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Behaviour and Language Development in Infants at Risk for ASD: The Role of Early Attention Preferences and Early Language Development

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Behaviour and Language Development in Infants at Risk for ASD: The Role of Early Attention Preferences and Early Language Development

by

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Abstract

The diagnostic criteria for Autism Spectrum Disorder (ASD) cluster around communicative, linguistic, and social deficits (DSM-IV; American Psychiatric Association, 1994). Identification of impairments associated with ASD relies on children's explicit behaviours, which only emerge reliably after the first year of life (De Giacomo & Fombonne, 1998). Thus, data on earlier social attention, language development, and ASD behaviours in children with ASD are limited. In this dissertation, I examine how early perceptual biases and ASD-like behaviours relate to language skills in a high-risk cohort of infant siblings of children with ASD (SIBS-A) and a low-risk cohort of infants siblings of typically developing children (SIBS-TD). Chapter 2 investigates whether biases to infant-directed (ID) speech and faces differ between SIBS-A and infant siblings of typically developing children (SIBS-TD), and to what extent early differences may be predictive of language skills and risk group membership. In this study, we found that both infant groups preferred ID to adult-directed (AD) speech and preferred faces to checkerboards; however, the magnitude of the preference was smaller in SIBS-A. SIBS-TD demonstrated higher expressive vocabulary than SIBS-A at 18 months. Vocabulary size correlated with early speech preferences, suggesting that a preference for ID speech early in development may facilitate later expressive language. Finally, infants' preference for faces contributed to determining group membership. Chapter 3 explores vocabulary development and ASD-related behaviours, and whether these skills are associated with one another and differ between high-risk and typically developing infants. Results from this study demonstrated that SIBS-A exhibited lower expressive vocabulary at 12 and 18 months as compared with SIBS-TD. SIBS-A

additionally demonstrated significantly more ASD-like behaviours that were also more severe in nature at 18 months. Moreover, expressive and receptive vocabularies were significantly correlated with ASD behaviours. The findings of these studies suggest that infants at heightened risk of ASD differ from SIBS-TD in their preferences for ID speech and faces, as well as vocabulary skills and ASD behaviours early in childhood. These early linguistic and behavioural differences may underlie deficits in later language development and social communication and have important implications for research examining early detection measures for ASD.

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For my family, whose faith and encouragement carried me.

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Chapter One: **Introduction**

Autism Spectrum Disorder (ASD) is a pervasive developmental disorder (PDD; American Psychiatric Association, 1994) that has been shown to affect as many as 1 in 91 children and adolescents in the U.S. (Kogen et al., 2009) and 1 in 161 reported in Canada (Fombonne, 2003). Autism/ASD (hereafter, ASD) is now recognized as the most common neurological disorder affecting children and one of the most common developmental disabilities (Fombonne, 2003). The diagnostic criteria for ASD cluster around communicative, linguistic, and social deficits (DSM-IV; American Psychiatric Association, 1994) that only emerge reliably after the first year of life (DeGiacomo & Fombonne, 1998). That is, impairments in these explicit productive abilities can only be identified once the child is at the developmental level in which they can demonstrate atypical patterns of behaviour. As a result, ASD is typically diagnosed later, around 3 to 5 years (Mandell, Novake, & Zubritsky, 2005; Ozonoff et al., 2009).

While the defining characteristics of ASD often present in the second to third year of life, studies have found that some behaviours consistent with ASD may be present even earlier (Short & Schopler, 1988; Stone, Hoffman, Lewis, & Ousley, 1994; Sullivan, Kelso, & Stewart, 1990). Several retrospective studies of early home videos have revealed behaviours indicative of ASD in children later diagnosed compared to those of typically developing children (Adrien et al., 1991; Adrien et al., 1992; Baranek, 1999; Bernabei, Camaioni, & Levi, 1998; Maestro, Casella, Milone, Muratori, & Palacio-Espasa, 1999; Mars, Mauk, & Dowrick, 1998; Massie, 1978; Osterling & Dawson, 1994; Rosenthal, Massie, & Wulff, 1980; Werner, Dawson, Osterling, & Nuhad, 2000; Zakien, Malvy, Desombre, Roux, & Lenoir, 2000). During the first year of life, children with ASD have been observed to demonstrate reduced social interaction, lack of social smiling and flat facial expression (Adrien et al., 1992), failure to orient to name (Bernabei et al., 1998; Osterling & Dawson, 1994; Maestro et al, 1999; Mars et al., 1998; Zakien et al., 2000), lack of pointing or showing (Osterling & Dawson, 1994; Mars et al., 1998), decreased orienting to faces (Bernabei et al., 1998; Osterling & Dawson, 1994; Maestro et al, 1999; Mars et al., 1998; Zakien et al., 2000), and lack of spontaneous imitation (Mars et al., 1998). Together these studies suggest the potential for identifying children at risk for a later ASD diagnosis early in development. Early identification and early intervention have been recognized as critical aspects of the management and treatment of children with ASD (American Academy of Pediatrics Committee on Children with Disabilities, 2001; National Research Council, 2001), and more recently, the American Academy of Pediatrics has recommended universal screening for ASD at both 18 and 24 months of age (Johnson, Myers, & Council on Children with Disabilities, 2007).

Significant improvements in language, cognitive abilities, and social skills have been reported for young children with ASD who participated in specialized early intervention (Bondy & Frost, 1995; Harris, Handleman, Gordon, Kristoff. & Fuentes, 1991; McEachin, Smith, & Lovaas, 1993; Rogers & Lewis, 1989; Strain, Hoyson, & Jamieson, 1985), especially when an intervention begins before five years of age (Fenske, Zalenski, Krantz, & McClannahan, 1985). Moreover, providing intervention by the age of two years ultimately lowers the cost of care and results in higher gains in a dependency-free life for adults with ASD (Motiwala, Gupta, Lilly, Ungar, & Coyte, 2006). However, despite practice guidelines recommending that clinicians monitor for early signs and use ASD-specific screening tools, clinical detection of ASD is still difficult prior to 24 months. There are currently a limited number of available diagnostic instruments for children younger than 24 months (Luyster et al, 2009; Zwaigenbaum, et al., 2005), and those that do assess early ASD in infants and toddler are still in development. Thus, there is a need for identifying potential early markers for atypical development.

One of the defining characteristics of ASD is a moderate to profound difficulty in the development and use of language. Indeed, this is one of the first concerns that parents of children with ASD have about their child's development (Rutter, 1970). Given the critical importance of early language development for later prognosis, a better understanding of developmental factors that underlie, facilitate, and predict language acquisition in individuals with ASD would shed light on the nature of this disorder and allow for the refinement of targeted early interventions. In this dissertation, I examine whether atypical processing of speech and ASD-like behaviours can be observed earlier in development. To this end, I examine whether infants who are at risk for ASD because they have an older sibling diagnosed with ASD (SIBS-A) process infant-directed (ID) and adult-directed (AD) speech differently than infant siblings of typically developing children (SIBS-TD) and whether this is related to later language abilities – specifically receptive and expressive vocabulary. I further explore whether the type of visual input (faces or checkerboards) impacts this relationship. I then examine whether a relationship exists between early atypical social behaviours and vocabulary development. Together these findings will help to further our understanding of the development of defining characteristics of ASD and may potentially help to identify atypical development prior to the age of 3 years.

1.1 Language and Social Communication in Children with ASD

Approximately half of the ASD population fails to develop productive language and when language does develop individuals with ASD often fail to use it functionally (Volkmar, 1991). Many of the children with ASD who do not speak at all also show difficulty in understanding language. Some children with ASD are reported to produce one-word utterances and then completely stop speaking (Lord, Shulman, & DiLavore, 2004), while others speak, but often by only repeating precisely what they have heard (echolalia; e.g., Cantwell, Baker, & Rutter, 1978;

Kanner, 1946). Other individuals with ASD have a restricted use of language using it only for the primary objective of obtaining a desired item (see Tager-Flusberg, 1996; Volkmar, 1991). These studies demonstrate the heterogeneity of language abilities in individuals with ASD and further demonstrate the importance of examining early linguistic abilities in order to potentially identify early indicators of atypical language development.

Children with ASD who are able to use language by age 5 years have a much greater chance of achieving levels of functionality than those who do not possess this skill (Johnson, Dziurawiec, Ellis, & Morton, 1999). Recent research has consistently found that early language skills (i.e., meaningful speech by 5 to 6 years of age) in children with ASD are an important predictor for later outcome (Luyster, Kadlec, Carter, & Tager-Flusberg, 2008). Indeed, these skills have been associated with later academic achievement and social competence (Howlin, Mawhood, & Rutter, 2000; Sigman & Ruskin, 1999; Venter, Lord, & Schopler, 1992). Furthermore, in typically developing children, vocabulary size at 25 months has been shown to predict cognitive and language skills in later childhood (Marchman & Fernald, 2008). Vocabulary knowledge is assumed to be a core aspect of general intelligence, and performance on standardized vocabulary tests is highly correlated with performance on more general tests of intelligence (Dunn & Dunn, 1981; Huttenlocher, Haight, Bryk, Seltzer, & Lyons, 1991).

In children with ASD, language skills at age 4 years have been shown to predict language development at age 10 to 13 (Sigman & Ruskin, 1999). In a more recent study, Charman and colleagues (Charman, Drew, Baird, & Baird, 2003) used data from a single administration of a parent report of vocabulary development (MacArthur–Bates Communicative Development Inventory; CDI; Fenson et al., 1993, 1994) from parents of children with ASD and found that, although the children as a whole demonstrated severe language delays, there was considerable variability in the size of their vocabularies. They also found that the children's actions on objects were relatively advanced compared with their ability to imitate and to initiate joint attention gestures. This particular study highlighted the amount of variability in the vocabularies in children with ASD and the prelinguistic skills that appear to be associated with language development in young children with ASD. Smith, Mirenda, and Zaidman-Zait (2007) found significant relations between vocabulary development and both verbal imitation skills and joint attention gestures in children with ASD. Their results confirmed the heterogeneity in language development in young children with ASD and, consistent with other reports, confirms that vocabulary skills are predictive of aspects of social-communicative development.

Several social communication skills have also been found to be predictive of language skill gain and outcome in children with ASD, including imitation (Charman et al., 2005; Toth, Munson, Meltzoff, & Dawson, 2006), responding to pointing (Sigman & Ruskin, 1999), the frequency of joint attention interactions (Charman et al., 2005), and the duration of those interactions (Adamson, Bakeman, & Deckner, 2004). Research on joint attention and ASD has illustrated how social communication skills can also be predictive of later language skills. Joint attention has been characterized as shared attention between social partners in relation to objects or events, which typically emerges by 9 to12 months of age (Adamson & Bakeman, 1985, 1991; Adamson & Chance, 1998; Brooks & Meltzoff, 2002; Bruner, 1983; Butterworth & Jarrett, 1991; Carpenter, Nagell, & Tomasello, Butterworth, & Moore, 1998). Joint attention skills are commonly believed to be a pre-requisite for acquiring expressive language (Kasari, Paparella, Freeman, & Jahromi, 2008).

Children with ASD who demonstrate joint attention behaviours, including imitation, social referencing, communicative gestures, and early productive language (Tomasello, 1995) have also been shown to demonstrate better-developed language skills (Dawson et al., 2004). In a study examining the contribution of early joint attention, imitation, and toy play to early language ability in young children with ASD, it was found that joint attention and immediate imitation contributed to language ability at the outset, while toy play and imitation were predictive of rate of development of communication skills over the next few years (Toth et al., 2006). Of particular importance, toy play and imitation abilities often involve shared attention and an active interest in and exploration of the environment, as well as higher level cognitive skills that are important for ongoing development of language and social communication abilities.

Joint attention occurs in a social context and presupposes social attention, toy play, imitation, and other social communication skills that require the child to actively attend to their immediate environment, observe the events and actions taking place, and subsequently reproduce these events and socially mediated actions at a later time. The ability to demonstrate these skills requires, amongst other important skills, an active interest in people and/or things. Thus, activities such as joint attention, imitation, and social toy play that contribute to language acquisition (Newland, Roggman, & Boyce, 2001; Toth et al., 2006), require children to engage in and maintain a social interaction, suggesting that the attentiveness to another person may support the development of language skills (Kasari et al., 2008). Social attention or social orienting (i.e., orienting to naturally occurring social stimuli in one's environment; Dawson, Meltzoff, Osterling, Rinaldi, & Brown, 1998) is important to later developing skills (Dawson et al., 2004). From birth, typically developing infants preferentially attend to social information from their own species, such as the sounds, movements, and features of the human face (Maurer & Salapatek, 1976; Morton & Johnson, 1991). Social cognition, which develops later in childhood, likely depends on this very early tendency to attend to faces (Rochat & Striano, 1999) and speech (Schultz & Vouloumanos, 2010). It has been argued that a failure to orient to social stimuli represents one of the earliest and most basic social impairments in ASD, and may contribute to the later-emerging social and communicative impairments (Dawson et al, 1998; Mundy & Neal, 2001). A failure to engage in social attention or orienting may impact the developmental pathway of young children with ASD by depriving them of appropriate social stimulation (Mundy & Neal, 2001). Thus, it is important to examine the social information very young children are attending to prior to a diagnosis of ASD (e.g., SIBS-A), and how it impacts later social and communicative development. In particular, there may be important potential differences between attention to social information in typically developing and high-risk infants that may underlie social and communicative behaviours that are characteristic of ASD. In fact, it is possible that these early measures of infant's attention to social information, such as speech and faces, may prove to be sensitive predictors of language development. As such, we examine attention to social information related to speech and faces in high and low risk infants at 6, 8, 12, and 18 months, and how this might predict language skills at 18 months.

1.2 Speech Preferences in Typically Developing Infants

From birth, infants demonstrate preferences for the communicative signals of their species, privileging natural speech over filtered speech (Spence & DeCasper, 1987) and synthetic sine-wave analogues of speech (Vouloumanos & Werker, 2004). This basic preference may form the foundation for attending to and processing linguistic information (Schultz & Vouloumanos, 2010). Over the first year of life infants are discriminating speech sounds (e.g., Eimas, Siqueland, Jusczyk & Vigorito, 1971; Werker & Tees, 1984), segmenting words from the speech stream (e.g., Saffran, Aslin, & Newport, 1996), learning about sound sequences (e.g., Jusczyk, Luce, & Charles-Luce, 1994), and recognizing highly familiar words (Bergelson & Swingley, 2012). In typically developing children, language development may be supported by their social interest in speech (Cooper & Aslin, 1990; Fernald, 1985; Glenn & Cunningham, 1983; Pegg, Werker & McLeod, 1992; Werker & McLeod, 1989). Typically developing infants demonstrate a preference for attending to ID speech over AD speech from the first days of life (Cooper & Aslin; 1990; Fernald, 1985). ID speech is a unique type of speech characterized by exaggerated intonation that is used when caretakers address infants (Fernald & Simon, 1984). Utterances directed towards infants generally have a higher pitch, a greater pitch range, and the overall length is generally shorter than AD speech (Fernald & Mazzie, 1991). In addition to presenting with a slower rate of speech, ID speech is more repetitive and more likely to have longer pauses. The significance of ID speech in language acquisition has been widely researched (Jusczyk, 1997; Kuhl et al., 1997; Kemler-Nelson, Hirsh-Pasek, Jusczyk, & Cassidy, 1989; Snow, 1977) and it is thought to serve several functions that facilitate in the development of fundamental abilities necessary for language learning over the course of infant development (Fernald & Mazzie, 1991; Hayashi, Tamekawa, & Kiritani, 2001).

