stereotyped interests and behaviors (APA, 2000). Common social impairments include a lack of reciprocal social interaction, poor eye contact, and difficulty engaging in joint attention. Communication deficits can include failure to develop verbal communication, use of echolalia (i.e., repeating others'

speech or lines from television shows or movies), verbal rituals (i.e., requiring others to say phrases in certain ways), and difficulty initiating or maintaining conversations with others.

A modification to the current diagnostic criteria in the Diagnostic and Statistical Manual of Mental Disorders, 5<sup>th</sup> Edition (DSM-5; APA, 2013) included lumping the social and communication symptoms into one domain. Impairments such as difficulty understanding non-verbal communication (e.g., reading facial expressions, understanding the prosody of speech) and difficulty engaging in reciprocal social interactions may be better described as difficulties in social-communication abilities rather than fitting neatly into one category or the other. In addition to social-communication deficits, many children with ASD exhibit a restricted, repetitive, or stereotyped pattern of behavior or interests. For some children, a restricted interest may manifest in an intense preoccupation with particular objects (e.g., trains, wheels on a toy car) or topic (e.g., knowledge of dinosaurs, transportation schedules, makes and models of cars). Other common restricted behaviors include a rigid insistence on sameness in daily routine (e.g., requiring a parent to drive a certain way to school or to the grocery store) or in the organization of household items (e.g., lining up toys, insisting that canned food labels all face in the same direction). Children who engage in repetitive behaviors (e.g., opening and closing a door) often become upset when told to stop performing the repetitive action. Commonly reported stereotyped behaviors include spinning, rocking, or hand flapping.

As previously noted in DSM-IV (APA, 2000), children with Autistic Disorder were considered to be more impaired (e.g., exhibiting limited language functioning or being nonverbal) than children with Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS) or Asperger syndrome. At present, the diagnostic criterion no longer distinguishes between children who did or did not have delayed language acquisition (previously a

requirement to meet the diagnostic criteria for Asperger syndrome). These children tended to be known as “higher functioning” and typically attended the regular education classroom, whereas children with Autistic Disorder were commonly placed in special education. Children with PDD-NOS were typically in mainstream classrooms, depending on their level of functioning, but some may have been in special education for part of the day, such as for academic subjects, and with typically developing peers for special subjects (e.g., physical education, art, music). Presently, the “spectrum” modifier in the new diagnostic category “Autism Spectrum Disorder” allows for the inclusion of children with a range of abilities across the areas of social-communication and restricted and repetitive behaviors. Currently, individuals meeting criteria for ASD must show impairments in social-communication abilities, as well as the presence of restricted and repetitive behaviors. If a child does not present with restricted and repetitive behaviors but does exhibit age-inappropriate levels of social-communication abilities, the child meets criteria for Social Communication Disorder, a new disorder included in DSM-5.

Gold standard diagnostic practices recommend that if a young child fails a screening test, a trained professional (e.g., a psychologist) should administer a battery of follow-up tests, which includes the Autism Diagnostic Interview-Revised (ADI-R; Lord, Rutter, & Le Couteur, 1994) and the Autism Diagnostic Observation Schedule (ADOS; Lord, Rutter, Goode, Heemsbergen, Jordan, Mawhood, & Schopler, 1989). Recent research has shown that children with ASD can be reliably differentiated from children without ASD and from children with mental retardation at 12 months of age (Osterling, Dawson, & Munson, 2002). This suggests that early identification is possible, which is important for parents and healthcare professionals to know, especially in light of the fact that early intensive behavioral intervention often is effective at improving several areas of functioning such as IQ, adaptive behavior, personality, and school



placement (i.e., advancing from special education to the regular education classroom; Eldevik, Hastings, Hughes, Jahr, Eikeseth, & Cross, 2009). In fact, timing is so important, that the earlier that children begin receiving intensive services, the better off they are at follow-up in terms of outcomes across a range of domains (i.e., cognitive, social, and adaptive functioning), with some potentially becoming indistinguishable from peers by middle school (Helt et al., 2008).

**Autism prevalence rates.** Current estimates of autism prevalence are as high as one in every 88 children, according to a recent report by the Center for Disease Control and Prevention (CDC, 2012). A prior report in 2009 by the CDC estimated that 1 in 110 children between the ages of three and 10 met criteria for ASD. The report indicated that there has been a 78% increase over the past five years and a 10-fold increase over the past decade. There is significant debate over the reasons for the increase in prevalence rate in recent years. One of the commonly agreed up reasons for the increase is the direct result of broadening the diagnostic criteria to include a "spectrum" of social behaviors, communication abilities, and restricted or repetitive interests/behaviors. By expanding the diagnosis to include children with impairments of a lesser severity than children with Autistic Disorder, many more children began receiving diagnoses of PDD-NOS and Asperger syndrome. In addition, screening measures and diagnostic practices were improving such that children who were being "missed" in the past were no longer being overlooked when given the gold standard diagnostic test battery including the ADI-R and ADOS, as well as a cognitive ability measure to estimate intellectual functioning.

An additional reason for the increase came to light following a study done in California by researchers at Columbia University that showed that children previously meeting criteria for intellectual disability (i.e., mental retardation) in the past were increasingly being more accurately diagnosed with ASD (King & Bearman, 2009). This change alone was estimated to

account for 26% of the increased number of cases. Another factor includes the increased awareness of the disorder among parents, healthcare professionals, and educators. Better dissemination of knowledge of the characteristics of the disorder, typically among parents, was estimated to increase the number of diagnoses by 16% (Liu, King, & Bearman, 2010).

Another 11% of the reported increase in ASD prevalence was attributed to social factors such as advanced parental age, specifically that of the mother. This finding is not unusual given that other studies have shown that advanced parental age is associated with other developmental disorders such as mental retardation. Grether, Anderson, Croen, Smith, and Windham (2009) reported that a 10-year increase in the age of the mother, especially for woman over the age of 40, dramatically increased their risk of having a child with ASD by 38% (Liu, Zerubavel, & Bearman, 2010). The mechanism behind the increase in rates of ASD in older parents is currently unknown, but it is the focus of several studies in progress such as the Early Autism Risk Longitudinal Investigation, which is funded by the National Institutes of Health and Autism Speaks.

In terms of etiology, autism spectrum disorder is believed to be the most highly heritable neurodevelopmental disorder in children (APA, 2000). The strong genetic influence in ASD has been documented in many twin and family studies (see Folstein & Rosen-Sheidley, 2001, for review). Heritability estimates as high as 60–70% have been reported in the literature (Veenstra-VanDer-Weele et al., 2004). Furthermore, Hallmayer and colleagues (2011) reported results from a study of 192 twin pairs (54 monozygotic [MZ], or identical, twins and 138 dizygotic [DZ], or fraternal, twins) of which at least one twin was diagnosed with ASD. Concordance rates for MZ twins ranged from 50% to 71% for male and female twin pairs, respectively, whereas concordance rates for DZ twins ranged from 31% to 36% for male-male and female-

female twin pairs, respectively. Another twin study by Rosenberg et al. (2009) also reported higher concordance rates for MZ twins (88%) versus DZ twins (31%) across the autism spectrum. This study included 277 twin pairs (67 MZ twins and 210 DZ twins) with at least one twin with autism spectrum disorder. Rosenberg and colleagues (2009) investigated not only the concordance rates of MZ and DZ twins, but also the concordance rates when taking comorbid conditions into consideration. More specifically, DZ twins were significantly more discordant on overall loss of skills, and there was a trend toward DZ twins being more discordant on intellectual disability (i.e., by parent report or by documented IQ), timing of achieving developmental milestones, and early loss of social skills compared with MZ twins.

Although ASD is believed to be highly heritable, environmental influences cannot be ruled out. Not surprisingly, environmental factors have been the subject of several recent studies of children with autism spectrum disorder (Hallmayer et al., 2011). A recent meta-analysis of 40 studies by Gardener, Spiegelman, and Buka (2011) reported that children were at higher risk for ASD if there were perinatal or birth complications (e.g., low birth weight, multiple births, maternal infection during pregnancy). The authors of the meta-analysis concluded that there was not enough information to indicate that one risk factor alone is responsible for an increased risk of developing ASD. Rather, it is likely the interaction of multiple perinatal factors that may be responsible for the elevated risk.

In summary, children with ASD are impaired in social and communication abilities and are characterized by restricted, repetitive, or stereotyped patterns of behavior and interests (APA, 2013). Prevalence estimates for this neurodevelopmental disorder have been reported to be as high as 1:88 children, according to the most recent report by the CDC (2012). Several factors have been identified as contributing factors (e.g., increased parent and clinician awareness,

broadening of the definition, advanced parental age), but these factors alone do not explain the 600% increase in rates over the past decade. ASD is a highly heritable disorder, as shown by the high concordance rates between monozygotic (MZ) twins versus dizygotic (DZ) twins.

However, genetics alone do not account for the high rates of the disorder, suggesting that environmental influences also are likely at play. A recent meta-analysis of perinatal factors suggested that birth complications might be contributing to the increased rates of the disorder; however, no single factor was isolated as being sufficient for the disorder (Gardener et al., 2011).

### **The Relation Between Autism Spectrum Disorder and Theory of Mind**

Impairments in individuals with autism spectrum disorder (ASD) go beyond social difficulties (e.g., difficulty starting or maintaining a conversation with a peer) to include cognitive components as well (e.g., difficulty understanding others' mental states). Deficits in social cognition in children with ASD have been routinely reported in the literature using theory of mind (ToM) tasks. In a review of the relation between ToM abilities and autism by Baron-Cohen (2000), the author noted that several studies have shown that children with autism had difficulties understanding others' mental states when compared with typically developing peers with comparable mental ages. During these tasks, children with autism reported what they believed was true rather than indicating what another person might be thinking (Baron-Cohen et al., 1985; Leekam & Perner, 1991; Perner, Frith, Leslie & Leekam, 1989; Reed & Peterson, 1990; Swettenham, 1996; Swettenham, Baron-Cohen, Gomez & Walsh, 1996). One explanation for the difference in ToM performance between typically developing children and children with ASD may be that children with ASD lack the understanding of where the knowledge needed to explain a given situation comes from. For example, Pratt and Bryant (1990) tested three- and four-year-olds on a "seeing leads to knowing" task to determine the plausibility of an event

occurring based on knowledge differences (i.e., having or lacking information). More specifically, this study showed that typically developing three- and four-year-old children easily understood that if a person was allowed to look inside a box, then that person would have knowledge of its contents; however, if the same person was not allowed to look inside the box, they would be unaware of its contents. In contrast, Baron-Cohen and Goodhart (1994) showed that children with autism responded at the level of chance when completing this task, which suggested that they did not understand the concept of “seeing leads to knowing” (i.e., that the person who was able to look inside the box would know the contents of the box).

Children with ASD also have difficulty performing other cognitive and emotional tasks related to the understanding of mental states when compared with typically developing peers. For example, a study by Tager-Flusberg (1992) showed that children with autism used fewer words than controls to describe the functions of the brain such as "thinking," "knowing," "hoping," and "imagining" and instead used more action words than controls such as "jump," "eat," or "move" when describing a picture by telling a story. The action word descriptions in this study can be thought of as being more literal, whereas the brain functions are more abstract. When investigating the emotional understanding of children with ASD, Harris, Johnson, Hutton, Andrews, and Cooke (1989) reported that typically developing children aged four to six were able to understand that emotions can be caused by internal thoughts and beliefs rather than by actions. For example, a child can be excited by the prospect of getting a toy that he was promised when he successfully completes his homework. In contrast, children with ASD have difficulty with this type of abstract emotional understanding (Baron-Cohen, Spitz, & Cross, 1993). In this study, children with ASD were able to understand more basic emotions such as happy and sad that are the result of actions; however, they had difficulty understanding emotions

such as surprise, which are related to more advanced cognitions and which are the result of beliefs rather than actions. Interestingly, children with intellectual disability did not differ from typically developing children in their ability to recognize any of the three emotions (i.e., happy, sad, or surprise), which suggests that this impairment may be unique to autism and not due to overall cognitive ability. Overall, this implies that children with ASD have difficulty understanding emotions when there is a more advanced underlying cognitive component involved (Baron-Cohen et al., 1993).

In addition to basic social cognitive impairments such as emotion recognition, children with ASD are impaired in their ability to comprehend higher-order cognitive demands such as understanding pragmatics, or the social use of language (APA, 2000; Baron-Cohen, 2000; Peterson et al., 2012). In this population, difficulties with pragmatic language such as understanding prosody are apparent in a social context even at a young age, and therefore can be used to measure higher-order social cognitive abilities in children with ASD. These children frequently have trouble taking turns in a conversation, staying on topic, smoothly transitioning to a new conversational topic, and understanding what is appropriate to say in a given context. In addition, children with ASD often have difficulty understanding humor, sarcasm, or irony because the content of the spoken words (i.e., a more literal understanding) does not match the intended meaning (i.e., more abstract social communication). Furthermore, the additional demands of understanding nonverbal communication when interacting with others such as reading body language proves challenging for many children on the autism spectrum. Several studies have provided support for the idea that this is a common area of weakness in children with ASD (Baron-Cohen, O'Riordan, Stone, Jones & Plaisted, 1999; Surian, Baron-Cohen & Van der Lely, 1996). In these studies children with ASD were unable to choose which responses

would not be appropriate to a given question or if there were certain comments that someone made in a short story that he or she should not have said. Results from these studies support the idea that these children have difficulty understanding the pragmatics of language, a proxy of the understanding of higher-order cognitive abilities in social context, which included understanding what is appropriate to say in a given situation.

**The debate over ToM impairments in ASD.** The debate over Baron-Cohen's (2000) claim that all children with ASD exhibit impairments in the understanding of theory of mind deserves some additional attention. Researchers who have argued that not all children with ASD exhibit these challenges often have pointed to the fact that many children with ASD, especially higher functioning children with Asperger syndrome, can pass first-order false belief tasks (i.e., understanding that another person has a different belief). Happé (1995) attempted to clarify this argument by noting that many children with ASD can eventually pass first-order false belief tasks, but this is often not accomplished until much later than typically developing children (i.e., nine years old versus four years old), with the earliest reported age of successful completion of this type of task being five-and-a-half years old. Given this information, the debate then turned to the ability of children with ASD to pass second-order false belief tasks (e.g., Tom thinks that Sally thinks that...). These tasks are often understood by typically developing children around the age of six (Happé, 1995). Much like the first-order debate, researchers have shown that some higher functioning individuals with autism or Asperger syndrome can complete these tasks by the teenage years (Bowler, 1992; Ozonoff, Pennington, & Rogers, 1991). Currently, many researchers agree that these abilities (i.e., passing first- and second-order false belief tasks) can be acquired, albeit at a delayed rate, by adolescence (Peterson et al., 2012; Wellman & Liu, 2004). Therefore, much of the most recent research on theory of mind understanding and autism

spectrum disorder has focused on the understanding of higher-order cognitive processes such as the understanding of the pragmatics of language given that only a few studies have investigated this area of social cognition in ASD. The present study attempted to add to the growing literature in this area.

In summary, children with ASD exhibit a variety of deficits in social cognition, ranging from the more basic understanding that others may have beliefs that are different from one's own to more the more advanced understanding of the social use of language such as pragmatics. These difficulties, evidenced by poor performance on cognitive measures such as theory of mind tasks, have been reported consistently in the literature (Baron-Cohen, 2000; Tager-Flusberg, 2000). Even children who are "higher functioning" in certain areas (e.g., communication or adaptive functioning abilities) still exhibit impairments in social cognition, especially when emotions are involved. This suggests that this cluster of abilities may be deficient in many, if not all, children with ASD. Furthermore, impairments in social-emotional functioning can have detrimental effects on the social competence of children with ASD (Sigman & Ruskin, 1999; Tager-Flusberg, 2000).

Lastly, as previously mentioned, autism spectrum disorder is a highly heritable neurodevelopmental disorder, as noted by the high concordance rates in MZ twins compared with DZ twins (APA, 2000). Given that impairments in social cognition, specifically the understanding of theory of mind (ToM), are common to many, if not all, children with ASD, there might be a genetic component to ToM as well. Therefore, studies such as the present one are important because they combine psychological and genetic contributions to the development of social cognitive abilities. A strength of this study was that it used a cross-sectional design, which included children with and without ASD, as well as a longitudinal design using twins,



which allowed for an assessment of the development of social cognitive abilities over time, as well as an investigation of the behavioral genetics of social cognitive abilities.

### **Benefits of Using a Twin Sample**

Behavioral genetics is the study of genetic and environmental influences on behavioral phenotypes. The use of a twin sample in behavioral genetic studies allows for the investigation of genetic influences on behavior (e.g., social cognition), parsing out influences due to genes and environment. There are two types of twins included in a twin sample: identical or monozygotic (MZ) twins, who share the same genetic makeup, and fraternal or dizygotic (DZ) twins, who share approximately 50% of their DNA with their co-twin (Plomin, DeFries, McClearn, & McGuffin, 2000). The difference between DZ twins and siblings who are not twins is that DZ twins share the same prenatal environment and are a member of the same birth cohort. When studying twins, heritability estimates are computed in order to determine the genetic and environmental influences on a particular behavior. For example, correlating the performance of MZ twins on ToM tasks and comparing that value to the correlation of the performance of DZ twins on the same tasks provided an estimate of the heritability of the understanding of theory of mind. Higher correlations between MZ twins imply a greater genetic contribution ( $h^2$ ) on a particular behavior. The equation for the correlation of MZ twins is:  $r_{MZ} = h^2 + c^2$  (Falconer, 1960). The equation for correlation of DZ twins (Falconer, 1960) is:  $r_{DZ} = (1/2)h^2 + c^2$  because DZ twins only share about 50% of their genetic makeup. Heritability is estimated by subtracting the DZ equation from the MZ equation and doubling the difference, which results in the following equation:  $h^2 = 2(r_{MZ} - r_{DZ})$ .

**Equal environments assumption (EEA).** When studying twins, it is imperative to include a discussion of the equal environments assumption (EEA). This assumption states that

MZ and DZ twins share their environments to the same extent, which allows for valid comparisons between the two groups. This assumption is crucial in the study of twins (Kendler, Neale, Kessler, & Heath, 1993). One exception is that more similar twins will select more similar environments, thus drawing influences from both genetics and environment, which should be taken into consideration when analyzing the behavioral differences between twins (DiLalla, 2002b). Neale and Cardon (1992) reported that some researchers believe that twin study samples are flawed based on the notion that MZ twins evoke a more similar response from those with whom they interact compared with DZ twins, which could affect twins' behavioral presentation. This would invalidate the EEA because of the overestimation of the heritability between twins; therefore, researchers should be aware of the implications of the special case of twin samples. Although violations of the EEA cannot be fully explained, several researchers in the field believe that the assumption is not being violated (Borkenau, Riemann, Angleitner, & Spinath, 2002; Derks, Dolan, & Boomsma, 2006; Klump, Holly, Iacono, McGue, & Wilson, 2000). Given the debate over the EEA, twin samples are best used with a longitudinal design in order to measure environmental influences over several points in time (Neale & Cardon, 1992).

### **Behavioral Genetics of Social Cognition**

Using twin studies is a useful way for researchers to investigate the genetic influence on the understanding of social cognition. In general, there are limited studies on the behavioral genetics (i.e., heritability and environmental influence) of social cognitive abilities. Results from a molecular genetics study were the first to show a connection between genes and social cognition (Skuse et al., 1997). This study reported that a locus on the paternal X chromosome was implicated in the understanding of social cognition in children, given that females performed significantly better than males on the social cognitive tasks included in the study. Recent studies by Skuse and colleagues (Good et al., 2003; Skuse, Morris, & Lawrence, 2003) reported that

impairments in social cognition in Turner's syndrome, a genetic disorder in females in which one of the X chromosomes is missing, is linked to genes on the paternal X chromosome in some cases, specifically Xp 11.3 when there is a partial monosomy. However, most single X chromosomes come from the mother in this disorder. In addition, the authors of these studies reported that these genes likely influence social cognitive abilities through connections to the amygdala, which is larger in individuals with Turner's syndrome compared with controls. Typically, decreased grey matter volumes are associated with impairment; however, in this case, the authors explained that hyperactivity of the amygdalae in their study was related to social difficulties. The two studies (Good et al., 2003; Skuse et al., 2003) were comprised of different age ranges as well, suggesting that the volume differences were independent of age. The bilateral amygdalae, which are part of the limbic system, are believed to be involved in the processing of negative emotional reactions and socially relevant information (Skuse et al., 2003). The link between emotional processing (via connections to the amygdala and the superior temporal sulcus) and social cognition is important given the fact that children with autism spectrum disorder have been shown to be impaired both structurally and functionally in both of these areas of the brain (Pelphrey, Shultz, Hudac, & Vander Wyk, 2011). After the molecular studies were published suggesting a link between genetics and social cognitive abilities, behavioral genetics studies using twin samples followed.

**Independent Pathway model.** The covariance between twins in behavioral genetics studies can be explained using the Independent Pathway model (Neale & Cardon, 1992). This model includes three main components: (A) genetic, (C) shared environmental, and (E) nonshared environmental effects. The genetic effects can be additive (A) or not, depending on whether there is an effect of alleles at the same location on a particular chromosome (additive) or

at different locations (non-additive), which is the case with genetic dominance. Shared environmental factors (C) make twins more similar, whereas nonshared environmental factors (E) result in twins appearing more dissimilar. Shared environmental influences include factors such as the number of family members living in the home and the socioeconomic status of the family. An example of a nonshared environmental factor is the difference in perceptions between twins following a move to another state. Twin A had an easier time getting along with his teacher at his old school compared with Twin B, which led to a more positive perception of his new teacher at the new school than Twin B. Thus, the same environmental influence (i.e., moving to a new school) resulted in different perceptions for each twin at the new school. It should be noted that the nonshared environmental component is inherently confounded with measurement error. Systems-level influences such as socio-cultural, political, and historical experiences are likely to influence measurement error and cannot be directly measured (Rutter, 2001). This may make the differences between twins appear larger than they actually are.

One of the first twin studies to investigate the genetic influence on social cognitive development using behavioral genetics was conducted by Hughes and Cutting (1999). This study included a sample of 119 same-sex twin pairs who were three years of age at the time of data collection. The children were given a series of eight false belief tasks, as well as the Vocabulary and Comprehension subtests of the Stanford-Binet. The authors used structural equation modeling to estimate the genetic and environmental influences between twins on social cognitive functioning. Intraclass correlations were .66 and .32 for MZ and DZ twins, respectively, which resulted in a heritability estimate of .66. Given that verbal ability also was shown to be genetic, a bivariate modeling approach was used to determine the amount of overlap between these two abilities. The authors reported that 67% of the genetic variance of theory of

mind was unrelated to verbal ability, suggesting that the overlap between these two abilities may not be as large as previously reported (Happé, 1995).

Another twin study by Scourfield, Martin, Lewis, and McGuffin (1999) provided further evidence that social cognitive abilities likely have a genetic basis. Intraclass correlations between MZ and DZ twins were reported, suggesting a genetic influence on social cognition. More specifically, correlations for MZ twins ranged from 69% to 74% for males and females, respectively, and correlations for DZ twins were 47% and 26% for males and females, respectively. Given the fact that the correlations for MZ and DZ twins were highly discrepant in females but not males, the authors noted that non-additive genetic effects were likely at play. In addition, males were found to have more difficulty with social cognitive tasks than females in general, and the overall performance improved as all children got older. Additionally, the genetic effects on performance for children under the age of 11 were greater than those for children over 11 years of age. Thus, this study provided support for the heritability of social cognition and the impact of age on the genetic influence of social cognitive task performance.

In contrast, a more recent study (Hughes et al., 2005) reported that the understanding of social cognition was driven by environmental factors more than by genes. Individual differences in the understanding of ToM were studied in a large sample of 1,116 same-sex five-year-old twins, who were a part of the Environmental Risk (E-Risk) Longitudinal Twin Study. The children were given the Vocabulary subtest from the Wechsler Preschool and Primary Scales of Intelligence-Revised (WPPSI-R; Wechsler, 1990) in addition to a series of ToM tasks with a forced-choice format that increased in difficulty level. The ToM tasks included first-order and second-order false belief tasks as well as a belief-desire reasoning task. The belief-desire reasoning task required the child to make an inference about a person's emotional state based on

a false belief attribution. Results from this study indicated that there was no difference between correlations for MZ and DZ twins ( $r = .53$ ), which suggested that environmental rather than genetic factors may be influencing the individual differences in ToM performance observed in this large sample. The shared environmental influences accounted for 48% of the variance, the nonshared environmental influence accounted for 45% of the variance, and genes only accounted for 7% of the variance. It is important to note that these genetic and environmental differences were the same for boys and girls. In contrast to the study by Hughes and Cutting (1999) that reported that the genetic influence of ToM and verbal abilities overlapped by 33%, the large study by Hughes and colleagues (2005) suggested that the genetic influence on verbal abilities completely overlapped with the genes that influenced ToM abilities. In addition, there was a strong overlap between verbal abilities and ToM abilities in terms of shared environmental factors. Similar to the previous study by Hughes and Cutting (1999), there were no sex differences in verbal ability or ToM ability.

Given the wide range of results reported in the few behavioral genetics studies of ToM abilities in children, it is important to consider the reasons that may be influencing these differences. One reason for the large disparity in outcomes (i.e., genetic versus environmental influences) between the more recent study by Hughes and colleagues (2005) and the study by Hughes and Cutting (1999) may be the markedly larger sample size used in the Hughes et al. (2005) study. A second reason could be that the E-Risk Longitudinal Twin Study included a disproportionately high number of families with low socioeconomic status, which could be influencing the environmental effects, especially the influence of nonshared environmental experiences, which are unique to each individual. Lastly, the Hughes and Cutting (1999) study consisted of three-year-olds and the larger Hughes et al. (2005) study was comprised of five-

year-olds, suggesting that genetic influences on ToM abilities might be stronger in younger children.

In conclusion, results from behavioral genetics studies of twins investigating the understanding of theory of mind are mixed. A few twin studies provide support for a strong genetic influence on ToM abilities (Hughes & Cutting, 1999; Scourfield et al., 1999), whereas a single, larger study by Hughes and colleagues (2005) reported a strong environmental influence and a negligible genetic influence on the understanding of ToM at the age of five. Further studies are needed to help clarify these highly discordant results.

### **The Role of Molecular Genetics in Social Cognition**

With advances in technology, the field of genetics has exploded in recent years. The molecular genetics of most disorders are complex, with the exception of a few rare disorders that have been linked to a single gene mutation (e.g., cystic fibrosis, sickle cell anemia). In the vast majority of psychiatric disorders, the genetic influences are often polygenetic and multifactorial, meaning that they result from the combination of many genes and environmental factors (e.g., prenatal environment, parental psychopathology). The relation between genes and the resulting behavioral phenotype is complicated; an understanding of biological terms (e.g., DNA, RNA, risk alleles) is essential to the comprehension of the field of molecular genetics.

**Biology of molecular genetics.** Each individual's genetic information is located within deoxyribonucleic acid (DNA) molecules ([http://www.ncbi.nlm.nih.gov/About/primer/genetics\\_genome.html](http://www.ncbi.nlm.nih.gov/About/primer/genetics_genome.html)). A particular gene is a stretch of DNA that encodes information from an individual's genetic makeup. Gene expression is the process by which that information produces an observable behavioral phenotype. This process is driven by the transcription of double-stranded DNA into single-stranded RNA, or ribonucleic acid, a complementary copy of DNA.

This single-stranded RNA, or messenger RNA (mRNA), is then translated into a protein after a series of processing steps (e.g., folding into a three-dimensional structure), which is necessary for the protein to become functional.

The structure of DNA and RNA are similar, but the molecules differ in a few important ways. For example, both are comprised of several smaller components such as nucleic acids, which are made up of several nucleotides. Each nucleotide has a five-carbon sugar (ribose), a nitrogenous base, and a phosphate group ([http://www.ncbi.nlm.nih.gov/About/primer/genetics\\_genome.html](http://www.ncbi.nlm.nih.gov/About/primer/genetics_genome.html)). DNA molecules are missing one oxygen atom, which results in the prefix “deoxy.” The structure of DNA is a double helix, consisting of two backbones, which wrap around each other. The four possible nitrogenous base pairs in DNA include adenine (A), thymine (T), cytosine (C), and guanine (G). These bases are found along the backbones of the helix. The two strands are connected by chemical bonds across base pairs. For example, the “A” on one strand is attracted to the “T” on the opposite strand. The structure of DNA and RNA are similar, but complementary, and the single-stranded RNA includes a base called “uracil” in the place of “thymine,” which is found in DNA. These base pair sequences provide instructions for how a given protein is synthesized, which is dictated by the order of the base pairs.

Genes are segments of DNA that code for a particular protein or proteins. Each individual receives two copies of each gene, one from their mother and one from their father. These copies can be similar or different. Different versions of the same gene are referred to as alleles. One gene may have several different alleles, but offspring will only receive two alleles for any gene, one from each parent ([http://www.ncbi.nlm.nih.gov/About/primer/genetics\\_genome.html](http://www.ncbi.nlm.nih.gov/About/primer/genetics_genome.html)). When the two inherited alleles are the same, they are referred to as “homozygous,” and when they are different, they are referred to as “heterozygous.” Although



humans share a large portion of their genetic makeup, only identical twins have exactly the same DNA. The special case of identical twins, as discussed in a previous section, allows for the ability to study genetic and environmental influences to a specific trait or disorder such as the understanding of social cognitive abilities in children with and without autism spectrum disorder in this study.

**Risk alleles.** An understanding of alleles is not complete without a discussion of what it means to have a “risk allele.” The presence of a risk allele results in a protein with an atypical function, which is often associated with an increased risk of developing a disease or disorder ([http://www.ncbi.nlm.nih.gov/About/primer/genetics\\_genome.html](http://www.ncbi.nlm.nih.gov/About/primer/genetics_genome.html)). For example, when a woman has a mutation in the BRCA1 gene, which is responsible for tumor suppression, the presence of this risk allele dramatically increases the likelihood of developing breast or ovarian cancer. It is important to note that “risk” is a statistical term that results from the comparison of groups of individuals with and without a specific condition (e.g., depression, anxiety) and should not be applied to individuals. A risk allele, or a common variation within genes, is also known as a single nucleotide polymorphism (SNP). SNPs occur when a single nitrogenous base change occurs in the DNA sequence. Although these are quite common, and often are benign, certain variants have been found to increase the risk of specific groups of people in developing common diseases or disorders.

Researchers study these risk alleles using candidate gene studies, which allows for causal inferences to be made from the level of genotype to the level of phenotype. Genes that are chosen for study in these studies are included based on their theoretical link to biological pathways underlying a given disease or disorder. Given this link, when an association is found between a risk allele and a particular condition, it is assumed that the gene is connected to the

protein produced in the identified biological pathway, which is the case with the BRCA1 gene involved in tumor suppression in breast and ovarian cancer. On the other hand, when an association is not found, evidence against the biological connection is implied ([http://www.ncbi.nlm.nih.gov/About/primer/genetics\\_genome.html](http://www.ncbi.nlm.nih.gov/About/primer/genetics_genome.html)).

**The neurotransmitters dopamine and serotonin.** The candidate genes in this study are common variations of the genes that synthesize the transporters and receptors used in conjunction with the neurotransmitters dopamine and serotonin. These neurotransmitters have been linked to human social behavior, specifically social cognition, in a recent study by Skuse and Gallagher (2011). The authors explained how these neurotransmitter systems influence social interactions through dopaminergic connections to reward pathways and serotonergic links to emotional regulation. More specifically, dopamine is associated with the rewards gained through building social relationships, and serotonin is related to emotional states associated with social interactions such as subjective feelings, physiological arousal, desire to interact with others, and the feelings associated with being included or excluded from a group. The authors specifically implicated the pathways of the “social brain” that underlie the ability to take another’s perspective, including the dorsomedial and dorsolateral prefrontal cortices, the paracingulate cortex, the bilateral temporoparietal junctions, and the amygdala (Skuse & Gallagher, 2011).

Genetic variations in the dopamine and serotonin genes have been shown to affect the levels of dopamine and serotonin available for use in the brain. The dopamine receptor D4 gene (DRD4) has a 48-base pair variable number tandem repeat (VNTR), or a location on a gene where a short sequence repeats itself, polymorphism in exon III in the D4 receptor. It is hypothesized that the presence of a common 7-repeat allele may lead to reduced expression of

the DRD4 gene, which reduces the intracellular concentration of the second messenger cyclic AMP, which in turn causes a cascade of signaling events (Lachowicz & Sibley, 1997). The final result is a decreased amount of dopamine available in the system for binding.

The serotonin transporter gene (5-HTTLPR) is a monoamine transporter protein that encodes the serotonin reuptake transporter (5-HTT). The 5-HTT is responsible for transporting the unused neurotransmitter serotonin from the synaptic space back into the presynaptic neurons so it can be released again. The promoter region of the SLC6A4 gene contains a polymorphism with “short” and “long” repeats in the 5-HTT-linked polymorphic region (5-HTTLPR). The short variation has 14 repeats of a sequence and the long variation has 16 repeats (Nakamura, Ueno, Sano, & Tanabe, 2000). In addition, the short variation leads to less transcription of SLC6A4, which leads to the creation of fewer 5-HTT proteins and lower 5-HTT expression, which increases extracellular concentrations of serotonin in the brain. Increased levels of serotonin are associated with overstimulation of nerves; the recycling of 5-HT by the transporters prevents this overloading (Canli & Lesch, 2007). Thus, the creation of more SERT proteins (i.e., the protein produced when the  $L_A$  allele is present) helps to maintain homeostasis at the synapse site. One study showed a decrease in grey matter in the perigenual anterior cingulate cortex and the amygdala for those who had the short/short allelic combination compared with those with the long/long genotype. A meta-analysis of the association between the 5-HTTLPR polymorphism and individuals with autism spectrum disorder (ASD) was conducted given the conflicting results reported in previous studies (Huang & Santangelo, 2008). This review and meta-analysis showed no overall relation between 5-HTTLPR and ASD even after separating out families with only one child with ASD from those with more than one affected individual.