According to Fernald (1992), the function of ID speech changes depending on the stage of language development. Early on, infants are biologically predisposed to respond differentially to ID speech. Once infants become more perceptually proficient, ID speech increases its role for modulating and regulating infants' attention. As infants begin to integrate the vocal activity of their caregivers with a range of other sensory input (facial expressions, joint attention, eye gaze), they learn about socioemotional meaning (Fernald, 1992). Compared with AD speech, the enhanced acoustic features of ID speech are more effective at engaging and maintaining attention, modulating arousal, communicating affect, and facilitating learning and information processing. These highly engaging features of ID speech could, in turn, affect language learning.

ID speech may also enable infants to more efficiently discover or use information in speech, even in situations when it conveys the same information (e.g., statistical information) as AD speech. In particular, ID speech may facilitate language acquisition by enabling faster or more efficient learning. Thiessen, Hill and Saffran (2005) presented a group of 6 to 8 month old infants with a set of nonsense words spoken with the intonation contours that are characteristic of ID speech, while another group of same aged infants heard the same sentences spoken with intonation contours characteristic of AD speech. Infants successfully distinguished words from syllable sequences spanning word boundaries after exposure to ID speech but not after hearing AD speech. The authors proposed that the exaggerated prosody of ID speech facilitates in the acquisition of purely statistical information. They further suggested that the structure of ID speech may highlight linguistic patterns, thereby facilitating language acquisition. For example, a vital component in the process of language acquisition is the language learner's ability to parse out the words embedded in the speech stream. In order to do so, infants must rely on linguistic cues. One such implicit, yet salient indication of word beginnings and endings is the frequency of various syllable combinations occurring together, known as the statistical structure of speech (Saffran, Aslin, & Newport, 1997). Researchers speculate that aspects of ID speech help infants use these statistical cues to segment speech (Fernald & Simon, 1984; Thiessen et al., 2005). For instance, the longer pauses at phrase boundaries in ID speech may make it easier for infants to locate word boundaries allowing infants to track the distributional properties at word edges.

Together the findings from studies exploring speech processing in typically developing children demonstrate that the exaggerated prosodic cues that characterize ID speech facilitate language acquisition across early development. Given this, it is possible that deficits in language and social development may be related to differences in attention to ID speech. Interestingly, we do not have data on early speech processing in children with ASD. Retrospective studies focus on observable language abilities in ASD that are often atypical in nature and/or delayed. With the range of language abilities in this population, it is possible that one contributing factor is the lack of a preference for ID speech. Indeed, perhaps some of the symptoms of ASD are associated with an underlying deficit in attending to information generated by one's species (including visual and auditory information), which subsequently may have cascading effects on development.

Support for this possibility can be found in studies with typically developing infants. That is, a number of relationships between this early speech processing and later language achievement have been identified in typically developing infants. For example, in a study examining infant speech discrimination, Tsao, Liu, and Kuhl (2004) demonstrated significant correlations between speech perception at 6 months of age and later language (word understanding, word production, phrase understanding). Based on their finding that speech perception performance at 6 months predicts language at 2 years, the authors concluded that phonetic perception may play an important role in language acquisition. In a longitudinal study, Bernhardt, Kemp, and Werker (2007) found that toddlers' performance on a word-object association task was related to the children's later language skills. Specifically, they found that performance at 17 or 20 months was significantly related to scores on some standardized tests of language comprehension and production up to two and a half years later. Further research has shown links between speech perception and language in clinical populations, including studies demonstrating deficits in phonetic discrimination skills amongst children diagnosed with reading disorders (Reed, 1989; Manis et al., 1997), dyslexia (Godfrey, Syrdal-Lasky, Millay & Knox, 1981), learning disabilities (Kraus et al., 1996; Bradlow et al., 1999), and specific language impairment (Leonard, McGregor & Allen, 1992; Tallal & Stark, 1981; Werker & Tees, 1987) compared to control groups. Thus, understanding how speech is processed in the first year of life may help us to identify whether linguistic cues, such as those observed in ID speech, contribute to language acquisition, and whether children at risk for developing ASD demonstrate atypical preferences.

1.3 Attention to Faces

1.3.1 Attention to Faces in Typically Developing Infants

While attention to ID speech has an important role in social development, interaction, and communication, infants can also learn about the social world from the faces of others. Faces are salient and preferred stimuli for TD infants in early development (e.g., Bushnell, Sai, & Mullen, 1989) and may support social development and communication. By attending to relevant social information, such as human faces, infants learn about the significance of facial affect and identity, and begin to pick up on social cues, such as eye gaze (Cohn & Tronick, 1983; Corkum & Moore, 1998; Pascalis, De Haan, Nelson, & De Schonen, 1998; Scaife & Bruner, 1975; Tronick, 1989). Recognition of individual faces is an integral part of interpersonal interactions and successful functioning within a social group, and is fundamental to the development of social communication.

In order to extract socially and emotionally relevant information from faces, infants must first attend to them. Even in the first weeks of life, newborn infants demonstrate a preference to attend to faces and face-like stimuli over non-face-like stimuli (e.g., geometric shapes, lines, scrambled faces, shading/colour manipulated faces; Macchi Cassia, Turati, & Simion, 2004; Farroni et al., 2005; Johnson et al., 1991; Simion, Macchi Cassia, Turati, & Valenza, 2001). Faces provide relevant information for the kind of social inferences that children are beginning to make over their first year (e.g., Carpenter et al., 1998; Corkum & Moore, 1998). Many researchers have speculated that the skills necessary to process and use social and emotional information gleaned from faces, and the ability to appropriately respond to such information, may have been critical for the evolution of social communication (Andrew, 1963; Ekman, 1992; Ekman & Oster, 1979).

1.3.2 Attention to Faces in ASD

Many developmental models of ASD, which focus on understanding the precursors of the social difficulties, suggest that there are differences in attention to socially relevant information, and that contribute to social impairments (e.g., Dawson et al., 2005; Gepner, de Gelder, & de Schonen, 1996; Schultz, 2005). Characteristic of ASD is an unusual pattern of eye contact, which may reflect the more prevalent deficits in communication and social interaction observed in this population. Behavioural studies with individuals with ASD have demonstrated that the use of gaze cues in social contexts, such as joint attention (Baron-Cohen, 1989; Leekam, Hunnisett, & Moore, 1998; Mundy, Sigman, Ungerer, & Sherman, 1986) or in inferring mental states (Baron-Cohen, Baldwin, & Crowson, 1997) is more challenging than for neuro-typical individuals. Falck-Ytter, Fernell, Gillberg and von Hofsten (2010) found that when children with ASD were looking at other people's faces, those who are better at socio-emotional behaviours than non-verbal communication tended to look more at the eyes, while those with the opposite profile tended toward looking more at the mouth. In a second study, they found that a similar pattern was observed for inverted faces for the mouth area, suggesting that information from this area is perceived on a featural (as opposed to a more gestalt or holistic) basis. When the children were shown a person performing manual actions they found that the children from the first study who looked more at the eyes, tended to look most at the face of the actor, while those who looked more at the mouth tended to look at the action itself (hand⁄objects). This finding was observed in both ASD and typical development. Based on these findings, the authors suggest that the underlying neurosystems responsible for socio-emotional and communicative symptoms of ASD differ in the manner that they affect social perception in ASD, and that this differentiation cannot be explained by views of ASD building on one single cognitive explanation (Baron-Cohen, Knickmeyer & Belmonte, 2005). That is, the socio-emotional versus non-verbal communication skills are separable. Taken together, studies examining attention to faces in an ASD population suggest that this social attention to faces is an important skill that contributes to social and communication skills.

Given the importance of faces in conveying social and emotional information starting soon after birth, interest in how infants attend to faces is partly fuelled by several developmental conditions, such as ASD. Research suggests that there are a number of attentional atypicalities observed in individuals with ASD (e.g., Burack, 1994; Courchesne et al., 1994; Plaisted, O'Riordan, & Baron-Cohen, 1998; Townsend & Courchesne, 1994; Wainwright-Sharp & Bryson, 1993). Several studies suggest that children with ASD demonstrate impairments in face recognition (Dawson et al., 2002; Klin et al., 1999; Marcus & Nelson, 2001), deficits in understanding facial information and using it in a socially beneficial way (Baron-Cohen, 1994; Hobson, 1989), and decreased attention to faces (Osterling, Dawson & Munson, 2002) relative to controls. Thus, it is important to understand the role of attention to faces in the development of social communication and language acquisition.

While there is a paucity of research on early speech processing, there are fewer studies that provide insight into how children with ASD processed and attended to faces over the first year of life. It has been shown that SIBS-A demonstrate a reduced sensitivity to gaze in social contexts, suggesting that the early developmental course in infants at heightened risk for ASD may contrast with that in typical development (Elsabbagh et al., 2009). This finding supplements work in the infant sibling literature showing that SIBS-A are less able to integrate social cues, including gaze, relevant in the context of joint attention by relying on eye gaze to direct attention (Presmanes, Walden, Stone, & Yoder, (2007). There are numerous developmental models of ASD that suggest that attention to socially relevant information plays a significant role in the development of social and communication skills (e.g., Dawson et al., 2005; Gepner et al., 1996; Schultz, 2005).

Research has suggested that children with ASD have problems with social and emotional information processing, as well as an absence of social interest in communication, particularly speech (Baron-Cohen, Tager-Flusberg & Cohen, 1993). For children with ASD, a lack of interest or engagement in social activities early in development, such as listening to speech sounds and looking at a speaker's face, could contribute to social and language impairments characteristic of ASD. Attention to speech is necessary for spoken language learning, and may also be necessary for other aspects of social, emotional, and cognitive development (Fogel & Thelen, 1987; Patterson & Werker, 2002, 2003). Importantly, attention to speech and faces is integral to the development of communication. It is this body of literature that motivated us to prospectively explore infant preferences for ID speech and faces, and investigate how these preferences related to later language skills in infants at risk for ASD.

1.4 Identifying ASD Behaviours Prior to Diagnosis

As previously mentioned, there is some evidence that the characteristics of ASD are apparent early in infancy (e.g., Dahlgren & Gillberg, 1989; Ohta, Nagai, Hara, & Sasaki, 1987; Rogers & DiLalla, 1990). Retrospective parent reports indicate that characteristics consistent with ASD present within the first year of life and include extremes of temperament and behaviour ranging from extreme passivity to marked irritability (Gillberg et al., 1990; Hoshino et al., 1987), poor or inconsistent eye contact (Gillberg et al., 1990; Hoshino et al., 1987; Rogers & DiLalla, 1990; Volkmar, Stier, & Cohen, 1985), and lack of response to the parents' voices or attempts to play and interact (De Giacomo & Fombonne, 1998; Gillberg et al., 1990; Hoshino et al., 1987; Ohta et al., 1987; Rogers & DiLalla, 1990; Volkmar et al., 1985). The most notable developmental issue that presents in early childhood, which typically first raises concerns from parents and health care providers, are delays in language development and stereotyped behaviour (Short & Schopler, 1988; Stone et al., 1994; Sullivan et al., 1990). Much of the research targeting early signs of ASD has come from retrospective parent reports and analysis of home video tapes of children who have already received a diagnosis. This body of work has guided current practice guidelines in early detection (Filipek et al., 2000), and provided a foundation for currently available autism screening tools (Robins, Fein, Barton, & Green, 2001; Stone, Coonrod, & Ousley, 2000; Bryson, Zwaigenbaum, McDermott, Rombough, & Brian, 2008; Zwaigenbaum et al., 2005). However, it is not clear that we have a complete account of the early behavioural signs of ASD. Knowledge about their time course and their underlying developmental mechanisms remain unclear. Furthermore, retrospective reports may be prone to errors and distortions of recall, especially when parents are asked to remember behaviours that occurred many years ago. When compared with systematic assessment by trained clinicians, free recall of early behaviours is limited and likely does not accurately represent the breadth of abilities and challenges (Stone et al., 1994). Parents of children already diagnosed with ASD may also tend to bias their responses toward behaviours that are consistent with the diagnosis.

While analysis of home videos has significant strengths, particularly relative to retrospective parental reports, this methodology does not always capture a range of children's behaviour across a variety of settings. Home videos from different families will subsequently vary in their quality of the recording, the activities and settings that were recorded, and the length of time the child is visible (Baranek, 1999). Behaviours may be elicited or prompted, rather than spontaneously occur, and if a child fails to behave as expected (or desired), parents may re-record taped segments until they obtain the desired response. Finally, studies that employ video recordings often use typically developing children as comparison groups as opposed to developmentally delayed controls, which limits the extent of our knowledge about ASD-specific deficits (e.g., Adrien et al., 1992; Baranek, 1999; Osterling & Dawson, 1994; Osterling, et al., 2002; Maestro et al., 2002). Both methodologies of collecting data related to early ASD symptomotology (retrospective parental reports and analyses of home videos) aid in the development of early identification and screening procedures (e.g., Filipek et al., 2000). However, these procedures both suffer from methodological pitfalls discussed above and must ultimately be validated empirically in prospective studies (Zwaigenbaum et al., 2007).

1.5 Symptoms in Infant Siblings

ASD appears to be highly genetically determined (Cook, 2001; Le Couteur et al., 1996; Pickles et al., 2000). Twin and family studies of individuals with ASD have indicated a large genetic contribution, with heritability estimates as high as 90% (Freitag, 2007; Gupta & State, 2007). Concordance rates for monozygotic twins range from 60% to 90% as compared to 2% to 10% for dizygotic twins and siblings, thereby establishing that genetic factors play a key role. In addition, risk for ASD is 60 to 100 times higher for siblings of individuals already diagnosed than the general population (see Rutter, et al., 1999 for review). A number of studies have explored varied subclinical behavioural manifestations of ASD in family members of individuals with the diagnosis. In particular, there is considerable evidence that both parents and siblings of children with ASD show a raised incidence of language delays, difficulties with sensory integration, and poor emotion regulation and communication (for a review, see Bailey, Palferman, Heavey, & Le Couteur, 1998; Yirmiya, Shaked, & Erel, 2001). Additionally, the recurrence risk in a younger siblings of children diagnosed with ASD (SIBS-A) is approximately 19% (Ozonoff et al., 2011) and the probability of having another sibling with some form of cognitive or language problem is around 15% (Ritvo, et al, 1989).