The 5-HTTLPR polymorphism has been extensively studied in psychiatric populations such as those with ASD, depression, and anxiety since the mid-1990s with mixed results (Wendland, Martin, Kruse, Lesch, & Murphy, 2006). A recent focus has shifted to further subdividing the short and long genetic variations in order to explain these conflicting findings. A functional variation was identified within the long variant, known as  $L_A$  or  $L_G$ .  $L_A$  was associated with higher levels of 5-HTT expression and lower levels of extracellular serotonin, whereas  $L_G$  was more similar to the short variant, or the S allele (Praschak-Rieder et al., 2007). Given this important distinction, the short and long variants were further subdivided into S,  $L_A$ , or  $L_G$  in this study in order to account for the specialized function of the  $L_A$  genotype. Abnormalities in the serotonin system have been linked to different disorders such as those with and without depression (Cannon et al., 2007). Increased levels of serotonin have been shown to decrease aggression and increase cooperation and vice versa in a primate study (Carver & Miller, 2006). Studies in humans and primates also have shown that increased levels of serotonin activity have positive effects on social interactions and cooperation, whereas the opposite also is true (Cools, Roberts, & Robbins, 2008).

**Dopamine and social cognition.** There is significant evidence to suggest that the neurotransmitter dopamine is related to the understanding of ToM in humans. Two studies using functional neuroimaging techniques or electroencephalography (EEG) in preschoolers have implicated the dorsomedial prefrontal cortex (dMPFC) in the development of ToM (Liu, Sabbagh, Gehring, & Wellman, 2009; Sabbagh, Bowman, Evraire, & Ito, 2009). Given that the dMPFC is a main target of dopamine projections, dopamine may be associated with developing and maintaining functioning of social reasoning in preschoolers (Popolo, McCarthy, & Bhide, 2004). Developmentally, this is important because the brains of preschoolers are maturing at a

rapid rate. Another study of 91 four- to five-year-old typically developing preschoolers showed that individual differences in rates of eye blinking, which was used as a proxy for dopamine functioning, successfully predicted the performance on ToM tasks (Lackner et al., 2010). These effects remained even after controlling for executive functioning (i.e., performance on a Stroop-like task), language ability, sex differences, and age. A recent study provided further support for the link between dopamine and the understanding of ToM (Lackner et al., 2012). In this study of 73 typically developing 42- to 54-month-olds, polymorphisms of the dopamine D4 receptor gene (DRD4) were associated with ToM performance.

Dopamine has also been shown to be associated with learning, which is a crucial component to the successful completion of ToM tasks. In an animal study by Schultz (2000), dopamine was released during situations in which the expected event did not match with an unexpected outcome. Thus, the author proposed that dopamine may be responsible for plasticity of neural cells, providing updated information based on expected versus actual outcomes and adjusting future expectations.

**Serotonin and social cognition.** Much like dopamine, there is a documented link between serotonin and social cognitive abilities in the literature; however, studies investigating this connection are limited in humans. Serotonin is linked to a wide range of behavioral and emotional functions including behavioral inhibition, appetite, aggression, mood, sleep, and navigating social situations (Rogers, 2011). Primate studies have shown elevated serotonin decreases aggression and increases cooperation and effectiveness in social situations and vice versa (Carver & Miller, 2006). In both animals and humans, greater serotonin activity positively influences social interaction and cooperation, whereas low serotonin levels have the opposite effect (Cools, Roberts, & Robbins, 2008).

In addition, a recent study by Bosia et al. (2011) reported that ToM abilities were deficient in a clinical population of 118 individuals with schizophrenia when compared with controls. In this study, prefrontal cortex dysfunction was related to difficulties taking another person's perspective. The authors also investigated the specific effect of a functional polymorphism of the serotonin 1A receptor (5-HT1A-R), which is involved in the regulation of both serotonin and dopamine transmission. More specifically, the 5-HT1A-R is involved in the decreased production of serotonin, and it directly influences the release of dopamine in the prefrontal cortex (Rollema et al., 2000). The authors reported that individuals with the C/C genotype performed better than those individuals with either the C/G or the G/G genotypes and concluded that this effect was likely related to interaction of serotonin and dopamine working together to influence social functioning. The findings from the study by Bosia and colleagues (2011) and an earlier study by Abu-Akel (2003) provided support for the influence of both serotonin and dopamine levels on ToM performance in individuals with schizophrenia after controlling for other cognitive abilities (i.e., IQ and executive functioning).

In conclusion, human and animal studies of dopamine and serotonin have provided strong evidence for a link between risk alleles associated with these neurotransmitters and performance on measures of social cognitive abilities (Abu-Akel, 2003; Bosia et al., 2011; Skuse & Gallagher, 2011; Lackner et al., 2010, 2012). Dopamine has been linked to reward pathways, and the bonds created during social interactions are inherently rewarding. Serotonin is associated with a wide variety of emotional states, including the desire to interact with other people. More genetic research on clinical populations typically impaired in the understanding of ToM such as those with ASD are needed in order to better understand the underlying biological pathways affecting these complex cognitive abilities. Given that behavioral traits and psychiatric disorders are

rarely caused by single gene mutations, it is crucial to investigate both the genetic and environmental influences of a particular condition, which was best accomplished in this study through the use of a twin sample. In addition, the long repeats of the DRD4 polymorphism and the short variant of the 5-HTTLPR polymorphism were the focus of the genetic analysis given their connection to the development of the social brain.

### **The Present Study**

The goals of this study were three-fold. First, I investigated whether performance on a theory of mind task at ages three and four predicted performance on theory of mind (ToM) tasks at follow-up (i.e., one time point between 6 to 10 years of age). The longitudinal portion of this study used a sample of typically developing twins, most of whom were tested in the past as part of SITSS (87%), but some of whom were newly recruited as part of the larger study (13%). The follow-up testing comprised five measures of ToM, which increased in difficulty and included the addition of an age-appropriate measure of ToM that assessed the understanding of nonliteral communication (i.e., sarcasm). Second, I compared the performance of typically developing twins to children with ASD on the ToM tasks, given that children with ASD have been shown to be impaired in processing social information, especially when emotional cues are incorporated. In addition to the ToM tasks, all children were administered the receptive and expressive subtests of the Clinical Evaluation of Language Fundamentals, 4<sup>th</sup> Edition (CELF-4) as a way to control for language differences between groups. This cross-sectional analysis allowed comparisons between groups to investigate the extent of impairment in social cognitive abilities in children with ASD. Third, I tested whether different genotypes were related to performance on ToM tasks to better understand the role of molecular genetics in the development of social cognition in both groups of children. Prior behavioral genetics studies have indicated that ToM is heritable,

and there is evidence from molecular genetics studies to suggest that risk alleles associated with the neurotransmitters dopamine and serotonin may be linked to deficits in social cognitive functioning. The current study added to the literature by expanding upon the role of dopamine and serotonin in the understanding of social cognition.

### **Hypotheses**

This study sought to add to the literature by investigating the psychological and genetic contributions to the development of social cognition through the use of a longitudinal and a cross-sectional design. The hypotheses for the current study were as follows:

Hypothesis 1 tested the development of social cognitive abilities in typically developing children over time.

1a) Performance at ages three and four was expected to predict performance on all five ToM tasks at follow-up (i.e., one time between ages 6 to 10). Thus, it was hypothesized that performance at ages three and four would be positively correlated with scores at follow-up on individual tasks.

1b) Younger children in the follow-up study were expected not to pass as many of the five ToM measures as older children in the follow-up study, thus generating a lower overall total score. Therefore, it was hypothesized that age and total score would be positively correlated in the follow-up sample.

Hypothesis 2 tested the group differences in ToM abilities in children with and without autism spectrum disorder.

2a) Children (ages 6 to 10) with ASD were expected not to score as high as typically developing children on each of the five measures of ToM included in the current study given their deficits in social cognitive abilities.



2b) Children with ASD were expected to have significantly more difficulties on the CBCL scales related to social abilities (i.e., social problems, thought problems) and emotional functioning (i.e., anxiety, social withdrawal) compared with typically developing children. Social problems are a core deficit in children with ASD, and emotional processing difficulties make navigating social situations even more difficult for these children.

2c) Autism symptom severity as measured by the social cognition, social communication, and social awareness treatment subscales on the SRS-2 was expected to be negatively correlated with performance on all five ToM tasks.

Hypothesis 3 tested whether genes in general and different genotypes of the DRD4 and 5-HTTLPR genes in particular would predict performance on ToM tasks.

3a) It was hypothesized that ToM would be heritable, which was assessed by comparing the correlations of MZ and DZ twins at all ages. It was expected that MZ twins would perform more similarly than DZ twins at all ages (behavioral genetics hypothesis).

3b) Participants who have at least one S or L<sub>G</sub> allele in the 5-HTTLPR polymorphism or six or more repeats in the DRD4 polymorphism would perform worse on ToM tasks at all ages than those with only 5-HTT L<sub>A</sub> alleles or only fewer than six repeats of the DRD4 polymorphism, respectively. Participants who have both risk genotypes (i.e., at least one risk allele in both genes, DRD4 and 5-HTTLPR) would be more impaired on ToM tasks than those with only one of the DRD4 or the 5-HTTLPR genes, as well as those without any risk alleles (molecular genetics hypothesis).

## CHAPTER 3

### METHODS

#### **Participants**

The first part of this study included twin pairs or triplets who participated in the Southern Illinois Twins and Siblings Study (SITSS; DiLalla, 2002a) at ages three and/or four, shortly after their birthdays each year, as well as new families who had not been tested previously. In total 115 families were targeted for participation, and 62 families participated in the current study, which resulted in a response rate of 54%, which is considered to be a high response rate given the nature of the study and the geographical area in which it is being conducted. Of those who did not participate, 23% were not interested for various reasons (four families lived too far away, three families were not interested/did not have time, three families had children who aged out of the study by the time they were contacted, and no reason was documented for two families), 28% were unable to be contacted, and the reason for not participating was unknown for the remaining 49% (the specific information for why families did not participate was not recorded until halfway through the study). We attempted to contact 23 new families, and the remaining 92 were families with multiples who participated in SITSS who were contacted three to six years after their initial participation. The children who were tested previously were between the ages of 6-10 at the time of follow-up. Of the 62 families in the follow-up study, which resulted in a total sample of 127 children, 69 children were tested at ages three and four (54%), five were tested at age three only (4%), 18 were tested at age four only (14%), 17 children were tested at age 5 as a part of SITSS but were not tested at ages three or four (13%), and eight new families participated, including seven twin pairs and a set of quadruplets (n = 18 children; 14%).

The second part of this study included 25 6- to 10-year-old boys who had previously been diagnosed with autism spectrum disorder (ASD). Given the fact that four times as many boys are diagnosed with ASD than girls, only boys were included in this sample. All but one of the children in the ASD sample were recruited through the Center for Autism Spectrum Disorders (CASD). The only child who was not recruited there was contacted using information initially gathered for SITSS. (This child and his co-twin were excluded from SITSS several years ago after he was diagnosed with ASD as a toddler.) Most, but not all, of the children were evaluated by clinicians at the CASD at SIU. The children who were evaluated there were administered the Autism Diagnostic Observation Schedule (ADOS; Lord et al., 1989) in order to verify their diagnosis, following an initial screening visit. The remainder received diagnoses from their primary care physicians; however, each child's diagnosis was verified by the staff at the CASD prior to including them in interventions or research studies. One set of MZ twins was included in the data collection; however, only one twin was included in the analyses in order to avoid violating statistical assumptions. All of the children tested for the current study were high-functioning as evident by their ability to attend a regular education classroom for most, if not all, of the school day. Although the cognitive level of the children was not formally assessed as part of this study, cognitive data were available for a portion of the children with ASD (see Table 1).

A series of ANOVAs and a Chi-square test were run to determine group differences in demographic variables. For these analyses, only one randomly selected twin was included when comparing the typically developing group to the children with autism spectrum disorder (ASD) to avoid violating the independence of samples assumption. There were no significant differences in age,  $F(1, 85) = 3.00, p = .09$ , SES,  $F(1, 74) = 1.36, p = .248$ , or race/ethnicity,  $X^2$

(2,  $N = 87$ ) = 2.75,  $p = .25$  between groups. The total sample was 86% Caucasian, 7% African American, and 7% "Other" or mixed race (see Table 1 for sample descriptive statistics).

## Measures

### Age 3 and 4 Testing

**Demographic questionnaire.** All families were mailed a demographics questionnaire (see Appendix A) that included questions related to family information such as marital status, income, family structure, age, education level, occupation, and race. The Hollingshead Index (Bonjean, Hill, & McLemore, 1967) was used to calculate a socioeconomic status (SES) score for each family based on occupation status, educational attainment, and family income. Education level was rated on a 5-point Likert-type scale, ranging from 1 = some high school or high school degree to 5 = advanced training beyond college degree. Each occupation also was rated on a 7-point Likert scale, ranging from 1 = unskilled laborer to 7 = high-level professional. Lastly, family income was scored on a 12-point Likert-type scale, with incomes ranging from 1 = less than \$5,000 to 12 = greater than \$55,000. Given that the scores are not on the same scale, all the education and occupation scores were put on a 12-point scale by multiplying the education scores by (12/5) and the occupation scores by (12/7) prior to averaging the scores. Because data on paternal education and occupation level were missing from four families, the maternal education, maternal occupation, and family income scores were averaged to generate a maternal SES score (see Table 1).

**DNA collection.** A laboratory at the University of Colorado has previously analyzed buccal cells collected for DNA analysis on most, but not all, of the twins who participated in the follow-up study as part of their participation in SITSS. As part of the present study, buccal cell samples were collected from all children with ASD, as well as from any twins whose DNA

samples had never been collected (i.e., new families) or were missing for any reason. Collection of buccal cells allowed for the investigation of the specific risk alleles implicated in social cognitive processing. In relation to the DRD4 gene, the number of repeats were indicated for each sample: two repeats and four repeats of the DRD4 polymorphism are common, non-risk variants; however, the presence of greater than or equal to six repeats in the DRD4 gene is considered “high risk” in clinical samples (Faraone et al., 2005). The high-risk allele results in less dopamine binding to receptors. Additionally, the promotor region of the 5-HTTLPR gene was classified as one of three alleles: S, L<sub>A</sub>, and L<sub>G</sub>. Both the L<sub>G</sub> and S alleles result in decreased serotonin transporter (5-HTT) mRNA levels and decreased 5-HTT transmission, which leaves more serotonin in the presynaptic cleft that typically would have been recycled by the transporters (Praschak-Rieder et al., 2007). Therefore, risk alleles disrupt neurotransmitter action in the brain, which can result in atypical behaviors in certain groups of individuals. Lastly, given the large number of genetic tests that were run in this study, a Bonferroni correction was done by dividing the p-value of .05 by 5 ( $p = .01$ ), which is the number of social cognitive tasks included in each analysis for each group.

**Theory of mind tasks.** All children were administered the same two similar false belief tasks at ages three and four (Gopnik & Astington, 1988). The original protocol was modified slightly to account for the fact that each twin was asked to reference their co-twin’s belief, rather than asking each child what a peer might be thinking about the situation presented. Each twin was tested separately in a room away from their co-twin. A total of 17 trained undergraduate research assistants coded these tasks from videotapes. Each child from a twin pair was coded by a different trained coder to reduce bias. Average inter-rater reliability for coders who have rated the age three and four protocol is .96 (range was .87-1.0).

*Theory of Mind I.* Each twin was shown a Playdoh container with incongruent contents (i.e., crayons) and asked what she thought was inside prior to seeing the contents. Next, the twin was shown the contents of the Playdoh container, the crayons were placed back into the container, and the lid was closed. Then, the twin was asked what she thought was inside the container *before* being shown the contents. (This will be referred to as the "memory question" from this point forward because the twin was asked to remember what she had initially believed was in the container versus what she now knows is inside the container.) Finally, the child was asked what she thought her co-twin would say was in the Playdoh container. (This will be referred to as the "contents question" from this point forward). Scoring ranged from 0 to 1 for both the memory question and the contents question. This scoring method was initially described by Wellman and Liu (2004). Specifically, a child received a score of 1 on the memory question if she said she remembered thinking there was Playdoh inside the container, but she received a score of 0 if she said there were crayons (or another incorrect answer) in the container. She received a score of 1 on the contents question if she said that her co-twin would think there was Playdoh in the container, and she received a score of 0 if she said her co-twin would think there were crayons (or any other incorrect answer) in the container.

*Theory of Mind II.* Each twin was shown a box of crayons with incongruent contents (i.e., blocks) and asked what he thought was inside prior to seeing the contents. Next, the box of crayons was opened to reveal the blocks, the blocks were returned to the crayon box, and the lid was closed. Then, the twin was asked what he thought was inside the container *before* being shown the contents (i.e., the "memory question"). Finally, the child was asked what he thought his co-twin would say was in the box of crayons (i.e., the "contents question"). Again, scoring ranged from 0 to 1 for both the memory question and the contents question, using an identical

scoring scheme to Theory of Mind I. See Appendix B for Theory of Mind Protocol (Age 3 and 4 Testing).