Recent data from prospective studies of developmental trajectories of high-risk infant siblings provide evidence of early difficulties in cognitive and language development and in social engagement and attention (Elsabbagh & Johnson, 2007; Orsmond & Seltzer, 2007; Yirmiya & Ozonoff, 2007). Symptoms such as developmental delays in cognition, language, and social engagement (Bryson et al., 2007; Sullivan et al., 2007; Zwaigenbaum et al., 2005) and in repetitive or atypical motor behaviour (Loh et al., 2007) have been identified as early as 12 to 14 months in SIBS-A who are later diagnosed ASD. In a longitudinal study (Zwaigenbaum et al., 2005), 65 SIBS-A were followed for 24 months, with data obtained on measures of visual orienting task, standardized measures of temperament, and cognitive and language development. The 24-month Autism Diagnostic Observation Schedules (ADOS; Lord et al., 2000) scores exceeded threshold for autism in 7 siblings and an additional 12 siblings exceeded threshold for ‘autism spectrum’. Their results are striking and suggest that by 12 months of age, siblings who are later diagnosed with ASD may be distinguished from siblings who are not later diagnosed and from SIBS-TD on the basis of a number of behavioural markers (atypical eye contact, visual tracking, disengagement of visual attention, orienting to name, among others), early characteristic temperament, and delayed expressive and receptive language (Zwaigenbaum et al., 2005).

Studies investigating language development in SIBS-A have revealed that delays in language (Gamliel, Yirmiya, & Sigman, 2007; Toth, Dawson, Meltzoff, Greenson, & Fein, 2007) and social communication (Presmanes, Walden, Stone & Yoder, 2007; Stone, McMahon, Yoder, & Walden, 2007; Sullivan et al., 2007) are relatively common in infant siblings. A recent longitudinal study revealed significantly lower early language scores (but not cognitive scores) in infants 14 to 54 months of non-diagnosed SIBS-A compared to the language scores of SIBS-TD (Gamliel, Yirmiya, Jaffe, Manor, & Sigman, 2009). The authors further observed that the rate of development was also significantly different, thus pinpointing language as a major area of difficulty for SIBS-A (Gamliel et al., 2009). In another study, Goldberg and colleagues (2005) found that 14 to 19 month old siblings of children with ASD not yet identified as having ASD obtained lower scores on the Early Social and Communication Scales (ESCS; Mundy, Delgado, Hogan, & Doehring, 2003; Seibert & Hogan, 1982) as compared to children demonstrating typical development. Siblings of children with ASD actually performed similarly to children with ASD on measures of responding to social interaction, initiating joint attention, and requesting behaviour. Further research demonstrated differences in social engagement, communication, and language in SIBS-A compared to SIBS-TD at 4 and 14 months of age (Yirmiya et al., 2006). These results suggested that while most SIBS-A were performing as well as the SIBS-TD, there were some differences in early social engagement and also with later cognitive and communication skills. Moreover, 6 out of 30 infants in the SIBS-A group revealed a language delay of 5 months.

Taken together, these studies provide evidence that delays in language and communication skills precede speech onset in children at risk for ASD, and they may be predictive of a later diagnosis. Furthermore, there are early behavioural features that are characteristic of ASD present prior to 24 months. Infant-sibling studies offer promise in furthering our understanding of the timing and emergence of atypical patterns of behaviour in language, as well as the emergence of ASD related behaviours. By following these high-risk infants longitudinally, early linguistic and/or behavioural atypicalities may surface. Furthermore, it is possible that these early measures of speech and communication may prove to be sensitive predictors of language and social development. Recent work by Curtin and Vouloumanos (2013) found that low-risk infants listen significantly longer to speech than to non-speech at 12 months, but SIBS-A show no preference. The relative preference for speech correlated positively with general cognitive ability at 12 months for both SIBS-A and SIBS-TD, but only in the SIBS-A group did relative preference for speech predict autistic-like behaviour at 18 months. Interestingly, this relative preference for speech predicted language abilities in only the SIBS-TD group. Together, these findings suggest that in infants at risk for ASD, a lack of a speech preference may underlie atypical social development.

1.6 Current Research

The main objective of this research is to identify potential precursors for atypical language development (i.e., expressive and receptive vocabulary scores) or behavioural manifestations of ASD in infants who are at elevated risk because they have an older sibling with a diagnosis. A further goal of this research is to determine whether early speech and face preferences, vocabulary, and/or ASD-like behaviours relate to later ASD behaviour expression and/or vocabulary development. Research has found that early language skills in children with ASD are an important predictor for later outcome (Dawson et al., 2004). A prospective study examining early language development has the potential to identify early behavioural indicators of ASD. Specifically, this research will examine early ASD-like behaviours, vocabulary development, and speech and visual preferences in this high-risk population, and explore how these patterns of development relate to language outcomes (CDI-II vocabulary scores, Fenson et al., 1993, 2007) and behavioural symptomotology (AOSI scores, Bryson et al., 2008; Zwaigenbaum et al., 2005) across two studies.

In the first study, we investigate whether preferences for ID speech and faces differ between SIBS-A and SIBS-TD, and whether speech and face preferences correlate with vocabulary outcomes and risk group membership. We predict that SIBS-A differ from typically developing infants in their preferences for ID speech and faces, and that a preference for ID speech correlates with later expressive language. The second research study explores whether a relationship exists in early infancy between vocabulary development and ASD-like behaviours. To this end, we examine these relationships in infants at 12 and 18 months of age using a parent report measure of language ability (MacArthur-Bates Communicative Development Inventory: Words and Sentences, Second Edition (CDI-II; Fenson et al., 1993, 2007) and the AOSI **(**Bryson et al., 2008; Zwaigenbaum et al., 2005**)** as a measure of ASD-related behaviours. By following both high- and low-risk infant siblings longitudinally, we are able to track performance and detect prospective ASD indicators in the early stages of development. We predict that vocabulary size and ASD-like behaviours vary by risk group. We further hypothesize that our measures of vocabulary and ASD-like behaviours will correlate with one another, providing further support for the relationship between these skills and behaviours in development.

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Droucker, D., Curtin, S., & Vouloumanos, A. (In press). Linking infant-directed-speech and face preferences to language outcomes in infants at risk for Autism Spectrum Disorder. *Journal of Speech, Language, and Hearing Research.*

Chapter Two: **Linking Infant-Directed-Speech to Language Outcomes in Infants at Risk for Autism Spectrum Disorder**

2.1 Introduction

Speech and faces are salient stimuli for typically developing (TD) infants in early development (e.g., Bushnell, Sai, & Mullen, 1989; Johnson, Dziurawiec, Ellis, & Morton, 1991; Vouloumanos & Werker, 2007). Attention to such species-specific information is integral to the development of social interactions and communication. By extracting species-specific communicative signals from their auditory environments, human infants can analyze speech signals more thoroughly; initial biases towards the processing of human speech sounds could underlie our capacity to process and produce speech (Shultz & Vouloumanos, 2010). Similarly, processing of faces may help infants orient towards, attend to, and assign value to socially relevant stimuli (e.g., Johnson et al, 1991). Indeed, the absence of speech and face biases may result in delays or impairments in language and social development. Children diagnosed with autism spectrum disorder (ASD) show apparent deficits in basic processing of the voices and faces of their own species (Behrmann, Thomas & Humphreys, 2006; Chawarska, Volkmar, & Klin, 2010; Kuhl, Coffey-Corina, Padden, & Dawson, 2005; Paul, Chawarska, Fowler, Cicchetti, & Volkmar, 2007; Whitehouse & Bishop, 2008). A failure to engage in species-typical activities such as looking at a parent's face or listening to speech sounds in the first year of life may help explain the profound impairments in social and language development shown by most children with the disorder. Further, studies have revealed impairments in social and communication skills in undiagnosed family members of individuals with ASD that are milder, but qualitatively similar to those shown by individuals with ASD (for a review see Gerdts & Bernier, 2011). In the current study, we propose to examine whether potential differences in early attentional biases to speech and faces between infant siblings of children with ASD and typically developing infants predict language outcomes and risk group membership.

2.1.1 Preferences for infant-directed speech in typically developing infants

From the first month of life, typically developing (TD) infants show a preference for hearing infant-directed (ID) speech over adult-directed (AD) speech (Cooper & Aslin; 1990; Fernald, 1985). Infants' heightened interest in ID speech may play an important functional role in their linguistic and socio-emotional development. On the linguistic front, the heightened prosodic structure of ID speech is thought to facilitate processing in domains such as word segmentation (Thiessen, Hill, & Saffran, 2005) and word learning (Graf-Estes, 2008). It can also enhance the learning of associations between speech and faces. For example, infants readily learn to associate even unfamiliar ID speech with a face, but do not associate speech lacking ID characteristics with a face (Kaplan, Bachorowski, Smoski, & Hudenko, 2002; Kaplan, Jung, Ryther, & Zarlengo-Strouse, 1996). Caregivers' use of ID speech may facilitate language acquisition (Kemler Nelson, Hirsh-Pasek, Jusczyk, & Cassidy, 1989; Jusczyk, 1997; Kuhl et al., 1997; also see Snow, 1977) over the course of infant development (Fernald, 1992; Hayashi, Tamekawa, & Kiritani, 2001) and across a variety of cultures (Fernald et al., 1989).

ID speech is also thought to facilitate emotional development. Specifically, ID speech has been linked with the transmission and reciprocity of emotional affect (Fernald & Simon, 1984; Fernald, 1985). While caregivers' production of ID speech may express their positive emotional state (Fernald & Simon, 1984), infants' reactions to ID speech may reflect their own positive emotional response. Werker and McLeod (1989) found that 4-5- and 7-9-month-old infants demonstrated greater affective responsiveness while watching a recording of ID speech than when presented with AD speech. Moreover, a person's use of ID speech may further signal their status as a potential caregiver to the infant. An unfamiliar adult who uses ID speech in their initial interaction is more likely to have a high level of interest in infant interaction and caregiving than someone who initially uses AD speech, which typically addresses the adult (e.g., Fernald, 1992). Consistent with this, Schachner and Hannon (2011) found that 5-month old infants look longer at an image of the face of a person who had just used ID speech than at an image of a novel person, but not when that person had used AD speech, instead preferring the novel person. ID speech may thus play an important role in maintaining parent-infant interactions and provide infants with a cue for selection of appropriate social partners.

Despite the role of ID speech in linguistic and emotional development, attention to ID speech changes over the course of development. Hayashi, Tamekawa, and Kiritani (2001) followed infants from 4-10 months of age and found that infants showed a U-shaped developmental shift in attentional biases for ID speech across three stages. At 4-5 months of age, infants attended to ID speech significantly longer than AD speech. Similarly, infants demonstrated the same reliable preference for ID speech at 9-10 months old. However, between 7 and 9 months, infants' preference for ID speech over AD speech decreased, with some infants demonstrating a preference for listening to AD speech. The authors speculated that these changes in ID speech preference reflected three distinct stages of development: an initial preference driven by innate emotional arousal elicited by prosodic and rhythmic characteristics, a subsequent decrease, and final renewal of interest in concert with infants' speech perception abilities. The authors argue that the ID speech preference returns at the same time as infants show awareness that speech signals have distinct linguistic structures. In particular, ID speech may more clearly exhibit the structure of the infant's native speech, such as highlighting word boundaries, and thus a preference for ID speech may be favorable during this period of linguistic development.

2.1.2 Preference for faces in typically developing infants

In addition to attention to ID speech, attention to faces is integral to social development, interaction, and communication. Infants learn much about the social world from the faces of others. They begin to recognize identity (Pascalis, De Haan, Nelson, & De Schonen, 1998), recognize and prefer faces from their own race (Kelly et al., 2005), perceive affect (Cohn & Tronick, 1983; Tronick, 1989), and follow gaze (Corkum & Moore, 1998; Scaife & Bruner, 1975). The ability to use the social and emotional information conveyed in faces and respond to it discriminatively may have been critical for the evolution of social communication (Andrew, 1963; Ekman, 1992; Ekman & Oster, 1979).

In order to extract socially and emotionally relevant information from faces, however, infants must first attend to them. Even newborns show a generalized bias to attend to faces and face-like stimuli (Cassia, Turati, & Simion, 2004; Farroni et al., 2005; Johnson et al., 1991; Simion, Macchi Cassia, Turati, & Valenza, 2001). Thus, faces are salient and preferred stimuli for TD infants in early development (e.g., Bushnell, Sai, & Mullen, 1989).

2.1.3 Is Preference for ID Speech and/or Faces Attenuated in Children Developing Atypically?

A lack of preference for speech and faces may be a reflection of atypical development. Indeed, while TD and developmentally delayed children prefer listening to their mother's ID speech over a noisy overlay composed of many voices and environmental noise, 4- to 6-year old children with ASD showed no preference (Klin, 1991). Moreover, children with ASD do not show a preference for ID speech versus non-speech analogues (Kuhl, Coffey-Corina, Padden, & Dawson, 2005), and those children with ASD who prefer listening to non-speech over ID speech are more likely to exhibit deficits in expressive language ability (Kuhl et al., 2005). A recent study examining ID speech preference in 6-month old infants at risk for ASD, because they have an older sibling diagnosed with ASD (SIBS-A), found that some infants in the group at risk displayed a clear, atypical preference to listen to AD speech (Nadig, Ozonoff, Singh, Young, & Rogers, 2007). These findings suggest that while TD children and children with developmental delays prefer listening to ID speech, children with ASD do not demonstrate similar preferences, and that these atypical preferences may arise in early development.

A preference for faces might also develop atypically in children with ASD. Recent studies show that children with ASD demonstrate decreased attention to faces (Osterling, Dawson & Munson, 2002), impairments in face recognition (Dawson et al., 2002; Klin et al., 1999; Marcus & Nelson, 2001), and deficits in understanding and using facial information in social contexts (Baron-Cohen, 1994; Hobson, 1989). A failure to develop attentional biases toward faces in children with ASD may negatively impact the development of social communication, and in particular language abilities.