*Creating total scores for ToM performance at ages three and four.* First, memory scores at ages three and four were computed by adding the two scores (memory from Theory of Mind I and memory from Theory of Mind II) at each age and then dividing by two. The same was done for ToM contents. This resulted in a total of four average scores: memory at age three, contents at age three, memory at age four, and contents at age four. These four average scores were used as dependent variables in the main analyses. The memory at age three score was not significantly correlated with the memory at age four score,  $r(69) = .12, p = .348$ , nor was the contents at age three score correlated with the contents at age four score,  $r(69) = -.02, p = .897$ , which suggests that performance at the younger age was not related to performance one year later.

## **Current Study**

**Demographic questionnaire.** The same demographic questionnaire used in the age three and four testing protocols in the younger twin study was included in the mailing packets to all study participants in the follow-up study. This was scored the same way that was described in the previous section in order to obtain an SES score for each family based on maternal education, occupation, and family income (see Table 1).

**DNA collection.** The same procedure used in SITSS (DiLalla, 2002a; DiLalla, Gheyara, & Bersted, 2013) was followed to obtain buccal cells from the participants in the current study. Most, but not all, of the twins had their DNA collected previously; therefore, buccal cells were collected from all of the children with ASD (without any problems), as well as those twins

whose DRD4 and/or 5-HTTLPR gene data were missing or never obtained. These samples were sent to the University of Colorado in two batches to be analyzed.

When only one child was selected from each family, the genotype breakdown for the sample used in all molecular genetic analyses was as follows: 23 individuals (27%) with no risk allele and 63 individuals (73%) with 1 or 2 risk alleles for the 5-HTTLPR gene; 51 individuals (59%) with fewer than six repeats and 35 individuals (41%) with six or more repeats for the DRD4 gene; and 15 individuals (17%) without any risk alleles in either gene, 44 individuals (51%) with at least one risk allele in either gene, and 27 individuals (31%) with at least one risk allele in both genes (see Table 11).

**Theory of mind tasks.** Five measures of ToM were administered in the same order to all study participants in the current study. All twins underwent a one-hour testing session as part of a larger emotion study that was approved by the Southern Illinois University School of Medicine in Springfield, IL. The administration procedure was designed such that the twins completed all five ToM measures, but each successive ToM task was separated by a different measure in an alternating pattern. The testing session took place in Lindegren Hall on the SIUC campus. For the children with ASD, the testing battery only included the five measures of ToM abilities and was completed in 15 minutes at the Center for Autism Spectrum Disorders at SIU. The group of testers, who included the principal investigator of the research lab, six graduate students (myself included), and one advanced undergraduate student, were trained to precisely follow a written script while administering the testing battery, and responses were written down verbatim during the testing sessions. Trained testers scored all ToM tasks from oral responses during the testing session, and I double-checked the scoring of the five ToM items for the total sample prior to



double data entry. The administration and scoring procedures for the five ToM measures are described below (see Appendix C for the Theory of Mind Protocol for the current study).

*Contents False Belief (Perner, Leekam, & Wimmer, 1987).* This task assessed each child's understanding that, despite a certain fact, someone might believe something differently. The administration of this single task was similar to the false belief tasks administered during the age three and four testing protocol for SITSS, but there were a few notable differences. Similar to Theory of Mind I and II in SITSS, each child was shown the Playdoh container, asked about its contents (without knowing what is inside), shown the unexpected contents (i.e., Q-tips), and then asked what was in the container. Next, each child was shown a toy figure and asked if he has ever seen inside the container. The child was then asked what he thought the toy figure would think was inside the container. Lastly, each twin was asked what he thought his co-twin would think was in the container, and every child with ASD was asked what a well-known peer at school (whose name was provided in advance by the parent without the child knowing) would think was in the container. The method for the children with ASD was selected as the best way to approximate the procedure used in the twin protocol. Scoring ranged from 0 to 1, with a score of 1 given if the child identified that the toy figure and his co-twin/peer thought Playdoh was inside the container before seeing the contents.

*Explicit False Belief (Siegal & Beattie, 1991; Wellman & Bartsch, 1989).* This task measured each child's ability to understand that someone's behavior might be different based on a mistaken belief. Each child was shown a toy figure and a sheet of paper with two pictures: a backpack and a closet. The child was told that the toy figure is looking for his mittens, which might be in the backpack or the closet. The examiner told the child that the mittens are in the backpack, but the toy figure thinks they are in the closet. The child was asked where he thought

the toy figure would look for the mittens (known as the "target question") and where the mittens really are (known as the "reality question"). To be correct, the child needed to answer both questions correctly (i.e., the toy figure will look in the closet, but the mittens are really in the backpack), which resulted in a score of 1. If the child was incorrect on either or both of the questions, he received a score of 0.

*Belief-Emotion (Harris et al., 2000).* This task assessed each child's understanding of the fact that someone may have a different emotion because of a false belief. Each child was shown a toy figure and a small, closed Cheerios box with bouncy balls inside. The examiner asked the child what she thought was inside the box. Next, the examiner spoke for the toy figure, saying that Cheerios are her favorite snack, and then has the toy figure leave. The child was then shown that there are bouncy balls inside the box instead of Cheerios. The toy figure was brought back, and the child was told that the toy figure has never seen inside the box. Then, the child was asked how the toy figure would feel when she gets the box *before* seeing inside (this is referred to as the "target question"). Next, the examiner opened the Cheerios box and let the toy figure see inside. Finally, the child was asked how the toy figure would feel after looking inside the box (this is referred to as the "emotion-control question"). Scoring ranged from 0 to 1, with a correct score indicated when the child responded "happy" to the target question and "sad" to the emotion-control question. The child was given a score of 0 if he responded to either or both of the questions incorrectly.

*Real-Apparent Emotion.* This measure tested each child's understanding of the fact that someone can show a different emotion than the one they are feeling. Each child was shown a piece of paper with three faces (i.e., happy, sad, and neutral) in order to test their understanding of these emotions. All children were able to correctly identify each of the three emotions on the

piece of paper. Next, a picture of a boy was shown from the back so that the facial expression is unknown. The examiner then told a story about a boy named Matt who feels one way inside, but looks a different way on his face. Matt's friend, Rosie, tells a joke about Matt that is not nice, which causes others to laugh; however, Matt does not want his friends to know that he feels bad, so he hides how he truly feels. The child was then asked two questions to test their memory of the story, one asking about what the other children did when Rosie told a mean joke about Matt, and another about what the children might do if they knew how it made Matt feel. Finally, the child was asked how Matt really felt after being teased, choosing between "happy," "sad," and "so-so" (this is referred to as the "target-feel question"), and how Matt tried to make his face look, again choosing between "happy," "sad," and "so-so" (this is referred to as the "target-look question"). Scoring ranged from 0 to 1, with a correct score attained if the "target-feel question" was more negative than the "target-look question" (i.e., the child stated that Matt felt worse than the face that he made in front of his friends).

*Understanding Sarcasm (Peterson et al., 2012).* This task measured each child's understanding of the fact that the meaning of words is not always to be taken literally. The understanding sarcasm task is a newly validated measure of understanding non-literal language, which has been shown to be sensitive to differences in social cognitive abilities at this age group (i.e., only 25% of typically developing nine-year-old children passed this task; Peterson et al., 2012). Given that the children included in this study were school-aged at the time of data collection, it was appropriate and useful to include this more difficult task in the current study.

Each child was shown a picture of the back of a girl's and boy's head, as well as a picnic scene. The examiner read a story aloud about how the boy and girl were planning on having a picnic, but after they got the food out, it rained and ruined the food. Then, the girl in the story

said, "It's a lovely day for a picnic" without any inflection or a sarcastic tone. The child was asked whether what the girl said was true (this is referred to as a "control question"), why the girl said it was a "lovely day for a picnic" (this is referred to as the "test question"), and if the girl was happy about the rain (this is referred to as the "comprehension question"). Scores ranged from 0 to 1. A correct "test question" included some mention of the words "sarcasm," or "irony," or a description of the use of nonliteral language (e.g., using the word "opposite" or saying the girl was trying to be mean or mocking the boy), which resulted in a score of 1.

*Total score for all five ToM measures.* Each of the five measures was scored from 0 to 1 and then scores were added together to create a single total ToM score, which ranged from 0 to 5. Each measure included a "control question" in order to assess whether the child understood the question being asked. To receive full credit, the child needed to answer the control question correctly in addition to the test question. If the child failed any control questions, but passed the accompanying test question, the child did not receive credit and got a score of 0 on that item. Previous studies have shown that it was rare for children to fail the control question, but then pass the test question (Peterson et al., 2012). Indeed, this only happened on one occasion on the real-apparent emotion task, which resulted in a score of 0 for this child in the ASD sample.

*Clinical Evaluation of Language Fundamentals, 4<sup>th</sup> Edition (CELF-4; Semel, Wiig, & Secord, 2003).* The CELF-4 was selected for use in the current study due to the fact that it has been shown to be sensitive to the language difficulties exhibited by children with intellectual disability or autism spectrum disorder. The CELF-4 can be administered to individuals aged 5-21 and consists of subtests measuring core language, receptive language, expressive language, language structure, language content, language memory, and working memory. For the purposes of the current study, only the receptive and expressive language subtests from Word Classes

were administered in order to obtain a measure of current language functioning. These two subtests assessed each child's understanding of relationships between words with minimal language demands. Results from these two subtests generated scaled scores for receptive and expressive language as well as a total language scaled score.

The CELF-4 was normed on 2,650 youth aged 5-21 years (200 from each age group from 5-17 and 50 from each age group from 17-21). The sample was diverse in terms of age, gender, race/ethnicity, and parental education. The test's previous edition (CELF-3) did not include students with disabilities in the norm sample; however, the current version's norm sample was comprised of youth in special education and youth who were diagnosed with speech-language disorders (9% and 7%, respectively).

Internal reliability estimates for subtests (.70-.91) and composite scores (.89-.95) were adequate in the overall sample, as well as for the clinical subgroups assessed (i.e., Language Learning Disorder, Mental Retardation, Autism, and Hearing Impairment). Test-retest reliability scores completed on a subsample of youth (N = 320 students) after a delay of approximately 16 days produced scores above .90. Inter-rater reliabilities for the seven subtests, which required subjective scorer judgment, ranged from .88-.99, which suggests that there is high inter-rater agreement. Support for validity for the CELF-4 comes from factor analyses that resulted in high correlations between the core language score and the individual indices, as well as between the receptive and expressive subscales and their respective composite scores. In addition, there were moderate correlations between the current edition and the previous edition. The receptive language score was used in the subsequent analyses to control for receptive language differences between groups; the expressive and total language scores were reported for descriptive purposes (Table 1).

***Child Behavior Checklist for ages 6-18 (CBCL/6-18; Achenbach & Rescorla, 2001).***

The CBCL/6-18 is a commonly used parent-report questionnaire to assess the emotional and behavioral functioning of children aged 6-18. The CBCL consists of 113 questions, which comprise 8 subscales (attention problems, social problems, aggressive behavior, somatic complaints, withdrawal symptoms, delinquent behaviors, thought problems, and anxious/depressed behavior). Parents rated children's behavior over the past six months on a 3-point scale, with 0 = Not True, 1 = Sometimes or Somewhat True, and 2 = Very True or Often True. Individual items were summed to create total scores for each of the subscales, and T-scores were generated separately for each subscale based on separate gender norms. The norm-referenced sample was diverse in terms of socioeconomic status and race/ethnicity. The manual listed information on reliability and validity, with high internal consistency (Cronbach's alpha .54-.96 for ages 4-11), test-retest reliability ( $r = .82-.95$ ), construct validity, and criterion-related validity. See Table 2 for sample means and standard deviations.

***Social Responsiveness Scale, Second Edition (SRS-2; Constantino & Gruber, 2012).***

The SRS-2 is a parent-reported measure that assesses the severity of social difficulties in children aged two-and-a-half to adulthood. The school-age form is designed for parents of children aged 4-18 and was included in the current study only for the ASD group as a measure of autism severity. The SRS-2, which was recently released from Western Psychological Services in October 2012, is an updated version of the commonly used Social Responsiveness Scale (SRS; Constantino & Gruber, 2005), which was designed to help distinguish children with autism spectrum disorder from those with other clinical diagnoses. The sensitivity, or the ability to accurately identify a diagnosis of ASD, of the SRS was reported to be .85, and the specificity, or the ability to exclude those without ASD, was reported to be .75. Additionally, internal

consistency of the SRS was reported to be .91-.97, the test-retest reliability was .84-.97, inter-rater reliability was .76-.95, and convergent validity was reported to be .35-.58 when compared with the ADOS and ADI-R (Constantino & Gruber, 2005).

Given that the SRS-2 was recently released in October 2012 and limited research has been conducted using this measure, reliability analyses were conducted on the sample of children with ASD using coefficient alpha in order to assess the consistency of scores within each of the five treatment subscales. When interpreting the results, the greater the consistency of the items within subscales, the higher the coefficient alpha should be. For the eight items that comprised the Social Awareness subscale,  $\alpha = .75$ ; for the 11 items that comprised the Social Cognition subscale,  $\alpha = .82$ ; for the 22 items that comprised the Social Communication subscale,  $\alpha = .92$ ; for the 11 items that comprised the Social Motivation subscale,  $\alpha = .87$ ; and for the 12 items that comprised the Restricted and Repetitive Behaviors subscale,  $\alpha = .88$ . These results indicate that there was high internal consistency for all of the treatment subscales, which is consistent with the data reported in the manual.

The SRS-2 is available for use in clinical and research settings as a screening measure or as a way to assess the impact of a treatment over time. It was normed on an ethnically diverse sample of 1,906 children. The SRS-2 consists of 65 questions, and the scoring generates a total score and five treatment subscale scores: social awareness, social cognition, social communication, social motivation, and restricted interests and repetitive behavior. Sample items from the SRS-2 include: "Is aware of what others are thinking or feeling," "Shows unusual sensory interests or strange ways of playing with toys," "Does not join group activities unless told to do so," "Walks in between two people who are talking, and "Talks to people with an unusual tone of voice." The total score and the social cognition score were used in analyses for

the current study. The parent who brought the child to the testing session completed this questionnaire, which took approximately 15 minutes. See Table 2 for sample means and standard deviations.

## **Procedure**

### **Typically developing twins**

**Three- and four-year-old testing.** At the beginning of each family's visit to the Play Lab at ages three and four, a trained graduate research assistant explained the purpose of the study, confidentiality, risks, and benefits to the parent(s) prior to obtaining consent. A separate consent form was signed to give permission to collection buccal cells, which allowed for the ability to acquire genetic information from the twins. The twins took turns being tested by a trained examiner in a separate room away from their co-twin, with each child's testing session lasting about 10-15 minutes. I was trained on and have tested twins at ages three and four; therefore, I am familiar with the administration of this protocol. As part of the testing block, the children were administered two similar false belief tasks (outlined by Wellman & Liu, 2004) to measure ToM abilities (see Appendix A for the Theory of Mind Protocol at Ages 3 and 4). After the twin testing sessions were completed, a 10-minute parent-child interaction was recorded from behind a one-way mirror without the presence of any lab members. Three separate buccal cell collections occurred over the course of the testing block, once prior to each twin's testing session, and a third prior to the parent-child interaction. Following completion of the testing session, the twins were each given gift bags and small presents (e.g., toy figures, books, activity coloring sets) as compensation for their participation. All tasks completed on the day of testing were coded later by trained raters and double entered by two different research assistants. Only the



data from the two ToM tasks from the test battery were used in the current study. Detailed scoring procedures were outlined above.

**Follow-up study.** As part of the current study, families of 6- to 10-year-old twins who completed the ToM tasks at ages three and/or four as part of SITSS (DiLalla, 2002a), as well as those families whose children participated at age five only, were contacted and asked to participate in this study. The data from the children who participated in SITSS at age five but not age three or four were still used in the larger study. These families were recruited and contacted through direct mailings and phone calls. In addition to these families, eight new families were included whose children had not previously participated in SITSS. A consenting procedure similar to the one used in the younger twin study was used in the current study to outline the purpose of the study, explain procedures used to maximize confidentiality, detail the risks and benefits of participation, and describe compensation. All of the children whose families participated in testing sessions at age three and four already had buccal cells analyzed following their initial visit(s) to the lab. Buccal cells were collected during the follow-up study from new children who were not previously part of SITSS. Children were administered a one-hour-long test battery, which comprised the five ToM measures, the receptive and expressive subtests of the CELF-4, as well as other measures to assess emotional development as part of a larger study. Two trained testers (chosen each test session from five graduate students, including myself, and two advanced undergraduate students) tested the twins, one by one tester and the other by another tester in two separate rooms.

**Compensation.** Funds were available to compensate the twin families with \$50 and a \$10, age-appropriate toy after completion of the follow-up study, thanks to a grant obtained by Dr. Lisabeth DiLalla through SIU's School of Medicine.

### **Children with autism spectrum disorder**

Twenty-five boys with autism spectrum disorder (ASD) were recruited through the Center for Autism Spectrum Disorders (CASD) on campus, which was also where the testing took place. All children whose parents agreed to their child's participation underwent a consenting procedure, which outlined the purpose of the study, confidentiality, risks, benefits, and compensation, prior to administering the five ToM measures and the receptive and expressive subtests of the CELF-4. This testing session took approximately 30 minutes, which included the five minutes needed to collect buccal cells, once at the beginning, once between the ToM tasks and the CELF-4 administration (using a counterbalanced order), and once after the testing protocol was completed. It should be noted that there were only minor issues noted during testing with this sample (i.e., inattention requiring repetition of test questions), but otherwise the testing protocol appeared to be well-tolerated by the children with ASD. In fact, many reported that they thought the ToM tasks were "fun" or "kinda like games." In addition, the parent who brought the child on the day of testing completed the Social Responsiveness Scale, Second Edition (SRS-2; Constantino & Gruber, 2012) and the CBCL (Achenbach & Rescorla, 2001), as well as a short demographic questionnaire which was used to generate an estimate of socioeconomic status for descriptive purposes.

**Compensation.** I was awarded a dissertation research grant offered by The Autism Program of Illinois (TAP) in the amount of \$1000. This award provided funding to process the genetic samples, to purchase the SRS-2 questionnaires, and to compensate the families who participated at the CASD. I compensated these families with a \$10 Walmart gift card and provided small toys (worth \$5 each) to all children with ASD following completion of the

protocol. This level of compensation was advised by a committee member familiar with this population as a reasonable compensation for the twin families from SITSS.

### **Power Analysis**

An a priori power analysis was completed with G\*Power 3 (Faul, Erdfelder, Buchner, & Lang, 2009) by including the projected minimum sample size of the current study (62 participants), given the available sample in the surrounding geographical area. This analysis indicated that there would be sufficient power to detect a medium to large effect size. This is consistent with past literature reporting significant differences with large effect sizes. The conventional value for power used in psychological research to reject the null hypothesis is 80% (Cohen, 1992), with alpha set at .05.

For hypotheses including bivariate correlations, I calculated that I would have 78% power to detect a large effect ( $\rho = .30$ ), given a sample size of 62 participants. For hypotheses requiring a  $\chi^2$  test, I calculated that I would have 79% power to detect a large effect ( $w = .35$ ) with a sample of 62 participants. Lastly, for hypotheses using MANOVA, I calculated that I would have 84% power to detect a medium effect ( $f^2 = .25$ ), given a sample size of 62 participants.

For the single behavioral genetic analysis in Hypothesis 3a, G\*Power 3 could not be used to estimate power unless the frequency of the single nucleotide polymorphism (SNP) in a specific sample to be studied is known, which it was not. Furthermore, Schmitz, Cherny, and Fulker (1998) indicated that 100 MZ and 100 DZ twins are typically required to detect significant genetic influence at the .50 level in behavioral genetic analyses. The current study did not include 200 twins; therefore, results from this genetic analysis should be replicated in a

larger sample. In sum, the current sample size would be sufficient to detect any medium to large relationship that existed for hypotheses 1, 2, and 3b.

## CHAPTER 4

### RESULTS

#### **Preliminary Analyses**

None of the variables used in the analyses required correction for skewness or kurtosis because either: 1) the variables were dichotomous; or 2) the variables were standardized using T-scores. For Hypotheses 1 and 3, one randomly selected twin was chosen from each of the twin families for the analyses in order to avoid violating the independence of samples assumption. In the analyses used in Hypothesis 2, one boy from each twin pair with at least one male was quasi-randomly selected (i.e., Twin 1 from each pair was selected unless Twin 1 was female and then Twin 2 was selected), resulting in a sample of 62 boys (37 typically developing boys and all 25 boys with ASD). There was no difference between groups for age of boys.

Group differences were significant for language ability such that typically developing twins outperformed children with ASD on receptive language,  $F(1, 67) = 35.63, p < .001$ , and expressive language,  $F(1, 67) = 42.89, p < .001$ . In addition, there were significant group differences in SES when only maternal education, occupation, and income were included (there was missing data from fathers),  $F(1, 44) = 6.51, p = .014$ . Because maternal education has been shown to be related to verbal ability in children, an ANCOVA was run to compare receptive language ability between groups after controlling for maternal education level and was significant,  $F(1, 37) = 17.96, p < .001$ . Since the group differences appear to be largely driven by language differences rather than differences in maternal education, maternal SES was reported for descriptive purposes only, especially given the concern of low power, and was not controlled for in any of the subsequent analyses. All typically developing twins were included in Hypothesis 3a for the behavioral genetics analyses (see Table 1 for total sample demographic information).

### **Development of Theory of Mind from Preschool to Middle Childhood**

Hypothesis 1 tested the understanding of social cognition in typically developing children over time. Hypothesis 1a stated that the average ToM performance from ages 3 and 4 would predict performance at follow-up (i.e., one time between ages 6 to 10). Hypothesis 1a was analyzed using Spearman's rho correlation coefficients, specifically a nonparametric partial correlation controlling for age, given the varying ages at follow-up. The variables included in the analyses were the two ToM scores (memory and contents) from ages 3 and 4 and all five ToM task scores at follow-up (see Table 3). The memory scores at ages three and four were not correlated with performance on any of the five ToM tasks at follow-up ( $p > .05$ ), with the exception that memory at age four was negatively correlated with the score on the real-apparent emotion task at follow-up,  $\rho(38) = -.33, p = .04$ . The contents score at age three was not correlated with the performance on any of the five tasks at follow-up ( $p > .05$ ), but the contents score at age 4 was negatively correlated with two of the follow-up ToM tasks, belief emotion and real-apparent emotion,  $p < .05$ . Thus, ToM ability at age 4, especially performance on the contents question, appears to be more related to future ToM performance than performance at age 3.

Hypothesis 1b stated that younger children in the follow-up sample would not pass as many of the five ToM measures as older children in the current study, thus generating a lower overall score. Hypothesis 1b also was tested using Spearman's rho correlation coefficients. Age (in months), the five individual ToM scores, and the total ToM score were the variables used in this correlational analysis. Results showed that age was only positively correlated with the total ToM score at follow-up for the combined sample,  $\rho(86) = .23, p = .03$ , but not any of the individual ToM tasks ( $p > .16$ ). However, when the groups were separated by diagnostic status,

age was positively correlated with performance on three out of the five tasks (contents false belief,  $\rho(61) = .33, p = .009$ , explicit false belief,  $\rho(61) = .40, p = .001$ , and real-apparent emotion,  $\rho(61) = .34, p = .007$ ) in the typically developing sample. Age was not correlated with performance on the belief emotion task,  $\rho(61) = .05, p = .70$  or the understanding sarcasm task,  $\rho(61) = .21, p = .10$ . There were no significant correlations between age and ToM task performance for any of the five ToM tasks ( $p > .25$ ) for the children with ASD (see Table 4). Thus, it appears that chronological age is important for the development of advanced social cognition in typically developing children but not children with ASD.

### **Group Differences in Social Cognitive Abilities**

Hypothesis 2 tested the group differences in ToM abilities of children with and without autism spectrum disorder. Hypothesis 2a stated that children with ASD would not score as high as typically developing children on all five measures of ToM included in the current study given their widespread deficits in social cognitive abilities. Only boys from the typically developing sample were included because all the children in the ASD sample were boys. Specifically, group differences in ToM performance in Hypothesis 2a were tested using a series of Chi-square tests. Age was not controlled for because age was not significantly different between the diagnostic groups as shown in the preliminary analyses section. This resulted in a subgroup of 62 boys: 25 ASD boys and 37 boys from the twin sample who were quasi-randomly selected (Twin 1 from each twin pair was selected unless Twin 1 was female and then Twin 2 was selected). Five separate Chi-square analyses were conducted using diagnostic status (i.e., typically developing or ASD) and the score from each of the five ToM tasks administered when boys were between the ages of 6 and 10. Results indicated that typically developing children outperformed children with ASD on all five tasks. (see Table 5).

### **Developmental progression of ToM abilities**

Figure 1 shows the results of a profile plot of the group differences broken down by ToM task. Although the twins outperformed the children with ASD on all tasks, the order effects between groups were not significant, with the exception of the understanding sarcasm task. These data are in contrast to previously published research suggesting that children with ASD develop social cognitive abilities in a different order than typically developing children (Wellman et al., 2011).

In addition, a reliability analysis was run in order to determine whether the performance on any of the five tasks was markedly different than any other. The total ToM score had a Cronbach's alpha of .60, which is fair, and all of the inter-correlations between tasks were positive, ranging from .23-.41. Importantly, the alpha did not improve if any of the items were deleted, suggesting that these items are all conceptually related to the broader construct of social cognition. However, the inter-correlations were low, which reflect the heterogeneous nature of this construct. Thus, the use of the total ToM score was used in subsequent analyses as a way to increase variability.

### **Language ability**

In order to assess group differences on language abilities, an analysis of covariance (ANCOVA) was used to compare groups on language ability after controlling for age. It should be noted that the CELF-4 was not collected on the first 20 twin families because this measure was added after data collection began; therefore, the sample for this analysis is smaller than the sample used in the other analyses that do not include the CELF-4. In addition, consistent with the other analyses investigating group differences, only boys were used in this analysis to minimize confounds. Thus, the total N for the typically developing sample with CELF-4 data



was 21. Results showed that there was a significant group difference on CELF-4 language ability,  $F(1, 51) = 23.60, p < .001, \eta = .32$ , such that typically developing children had higher receptive language scaled scores compared to the children with ASD even after controlling for age (see Table 6).

### **Theory of mind**

In order to test whether age and/or language ability better predicted total ToM score (i.e., the sum of the performance on all five of the tasks), a hierarchical regression was used. CELF-4 receptive language scores and age were included as the control variables in Step 1 and group was entered in Step 2 to predict overall ToM ability. During the first step ( $F(2, 45) = 35.42, p < .001$ , adjusted  $R^2 = .61$ ), receptive language ( $\beta = .88, p < .001$ ) and age at follow-up ( $\beta = .43, p < .001$ ) were significantly related to the total ToM score, such that children with better receptive language ability and older children scored higher on the ToM total score. In the second step, diagnostic group was added to determine its effect after controlling for receptive language and age. At this step ( $\Delta R^2 = .03, p = .059$ ), group ( $\beta = -.22, p = .059$ ) was not predictive of ToM total score at follow-up after accounting for receptive language ability and age, ( $F(3, 45) = 26.40, p < .001$ , adjusted  $R^2 = .63$ ) (see Table 7).

### **Social and emotional functioning**

Social-communication problems and restricted and repetitive behaviors are core deficits in children with ASD, and emotional processing difficulties make navigating social situations even more difficult for these children. Thus, hypothesis 2b stated that children with ASD would have significantly more difficulties on the CBCL scales related to social abilities (i.e., Social Problems, Thought Problems, Withdrawal) and emotional processing abilities (i.e., Anxious/Depressed) but not other CBCL problem scales (i.e., Aggression, Delinquency,

Somatization) compared with typically developing children. It should be noted that the CBCL Thought Problems scale is believed to better measure unusual or odd behavior than thought processing difficulty. In addition to these four CBCL subscales, Attention Problems was added as a dependent variable following data collection due to the behavioral observations obtained during the testing sessions indicating that several children with ASD struggled with inattention throughout the tasks. Hypothesis 2b was analyzed using a MANCOVA, controlling for language differences, to test whether groups significantly differed on the hypothesized CBCL scales. Diagnostic status (typically developing or ASD) was used as the independent variable, receptive language score was used as the covariate, and scores on the CBCL scales were used as the dependent variables. Results from the MANCOVA indicated that children with ASD were more impaired on the Thought Problems subscale, and trends in the hypothesized direction were noted in the Attention Problems and Anxious/Depressed subscales. There were no significant group differences on the Social Problems and Withdrawn/Depressed subscales (see Table 8).

### **Autism severity and ToM performance**

Hypothesis 2c, which was analyzed only using children with ASD, stated that autism symptom severity, as measured by the treatment subscales on the SRS-2, would be negatively correlated with ToM abilities. Hypothesis 2c was analyzed using Spearman's rho correlation coefficients and included the scores on the five treatment subscales (i.e., social cognition, social communication, social awareness, motivation, and restricted/repetitive behaviors) and the scores on the five individual ToM items. In addition, a bivariate correlation was conducted using the total SRS-2 score and the total ToM score as variables.

Results from the nonparametric correlations (see Table 9) showed that lower scores on explicit false belief,  $\rho(25) = -.46, p = .021$ , and real-apparent emotion,  $\rho(25) = -.56, p = .006$ ,

were associated with higher parent-rated social cognitive difficulties. In addition, lower scores on the real-apparent emotion task were associated with higher parent-rated social communication difficulties,  $\rho(25) = -.52, p = .008$ , and restricted/repetitive behaviors,  $\rho(25) = -.44, p = .028$ . Given the nature of nonparametric tests, low statistical power may be influencing the ability to detect significant relationships between these variables. Results from the bivariate correlation between total ToM score and Total SRS-2 score indicated that there is a negative relationship between these variables,  $r(25) = -.44, p = .028$ , suggesting that children with greater social impairments do not pass as many ToM tasks. It should be noted that correlations between the SRS-2 treatment subscales and the understanding sarcasm task could not be computed due to the fact that none of the children with ASD in the current study passed this task.

An exploratory analysis examined the correlation between the CBCL scales that were hypothesized to be related to ASD behaviors (Anxious/Depressed, Attention Problems, Social Problems, Thought Problems, and Withdrawal) and scores on Social Cognition, one of the treatment subscales on the SRS-2 measuring parent-reported social cognitive abilities. Results showed that there were strong correlations between the scores on the Social Cognition subscale from the SRS-2 and the Attention Problems ( $r = .62, p = .001$ ), Social Problems ( $r = .59, p = .002$ ), Thought Problems ( $r = .57, p = .003$ ), and Withdrawal subscales ( $r = .60, p = .001$ ) from the CBCL. A non-significant correlation was found for the Anxious/Depressed subscale ( $p = .058$ ).

### **Genetic Effects on Social Cognition**

Hypothesis 3 tested whether broad heritability ( $h^2$ ) and different genotypes of the DRD4 and 5-HTTLPR genes in particular would be related to performance on ToM tasks. Hypothesis 3a stated that there would be significant heritability for ToM task performance. Specifically,

Hypothesis 3a analyzed heritability of ToM abilities by using intraclass correlations to correlate Twin 1's preschool ToM performance with Twin 2's preschool ToM performance, separately for MZ and DZ twins. Similarly, Twin 1's school-aged ToM score was correlated with Twin 2's school-aged ToM score, for each ToM task administered at follow-up and separately for MZ and DZ twins. Then, the intraclass correlations for each twin type were transformed into z-scores using Fisher's r-to-z transformation method. Next, t-tests were used to compare the z-scores for MZ and DZ twins to test whether they were significantly different from each other. Finally, heritability was estimated using the following formula outlined by Falconer (1960):

$$h^2 = 2(r_{MZ} - r_{DZ}).$$

### **Behavioral genetic hypotheses**

Results using Falconer's estimates of heritability are reported in Table 10. There were no significant heritability estimates; however, there were two heritability estimates that were significant at the  $p < .10$  level: the school-age contents false belief and understanding sarcasm tasks. In a few instances, the MZ correlation was negative (e.g., contents at age four); thus, the Falconer heritability estimate could not be computed, and the resulting heritability estimate was zero. This pattern ( $DZ > MZ$ ) suggests that the effect is driven by environmental factors rather than genetic factors. For the contents false belief and the understanding sarcasm tasks at follow-up, the correlation between MZ twins was more than two times greater than the correlation for DZ twins. As a result, the MZ correlation is substituted for  $h^2$ , and it is believed that dominant genetic effects are at play in this situation. There was no evidence for heritability for the other ToM measures.

### **Molecular genetic hypotheses**

Hypothesis 3b used three sets (one for each gene [5-HTTLPR and DRD4] and one using

both genes) of five Chi-square analyses -- 1) total sample; 2) twins only; 3) ASD only; 4) males only; and 5) females only -- to test whether children with risk alleles performed worse on ToM tasks compared to those without risk alleles. One child was selected from each family to avoid violating the independence of sample statistical assumption. A breakdown of the frequency of genotypes by diagnostic group can be seen in Table 11. Specifically, the first set of five Chi-square analyses tested whether children with the S or L<sub>G</sub> allele (versus 2 copies of the L<sub>A</sub> allele) of the 5-HTTLPR polymorphism performed worse on ToM performance, analyzed for each ToM task separately (see Table 12). The second set of five Chi-square analyses were conducted to test whether children with six or more repeats (versus both alleles with fewer than six) in the DRD4 polymorphism performed worse on ToM performance, analyzed for each task separately (see Table 13). Lastly, the third set of five Chi-square analyses tested whether children with at least one risk allele for both genes (5-HTTLPR and DRD4) performed worse than those who had at least one risk allele for one gene but not the other (5-HTTLPR or DRD4) who in turn would perform worse than those individuals with no risk alleles on each of the ToM tasks (see Table 14).