In recent prospective studies of developmental trajectories, SIBS-A show early difficulties in cognitive and language development and in social engagement (Elsabbagh & Johnson, 2007; Orsmond & Seltzer, 2007; Yirmiya & Ozonoff, 2007). Twin and family studies have shown that unaffected family members of individuals with ASD demonstrate subclinical features that are characteristic of ASD (for a review see Gerdts & Bernier, 2011). Both parents and siblings of children with ASD show a raised incidence of language delays, difficulties with sensory integration, and potential difficulties with emotion regulation and communication (for a review see Bailey, Le Couteur, Palferman, & Heavey, 1998; Yirmiya, Shaked, & Erel, 2001). Thus, poor language outcomes may be part of the subclinical phenotype seen in family members of children diagnosed with ASD and part of the broader autism spectrum.

2.1.4 The Current Study

The purpose of this study is to investigate whether early speech and face preferences differ in infants at risk of ASD, and to what extent early differences may be predictive of language delays and risk group membership. In order to investigate early species-typical attention to faces and ID speech and its relationship to language development, we examined preferential attention to speech and face information in two populations of infants: SIBS-A and SIBS-TD. Specifically, we tested infants' preferences to ID speech and faces in the first year and a half of life and the extent to which infants' attentional biases predicted language outcome and risk group membership at 18 months of age. We predicted that while SIBS-TD would orient longer to ID speech and faces throughout development, SIBS-A would demonstrate atypical preferences: either no attentional biases, or preferences for AD speech and a non-face visual display (a checkerboard). If SIBS-A do not display the same preferences as SIBS-TD by either attending longer to AD speech and the checkerboard, or by demonstrating no preference for ID speech or faces at all, they may be at risk for language delay and/or a language disorder. Thus, it is important to investigate early attentional biases to faces and speech, and the extent to which such biases predict language development and risk group membership.

2.2 Methods

2.2.1 Participants

Participants were recruited at 6 months of age and consisted of 36 healthy full term infants (22 female) with at least one older sibling. High-risk infants with an older sibling diagnosed with ASD (SIBS-A; N=14; Female = 6) were recruited from local ASD treatment agencies and pediatric clinics in all four quadrants of the City of Calgary including Early Child Development Team through Alberta Health Services, the Society for Treatment of Autism, Renfrew Educational Services, Parent-Link, and other local service organizations. Individuals working at these agencies referred families to our study, posted flyers in their office and information about our study on the agency website. Low risk infant siblings (SIBS-TD; N = 22; Female = 16) of children with no diagnosis were recruited from the Child and Infant Learning and Development (Ch.I.L.D.) research group at the University of Calgary. All infants resided in the Southern region of Alberta in North America. Longitudinal data was collected from infants at 6, 8, 12, and 18 months of age. Although a larger sample size was recruited for this study, only those infants who participated at all four visits were included in the analysis (see Table 1 for the number of participants contributing data). Exclusionary criteria included the presence of a neurological disorder of known etiology, significant sensory or motor impairment, major physical abnormalities, and history of serious head injury and/or neurological disease. Hearing status of the infants was confirmed using an otoacoustic emissions screening procedure at each session. There were no infants who were unable to be tested due to impaired hearing status.

The primary language of the sample for this experiment was English. Those parents who reported speaking two or more languages in the home environment indicated that English was the primary language, spoken equal to or greater than 80% of the time. Second languages reported by parents were: Arabic (N=1), Cantonese (N = 1), Farsi (N = 1), Ilokano (N = 1), Tagalog (N = 1), Spanish (N = 2), and French (N = 8). Educational backgrounds of parents included a high school diploma (maternal N = 3; paternal N = 6), college or technical degree (maternal N = 15; paternal N = 10), some university (maternal N = 1; paternal N = 1), university degree (maternal N = 11; paternal N = 12), and post graduate degree (maternal N = 7; paternal N = 8). Although we did not collect additional demographic information, consistent with other studies from the Speech Development Lab at the University of Calgary, the sample was likely to be predominantly middle class and of European American descent, and representative of the cultural demographic in Calgary. Specifically, 22.2% of the population in Calgary consists of visible minorities (Statistics Canada, 2007), including Chinese (6.2%), South Asian (5.4%), Aboriginal (2.5%), Filipino (2.4%), Black (2.0%), Latin American (1.3%), Southeast Asian (1.5%), Arab (1.1%), West Asian (0.6%), Korean (0.6%), Japanese (0.4%), and visible minorities not included in Census questionnaires (0.2%).

The participants were given a t-shirt or toy as a thank-you gift but were otherwise not compensated for their participation. This study was reviewed and approved by the University of Calgary Ethics Review Committee. Informed consent was obtained in person by trained research staff at the first lab visit.

Table 2.1 Means and standard deviations for each group across all tasks

			SIBS-TD		SIBS-A		TOTAL	
			Mean	SD	Mean	SD	Mean	SD
CDI-II (Raw Expressive Scores)	12 months		8.2	(7.8)	6.6	(5.8)	7.5	(6.9)
		N	*20*		*14*		*34*	
	18 months		82.1	(78.7)	29.3	(25.7)	62.9	(69.0)
		N	*21*		*12*		*33*	
Mullen (Standard Scores)	12 months		104.8	(11.0)	101.7	(12.8)	103.7	(11.6)
		N	*22*		*13*		*35*	
Experimental tasks between 6 and 18 months: Looking time (measured in seconds)*	ID Speech		19.8	(3.6)	22.1	(4.8)	20.7	(4.2)
	AD Speech		17.7	(4.5)	20.3	(4.1)	18.7	(4.4)
	Face		23.3	(3.5)	24.2	(4.0)	23.7	(3.7)
	Checkerboard		14.2	(4.5)	18.1	(5.0)	15.7	(5.0)
		N	*22*		*14*		*36*	

Note: Mean looking times reported for the experimental tasks represent data from ages 6, 8, 12, and 18 months collapsed together for each speech condition across the visual stimulus types and each visual condition across the speech stimuli types.

2.2.2 Apparatus

Testing took place in a 2.74 m by 1.82 m sound attenuated room, which was dimly lit by overhead lighting. Infants sat on a parent's lap facing a Smartboard monitor (122 cm wide x 91.5 cm high) mounted on the wall that was approximately 1.5 m away. Images were projected onto the Smartboard via a NEC LT245 projector. The audio stimuli were delivered at 65 dB +/- 5 dB, over a BOSE 101 speaker, located directly below the monitor. Infant behaviour was recorded using a Sony DCRDVD92 digital video camera. As a masking control during testing, the parent wore Bose True Noise-cancellation Headphones over which vocal music was played from a Teac CD-P1250 CD player. Habit X 1.0 (Cohen, Atkinson, & Chaput, 2004) run on a Macintosh Power PC G5 was used to present stimuli and collect looking time data.

2.2.3 Procedure

At each visit, infants participated in a sequential looking preference procedure (SLP; Cooper & Aslin, 1990; 1994; Pegg, Werker & McLeod, 1992). Parents were asked to keep infants centered and oriented forward. The experimenter, who was blind to the auditory stimuli, monitored infants' looking times via a closed circuit television system from an adjacent testing room. The experiment began with a pre-test control trial in which infants heard classical music paired with a checkerboard in order to direct infants' attention to the monitor. Infants were then exposed to the speech passages paired with a checkerboard or face visual display. A full trial consisted of tokens separated by 300 to 450 ms of silence, for 40 seconds. The presentation of visual and auditory stimuli was counter-balanced across participants. Coders who were blind to the experimental conditions coded infants' looking time from the video to determine reliability. Looking time to the visual stimulus in each condition was measured in seconds.

2.2.4 Stimuli

Two sets of speech stimuli were used to test for auditory preference. The stimuli consisted of two 40-second samples of ID speech and two 40-second samples of AD speech recorded by a female, native English-speaking adult. The ID speech samples described scenes from a child's picture book while the AD speech samples described a personal event (travel to a foreign country).

Overall pitch measurements were taken from each sample every 8 seconds using Praat (Boersma & Weenink, 2012). The mean pitch for AD speech was 212.0 Hz (range: 75.2 Hz -

498.3 Hz) and for ID speech was 293.2 Hz (range: 81.5 Hz - 524.77 Hz). The pitch differed significantly for the two speech types (equal variance not assumed), $t(11.85) = 8.97$, $p < .001$. There was a total of 19 sentences across the two 40-second AD speech samples and a total of 10 sentences across the two 40-second ID samples. The number of words per sentence differed significantly between the AD ($M = 15.58$, $SD = 7.99$) and the ID ($M = 10.22$, $SD = 4.85$) speech (equal variance not assumed), $t(29.92) = 2.02$, $p = .05$.

Each auditory stimulus sample was paired with either a photograph of a neutral black and white female face (NimStim face set; Tottenham et al., 2009) or a black and white checkerboard. Infants were presented with a total of 4 trials: 2 AD speech and 2 ID speech trials, each paired with either a face or a checkerboard.

2.2.5 Language Questionnaire: Communicative Development Inventory

Parents completed the MacArthur-Bates Communicative Development Inventory: Words and Sentences, Second Edition (CDI-II-II) at 12 and 18 months of age (Fenson et al., 1993). This is a standardized parental report measure of vocabulary development of children between 8 and 37 months of age.

2.2.6 Overall Functioning: Mullen Scales of Early Learning

Global cognitive development was assessed using the Mullen Scales of Early Learning (MSEL; Mullen, 1995) at 12 months. The Mullen is a comprehensive assessment of language, motor, and perceptual abilities for children of all ability levels. T scores, percentile ranks, and age equivalents are computed for the five subscales separately (Gross Motor, Visual Reception, Fine Motor, Expressive Language, and Receptive Language), and for a Composite score based on four subscale scores (fine motor, visual reception, expressive language, receptive language) and standardized for children aged 0–69 months. Groups did not differ on Mullen composite score (SIBS-TD: M = 104.8, SD = 10.9, SIBS-A: M = 101.7, SD = 12.8, $F(1,33) = .59$, $p = .448$). Of note, one SIBS-A infant could not complete a subtest from the Mullen contributing to the composite score; therefore, this infants' composite score was not considered in the analysis.

2.3 Results

2.3.1 Does Looking Time for ID Speech/AD Speech and Face/Checkerboard Differ Between Risk Groups?

We first examined infants' looking behaviour with a 2 (speech type: ID speech, AD speech) x 2 (visual stimuli: Face, Checkerboard) x 4 (age: 6, 8, 12, 18) x 2 (risk group: SIBS-TD, SIBS-A) mixed factor analysis of variance (ANOVA). Since not all infants completed the task at each age, our complete sample size was 36 (SIBS-TD: N = 22; SIBS-A: N = 14). Our findings yielded a main effect of speech type, $F(1,34) = 12.38$, $p = .001$, $\eta_p^2 = .267$, and visual stimulus, $F(1,34) = 180.33$, $p < .001$, $\eta_p^2 = .841$. We further found a visual stimulus by risk group interaction, $F(1,34) = 7.05$, $p = .012$, $\eta_p^2 = .172$, an age by risk group interaction, $F(3,102) = 2.82$, $p = .043$, $\eta_p^2 = .077$, and a speech type by age interaction, $F(3,102) = 4.85$, $p = .003$, $\eta_p^2 = .125$. No other factors or interactions were significant, however, there was a trend towards an effect of risk group, $F(1,34) = 3.41$, $p = .073$, $\eta_p^2 =.091$. We next examined each of the interactions in turn.

To understand the source of the visual stimulus by risk group interaction, pairwise comparisons were conducted comparing looking times to the visual stimuli collapsed across ages. The SIBS-TD group looked longer when the visual stimulus was a face rather than a checkerboard (face: M = 23.34, SD = 3.48; checkerboard: M = 14.16, SD = 4.50); $t(21) = 12.78$, $p < .001$, $r = .94$, as did the SIBS-A group (face: M = 24.27, SD = 3.96; checkerboard: M = 18.12, SD = 5.02); $t(13) = 7.00$, $p < .001$, $r = .89$. Thus, both our SIBS-TD and SIBS-A looked significantly longer at face stimuli than at the checkerboard, but the magnitude of the difference was greater for the SIBS-TD. Specifically, SIBS-A looked significantly longer at the checkerboard than the SIBS-TD, $F(1,34) = 6.07$, $p = .019$, $r = .39$, while there was no difference in looking time to faces between the SIBS-A and SIBS-TD groups, $F(1,34) = .553$, $p = .462$, $r = .13$.

The age by risk group interaction demonstrated that the SIBS-A group's overall looking time (8 months: $M = 23.11$, $SD = 4.93$; 18 months: $M = 22.52$, $SD = 6.33$) was greater than that of the SIBS-TD group (8 months: $M = 18.81$, $SD = 5.52$; 18 months: $M = 17.52$, $SD = 5.96$), at 8 months $F(1,34) = 5.62$, $p = .023$, $r = .38$, and 18 months of age, $F(1,34) = 5.73$, $p = .022$, $r = .38$. There were no significant differences between risk groups' looking times at 6 and 12 months.

We next examined the speech type by age interaction. Our findings reveal that infants show a trend for looking longer at ID speech than AD speech at 8 months (ID speech: $M = 21.12$, $SD = 5.62$; AD speech: $M = 19.85$, $SD = 6.39$), $t(35) = 1.812$, $p = .078$, $r = .29$), and look significantly longer at ID speech than AD speech at 12 months (ID speech: $M = 20.85$, $SD = 6.09$; AD speech: $M = 19.27$, $SD = 5.78$ $t(35) = 2.78$, $p = .009$, $r = .43$) and 18 months (ID speech: $M = 21.52$, $SD = 7.47$; AD speech: $M = 17.41$, $SD = 7.15$), $t(35) = 3.71$, $p = .001$, $r = .53$). There were no reliable differences at 6 months (ID speech: $M = 19.23$, $SD = 5.12$; AD speech: $M = 18.34$, $SD = 5.38$).

We predicted that our risk groups would differ in their preferences for speech, with SIBS-A showing atypical looking behaviour. Our SIBS-TD group showed a trend for looking longer during the ID speech sample than the AD sample at 8 months (ID: $M = 19.66$, $SD = 4.88$; AD: $M = 17.97$, $SD = 6.69$; $t(21) = 2.04$, $p = .054$, $r = .41$), and 12 months (ID: $M = 20.86$, $SD = 5.34$; AD: $M = 19.5$, $SD = 5.85$; $t(21) = 1.97$, $p = .062$, $r = .39$), and reliably longer to ID at 18 months

(ID: *M* = 19.3, *SD* = 6.96; AD: *M* = 15.74, *SD* = 6.71; $t(21) = 2.49$, $p = .021$, $r = .48$). The SIBS-A group showed a trend for looking longer during the ID speech sample than the AD sample at 12 months (ID: *M* = 20.83, *SD* = 7.35; AD: *M* = 18.91, *SD* = 5.85; $t(13) = 1.9$, $p = .08$, $r = .47$), and reliably longer at 18 months (ID: *M* = 25.01, *SD* = 7.09; AD: *M* = 20.02, *SD* = 7.27; $t(13) = 2.77$, $p = .016$, $r = .61$).

2.3.2 Does Language Outcome Differ Between Risk groups?

Using only those infants whose scores were included in the ANOVA above and for whom we had CDI-II scores (12 months: N = 34; 18 months: N = 33), we next examined CDI-II expressive language scores at 12 and 18 months of age (see Table 1). SIBS-TD scored significantly higher on the CDI-II at 18 months compared to SIBS-A, $F(1,32) = 5.05$, $p = .032$, $r = .37$. There were no differences in CDI-II scores at 12 months, $F(1,32) = .38$, $p = .542$, $r = .11$. However, infants' CDI-II scores at 12 months significantly correlated with CDI-II scores at 18 months, $r(31) = .698$, $p < .001$. Of note, both SIBS-TD and SIBS-A exhibited a range of vocabulary skills as demonstrated by the CDI-II raw score standard deviations at 12 and 18 months. Variability in vocabulary observed in our data is consistent with the literature. For instance, 16-month-old infants comprehend between 100 and 200 different words, and produce up to 50 words (Bates et al., 1994). Furthermore, although the mean vocabulary size at 18 months in a typical population is over 100 words, the standard deviation is 111 (102% of the mean; Fenson et al. 1993). Indeed, data on vocabulary acquisition reveals large standard deviations in word production during early language development (Fenson et al., 1993).

2.3.3 Does Attention to Speech and Faces in the First Year Correlate with Language Outcome (CDI-II scores) at 18 months?

We next explored whether overall looking behaviour between 6 and 12 months of age was correlated with later expressive language by looking at CDI-II scores at 18 months. Again, we only included those infants who had looking data at all time points in our analysis and CDI-II scores (N = 33). Our findings revealed that CDI-II scores were negatively correlated with overall looking time to the checkerboard image at 18 months, $r(33) = -.429$, $p = .013$, and negatively correlated with overall looking time to AD speech at 18 months, $r(33) = -.511$, $p = .002$. However, when we explored these correlations by risk group, we found that this correlation only held for the SIBS-TD group: SIBS-TD CDI-II scores at 18 months were negatively correlated with overall looking time to AD speech, $r(21) = -.573$, $p = .007$, but interestingly their CDI-II scores negatively correlated with looking time to faces, rather than checkerboards, between 6 and 12 months, $r(21) = -.524$, $p = .015$. SIBS-A showed no significant correlations.

2.3.4 Does Preference for ID Speech and Faces in the First Year Correlate with Language Outcome (CDI-II scores) at 18 months?

We further examined whether a relative preference for ID speech versus AD speech and a relative preference for faces versus checkerboards between 6 and 12 months was predictive of CDI-II scores at 18 months. We averaged the looking time to ID across ages and visual stimuli and we averaged the looking time to AD across ages and visual stimuli. Similarly, we averaged the looking time to faces across ages and speech type and averaged the looking time to checkerboards across ages and speech type. Preference for speech type was then determined by subtracting looking time to AD speech from looking time to ID speech and preference for visual stimuli was determined by subtracting looking time to the checkerboard from looking times to the face. A positive difference score in both cases would demonstrate a preference for ID speech and faces. Our findings revealed that a relative preference for ID speech between 6-12 months predicted CDI-II scores at 18 months, $r(33) = .351$, $p = .045$. There was no correlation observed for faces and CDI-II scores. We next looked at each risk group individually. We again found that a relative preference for ID speech predicted CDI-II scores for SIBS-TD, $r(21) = .504$, $p = .020$, but not for SIBS-A. However, we now observed that for SIBS-A, a relative preference for faces over checkerboards between 6 and 12 months predicted CDI-II scores at 18 months, $r(12) = .642$, $p = .024$. There were no other significant correlations.

2.3.5 Does Preference for ID Speech and/or Faces Predict Risk Group Membership?

To examine whether ID speech or face preferences might predict risk group, we conducted a Binary Logistic Regression using data from 6 to 18 months. We entered into the model the difference scores used as a measure of preference for ID speech (subtracting looking time to AD speech from looking time to ID speech) and faces (subtracting looking time to the checkerboard from looking times to the face) and the risk group membership status. We found that that only the preference for faces significantly improved the model's ability to predict group membership (see Table 2). Whereas the constant alone correctly classified 51% of cases, the inclusion of the face preference information improved classification to 62% of cases. This finding suggests that infants' preference for faces contributes to group membership.

Table 2.2 Results for the binary logistic regression analysis

					95% CI for Odds Ratio		
Variable	**B(SE)**	**Wald's x^2**	**df**	***p***	**Lower**	**Odds Ratio**	**Upper**
Included							
Constant	1.46 (0.59)	6.20	1	0.013			
Face–Checkerboard	-0.18* (0.07)	6.91	1	0.009	0.73	0.84	0.96
ID Speech–AD Speech	-0.08 (0.65)	2.67	1	0.102	0.72	0.86	1.03

Note: For the beta and X^2 columns *SD*'s appear in parentheses. R^2 = .12 (in Hosmer & Lemeshow), .15 (in Cox & Snell), .20 (in Nagelkerke). Model X^2 (2) = 10.4, $p < .01$.

2.4 Discussion

Early attention to speech and faces is crucial for the acquisition of linguistic and social skills. The goal of this study was to investigate early preferences for ID speech and faces in both typically developing and at-risk populations, and to determine the extent to which they predict language development and risk group membership. Overall, both SIBS-A and SIBS-TD infants preferred looking at faces over checkerboards, with a greater difference in looking time in SIBS-TD than in SIBS-A. With regard to speech preferences, we found that while SIBS-TD trended towards a more reliable preference for ID speech prior to 12 months of age, SIBS-A did not demonstrate a clear preference until 18 months. Furthermore, we observed that SIBS-TD exhibited higher expressive language scores at 18 months than SIBS-A. Collapsed across groups, language outcomes correlated with early speech preferences, suggesting that a preference for ID speech early in development may facilitate later expressive language. When we explored this effect by group, we found that a relative preference for ID speech predicted CDI-II scores for SIBS-TD, but not for SIBS-A. Interestingly for SIBS-A, a relative preference for faces over checkerboards in early development predicted CDI-II scores at 18 months. Finally, infants' preference for faces–but not ID speech–contributed to determining group membership.

2.4.1 Expressive Language Ability at 12 and 18 months of age

SIBS-A had significantly lower CDI-II scores at 18 months than SIBS-TD. This suggests that overall our SIBS-A do not have typical productive vocabularies. This finding is consistent with studies examining the language abilities of infant siblings. That is, there are increased incidents of language delay in siblings of children with ASD (see Bailey et al., 2001). It is possible that poor language outcomes or delays may be part of the broader phenotype seen in family members of children diagnosed with ASD. Indeed, consistent with our findings, there is a 15% probability of cognitive or language difficulties in siblings of children with ASD (Elsabbagh & Johnson, 2010; Ritvo, et al., 1989; Zwaigenbaum, et al., 2005).

2.4.2 Attention to Speech and Faces in the First Year Correlates with Language Outcomes in SIBS-TD

Overall looking time to AD speech and checkerboards negatively correlated with CDI-II scores at 18 months. However, it appears that the SIBS-TD were driving the negative correlation between AD speech and CDI-II scores. This suggests that greater overall attention to AD speech is associated with lower expressive language in a typically developing population. Given the early preference for ID speech (Pegg et al., 1992), this could be an indicator of atypical language development in the general population. Interestingly, longer looking times to faces also correlated negatively with CDI-II scores in the SIBS-TD. Attention to visual and auditory information may compete such that attention to species-specific visual information detracts infants' attention from speech information. Thus, within the typically developing population, greater attention to AD and face stimuli might affect typical language development.

2.4.3 Visual Preference in Infants At-Risk for ASD and Language Outcomes

Despite evidence indicating that children with ASD demonstrate decreased attention to faces relative to TD children (Osterling et al., 2002), our results demonstrate that both high and low risk infants looked equally at faces, and preferred to look at faces than the checkerboard. However, SIBS-TD exhibited a greater relative preference for faces than did SIBS-A. This suggests that, while still reliable, our SIBS-A group's preference for faces over checkerboards is not as robust as in the SIBS-TD group. Indeed, SIBS-A tended to look longer at the checkerboard than did SIBS-TD. Importantly, face preference in the SIBS-A group predicted CDI-II scores at 18 months and face preference was a significant predictor of group membership. Thus differential face processing in SIBS-A has implications for language development and potential for identifying ASD status.

We speculate that information transmitted through the face becomes more salient to infants as they begin to integrate speech signals with other sensory input (e.g., facial expressions, joint attention, eye gaze; Fernald, 1992). Thus, TD infants preferentially attend to faces as they learn about socioemotional meaning, which may be supported through the preference for the emotionally laden, highly prosodic ID speech. However, in our SIBS-A group, attentional biases to faces appear to better support later language development. This suggests that while a preference for ID speech may be more supportive in facilitating language development in SIBS-TD, attentional biases for faces may facilitate language learning in SIBS-A. While attending to ID speech sets the stage for finding words to attach to meanings generating larger vocabularies in typically developing infants, infants who do not exhibit the same early ID speech preferences may rely on other sources of information to learn about language. In the case of infants at risk for ASD, attending to faces may partly compensate for a lack of a preference for ID speech, and facilitate mappings between words and referents in the environment. That is, some interest in species-specific social information, in this case faces (e.g., Johnson et al., 1991), may compensate for other delayed or non-existent biases.

2.4.4 Preference for ID Speech Predicts Language Outcomes in Typically Developing Infants

Data from the speech analyses revealed that while SIBS-TD trended towards a preference for ID speech at 8, 12 and 18 months, SIBS-A did not exhibit a clear preference for ID speech over AD speech until 18 months of age possibly reflecting a delay in the development of an ID speech bias in SIBS-A. Consistent with this interpretation, our findings indicated that as looking time to ID speech increased, so did scores on the expressive CDI-II for the SIBS-TD. However, this correlation was not observed in SIBS-A. If the role of ID speech in linguistic and emotional development changes throughout early development (Hayashi et al., 2001), then a delay could impact learning. That is, if ID speech highlights the structure of the infant's native language, such as highlighting word boundaries, then typical linguistic development may rely on a preference for ID speech emerging prior to the end of the first year of life.

2.4.5 Determining Group Membership Based on Early Preferences

Infants' preference for faces, but not speech type, contributed to determining group membership, although this only improved classification by 11%. This may be explained by the heterogenous nature of the SIBS-A risk group (e.g., Ritvo et al., 1989), whose individual outcomes are still unknown. It may be the case that infants in the SIBS-A group will not develop a language disorder nor will they receive a diagnosis of ASD. However, risk group membership may still be helpful for determining which high-risk individuals may benefit from intervention prior to any outcome diagnosis. Following infants until a diagnosis may be made would further validate the risk group membership results of the logistic regression analysis.

2.4.6 Conclusions

The current study sheds light on how preferences for species relevant information, such as speech and faces, relate to language development in the siblings of children diagnosed with ASD. These results provide some evidence that lack of attending to ID speech and faces may be related to later delays in language development, and may be helpful for determining which high-risk individuals would benefit from intervention prior to any outcome diagnosis.

Chapter Three: **Early Behaviour and Language Development in Infants at Heightened Risk for Autism Spectrum Disorder**

Understanding the developmental trajectory of ASD-specific symptoms (e.g., social attention, language, behavioural manifestations) in infants at heightened risk for ASD involves identifying the patterns of development in high-risk infants that are distinct from typically developing infants who do not demonstrate such behaviours. Atypicalities may exist in underlying perceptual processes that are not apparently or directly linked to later manifestations of ASD, which may in fact be fundamental to later social and communicative impairment. A bias or preference for speech and faces is a first step in the development of social communication. By attending to the highly engaging features of infant directed (ID) speech that facilitate language learning, early preferences emerge that draw the infant into social communication, while they are concomitantly learning about other relevant social information conveyed through attention to faces. Differences in infants' attention to such socially relevant information may negatively impact on social communication and language development. Early variation in the development of abilities that are impaired in children with ASD (e.g., language, social interaction skills, behavioural symptoms) may then limit the child's opportunities to learn from social experiences. Reduced opportunity for social engagement, together with impaired language and social interaction skills may have downstream effects on the pattern of development that eventually leads to further manifestations of ASD. While in the previous study, we identified differences between low- and high-risk infants on factors related to social attention (e.g., attention to faces and speech) and how perceptual preferences predict vocabulary size, further investigation of autistic behaviour and vocabulary development will provide a more comprehensive understanding of early abilities in infants at risk for ASD.

3.1 Introduction

During early development, fundamental cognitive abilities begin to emerge that allow the infant to learn about and interact with the environment. As different abilities develop, acquired skills support further learning. For instance, the pre-linguistic child may first only use non-verbal cues, such as pointing or showing, for functional communication during a social interaction, but over time, these non-verbal cues become integrated with expressive language to supplement communication (Bates & Dick, 2002; Capone & McGregor, 2004). Social communicative behaviours, such as joint attention, imitation, reciprocal social games (e.g., peek-a-boo), and imitative play with toys, emerge and evolve from the first through the second year of life (e.g., Carpenter, Mastergeorge, & Coggins, 1983; Crais, Campbell, & Cox, 2004). These behaviours are associated with both language and cognitive development (Bates, Camaioni, Benigni, Bretherton, & Volterra, 1979; Carpenter, Nagall, & Tomasello, Butterworth, & Moore, 1998). The rapidly developing cognitive abilities also manifest behaviourally in how an infant plays within, explores, and moves about the environment (Toth, Munson, Meltzoff, & Dawson, 2006). Thus, for typically developing children, the emergence of social communication and the development of language and cognitive abilities work together to help the child navigate the social world.

Autism Spectrum Disorder (ASD) is characterized by social and communicative impairments, and repetitive behaviours (Volkmar, Chawarska, & Klin, 2005). Indeed, the diagnostic criteria for ASD cluster around communicative, linguistic, and social deficits (DSM-IV; American Psychiatric Association, 1994). Identification of impairments associated with ASD relies on children’s explicit behaviours, which only emerge reliably after the first year of life (DeGiacomo & Fombonne, 1998). That is, many of these behaviours manifest later in development, and as a result, ASD is typically diagnosed around 3 to 5 years (Mandell, Novake, & Zubritsky, 2005; Ozonoff et al., 2009). While ASD may not be diagnosed until late in the preschool years (Howlin & Moore, 1997), marked neurodevelopmental abnormalities may be present at birth and continue to evolve from the earliest months of life. During the first year of life, children with ASD are retrospectively distinguished by reduced social interaction (Adrien et al., 1992), reduced or absent social smiling (Adrien et al., 1992), lack of facial expression (Adrien et al., 1992), failure to orient to name (Zakien, Malvy, Desombre, Roux, & Lenoir, 2000; Maestro, Casella, Milone, Muratori, & Palacio-Espasa, 1999; Bernabei, Camaioni, & Levi, 1998; Mars, Mauk, & Dowrick, 1998; Osterling & Dawson, 1994), lack of pointing and/or showing (Mars et al., 1998; Osterling & Dawson, 1994), decreased orienting to faces (Zakien et al., 2000; Maestro et al., 1999; Bernabei et al., 1998; Mars et al., 1998; Osterling & Dawson, 1994), and lack of spontaneous imitation (Mars et al., 1998). Empirical findings with children diagnosed with ASD have demonstrated that they do not pattern with typically developing children in a number of developmental domains, and this atypical patterning might be exhibited in early infancy.