The genotypes were scored dichotomously for the first two sets of analyses (5-HTTLPR and DRD4), with a score of 0 indicating that there was no risk allele present and a score of 1 indicating that there were 1 or 2 risk alleles present. For the third analysis using both 5-HTTLPR and DRD4, the genotypes were scored trichotomously. Specifically, children with at least one risk allele in both genes (i.e., 5-HTTLPR and DRD4) were coded with a 2, children with only one risk allele in one or the other gene (but not both) were coded with a 1, and children without any risk alleles were coded with a 0. The task performance for all analyses was scored dichotomously such that a score of 0 indicated that the child failed the task and a score of 1

indicated that the child passed the task. Each of the three main molecular genetic analyses was run five times using different samples: total sample (N = 86); all twins (N = 61); all children with ASD (N = 25); males only (N = 62); and females only (N = 33). In addition, three ANOVAs were run to test for genetic effects on the total ToM score for each gene separately and for the triallelic combination of risk alleles using both 5-HTTLPR and DRD4 polymorphisms. Genotype data were unavailable for one of the male twins; however, all of the gene data were available for the remainder of the sample.

### **5-HTTLPR polymorphism**

Significant results from the Chi-square analyses for the 5-HTTLPR genotype are reported in Table 12. In the twin and female samples, there was a significant genotype effect for the contents false belief task. In the male sample, there was a significant genotype effect for the belief emotion task; however, this did not survive Bonferroni correction. There were no other significant genotype effects for the 5-HTTLPR polymorphism.

Results from the ANOVA showed that there was no significant difference between those with and without 5-HTTLPR risk alleles on total ToM task performance, (see Table 15).

### **DRD4 polymorphism**

Significant results from the Chi-square analyses for the DRD4 genotype are reported in Table 13. In the total sample, there were no significant effects for any ToM task. Analysis of the subsamples indicated that the genotype effects in the twin sample for the explicit false belief task and the belief emotion task were significant at the  $p < .10$  level and in the hypothesized direction. In the ASD and male only samples, there was a significant genotype effect for the contents false belief task. There were no other significant genotype effects for the DRD4 polymorphism.

Results from the ANOVA showed that there was no significant difference between those with and without DRD4 risk alleles on total ToM task performance (see Table 15).

#### **Triallelic combination of risk alleles from 5-HTTLPR and DRD4**

Significant results from the Chi-square analyses including both genes (5-HTTLPR and DRD4) are reported in Table 14. In the total sample, there were no significant effects for any of the ToM tasks. Analysis of the subsamples indicated that there was a significant genotype effect in the ASD sample for the contents false belief task. There were no other significant effects for the triallelic combination.

Lastly, the results from the ANOVA showed that there was no significant genotype effect between risk allele groups when using both genes on the total ToM score (Table 15).

## CHAPTER 5

### DISCUSSION

This study adds unique and important contributions to the growing literature on the psychological and genetic factors important for the development of social cognition in children with and without autism spectrum disorder. The inclusion of children from a longitudinal study provided an opportunity to analyze the development of social cognitive abilities over time, and the inclusion of twins allowed for the examination of heritability of social cognition in this sample. In the twin sample, age was related to ToM task performance; however, receptive language ability, not age, was strongly related to task performance in children with ASD. Analysis of group differences showed that children with ASD were markedly impaired on all ToM tasks compared with typically developing peers. Furthermore, it appears that ToM abilities progress differentially based on group membership, and more advanced skills such as the understanding of non-literal language, or pragmatics, may not develop in children with ASD until after the age of 11. Parents of children with ASD reported social, emotional, and behavioral difficulties on the CBCL in the hypothesized areas (i.e., social problems, thought problems, withdrawn/depressed, and attention problems), which suggests that the CBCL provides useful supporting information for clinicians who are assessing children with ASD. In addition, the newly released SRS-2 appears to be a valid and useful measure of autism severity given its strong negative relation to ToM task performance and positive relation to social, emotional, and behavioral characteristics commonly seen in children with ASD.

This study was one of the first of its kind to analyze genetic differences in children with and without autism spectrum disorder using both behavioral and molecular genetic approaches. Although the behavioral genetic analyses were limited due to small sample size, the findings suggest that some areas of social cognition are more influenced by genetic influences than are



other cognitive skills. Moreover, molecular genetic analyses suggest that the more basic ability to understand that others have thoughts, beliefs, and mental states that are different than one's own (as measured by the contents false belief task) may be influenced by the underlying biological pathways that regulate the production of serotonin and dopamine, two neurotransmitters that have been linked to social cognitive abilities in previous studies of animals and humans.

### **Development of Theory of Mind from Preschool to Middle Childhood**

Despite expectations that performance on false belief tasks during preschool would be positively related to advanced ToM abilities during middle childhood, findings did not support this hypothesis. In addition, the few significant results reported were in the opposite direction, with children who failed false belief tasks during preschool being more likely to pass the two emotion tasks during middle childhood. When taking into account the order in which typically developing children progressed through the tasks (i.e., contents false belief > belief emotion > real-apparent emotion > explicit false belief > understanding sarcasm), it could be that children who did not pass the simplest task (contents false belief) at age 4 showed the most improvement by middle childhood on the next two tasks (belief emotion and real-apparent emotion) in the developmental progression. Most research in this area has reported that children are largely able to pass this type of task between the ages of 5 and 6. Therefore, another explanation for these results could be that children in the preschool sample who were tested shortly after their third and/or fourth birthdays may have not yet mastered the ability to pass false belief tasks given their young age. Thus, it is possible that there was more variability in the youngest age groups (e.g., ages three and four) than during middle childhood, and more significant and positive correlations between preschool and school-age performance may have been found if children were tested

shortly after their fifth birthday instead of their third or fourth birthdays. The finding that children who failed the contents false belief task at age 4 did better on the two tasks with an emotional component at follow-up suggests that ToM abilities develop in a non-linear pattern over time in typically developing children (Wellman & Liu, 2004; Wellman et al., 2011).

As expected, chronological age was positively related to performance on ToM tasks in typically developing children, which suggests that children develop more advanced ToM abilities along with increased knowledge and higher-order reasoning abilities, skills also known to improve with age (Leslie, Friedman, & German, 2004). However, age was not related to performance on the belief emotion and understanding sarcasm tasks in the twin sample. Low power may have affected the ability to find significant results on the understanding sarcasm task, especially since only a small proportion (14.5%) of the typically developing children passed the understanding sarcasm task given its difficulty level for school-age children.

In contrast, age was not related to any of the ToM tasks in the ASD sample. One possible explanation is that other factors such as language abilities are responsible for progressive development of social cognition in children with ASD rather than age-related brain maturation. This finding is consistent with other studies assessing advanced ToM abilities in children with ASD that included a measure of receptive verbal abilities (Fisher, Happe, & Dunn, 2005; Peterson et al., 2012; Scheeren, Rosnay, Koot, & Begeer, 2013). Studies investigating the development of advanced social cognitive abilities over time such as the current study are especially important because so much of the previous research on ToM abilities in children has focused on false belief tasks (Wellman et al., 2001). Therefore, this study provides more information regarding ToM abilities in school-aged children when social demands begin to increase.

### **Group Differences in Social Cognitive Abilities**

As expected, and consistent with most (Beaumont & Sofronoff, 2008; Brent, Rios, Happe, & Charman, 2004; Peterson et al., 2012; Sobel, Capps, & Gopnik, 2005; Wellman et al., 2001), but not all (Scheeren et al., 2012) of the previous research in the area of advanced ToM abilities, children with ASD performed worse on all of the ToM tasks when compared with typically developing children. This study included five advanced ToM tasks that were previously included in other studies for the sake of generalizability, including two tasks that included an emotional processing component. The findings that children with ASD performed worse on all of the tasks provides support for social information processing theory, which states that difficulties in social competence are related to poor social skills and difficulty processing emotional information, and the ToM theory of ASD (Baron-Cohen, 2000), which states that all children with ASD have deficits in social cognitive abilities.

Analysis of the order in which children progressed through the ToM tasks did not reveal a differential pattern based on group membership, with the exception of the sarcasm task. As predicted, the typically developing children performed significantly better than children with ASD; however, there were no other detectable order effects based on type of task, except for the fact that both groups performed worse on the sarcasm task than predicted. Further studies using these tasks are needed to better understand the developmental progression of social cognitive abilities in typically developing children and children with ASD.

An interesting qualitative finding was noted during the data collection of the boys with ASD. A moderate proportion of children (38.5%) who were completing the real-apparent emotion task (i.e., asking the child to state how the boy in the story looked on his face and how he felt inside) flipped over the stimulus sheet that showed a picture of a boy from behind, as if to

see how he looked on his face. Although data on whether any of the twins performed this action was not collected, a retrospective report suggested that few, if any, of the children in the twin sample performed this action. One possible explanation for this finding in the ASD population is that children with poor social skills often have difficulty processing emotional information such as facial expressions. As such, these children are often taught in social skills groups to look at others' faces as a way to infer others' mental states. This finding is consistent with a previous study by Baron-Cohen and Goodhart (1994) on "seeing leads to knowing." The authors postulated that children with ASD do better on tasks such as the real-apparent emotion task than false belief tasks because they have likely been "trained" to know what a person's face looks like in certain situations (happy, sad, so-so). Thus, a face is something that you can see to gather information unlike someone's mind, an abstract concept, which cannot be seen. However, this also suggests that these children lack abstract reasoning abilities given their lack of understanding of how pictures work, which is consistent with prior research showing that children with ASD are able to pass concept identification but not concept formation tasks (Minschew, Meyer, & Goldstein, 2002).

For the children with ASD, the two false belief tasks were second and third (explicit and contents, respectively) in the developmental progression, the belief emotion task was fourth, and the understanding sarcasm task was the most difficult, with none of the children with ASD passing this task. This differential pattern suggests that not only are children with ASD delayed in their ability to acquire social thinking abilities, but also the order in which these skills develop over time are different. This finding is especially important to consider when designing and implementing social thinking curricula, especially if skills are taught using a stepwise approach.

In sum, the finding that the children with ASD performed worse on all five tasks of advanced ToM abilities is consistent with the ToM theory of ASD.

When comparing groups, children with ASD performed significantly worse on measures of receptive and expressive language, a pattern that has been consistently shown in previous research in this area despite no differences in cognitive abilities (Abu-Akel, 2003; Lackner et al., 2010; Ozonoff et al., 1991). It should be noted that these differences were seen in this study despite the fact that the five advanced ToM tasks were chosen in part because they required limited language abilities and included pictures to supplement the oral instructions. Most interesting, however, is that, after controlling for age and receptive language ability, the group differences on total ToM task performance were no longer significant. This finding suggests that children with ASD should be given intensive receptive, expressive, and/or pragmatic language instruction before, or in conjunction with, interventions seeking to improve social thinking skills.

Similarly, parent-reports of problem behaviors indicated that children with ASD had greater social, emotional, and behavioral challenges than typically developing peers. Children with ASD were reported to have greater difficulties in specific areas related to a diagnosis of ASD that can negatively impact the ability to engage in successful social interactions with peers. Interestingly, the group differences in social problems were not significantly different despite a trend in the hypothesized direction. This could be related to low power or the fact that the children with ASD in the current study were in regular education classrooms and, as such, did not exhibit as many of the aberrant social behaviors included on the CBCL. Clinical elevations on the Thought Problems and Withdrawn/Depressed scales suggest that the items on these scales may better represent the social, emotional, and behavioral difficulties of higher functioning children with ASD who spend the majority of the day in regular education classrooms. Due to

the limited scope of this study, teacher-reported problem behaviors were not included, but these data should be collected in future studies on this topic to determine whether similar patterns emerge.

The SRS-2 was included in the current study as a measure of autism severity given the strong psychometric properties and clinical utility of the previous edition of the SRS. It was hypothesized that the total score on the SRS-2, as well as the scores on the five treatment subscales on the SRS-2 (social awareness, social cognition, social communication, social motivation, and restricted and repetitive behaviors), would be negatively related to ToM task performance. Although the sample was small, several strong correlations emerged when comparing parent-reported problem behaviors with advanced ToM abilities as measured by performance during laboratory tasks. Performance on the social cognition subtest from the SRS-2 was negatively related to performance on the explicit false belief and real-apparent emotion tasks. Given that these tasks were the easiest two tasks for the children with ASD, this may indicate that parent-report of social cognitive ability is only useful for assessing lower-level abilities. The limited significant findings within this subscale also might indicate that parents cannot predict their child's thoughts based on their behaviors; therefore, self-report or direct observation may be more useful. However, given the strong correlations between autism severity level and ToM task performance, the SRS-2 may be a useful tool for assessing social cognitive abilities through parent report when more time-intensive laboratory testing is not available. Furthermore, given that the SRS-2 is new and, therefore, not well-studied, strong correlations of the treatment subscale scores and total score with ToM task performance enhance its validity. Due to limited time and resources, this measure was only given to parents of

children with ASD. In future studies, the SRS-2 should be completed by parents of typically developing children given that autism severity is measured dimensionally on the SRS-2.

### **Genetic Effects on Social Cognition**

It was expected that ToM abilities would be heritable, with higher correlations between MZ twins compared with DZ twins, given some previous support for this pattern in the literature (Hughes & Cutting, 1999; Scourfield et al., 1999). The heritability hypothesis was not fully supported in this study, which suggests that environmental factors may be more likely than genetic factors to influence the development of advanced ToM abilities (Hughes et al., 2005), or that the sample was too small to detect an effect. There were some noteworthy trends in the hypothesized direction (contents false belief and understanding sarcasm) that were likely affected by power. Therefore, it is possible that some of the advanced ToM abilities included in this study, specifically the easiest and most difficult tasks, may be more influenced by genetic factors than environmental factors at this age.

However, the finding that environmental experiences may be accounting for more of the variation in social cognitive abilities in middle childhood may not be surprising. Briley and Tucker-Drob (2013) recently published a meta-analysis of the heritability of cognitive development, which included data from 16 longitudinal twin and adoption studies. Previous literature has shown that heritability is lower for general cognitive ability in early childhood and increases linearly with age (Plomin, 1999; Spinath, Ronald, Harlaar, Price, & Plomin, 2003). Specifically, the heritability for preschool-aged children is estimated to be 20–30%, rising to 40% in middle childhood (9 years of age), 55% in adolescence, and increasing to about 66% during early adulthood. Thus, it is likely that this pattern holds for social cognitive abilities as well. Plomin & DeFries (1985) coined the terms “innovation” and “amplification” to help

describe the phenomenon that heritability of cognitive abilities increase with age. Amplification, or the idea that early genetic factors related to the development of cognitive abilities may become more important later in the selection of environments, may account for the size of the heritability estimate seen in this study for the contents false belief task, the most basic ToM ability. The findings from the meta-analysis by Briley and Tucker-Drob (2013) are consistent with the theory that the genetic influences that are present in early childhood are primarily novel, and they become amplified as children get older. On the other hand, innovation, or the increase in heritability due to novel genetic factors not previously present or activated, may be influencing the trend noted for the understanding sarcasm task. Mastering this task requires an understanding of non-literal language, which is a novel skill that begins to develop in middle childhood and adolescence. Thus, according to the theory by Plomin and DeFries, the genes influencing the development of an understanding of sarcasm are being “turned on” for the first time. Follow-up studies are needed to test this theory.

For a few of the ToM tasks, the correlation for MZ twins was lower than the correlation of the scores on the same task for the DZ twins, which underscores the importance of environmental influences on behavior. One possible explanation for this finding is that early environmental factors (e.g., prenatal environment) may affect brain development in such a way that the genetic similarities in MZ twins are overshadowed by environmental differences. Another possibility is that MZ twins may not be learning anything new from their genetically identical co-twin if they are sharing friends or participating in similar activities together. DZ twins, on the other hand, who are genetically more like non-twin siblings, may be choosing different environments (e.g., peer groups or activities), thus exposing themselves to different attitudes, beliefs, and desires. Previous studies of theory of mind in twins with and without other



siblings have shown that twins without other siblings perform worse than twins with at least one sibling and have comparable scores to only children without siblings (Cassidy, Fineberg, Brown, & Perkins, 2005). In addition, performance was boosted when the sibling was older and of the opposite sex, which suggested that the more different the experiences of the older sibling are, the better the child's performance on theory of mind tasks.

It was expected that results from the molecular genetic analyses would show that children with risk alleles in either or both of the 5-HTTLPR and DRD4 genes would perform worse on ToM tasks than those without risk alleles based on previous findings (Lackner et al., 2010, 2012; Skuse & Gallagher, 2011). Findings from the current study indicated that serotonin and dopamine risk alleles may not play an influential role in the developmental of advanced ToM abilities. There were some limited significant findings, specifically related to performance on the contents false belief task in relation to the DRD4 polymorphism for children with ASD. Additionally, there were a few trends in the hypothesized direction such that individuals with risk alleles in either or both of the 5-HTTLPR or DRD4 polymorphisms performed worse than those with fewer or no risk alleles. In general, the findings in the current study provide support for the idea that biological pathways involving dopamine in the brain are necessary for the understanding of advanced ToM abilities in boys and in children with ASD. This is consistent with the findings of Lackner and colleagues, who reported a link between DRD4 risk alleles and performance on theory of mind tasks in preschoolers (Lackner et al., 2010; Lackner et al., 2012). Future studies with larger sample sizes are needed to further investigate whether serotonin and dopamine are related to the development of higher-order cognitive processes such as advanced ToM abilities.

### **Clinical Implications**

The field of social thinking has become increasingly more important given the dramatic rise in prevalence rates of ASD in recent years. Despite the gravity of this problem, the number of evidence-based interventions used to treat difficulties in social cognition is not keeping up with the demand. However, a few high-quality interventions have demonstrated a strong evidence base recently, including the Social Thinking curriculum (Winner, 2002) and the UCLA PEERS program (Laugeson, Frankel, Mogil, & Dillon, 2009). The Social Thinking program attempts to teach high-functioning children with ASD social decision-making skills in vivo rather than through typical social skills groups. A brief report measuring the effectiveness of the Social Thinking intervention examined the outcomes of six males with Asperger syndrome following the implementation of this approach, which occurred over the course of eight weeks (Crooke, Hendrix, & Rachman, 2008). Findings indicated significant improvements across several areas assessed, including the integration of what one hears and sees others say and do, initiation of social overtures, and use of appropriate verbal responses, as well as decreases in inappropriate verbal and nonverbal responses.

The other widely used evidence-based social skills intervention for children with ASD is the UCLA Program for the Education and Enrichment of Relational Skills (PEERS), which is a 14-week program targeted for middle school and high school students who are interested in learning how to develop and maintain friendships. Research has shown that this program is efficacious and long-lasting, as evidenced by improvements in social responsiveness, social communication, social cognition, cooperation, as well as decreases in restricted and repetitive behaviors in adolescents with ASD, as reported by teachers (Laugeson et al., 2009). These effects persisted at a 14-week follow-up assessment (Laugeson, Frankel, Gantman, Dillon, &

Mogil, 2012). Modifications to current social skills curricula and future interventions should incorporate developmentally sensitive research findings related to social cognition such as the findings from the current study in order to maximize benefits for children who participate in them.

However, interventions such as these may not be sufficient for symptom reduction. Given the findings from this and other related studies on advanced cognitive abilities in children with ASD, more rigorous assessment and treatment may be required for optimal gains. Specifically, more advanced tools, such as genotyping, as well as a thorough speech and language evaluation, may help determine the level of vulnerability with which each child is presenting prior to beginning an intervention. Studies on monogenic disorders, such as Fragile X disorder, the most common known genetic cause of autism, are helping to progress the field by increasing knowledge of how genetic variations may influence the development of human behaviors (Losh, Martin, Klusek, Hogan-Brown, & Sideris, 2012). More detailed information related to an individual's strengths (e.g., increased verbal skills) and weaknesses (e.g., presence of a risk allele) could be used to design tailored treatments with more targeted goals.

### **Strengths and Limitations**

The current study has many important strengths that are noteworthy. First, the inclusion of a longitudinal study of twins allowed for the ability to conduct longitudinal and behavioral genetic analyses within the same study. Second, the sample included a high-functioning group of children with ASD in middle childhood, which is an important age for social development and one that has only recently begun to be investigated. Lastly, the inclusion of a range of advanced ToM tasks that are low in verbal demands increases the likelihood that group differences are not confounded by differences in the ability to understand or complete the tasks.

Although the current study has many important strengths, it is not without limitations. First, the sample size was not large enough to conduct behavioral genetics analyses with sufficient power. Additionally, the longitudinal analysis of ToM performance from preschool to school-age was also under-powered due to the low response rate of families over time. However, the current sample size was sufficient to detect moderate to large effects when analyzing group differences.

Second, due to time and resource limitations, other cognitive measures such as intelligence tests, memory tests, and/or executive functioning measures were not given as a way to test and control for potential group differences in these areas and to illustrate the dissociation between cognition and social cognition. However, previous studies have reported that, even after controlling for IQ and executive functioning abilities, children with ASD are still more impaired than their typically developing peers (Abu-Akel, 2003; Lackner et al., 2010). Given the limited number of studies on advanced ToM abilities in high-functioning children with ASD, however, a brief cognitive screening and/or other neuropsychological measures (e.g., memory) should be included to further clarify the neurocognitive factors important for the development of social cognitive abilities. Specifically, memory tests would be important to include in future studies of social cognitive abilities given that specific regions of the brain associated with memory overlap with the areas of the brain associated with four forms of self-projection: episodic memory, prospection, theory of mind, and navigation (Buckner & Carroll, 2007). The authors suggested that in order to successfully engage in social interactions, an individual must think about what might happen in the future (prospection), remember their own past behaviors, take the perspective of others, and then navigate, or “find their way” through a social interaction. Thus,

from a cognitive psychology perspective, ToM is believed to be a type of self-projection that relies on remembering what happened in the past in order to plan future behavior.

Third, the parents of typically developing children were not asked to complete the SRS-2 due to time and resource limitations despite the fact that data from this measure would have been useful in further analyzing group differences, as well as providing data to support the validity of the SRS-2. Fourth, it is possible that differences in the length of the testing protocols for each group could have confounded the results. However, this is very unlikely given that the children with ASD consistently performed worse on ToM and language tasks despite the reduced time required to complete their testing compared to the twins. Moreover, considering that the children with ASD in this study were chosen because they spent much of their day in the regular education classrooms, it also is unlikely that differences in the length of the testing battery of this magnitude would have affected their performance dramatically.

Lastly, the age range of children included in this study limited the opportunity to analyze performance on the understanding sarcasm task in both groups given its level of difficulty for school-age children. Moreover, a previous study by Ozonoff et al. (1991) reported that children with high-functioning ASD were able to pass more advanced ToM tasks by adolescence; however, the upper end of the age range in this study was pre-adolescence, which precluded the ability to test this relationship. Thus, these tasks should be administered to an older sample of children with and without ASD in future studies in order to determine the approximate age at which children develop an understanding of non-literal language such as sarcasm or irony.

### **Future Directions**

The current study used explicit/direct tasks to measure advanced ToM abilities. However, future studies should include implicit/indirect tasks in addition to explicit tasks as a

way to eliminate language demands given the difficulties in receptive and expressive language in many children with ASD. Although typically used with infants and younger children, implicit tasks have the benefit of being less cognitively demanding, which would allow for the analysis of more fine-grained differences in social cognitive abilities once language and cognitive demands have been minimized (Apperly & Butterfill, 2009). In addition, researchers including both implicit and explicit tasks would have the opportunity to compare the performance on these two tasks to each other as a way to see if they are truly measuring the same broader construct of social cognition. If they are related, the findings from the studies of younger children using implicit tasks would have more support, and interventions for young children could begin earlier once abnormalities in the development of social cognitive abilities are identified.

This study included five advanced ToM tasks that were selected because they tapped related but unique aspects of social cognition and required limited language demands on the part of the examinee. Previous studies on this topic, albeit limited, have used a surprisingly large number of different types of tasks to measure the construct of social cognition including strange stories, vignettes, pictures, and silent films. Future studies should incorporate as many of the non-redundant tasks related to social cognition as possible to explore the factor structure of the construct of social cognition using factor analytic techniques. The factors identified in such a psychometric study should be used in future studies on this topic.

Lastly, given that children with ASD were able to pass the real-apparent emotion task more easily than any other task and nearly 50% of the time, future studies should include a more advanced emotion such as surprise to increase the difficulty level of the task for both children with ASD and typically developing peers.

## **Conclusion**

The ability for children to integrate information about others' perspectives, emotional states, and behaviors is essential during successful social interactions with peers. Findings from the current study suggest that children with autism spectrum disorder (ASD) have more difficulty completing tasks that require them to take another's perspective than do typically developing peers. Neurobiological and genetic factors may be influencing the observed group differences, including the delayed onset of skill development, as well as the differential order in which these skills emerge. Furthermore, age is a crucial factor in the development of more advanced social cognitive abilities in typically developing children, whereas verbal ability appears to be an influential factor in children with ASD. Analysis of the development of these skills over time provides useful information for both groups about how and when to intervene when children are exhibiting social difficulties in middle childhood. Thus, this study provides support for the fact that implementing developmentally appropriate early interventions may be the most promising way to improve the quality of social interactions in typically and atypically developing children.

Table 1

*Sample Demographic Information*

	N	Twins (N= 127)			ASD (N = 25)		
		<i>M</i>	<i>SD</i>	Range	<i>M</i>	<i>SD</i>	Range
Age	62	8.07	1.56	6-11	8.69	1.29	6-11
SES	62	8.21	1.68	4-11	7.68	2.18	4-10
<b>Language Ability</b>							
Receptive	43	10.99 <sup>a</sup>	2.31	3-17	6.46	4.21	1-13
Expressive	43	11.34 <sup>a</sup>	2.53	5-16	6.23	3.79	1-13
<b>Intellectual Ability</b>							
Verbal IQ	9	--	--	--	86.56	6.25	77-96
Non-verbal IQ	12	--	--	--	96.92	15.93	63-112
FSIQ	10	--	--	--	90.00	9.50	70-107

*Note.* SES: socioeconomic status; Language ability was measured using the Clinical Evaluation of Language Fundamentals, 4<sup>th</sup> Edition (CELF-4).

<sup>a</sup> Group differences were significant for receptive and expressive language abilities ( $ps < .001$ ) but not for age and SES ( $ps > .05$ ).



Table 2

*Means and Standard Deviations for Parent-Reported Problem Behaviors*

CBCL <sup>a</sup>	Twins (n = 62)			ASD (n =25)		
	<i>M</i>	<i>SD</i>	Range	<i>M</i>	<i>SD</i>	Range
Attention	52.85	6.57	50-78	64.84	12.36	51-100
Social	52.73	5.05	50-73	58.80	8.34	50-85
Thought	53.72	6.38	50-73	64.12	10.05	50-83
Anxious/Depressed	53.68	6.34	50-75	57.80	8.95	50-78
Withdrawal	52.17	4.20	50-73	59.84	8.80	50-79
SRS-2 <sup>b</sup>						
Social Cog	--	--	--	67.00	12.25	42-90
Social Aware	--	--	--	67.31	12.41	42-90
Social Comm	--	--	--	68.15	12.97	45-90
Motivation	--	--	--	63.23	12.65	46-90
RRBs	--	--	--	71.12	13.19	48-90
SRS-2 Total Score	--	--	--	69.65	12.50	46-90

<sup>a</sup> Child Behavior Checklist, Parent-report; T-score < 60 = Within normal limits; 60-69 = Borderline; >70 = Clinical. Group differences exist for all subscales of CBCL listed above; <sup>b</sup> Social Responsiveness Scale-2, Parent-report; Subscales: Social Cognition, Social Awareness, Social Communication, Motivation, Restricted/Repetitive Behaviors; T-score < 59 = Within normal limits; T-score: 60-65 = Mild range, mild to moderate social impairment, T-score: 66-75 = Moderate range, clinically significant impairment, T-score > 76 = Severe range, consistent with Autistic Disorder.

Table 3

*Partial Nonparametric Correlations Controlling for Age for Preschool and School-Age ToM*

	Memory	Memory	Contents	Contents
	Age 3	Age 4	Age 3	Age 4
School-age ToM Task	(n =33)	(n =38)	(n = 33)	(n = 38)
Contents False Belief	-.24	.01	-.12	-.09
Explicit False Belief	.13	.26	.23	.29 <sup>+</sup>
Belief Emotion	-.03	-.16	.23	-.31*
Real-Apparent Emotion	.18	-.33*	.27	-.37*
Understanding Sarcasm	-.08	.17	.22	-.22

<sup>+</sup>  $p = .069$ , \*  $p < .05$

Table 4

*Partial Nonparametric Correlations between Age and Performance on ToM Tasks*

	Age in months		
	Twins	ASD	Total Sample
	N = 62	N = 25	N = 86
ToM 1	.33**	.06	.15
ToM 2	.40**	-.22	.11
ToM 3	.05	.24	-.00
ToM 4	.34**	-.13	.14
ToM 5	.21 <sup>+</sup>	--	.14
Total ToM	.51**	-.02	.23*

*Note.* ToM 1: Contents false belief; ToM 2: Explicit false belief; ToM 3: Belief emotion; ToM 4: Real-apparent emotion; ToM 5: Understanding sarcasm. None of the children with ASD passed the understanding sarcasm task; therefore, a correlation could not be computed.

<sup>+</sup>  $p < .10$ , \*  $p < .05$ , \*\*  $p < .001$ .

Table 5

*Chi-square Analyses for Group by ToM Task Performance (N = 62) with Males Only.*

5a. Contents False Belief

	Fail	Pass	Total
Twin	6 (16%)	31 (84%)	37
ASD	15 (60%)	10 (40%)	25
Total	21 (34%)	41 (66%)	62

5b. Explicit False Belief

	Fail	Pass	Total
Twin	6 (16%)	31 (84%)	37
ASD	14 (56%)	11 (44%)	25
Total	20 (32%)	42 (68%)	62

5c. Belief Emotion

	Fail	Pass	Total
Twin	5 (14%)	32 (86%)	37
ASD	16 (64%)	9 (36%)	25
Total	21 (34%)	41 (66%)	62

5d. Real-Apparent Emotion

	Fail	Pass	Total
Twin	6 (16%)	31 (84%)	37
ASD	13 (52%)	12 (48%)	25
Total	19 (31%)	43 (69%)	62

5e. Understanding Sarcasm

	Fail	Pass	Total
Twin	28 (76%)	9 (24%)	37
ASD	25 (100%)	0 (0%)	25
Total	53 (85%)	9 (15%)	62

Group x Contents false belief task was significant,  $X^2(1, N = 62) = 12.77, p < .001$ .

Group x Explicit false belief task was significant,  $X^2(1, N = 62) = 10.81, p = .001$ .

Group x Belief emotion task,  $X^2(1, N = 62) = 16.98, p < .001$ .

Group x Real-apparent emotion task,  $X^2(1, N = 62) = 8.99, p = .003$ .

Group x Understanding sarcasm task,  $X^2(1, N = 62) = 7.11, p = .008$ .

Table 6

*Group Differences on Receptive Language Functioning After Controlling for Age (Males Only)*

	Twins (N = 29)	ASD (N = 25)		
	<i>M (SD)</i>	<i>M (SD)</i>	<i>F</i>	<i>p</i>
Receptive Lang	11.24 (1.64)	6.56 (4.26)	23.60	< .001

Table 7

*Hierarchical Regression Analysis Predicting Total ToM Score from Language Ability, Age, and Diagnostic Group (N = 46)*

Total ToM Score at Follow-Up				
Predictor	<i>B</i>	<i>SE B</i>	$\beta$	$\Delta R^2$
Step 1				.62***
Receptive Lang	.34	.04	.88***	
Age	.04	.01	.43***	
Step 2				.03 <sup>+</sup>
Diagnostic Group	-.66	.34	-.22 <sup>+</sup>	

*Note.* For diagnostic group classification purposes 0 = Twin, 1 = ASD.

Full model:  $F(3, 45) = 26.40$ ,  $p < .001$ , adjusted  $R^2 = .63$ .

<sup>+</sup>  $p = .059$ , \*\*\*  $p < .001$ .

Table 8

*MANCOVA Results for Groups Differences on CBCL Subscales for Males Only*

CBCL Subscale	Twins	ASD	<i>F</i>	<i>p</i>	$\eta^2$
	( <i>n</i> = 37)	( <i>n</i> = 25)			
	<i>M</i> ( <i>SD</i> )	<i>M</i> ( <i>SD</i> )			
Anxious/Depressed	54.41 (6.46)	57.80 (8.95)	3.80	.057	.048
Attention Problems	54.14 (6.79)	64.84 (12.36)	3.76	.058	.243
Social Problems	55.57 (6.88)	58.80 (8.34)	.49	.487	.044
Thought Problems	54.70 (5.46)	64.12 (10.05)	6.58	.013	.274
Withdrawn/Depressed	53.89 (5.29)	59.84 (8.80)	2.41	.126	.156



Table 9

*Nonparametric Correlations Between ToM Task Performance and Parent-Rated Problem Behaviors*

	Parent-Rated Problem Behaviors				
	Social Awareness	Social Cognition	Social Comm	Motivation	RRBs
ToM Task					
Contents False Belief	-.31	-.12	-.28	-.15	-.05
Explicit False Belief	-.34 <sup>+</sup>	-.46*	-.34 <sup>+</sup>	-.20	-.26
Belief Emotion	-.24	-.26	-.18	.01	-.35 <sup>+</sup>
Real-Apparent Emotion	-.38 <sup>+</sup>	-.54**	-.52**	-.30	-.44*
Understanding Sarcasm <sup>a</sup>	--	--	--	--	--

*Note.* Social Comm = Social Communication; RRBs = Restricted and Repetitive Behaviors.