The course of language and communication development in individuals with ASD varies greatly. Language delay is often parents' primary concern when children are referred for possible ASD diagnosis (Lord, Risi, & Pickles, 2004). Social communication and language skills have been found to be predictive of language skill gain and outcome (Adamson, Bakeman, & Deckner, 2004; Charman et al., 2005; Sigman & Ruskin, 1999; Toth et al., 2006), as well as social function in children with ASD (Rutter, 1970; Venter, Lord & Schopler, 1992). In typically developing infants, vocabulary size is strongly related to early lexical and grammatical development (Bates, Bretherton & Snyder, 1988) and to linguistic and cognitive abilities at 4 years of age (Feldman et al., 2005). While there is little research investigating outcome predictors in children with ASD who are either nonverbal or demonstrate very delayed language at the time of diagnosis (e.g., Smith, Mirenda, & Zaidman-Zait, 2007), in general language development is associated with better long-term outcomes (Howlin, Goode, Hutton, & Rutter, 2004; Lord, Risi, & Pickles, 2004; Szatmari, Bryson, Boyle, Streiner, & Duku, 2003). Understanding of the sources of heterogeneity in language development of children with ASD is important yet our knowledge is limited. Most of what is known has largely relied on data from small, cross-sectional samples. Few studies have involved longitudinal data sets that allow examination of language development over time (e.g., Curtin & Vouloumanos, 2013; Howlin et al., 2004; Smith, et al., 2007). Even children with very delayed language or atypical language early in development can show significant progress as they mature (Howlin et al., 2004). It is currently unknown how language may be related to the developmental course of other ASD-related behaviours. As such, assessing language ability, specifically through measures of expressive and receptive vocabulary and gesture use, is important to understanding the developmental progression of ASD and potential language delays (American Academy of Pediatrics Committee on Children with Disabilities, 2001).

Previous research on early detection of ASD has largely explored neurodevelopmental markers, such as in studies that utilize MRI and EEG technology (e.g., Kuhl, Coffey-Corina, Padden, & Dawson, 2005; Boddaert & Zilbovicius, 2002; Pierce, Muller, Ambrose, Allen, & Courchesne, 2001; Critchley et al., 2000; Schultz et al., 2000), or retrospective parent reports of development (e.g., Werner, Dawson, Osterling, & Nuhad, 2000; Baranek, 1999; Osterling & Dawson, 1994). However, more recent studies have begun to focus on identifying earlier behavioural indicators to help with early identification (Zwaigenbaum et al., 2005). For instance, infants who go on to receive a later diagnosis of ASD may show differences in their attention to social information (e.g., attention to faces and speech). Indeed, children diagnosed with ASD show apparent deficits in basic processing of the voices and faces of their own species (Behrmann, Thomas & Humphreys, 2006; Chawarska, Volkmar, & Klin, 2010; Kuhl et al., 2005; Paul, Chawarska, Fowler, Cicchetti, & Volkmar, 2007; Whitehouse & Bishop, 2008). A failure to engage in such social activities as looking at a parent's face or listening to speech sounds early in life may help explain the profound impairments in social and language development shown by most children with the disorder. Attention to speech is integral to the development of social communication. As infants begin to integrate the vocal activity of their caregivers with a range of other sensory input (facial expressions, joint attention, eye gaze), they learn about socioemotional meaning (Fernald, 1992). Atypical processing of this information might be evident early in development; however this is not easily observable without specific tasks targeted to examine responses to social stimuli.

Behaviours that emerge in infancy and early childhood may also present differently in children who are later identified with ASD than those in typically developing children. Indeed, behaviours that serve as early identification markers, such as delayed visual disengagement of attention, non-response to name, or limited tendency to imitate actions performed on objects, may not simply be early developing symptoms of ASD, but rather behaviours that reduce opportunities to learn from social experiences. In turn, this may instigate an atypical pattern of development that eventually leads to further behaviours that are associated with ASD. Prospectively assessing behaviours in infants at risk for ASD, identifying such behaviours, and observing how they differ in an ASD population may contribute to earlier identification.

A number of studies have explored varied behavioural manifestations of ASD in family members of individuals with the diagnosis. There is considerable evidence that both parents and siblings of children with ASD show an increased incidence of language delays (Constantino, Zhang, Frazier, Abbacchi, & Law, 2010) and deficits in social communication and play skills (Toth, Dawson, Meltzoff, Greenson, & Fein, 2007). Moreover, recurrence risk in younger siblings of children diagnosed with ASD is approximately 19% (Ozonoff et al., 2011), and the probability of having another sibling with some form of cognitive or language problem is around 15% (Ritvo et al, 1989). Recent data from prospective studies of developmental trajectories of high-risk infant siblings (SIBS-A) provide evidence of early difficulties in cognitive and language development and in social engagement (Elsabbagh & Johnson, 2007; Orsmond & Seltzer, 2007; Yirmiya & Ozonoff, 2007). Symptoms such as developmental delays in cognition, language, and social engagement (Bryson et al., 2007; Sullivan et al., 2007; Zwaigenbaum et al., 2005) and in repetitive or atypical motor behaviour (Loh et al., 2007) have been identified as early as 12 to 14 months in SIBS-A who are later diagnosed ASD.

The high-risk infant sibling research suggests that there are marked behavioural and linguistic indicators in the early development of ASD. As early as 14 months of age, SIBS-A demonstrate differences in early social engagement, as well as later cognitive and communication skills compared with SIBS-TD (Yirmiya et al., 2006). Language delays of up to 5 months have also been observed. Longitudinal research has revealed that SIBS-A who are later diagnosed with ASD may be distinguished from SIBS-A who are not later diagnosed and from SIBS-TD on the basis of a number of behavioural markers (e.g., atypical eye contact, visual tracking, disengagement of visual attention, orienting to name), early characteristic temperament, and delayed expressive and receptive language (Zwaigenbaum et al., 2005). Further studies have shown that early social communicative behaviours (e.g., social interaction, joint attention, requesting) of SIBS-A differ from those of SIBS-TD but not from the behaviours displayed by young children with ASD. Overall, there is evidence of deficits in social and communicative behaviours of siblings of children with ASD.

Early identification is critical to treatment and prognosis for individuals with ASD (Dawson, 2008; National Research Council 2001). However, detection of ASD-related behaviours in the first year of life may be challenging. Infant-sibling studies offer promise in furthering our understanding of the timing and developmental patterns of language and social communication, and the emergence of autistic-like behaviours. The current study explores language development (CDI-II; Fenson et al., 1993, 2007) and autistic-like behaviours (AOSI; Zwaigenbaum, et al., 2005) at 12 and 18 months of age in siblings of children with ASD who were matched to a group of siblings of typically developing children on the Mullen Scales of Early Learning (MSEL; Mullen, 1995). The goal of this study was to examine whether high-risk infants demonstrated atypical language development and/or ASD related behaviours as compared with low risk infants from 12 to 18 months of age, and to determine whether relationships existed between language and autistic-like behaviour early in development. We predicted that SIBS-A would differ from SIBS-TD in their early autistic-like behaviours (AOSI) and their development of language (CDI-II) over the first year and a half of life.

3.2 Methods

3.2.1 Participants

Participants consisted of 67 healthy full term infants (34 female) with at least one older sibling. High-risk infants with an older sibling diagnosed with ASD (SIBS-A; N = 36; Female = 16) were recruited from local ASD treatment agencies and paediatric clinics in all four quadrants of the City of Calgary including the Early Child Development Team through Alberta Health Services, the Society for Treatment of Autism, Renfrew Educational Services, Parent-Link, and other local service organizations. Individuals working at these agencies referred families to our study, posted flyers in their office and included information about our study on their agency's website. Low risk infant siblings (SIBS-TD; N = 31; Female = 18) of children with no diagnosis were recruited from the Child and Infant Learning and Development (Ch.I.L.D.) research group at the University of Calgary. Participants were part of a larger longitudinal study which included additional tests and data collected at several ages. For this study, data from 12- and 18-month visits were examined. Exclusionary criteria included the presence of a neurological disorder of known etiology, significant sensory or motor impairment, major physical abnormalities, and history of serious head injury and/or neurological disease. Hearing status of the infants was confirmed using an otoacoustic emissions screening procedure at each session. There were no infants who were unable to be tested due to impaired hearing status.

Participant groups were matched on age and performance on the MSEL (Mullen, 1995). Global development was assessed using the MSEL at 12 months. The MSEL is a comprehensive assessment of language, motor, and perceptual abilities for children of all ability levels. T scores, percentile ranks, and age equivalents are computed for the five subscales separately (Gross Motor, Visual Reception, Fine Motor, Expressive Language, and Receptive Language), and for a Composite score based on four subscale scores (fine motor, visual reception, expressive language, receptive language) and standardized for children aged 0–69 months. Groups did not differ on MSEL composite score (SIBS-TD: M = 102.85, SD = 11.04, SIBS-A: M = 98.48, SD = 12.60, $F(1,39) = 1.39$, $p = .245$). Only those infants from whom we collected CDI-II and AOSI data at both 12- and 18-month visits were included in this current analysis (see Table 2). A subset of participants from the first study reported in Chapter 2 participated in the current study (N = 26: SIBS-TD = 16; SIBS-A = 10). That is, their CDI-II expressive language scores were used in both studies.

The primary language of the sample for this study was English. The majority of participants reported speaking only English in the home environment. However, a total of 15 participants reported speaking two or more languages including: Arabic (N=1), Cantonese (N = 1), Farsi (N = 1), Ilokano (N = 1), Tagalog (N = 1), Spanish (N = 2), and French (N = 8). Of note, these participants indicated that English was the primary language, spoken equal to or greater than 80% of the time. Educational backgrounds of parents included those who did not earn a high school diploma, high school diploma, college or technical degree, some university, university degree, post graduate degree, and unknown/not reported (Table 1). Additional demographic information was not collected from participants. However, the participant sample was consistent with other studies in the Speech Development Lab at the University of Calgary, and was largely comprised of middle class and of European American descent. This sample is representative of the cultural demographic in Calgary. According to Statistics Canada (2007), 22.2% of the population in Calgary are composed of visible minorities including Chinese (6.2%), South Asian (5.4%), Aboriginal (2.5%), Filipino (2.4%), Black (2.0%), Latin American (1.3%), Southeast Asian (1.5%), Arab (1.1%), West Asian (0.6%), Korean (0.6%), Japanese (0.4%), and visible minorities not included in Census questionnaires (0.2%).

Table 3.1 Sample size of maternal and paternal parent education

	High School but no diploma	High School Diploma	College/ Technical Degree	Some University	University Degree	Post-graduate Degree	Unknown/Not reported	Total
Maternal	0	6	27	1	21	11	1	67
Paternal	1	7	26	1	21	11	0	67

Attrition rate was low and varied for each task depending on whether the infant was able to complete all tasks at each visit. As such, sample size varied for each task (see Table 2). The participants were given a t-shirt or toy as a thank-you gift but were otherwise not compensated for their participation. This study was reviewed and approved by the University of Calgary Conjoint Health Research Ethics Board. Informed consent was obtained in person by trained research staff at the first lab visit.

3.2.2 Measures

3.2.2.1 Language: MacArthur-Bates Communicative Development Inventory, Second Edition

At the 12 and 18 months visits, parents completed the CDI-II (Fenson et al., 1993, 2007), a parent report measure of vocabulary development. The CDI-II measures early language skills of children between 8 and 37 months of age and has a large normative sample. This measure of early communication has been used widely in research with typically developing and some atypical samples (Fenson et al., 2007). Parents completed the Words and Gestures form when their infants were 12 and 18 months old. The Words and Gestures form assesses vocabulary production and comprehension, as well as gesture use. It is norm referenced and computes raw scores and percentile ranks for three composites: Understands (Receptive Language), Understands and Says (Expressive Language), and Gestures. The CDI-II was normed on an original sample of 659 children 8 to 16 months of age in 1988 to 1989 across three states. In the updated sample, a larger population of 1089 children aged 8 to 18 months was accessed across eight states, and included a larger range in ethnic diversity and educational background. Demographic information is reported in the MacArthur-Bates Communicative Development User's Guide and Technical Manual, Second Edition (Fenson et al., 2007). Measures of reliability show high internal consistency and test-retest reliability as summarized in the technical manual (Fensen et al., 2007). For instance, the vocabulary measures for the Words and Gestures form demonstrated high internal consistency with alpha coefficients of .95 and .96 for receptive and expressive vocabulary, respectively. Correlations on test-retest reliability measures were in the upper .80's for comprehension and in the mid-.80's for expression after the 10 month period. On measures of concurrent validity, the correlations between laboratory measures and inventory scores are generally high, with the highest (e.g., r = .73) on the Expressive One Word Picture Vocabulary Test (EOWPVT; Gardner, 1981, Dale, 1991).

3.2.2.2 Autistic Behaviours: Autism Observation Scale for Infants

The AOSI (Zwaigenbaum, et al., 2005) is an 18-item direct observational measure of autistic symptomotology developed to detect and monitor early signs of autism in infants aged 6 to 18 months. Infants completed the AOSI task at 12 and 18 months of age. Throughout the administration of the standard set of semi-structured activities, the infant is engaged in play while the examiner conducts a set of systematic presses to elicit particular target behaviours (e.g., eye gaze shift, anticipation of a routine, visual tracking, response to name, atypical motor and sensory behaviours). The relative presence or absence of these ‘‘pressed for’’ behaviours is rated, as are an additional set of behaviours, which are targeted for observation throughout the entire assessment. The AOSI provides a Total Score summing the ratings across all items, and a

Total Number of Markers, summing the total number of endorsed items (items that were rated as above “0”).

Inter-rater reliability both for the AOSI Total Scores and Total Number of Markers is good to excellent (.68 - .94) at 6, 12 and 18 months (Bryson, Zwaigenbaum, Mcdermott, Rombough, & Brian, 2008). Test-retest reliability of the AOSI at 12 months of age is within acceptable limits, (.61 and .68, for Total Scores and Total Marker counts, respectively). Preliminary data indicate that a Total Score on the AOSI of 9 or more at 12 months is predictive of an independent blind “gold standard” diagnosis of autism or ASD at 3 years of age (Bryson et al., 2008). They found that SIBS-A diagnosed with autism at 3 years old received mean AOSI Total Scores of 10.9 (SD = 6.8; range = 0 - 24) at 12 months and 13.2 (SD = 7.5; range = 3 - 25) at 18 months. Moreover, data from Zwaigenbaum and colleagues (2005) suggest that Number of Markers on the AOSI can distinguish between siblings with and without autism as early as 12 months of age. The presence of seven or more Markers at 12 months prospectively identified 6 out of 7 SIBS-A diagnosed with autism at 24 months, compared with 2 out of 58 SIBS-A that received no diagnosis and 0 out of 23 SIBS-TD. Thus, using a cut-off point of 7 Markers, sensitivity and specificity of the AOSI for autism are 84% and 98%, respectively.

The AOSI was administered in a 2.74 m by 1.82 m quiet room, which was lit by overhead fluorescent lighting. The infant was seated in a high chair beside his/her caregiver and facing the experimenter with a table positioned in the middle. The infant was presented with infant friendly toys according to the standardized format of the assessment kit. The caregiver was encouraged to assist in making the infant comfortable, but otherwise asked to assume an observer role. Caregivers were instructed not to comment on or direct attention to any of the toys or activities during administration. The administration time was approximately 20 minutes. The infant’s behaviour was coded online during administration and video recorded using a Sony DCRDVD92 digital video camera for subsequent offline coding. Twenty percent of the videos were coded from the video to determine reliability by a second, blind coder.

3.3 Results

We examined whether vocabulary is associated with ASD-like behaviours at 12 and/or 18 months of age. First, we tested for group differences for each of the factors of interest: expressive language, receptive language, and gestures using the CDI-II and ASD-related behaviours assessed with the AOSI. We then examined how linguistic development relates to ASD-like behaviours.

3.3.1 Language

To determine whether there were group differences, we examined infants' scores on the three CDI-II composites (Expressive Language, Receptive Language, and Gestures) at 12 and 18 months of age (see Table 2). First, we explored expressive language with a one-way ANOVA for each the 12 and 18 month visits, which revealed significant differences in vocabulary size between groups at 12 ($F(1,58) = 3.99$, $p = .05$, $\eta_p^2 = .25$) and 18 months of age ($F(1,53) = 4.43$, $p = .04$, $\eta_p^2 = .27$). These findings show that SIBS-TD have a larger vocabulary size than SIBS-A at both 12 (SIBS-TD: $M = 7.89$, $SD = 7.22$; SIBS-A: $M = 4.69$, $SD = 5.14$) and 18 months of age (SIBS-TD: $M = 73.59$, $SD = 71.49$; SIBS-A: $M = 38.07$, $SD = 52.61$).

Next, we used a one-way ANOVA to explore group differences on the CDI-II Receptive Language at both the 12 and 18 month visits. Results indicated significant differences between groups at 18 ($F(1,41) = 6.87$, $p = .01$, $\eta_p^2 = .37$) but not 12 months of age ($F(1,58) = 0.13$, $p = .72$). These results suggest that SIBS-TD demonstrate better receptive language skills than SIBS-A at 18 months (SIBS-TD: $M = 211.38$, $SD = 87.83$; SIBS-A: $M = 134.29$, $SD = 103.55$).

Finally, we examined whether there were any differences in gesture use between groups at 12 and 18 months. A one-way ANOVA revealed no significant differences between groups at 12 ($F(1,57) = 1.90$, $p = .17$) or 18 months of age ($F(1,40) = 0.57$, $p = .46$). This finding suggests that gesture use is similar across risk groups from a year to a year and a half.

3.3.2 Autistic Behaviours

We next examined infants' performance on the AOSI Total Score and Number of Markers. Findings from the one-way ANOVA indicated that SIBS-TD had a significantly lower overall score on the AOSI Total Score at 18 ($F(1,59) = 5.30$, $p = .03$, $\eta_p^2 = .28$), but not 12 months ($F(1,59) = 0.80$, $p = .37$) compared to SIBS-A (see Table 2). Likewise, when AOSI Markers were examined using a one way ANOVA, findings revealed significant differences between groups at 18 ($F(1,58) = 5.95$, $p = .02$, $\eta_p^2 = .25$), but not 12 months of age ($F(1,60) = 2.10$, $p = .15$). Overall, SIBS-A demonstrate a greater number of ASD-related behaviours (i.e., AOSI Number of Markers; SIBS-TD: $M = 2.93$, $SD = 2.93$; SIBS-A: $M = 4.32$, $SD = 2.34$), which are also more severe in nature (i.e., AOSI Total Score; SIBS-TD: $M = 4.48$, $SD = 3.96$; SIBS-A: $M = 6.84$, $SD = 4.03$) then SIBS-TD at 18 months old.

Table 3.2 Means and standard deviations for each group across all tasks

		12 months		18 months	
Group		**Mean (N)**	**SD**	**Mean (N)**	**SD**
CDI-II Expressive	**SIBS-TD**	7.89 (28)	7.22	73.59 (27)	71.49
Language	**SIBS-A**	4.69 (32)	5.14	38.07 (28)	52.61
CDI-II Receptive	**SIBS-TD**	67.54 (28)	47.47	211.38 (26)	87.83
Language	**SIBS-A**	61.91 (32)	70.56	134.29 (17)	103.55
CDI-II Gestures	**SIBS-TD**	25.00 (28)	7.96	43.88 (25)	7.88
	SIBS-A	21.77 (31)	9.80	51.47 (17)	49.66
AOSI Total Score	**SIBS-TD**	4.37 (30)	3.79	4.48 (29)	3.96
	SIBS-A	5.29 (31)	4.24	6.84 (32)	4.03
AOSI Number of	**SIBS-TD**	2.67 (30)	1.99	2.93 (29)	2.93
Markers	**SIBS-A**	3.5 (32)	2.49	4.32 (31)	2.34

Note: Values reported above represent mean raw scores for the CDI-II and AOSI.

These findings suggest that early in development, younger siblings of children with ASD are at risk for atypical development themselves. In particular, expressive language in SIBS-A differed significantly from SIBS-TD at both 12 and 18 months, while receptive language began to develop differently within the second year, as did ASD behaviours. Our findings are consistent with reports of atypical language development among siblings of children with ASD (Constantino et al., 2010), as well as differences in behavioural manifestations of ASD (e.g., social communication, play skills, eye gaze, social reciprocity; Toth et al., 2007; Goldberg et al., 2005; Zwaigenbaum et al., 2005), but only at 18 months of age.

3.3.3 Do Relationships Exist Between Language (CDI-II) and Autistic Behaviours (AOSI) at 12 and 18 months?

Our primary question is whether a relationship exists between atypical behaviours associated with ASD and vocabulary development. We explored whether autistic behaviour was correlated with language skills by looking at scores on the CDI-II and AOSI at 12 and 18 months of age. It was hypothesized that positive correlations would be observed between measures of communication (receptive and expressive language and gestures) across and within testing time points. This was also hypothesized to be the case for the AOSI total scores and total markers. For our main question we hypothesized that measures of communication would negatively correlate with the measures of ASD like behaviours. That is, lower CDI-II scores would correlate with higher AOSI scores, suggesting that vocabulary skills are related to the expression of ASD-like behaviours. Only infants who participated in both measures (CDI-II and AOSI) at both 12 and 18 months of age were included in the analysis (SIBS-TD: N=20; SIBS-A: N=21).

3.3.3.1 Correlations Within Measures

Our findings revealed correlations between testing age at 12 and 18 months on the CDI-II Expressive Language composite, $r(40) = .667$, $p < .001$, the Receptive Language composite, $r(40) = .712$, $p < .001$, and the Gestures composite, $r(40) = .776$, $p < .001$. At 12 months, the Gesture composite correlated with the CDI-II Expressive and Receptive composites ($r(40) = .420$, $p = .008$ and $r(40) = .570$, $p < .001$, respectively; see Table 3). We also found correlations between the CDI-II Expressive Language and Receptive Language ($r(40) = .691$, $p < .001$), as well as Gestures ($r(40) = .587$, $p < .001$) at 18 months. Receptive Language was correlated with Gestures at 18 months, $r(40) = .724$, $p < .001$. Finally, correlations were found between the

AOSI Total Score and Number of Markers composites at 12 and 18 months ($r(40) = .971$, $p < .001$ and $r(40) = .970$, $p < .001$, respectively).

3.3.3.2 Correlations Between Measures

At 18 months, the AOSI Total Score was negatively correlated with both the Expressive Language and Receptive Language composites ($r(40) = -.376$, $p = .017$ and $r(40) = -.463$, $p = .006$, respectively). Likewise, the AOSI Total Number of Markers was negatively correlated with both CDI-II Language composites at 18 months (Expressive Language: $r(40) = -.358$, $p = .023$; Receptive Language: $r(40) = -.434$, $p = .010$, respectively). Finally, 12 month CDI-II Expressive Language scores were negatively correlated with AOSI Total Scores at 18 months old ($r(40) = -.335$, $p = .034$).

Table 3.3 Significant correlations between tasks for 12 and 18 months collapsed across group

Experimental task		**CDI-II Expressive Language**		**CDI-II Receptive Language**		**CDI-II Gestures**		**AOSI-Total Score**		**AOSI Number of Markers**	
		12	**18**	**12**	**18**	**12**	**18**	**12**	**18**	**12**	**18**
CDI-II Expressive Language	**12**		.667*			.420**			-.335*		
	18			.421*	.691**	.372*	.587**	-.337*	-.376*	-.375*	-.358*
CDI-II Receptive Language	**12**				.712**	.570**	.498**				
	18					.580**	.724**		-.463**		-.434*
CDI-II Gestures	**12**						.776**				
	18										
AOSI Total Score	**12**									.971**	
	18										.970**
AOSI Number of Markers	**12**										
	18										

Note: * indicates significance at p <.05 and ** indicates significant at p <.01

These findings suggest that early emerging language skills, including expressive and receptive vocabulary and use of gestures at one year of age are related to later language skills at a year and a half. Furthermore, 12-month expressive and receptive vocabulary scores are related to ASD-like behaviours at 18 months. Conversely, early ASD behaviours at 12 months are highly related to expressive language at both 12 and 18 month, demonstrating a relationship between vocabulary knowledge and ASD behaviours. The question still remains as to whether lower vocabulary development predicts autistic-like behaviour or if autistic-like behaviours predict lower vocabulary. This inter-relatedness may also be simply indicative of the overall development of the disorder; however it's difficult to determine whether high-risk siblings as a group present with atypical language and ASD-behaviours or whether those who are later diagnosed with ASD are driving the effect.

3.4 Discussion

We examined whether infants of 12 and 18 months exhibited differences in their language abilities and their presentation of autistic-like behaviours by virtue of having an older sibling with ASD or not. We found that infants with an older sibling with ASD exhibited lower expressive vocabulary at 12 and 18 months as compared with SIBS-TD. They also demonstrated significantly more ASD-like behaviours (e.g., Number of Markers) that were also more severe in nature (e.g., Total Score). When we investigated whether relationships existed between ASD behaviours and vocabulary, we found that expressive and receptive vocabulary are significantly correlated with ASD behaviours.

Recent research has found that high-risk infant siblings tend to demonstrate atypical developmental trajectories (Gamliel, Yirmiya, Jaffe, Manor, & Sigman, 2009). Early linguistic and behavioural characteristics that are milder but qualitatively similar to the defining features of

ASD, often referred to as the broader autism phenotype (Bolton et al., 1994), may be present in this high-risk population. Thus, the group differences in the presentation of ASD-like behaviours and language in our high-risk infants may be related to a broader autism phenotype, or may be better accounted for by those that receive a later diagnosis of ASD that are driving group differences. It's important to note that the SIBS-A group is a highly heterogenic group, comprised of four possible subsets: infants who go on to receive a later diagnosis of ASD; those who will not meet criteria for ASD; and those who will demonstrate later language impairments with or without a concurrent ASD diagnosis. While delays in the development of communication abilities beginning at 12 months have been documented in high-risk infants who develop ASD (Landa & Garret-Mayer, 2006; Yirmiya et al., 2006; Zwaigenbaum et al., 2005), the findings are not as consistent for the high-risk infants who do not develop ASD (e.g., Goldberg et al., 2005; Toth et al., 2007). As such, it is difficult to determine the basis for the group differences in our data at this time. However, findings from this high-risk population have yielded important insights into how language ability and behavioural signs of ASD relate to one another in the development of infants who are at heightened risk for ASD and/or language impairment.

Our results show that within the first year and a half of life, siblings who are at risk for ASD may be distinguished from siblings of typically developing children on the basis of lower expressive and receptive language and increased number and severity of behaviours that are consistent with ASD as measured by the AOSI. The finding that SIBS-A had significantly lower Expressive Language scores at both 12 and 18 months than SIBS-TD (see Table 2) indicate that overall, high-risk infants demonstrate divergent expressive vocabulary development over the first year and a half of life in comparison to our TD infant siblings. In addition to differences observed in expressive language, SIBS-TD also demonstrated significantly higher receptive language abilities at 18 months. Together, these findings corroborate evidence from studies that show increased incidents of language delay in siblings of children with ASD (see Bailey et al., 2001). There is a great deal of evidence of increased rates of language deficits among SIBS-A, according to measures of verbal reasoning and expressive and receptive language (August, Stewart, & Tsai, 1981; Bolton et al., 1994; Fombonne, du Mazaubrum, Cams, & Gramdjean, 1997; Leboyer, Plumet, Goldblum, Perez-Diaz, & Marchaland, 1995; Plumet, Goldblum, & Leboyer, 1995), as well as impairments in syntax, phonological processing, and pragmatics (Bishop, Maybery, Wong, Maley, & Hallmayer, 2006; Plumet et al., 1995). Findings from the current study suggest that there may be fundamental differences in early language acquisition, which may underlie language deficits observed in siblings of children with ASD (see Bailey et al., 2001). Again, differentiation of sibling subgroups at 24 months or later is necessary to determine whether group differences are being driven by those infants who later meet diagnostic criteria for ASD and/or a language impairment, or whether the high-risk infants who do not develop ASD also present with early differences.