<sup>a</sup> None of the children with ASD passed the understanding sarcasm task; therefore, correlations could not be computed.

<sup>+</sup>  $p < .10$ , \*  $p < .05$ , \*\*  $p < .01$ .

Table 10

*Falconer's Estimates of Heritability for ToM scores*

Variable	$r_{MZ}$	$r_{DZ}$	$h^2$	$z$
	N = 12	N = 28		
<b>Preschool Scores:</b>				
Contents Age 3	.32	.21	.22	.31
Contents Age 4	-.34	.11	0	-1.20
Memory Age 3	-.24	.11	0	-.91
Memory Age 4	.47*	.07	.47	1.13
<b>School-age scores:</b>				
	N = 18	N = 47		
Contents False Belief	.46*	.08	.46	1.40 <sup>+</sup>
Explicit False Belief	.29	.46***	0	-.67
Belief Emotion	-.21	.06	0	-.91
Real-Apparent Emotion	.23	.12	.22	.38
Understanding Sarcasm	.38*	-.02	.38	1.41 <sup>+</sup>

Note.  $h^2 = 2(r_{MZ} - r_{DZ})$ . If  $r_{MZ}$  is greater than 2 times larger than  $r_{DZ}$ ,  $h^2 = r_{MZ}$ .  
<sup>+</sup>  $p = .08$ , \*  $p < .05$ , \*\*  $p = .001$

Table 11

*Frequency of Genotypes by Diagnostic Group*

Genotype	Twins (N = 37)	ASD (N = 25)
5HTTLPR Risk Allele	12 (32%)	21 (84%)
5HTTLPR No Risk Allele	25 (68%)	4 (16%)
DRD4 Risk Allele	12 (32%)	8 (32%)
DRD4 No Risk Allele	22 (59%)	17 (68%)
Risk Allele in Both Genes	11 (30%)	8 (32%)
Risk Allele in Only One Gene	16 (43%)	13 (52%)
No Risk Allele in Either Gene	9 (24%)	4 (16%)

Table 12

*Chi-square Analyses of 5-HTTLPR Risk Alleles by ToM Performance.*

## 12a. Contents False Belief (Twin sample; N = 61)

	Fail	Pass	Total	X <sup>2</sup>	p
No Risk Allele	7 (58.3%)	5 (41.7%)	12	5.15	.023
1 or 2 Risk Alleles	12 (24.5%)	37 (75.5%)	49		
Total	19 (31.1%)	42 (68.9%)	61		

## 12b. Belief Emotion (ASD sample; N = 25)

	Fail	Pass	Total	X <sup>2</sup>	p
No Risk Allele	1 (25%)	3 (75%)	4	3.14	.076
1 or 2 Risk Alleles	15 (71%)	6 (29%)	21		
Total	16 (64%)	9 (36%)	25		

## 12c. Contents False Belief (Females only; N =33)

	Fail	Pass	Total	X <sup>2</sup>	p
No Risk Allele	5 (71%)	2 (29%)	7	5.80	.016
1 or 2 Risk Alleles	6 (23%)	20 (77%)	26		
Total	11 (33%)	22 (67%)	33		

## 12d. Belief Emotion (Male only sample; N = 62)

	Fail	Pass	Total	X <sup>2</sup>	p
No Risk Allele	2 (10%)	19 (90%)	21	4.40	.036
1 or 2 Risk Alleles	14 (34%)	27 (66%)	41		
Total	16 (26%)	46 (74%)	62		

Table 13

*Chi-square Analyses of DRD4 Risk Alleles by ToM Performance.*

## 13a. DRD4 Risk Allele by Explicit False Belief (Twin sample; N = 61)

	Fail	Pass	Total	$X^2$	$p$
No Risk Allele	4 (33%)	8 (67%)	12	3.04	.081
1 or 2 Risk Alleles	30 (61%)	19 (39%)	49		
Total	34 (56%)	27 (44%)	61		

## 13b. DRD4 Risk Allele by Belief Emotion (Twin sample; N = 61)

	Fail	Pass	Total	$X^2$	$p$
No Risk Allele	3 (30%)	7 (70%)	10	3.21	.073
1 or 2 Risk Alleles	31 (61%)	20 (39%)	51		
Total	34 (56%)	27 (44%)	61		

## 13c. DRD4 Risk Allele by Contents False Belief (ASD sample; N = 25)

	Fail	Pass	Total	$X^2$	$p$
No Risk Allele	7 (41%)	10 (59%)	17	7.84	.005
1 or 2 Risk Alleles	8 (100%)	0 (0%)	8		
Total	15 (60%)	10 (40%)	25		

## 13d. DRD4 Risk Allele by Contents False Belief (Males only; N = 61)

	Fail	Pass	Total	$X^2$	$p$
No Risk Allele	10 (48%)	11 (52%)	21	3.70	.055
1 or 2 Risk Alleles	29 (73%)	11 (27%)	40		
Total	39 (64%)	22 (46%)	61		

Table 14

*Chi-square Analyses of Trichotomous Risk Alleles by ToM Performance.*

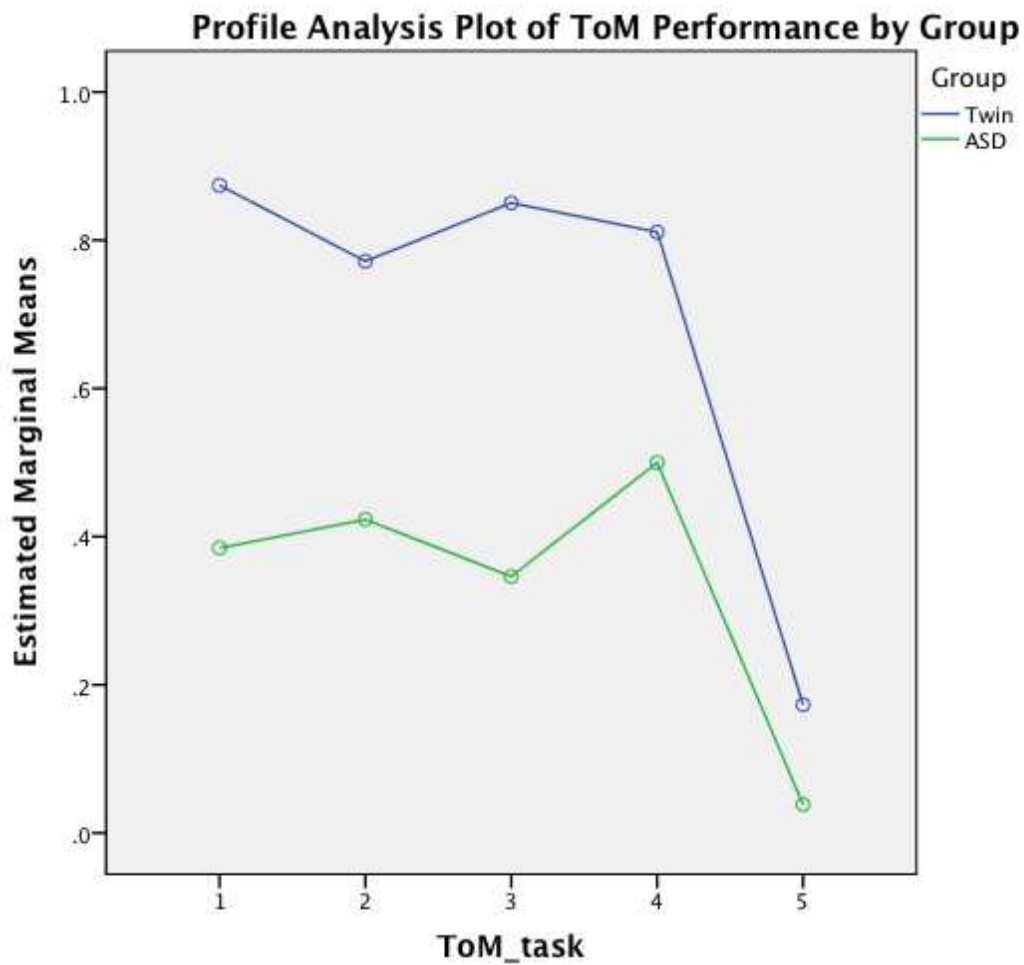
14a. Trichotomous Risk Alleles by Contents False Belief (ASD sample; N =25)

	Fail	Pass	Total	$X^2$	$p$
No Risk Allele	2 (50%)	2 (50%)	4	8.01	.018
1 or 2 Risk Alleles in Either Gene	5 (38%)	8 (62%)	13		
1 or 2 Risk Alleles in Both Genes	8 (100%)	0 (0%)	8		
Total	15 (60%)	10 (40%)	25		

Table 15

ANOVA Results for *Genotype on ToM Performance (N = 86)*

Gene	ToM Performance	
	<i>F</i>	<i>p</i>
5-HTTLPR	.79	.376
DRD4	.40	.529
5-HTTLPR & DRD4	.59	.557



*Figure 1.* Profile analysis of group differences in task performance.

*Note:* Task 1: Contents false belief; Task 2: Explicit false belief; Task 3: Belief-emotion; Task 4:

Real-apparent emotion; and Task 5: Understanding sarcasm.



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## APPENDICES

## Appendix A

**Demographic Information Sheet**

Date \_\_\_\_\_ ID Number \_\_\_\_\_

Age of Children \_\_\_\_\_ DOB \_\_\_\_\_

Your relationship to the children (Mother or Father; please note if adoptive parent): \_\_\_\_\_

Your Age: \_\_\_\_\_

Marital Status:

Single, never married \_\_\_\_\_ Married \_\_\_\_\_ Divorced/Separated \_\_\_\_\_

Widowed \_\_\_\_\_ Living with a significant other \_\_\_\_\_

Approximate Total Family Income:

\_\_\_\_\_ Less than \$5,000 \_\_\_\_\_ \$20,000-25,000 \_\_\_\_\_ \$40,000-45,000

\_\_\_\_\_ \$5,000-10,000 \_\_\_\_\_ \$25,000-30,000 \_\_\_\_\_ \$45,000-50,000

\_\_\_\_\_ \$10,000-15,000 \_\_\_\_\_ \$30,000-35,000 \_\_\_\_\_ \$50,000-55,000

\_\_\_\_\_ \$15,000-20,000 \_\_\_\_\_ \$35,000-40,000 \_\_\_\_\_ Over \$55,000

Race of Child's Parents: Mother \_\_\_\_\_ Father \_\_\_\_\_

Race of Children in Study: \_\_\_\_\_

	Occupation	Finished High School?	Attended College?	Years of College (undergraduate & graduate)	College Degrees (AA, BA, etc.)
Self		Yes No If yes, please continue -->	Yes No If yes, please continue -->		
Spouse or Significant Other <b>IF</b> living in home with children		Yes No If yes, please continue -->	Yes No If yes, please continue -->		

Siblings of Children in the Study

Please start the list with the **OLDEST** sibling and move to the **YOUNGEST**.  
(Please do not include the children in the study)

	First Sibling	Second Sibling	Third Sibling	Fourth Sibling	Fifth Sibling
Birth Date					
Circle any that may apply	Half-sibling Step-sibling Adopted	Half-sibling Step-sibling Adopted	Half-sibling Step-sibling Adopted	Half-sibling Step-sibling Adopted	Half-sibling Step-sibling Adopted

Please list everyone in your household and their relation (e.g., father, grandmother, etc.) to the children in the study. (First names only, example: Ben – grandfather)

We are interested in whether changes in the family, such as divorce or remarriage, affect your children's behaviors. Therefore, the following item will help us to understand when these things may have happened in your family and how they may influence your children.

If applicable, please indicate if you have ever been divorced or remarried and the year this occurred.

Not applicable\_\_\_\_\_

Divorced\_\_\_\_\_

Year\_\_\_\_\_

Year\_\_\_\_\_

Remarried\_\_\_\_\_

Year\_\_\_\_\_

Year\_\_\_\_\_

## Appendix B

**Theory of Mind Protocol (Age 3 and 4 Testing)**Theory of Mind I (Playdoh container filled with crayons):

Show the child the box and say: **“What do you think is in this Playdoh container?”** Then show the child the contents of the box. **“What are these?”** (Name the items if the child does not know or gives an incorrect response), then say: **“When I first showed you this Playdoh container before I opened it, what did you think was in it?”** Let the child respond, and then say: **“If I show this container to (name of child’s twin or brother/sister), what would s/he say is in it?”**

Theory of Mind II (Crayon box filled with blocks):

Show the child the box and say: **“What do you think is in this crayon box?”** Then show the child the contents of the box. **“What are these?”** (Name the items if the child does not know or gives an incorrect response), then say: **“When I first showed you this crayon box before I opened it, what did you think was in it?”** Let the child respond, and then say: **“If I show this box to (name of child’s twin or brother/sister), what would s/he say is in it?”**

## Appendix C

**Theory of Mind Protocol (Current Study)**Task 1: Contents False Belief

Tester shows child a clearly identifiable Playdoh container filled with crayons inside the closed container and says, “**Here’s a Playdoh container. What do you think is inside this Playdoh container?**” The Playdoh container is then opened: “**Let’s see...it’s really Q-tips inside!**” The container is closed and the tester then asks, “**Okay, what is in the Playdoh container?**”

Next, a toy figure of a boy is produced, and the tester says, “**Peter has never ever seen inside this Playdoh container. Now here comes Peter. So, what does Peter think is in the box? Playdoh or Q-tips? Did Peter see inside this box? If I were to ask (name of co-twin or a familiar peer at school) what was inside this container, what would s/he say?**”

Task 2: Explicit False Belief

The child is shown a toy figure of a boy and a sheet of paper with a backpack and a closet drawn on it. “**Here’s Scott. Scott wants to find his mittens. His mittens might be in his backpack or they might be in the closet. Really, Scott’s mittens are in his backpack. But Scott thinks his mittens are in the closet.**”

The tester then asks the child, “**So, where will Scott look for his mittens? In his backpack or in the closet? Where are Scott’s mittens really? In his backpack or in the closet?**”

Task 3: Belief-Emotion

The child is shown a toy figure of a boy and a clearly identifiable individual-size Cheerios box with rocks inside the closed box. “**Here is a Cheerios box, and here is Teddy.**

The tester pretends to be Teddy and says, **“Teddy says, ‘Oh good, because I love Cheerios. Cheerios are my favorite snack. Now I’ll go play.’”** The tester then puts Teddy out of view of the child.

Next, the tester opens the Cheerios box, and the contents are shown to the child: **“Let’s see...there are really rocks inside and no Cheerios! There’s nothing but rocks.”** The Cheerios box is closed, and the tester says, **“Okay, what is Teddy’s favorite snack?”**

The tester brings Teddy back out, and the tester says, **“Teddy has never ever seen inside this box. Now here comes Teddy. Teddy’s back, and it’s snack time. Let’s give Teddy this box. So, how does Teddy feel when he gets this box? Happy or sad?”** The tester opens the Cheerios box and lets the toy figure look inside and says, **“How does Teddy feel after he looks inside the box? Happy or sad?”**

#### Task 4: Real-Apparent Emotion

A child is shown a sheet of paper with three faces drawn on it—a happy, a neutral, and a sad face—to check that the child knows these emotional expressions. Then that paper is put aside, and the task begins with the child being shown a cardboard cutout figure of a boy drawn from the back so that the boy’s facial expression cannot be seen. The tester then says, **“This story is about a boy. I’m going to ask you about how to boy really feels inside and how he looks on his face. He might really feel one way inside but look a different way on his face. Or, he might really feel the same way inside as he looks on his face. I want you to tell me how he really feels inside and how he looks on his face.”**

Then, the tester reads the following story: **“This story is about Matt. Matt’s friends were playing together and telling jokes. One of the older children, Rosie, told a mean joke about Matt and everyone laughed. Everyone thought it was very funny, but not Matt. But,**

**Matt didn't want the other children to see how he felt about the joke, because they would call him a baby. So, Matt tried to hide how he felt.**" The tester then asks the following questions: **"What did the other children do when Rosie told a mean joke about Matt? In the story, what would the other children do if they knew how Matt felt?"**

Next, the tester shows the child the emotion pictures and asks, **"So, how did Matt really feel when everyone laughed? Did he feel happy, sad, or so-so? How did Matt try to look on his face when everyone laughed? Did he look happy, sad, or so-so?"**

#### Task 5: Understanding Sarcasm

The tester shows the child a colored line drawing of the back of a boy's and a girl's head, raindrops, and a wet cake and other food on a picnic blanket.

Next, the tester reads the following story to the child without any special intonation or emphasis: **"The girl and boy are going on a picnic. It is the boy's idea. He says it will be a lovely sunny day. But when they get the food out, big storm clouds come. It rains and the food gets all wet. The girl says: 'It's a lovely day for a picnic.'"**

The tester asks the child the following questions: **"Is it true, what the girl said? Why did the girl say 'It's a lovely day for a picnic'? Was the girl happy about the rain?"**



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