Prior studies have shown that CDI measures of expressive vocabulary in infancy predict language outcomes through age 4 years (Feldman et al., 2005; Bornstein & Haynes, 1998). Performance on standardized vocabulary tests is highly correlated with performance on more general tests of intelligence (Dunn & Dunn, 1997). There is also evidence for associations between children's vocabulary development and phonological working memory (Gathercole & Baddeley, 1990). One study explored vocabulary size in 2-year-old children as it relates to later language, intelligence, and working memory (Marchman & Fernald, 2008). They found that vocabulary size (as measured by the CDI) at 25 months is predictive of language and cognitive ability at 8 years of age. This body of research suggests that children's early vocabulary is linked to cognitive and linguistic outcomes later in childhood (Bates & Goodman, 1999; Dixon & Marchman, 2007; Marchman & Fernald, 2007).

Attentional biases, such as to speech and other salient social information, are also an important factor in the development of language. Typically developing infants show communicative behaviours even from the first months of life, including recognizing their mothers' voice, synchronizing their patterns of eye gaze, movements, facial expressions of affect, as well as vocal turn taking (Fernald, 1992). Several studies have found that, as early as 12 months of age, young children with ASD are less responsive to their names or to someone speaking compared to other children (Lord, 1995; Osterling & Dawson, 1994), and they are less responsive to the sound of their mother's voice (Klin, 1991). High-risk infants siblings have also been shown to demonstrate fewer responses when they hear their name called (Brian et al., 2008; Nadig et al., 2007; Zwiagenbaum et al., 2005) and to bids for joint attention (Presmanes, Walden, Stone, & Yoder, 2007). Failing to participate in social exchanges may provide fewer communication opportunities for the infant. If this were the case, high-risk infants may show lower receptive and expressive language, as seen in the data presented. It is also possible that lower vocabulary in high-risk infants is associated with underlying differences in attention to language or maybe it is associated with social communication atypicalities that make it difficult for a child to attend to significant people or to participate in language learning interactions. Either way, a failure to participate in social and communication exchanges early in life may have detrimental effects on emerging communication skills.

While language and speech delays are the most common developmental issue that initially prompts parents to seek counsel from health care providers, other behavioural manifestations of ASD present early in infancy. Data obtained from the AOSI revealed group differences in early behavioural manifestations of ASD at 18 months. That is, SIBS-A had significantly higher AOSI scores at 18 months than SIBS-TD suggesting that high-risk infants demonstrate early manifestations of ASD related behaviours as measured by the AOSI in the first year and a half of life. Interestingly, not only are AOSI Total Scores different between groups, suggesting that the overall magnitude of the autistic-related behaviours is significantly higher in SIBS-A, but there are also a higher number of early markers observed in this high risk population. These data indicate that SIBS-A are demonstrating a greater range of ASD behaviours, which are also presenting as more severe than in SIBS-TD. During the first year of life, children with ASD present with many behaviours that are captured by the ASD measure used in the current study. For instance, infants later diagnosed with ASD demonstrate reduced social interaction, absence of social smiling, lack of facial expression, failure to orient to name, lack of pointing/showing, decreased orienting to faces, lack of spontaneous imitation, and abnormal muscle tone, posture and movement patterns (Adrien et al., 1992; Bernabei et al., 1998; Maestro et al., 1999; Mars et al., 1998; Osterling & Dawson, 1994; Zakien et al., 2000). Based on the literature and consistent with our data, there is considerable evidence that the impact of ASD can be evident beginning in infancy. However, it will be important to tease apart whether our SIBS-A demonstrate ASD-like behaviours that are early indicators for a later diagnosis. If SIBS-A who are typically developing at 24 months (and at later follow-up) were found to demonstrate these early behavioural manifestations of ASD, this would lend support to the broader autism phenotype account.

The correlations observed between autistic-like behaviours and later language suggests that early behavioural atypicalities may influence later language outcomes midway through their second year. ASD-like behaviours, such as those captured by the AOSI, may contribute to language development. For instance, early behavioural manifestations of ASD-like behaviours (such as atypical eye contact or reduced tendency to orient to faces) may not simply be correlates of later signs of ASD, but rather these behaviours may interfere with opportunities to learn from social experiences, which may have cascading effects on development contributing to further manifestations of ASD (Zwaigenbaum et al., 2005). Atypical development in social behaviour may also be mediated by early developing neural systems (e.g., visual orienting, engagement of attention) that may not be directly linked to later diagnostic characteristics of ASD but are fundamental to later social and communicative impairment (Zwaigenbaum et al., 2005). As previously mentioned, attention to social information might facilitate the acquisition of communication skills by increasing opportunities for engaging in communicative opportunities.

It is also possible that early language may influence the expression of atypical behaviours that are characteristic of ASD at 18 months of age. Impairment in the early developing mechanisms involved in language acquisition may beget deficits in other pro-social skills typically affected in individuals with ASD (Zwaigenbaum et al., 2005). Again, reduced opportunities to participate in social exchanges through communication have the potential to initiate an atypical pattern of development that eventually may lead to further manifestations of ASD. Our findings reflect the reciprocity in skill development between early manifestations of ASD and vocabulary development.

A final note on the data observed on the CDI-II Gestures domain. Gestures in infancy play a fundamental role in pre-linguistic communication and tend to develop before and in conjunction with early words (Acredolo & Goodwyn, 1985). As such, there is a strong association between the use of early gestures and language comprehension in early infancy in both typical and atypical development (Bates, Benigni, Bretherton, Camaioni, & Volterra, 1979; Thal, Tobias, & Morrison, 1991; Zwaigenbaum et al., 2005). However, gesture use in infants at-risk for ASD may in fact be more atypical in nature, rather than simply delayed. For example, it is possible that they may produce gestures (e.g., point) but do not direct them to a person, as is measured on the Autism Diagnostic Observation Schedules (ADOS; Lord et al., 2000). At 15 months of age, SIBS-A have been shown to use directed gestures at a lower rate and produce gestures that are not directed to others more often as compared to SIBS-TD (Mitchell, Roberts, Brian, & Zwaigenbaum, 2011). This research suggests that frequency and range of gesture use alone may not capture atypical patterns of early non-verbal communication in a high-risk population. While we did not observe group differences in our data, The CDI-II Gesture data provides information about gesture used based on dichotomous responses (e.g., “Yes/No”) for each gesture item. Thus, we were not able to assess how gestures are being used socially. That said, it may be pertinent to assess qualitative gesture use, in addition to the full range of non-verbal communication techniques used by SIBS-A.

In sum, our initial findings from a longitudinal study of infants at high-risk for ASD (siblings of children with the disorder) indicate that lower vocabulary and behavioural atypicalities that are characteristic of ASD may be observed in the first year and a half of life in SIBS-A. Our results indicate that by 12 months of age, siblings who are at increased risk for ASD may be distinguished from low-risk controls on the basis of expressive language. By 18 months of age, group differences also show up in receptive language and ASD related behaviours. The current study sheds light on how early ASD-like behaviours relate to language development in the siblings of children diagnosed with ASD. In the literature and in the current study, there is support for related skill development relative to the defining characteristics of

ASD. Specifically, our study provides evidence that this high-risk population demonstrates deficits in receptive and expressive language, as well as significantly more autistic-like behaviours that were also more intense in nature. These results provide some evidence that language may be related to later behavioural makers of ASD and vice versa, and may be helpful for determining which high-risk individuals would benefit from intervention prior to any outcome diagnosis. While this research provides evidence of early differences in ASD-related behaviours and language development (i.e., vocabulary) in SIBS-A, it does not yet address how these domains of development impact on one another. Furthermore, it does not speak to whether there are primary impairments in SIBS-A that are characteristic of the broader autism phenotype or whether there are early manifestations of ASD prior to a diagnosis.

Chapter Four: **General Discussion**

The findings in this dissertation indicate that infants at-risk for ASD exhibit subtle deviations in their perceptual preferences and their general behaviours from low-risk infants. That is, their preferences for visual stimuli are somewhat attenuated, their preference for ID speech is slower to develop, their vocabulary knowledge is generally lower, and they demonstrate more autistic-like behaviours. Results from the first study revealed that both low- and high-risk infants preferred looking at faces over checkerboards, but this preference was weaker in SIBS-A than in SIBS-TD. We further found that SIBS-TD showed earlier preferences for ID speech than SIBS-A did, but both demonstrated an ID speech bias at 18 months. Our high-risk siblings also exhibited lower expressive language scores at 18 months when compared with low-risk siblings. Taken together, these results reflect the importance of early attentional biases to ID speech that may give infants a leg-up in vocabulary development.

We found that language outcomes at 18 months correlated with early speech preferences when we collapsed the group data. This finding suggests that a preference for ID speech early in development may facilitate later expressive language at least in terms of vocabulary growth. In the SIBS-TD group, a relative preference for ID speech predicted CDI scores, while we found that in the SIBS-A group, a relative preference for faces over checkerboards in early development predicted CDI scores at 18 months. Finally, using logistic regression analyses, we found that infants' preference for faces contributed to determining group membership, while this was not the case when we looked at ID speech preference. Together, the findings from this study suggest that attending to and preferring speech directed at infants contribute to later expressive language abilities.

In our second study, we found that the SIBS-A group exhibited lower expressive language at 12 and 18 months as compared to SIBS-TD. They also demonstrated significantly more ASD-like behaviours (e.g., Number of Markers) that were also more severe in nature (e.g., Total Score) by 18 months of age. When we investigated whether relationships existed between ASD behaviours and vocabulary, we found that expressive language and ASD behaviours are significantly correlated with each other. While this relationship is not surprising given the characteristics that define ASD, it does provide converging evidence for the early emergence of atypical development in this high-risk population. Moreover, it corroborates the importance of exploring early speech and vocabulary development, and atypical behaviours in order to identify potential markers for ASD and to further understand the developmental progression of this disorder.

Several studies have shown that SIBS-A who do not meet diagnostic criteria for ASD at 24 months or later may nevertheless demonstrate other clinical abnormalities in their language development and ASD related behaviours. This is known as the “broader autism phenotype” (Bolton et al., 1994), which characterizes the presence of subclinical characteristics related to the core deficits of ASD and occur at an elevated rate in first-degree relatives of persons with ASD. For example, Gamliel, Yirmiya, and Sigman (2007) reported that 11 of 39 of their high-risk infants siblings demonstrated developmental impairments that were not identified as ASD at 24 months. Sullivan and colleagues (2007) also reported 8 out of their 51 SIBS-A sample as demonstrating developmental atypicalities at 36 months. There is general consistency in the literature that developmental differences in infants at high-risk for ASD appear by 12 months as demonstrated on standardized development measures (Brian et al., 2008; Gamliel et al., 2007; Stone, McMahon, Yoder, & Walden, 2007; Sullivan et al., 2007). It is unclear whether findings from the two studies presented in this thesis are attributable to early ASD presentation in SIBS-A. Until such a time when we are able to identify sub-cohorts of this high-risk sample (e.g., SIBS-A who do not receive a later diagnosis of ASD or language impairment; SIBS-A who receive a later diagnosis of ASD; SIBS-A who are later identified with a language impairment; SIBS-A who receive a later diagnosis of ASD with language impairment), we are unable to distinguish whether our findings represent sub-clinical features related to a broader autism phenotype or early symptoms characteristic of ASD. Furthermore, there is considerable range of severity in the various symptoms seen in children with ASD. Even in infancy, deficits in language, social skills, and/or behavioural symptoms may be affected to varying degrees at different developmental rates. As such, we cannot currently know what or who may be driving the effects observed in our studies.

4.1 Research in Language and ASD

Within the first years of life, typically developing infants begin to glean relevant social and emotional information necessary for meaningful social interactions (Fernald, 1992). During these stages of development, infants can acquire valuable social information from speech and faces that may be fundamental to the development of communication skills (e.g., Bushnell, Sai, & Mullen, 1989: Vouloumanos & Werker, 2007). Infants and children are exposed to meaning-relevant communication through face-to-face interaction as well as through observation of others' language use. This communication is crucial for participation in and understanding of social interaction, as well as learning about the world in general (Garvey, 1984; Ochs, 1986). It is also through such social interactions that infants and young children begin to learn about different aspects of language. For instance, adults begin to shape infants' developing language ability by talking to infants and responding to their babbling and cooing, which fosters reciprocal social communication (Bruner, 1975; Tomasello, 1992; Trevarthen, 1979). This social exchange provides an infant with early experience with the back-and-forth of dialogue that is important for full-fledged communicative competence.

Speech and language also facilitate the communication of intentions. A speaker's intentions can be conveyed through syntax and semantics within a given language, and also through other means, such as prosody. Prosodic cues normally used for conveying intentions in AD speech are frequently exaggerated when talking to an infant. ID speech is effective in eliciting infants' attention (Fernald & Simon, 1984; Werker & McLeod, 1989) and communicating affective intentions (Fernald, 1992; Fernald, et al., 1989), while also providing enhanced social information (e.g., Fernald, 1993) and promoting infants' language learning overall. For instance, Schachner and Hannon (2011) found that infants prefer to attend to speakers of ID speech, even during intervals when these speakers are silent, and thus concluded that infants prefer not just ID speech, but also the people who provide it. The authors speculated that this capacity compels children to socialize with adults that are most likely to provide care. Interestingly, children with ASD tend not to show a preference for ID speech, instead preferring to listen to a non-speech analogue (Kuhl, Coffey-Corina, Padden, & Dawson, 2005). Taken together, this research suggests that infants' preference for ID speech (e.g., Cooper & Aslin, 1990, 1994; Fernald & Kuhl, 1987) is helpful for language learning, as well as for promoting social exchanges between caregivers and infants.

By attending to the highly engaging features of ID speech that facilitate language learning, early preferences emerge that draw the infant into social communication, while they are concurrently learning about other relevant social information conveyed through face perception. For SIBS-A, however, this may not be the case. For infants who do not rely on early ID speech preferences to further expand and develop their vocabulary, other cues may be more supportive in their language learning. Our data indicate that while both low- and high-risk groups look equally at faces, SIBS-TD showed a greater relative preference for faces than did SIBS-A, suggesting a more robust finding amongst the SIBS-TD group. Indeed, looking time to the checkerboard visual stimuli was longer in the SIBS-A group than in SIBS-TD. Of note, our data highlighted differential attention to faces in SIBS-A, based on the finding that face preference in the SIBS-A group predicted CDI scores at 18 months and face preference was a significant predictor of group membership. Attention to species-specific social information, such as faces, may compensate for deficits in other biases that support to language learning. Our findings suggest that language learning is supported by both social and linguistic cues that are available to infant as the appropriate cognitive abilities emerge. That is, a highly socially based cues, such as faces, may be better at supporting the development of expressive language in high-risk infants while in low-risk infants, attentional biases toward faces and ID speech together facilitate vocabulary development. It's noteworthy that SIBS-A may rely on a different set of cues to support vocabulary development, while SIBS-TD demonstrated higher overall vocabulary at 18 months. These data have implications for language development and possibly social development given the inter-relatedness between the two simultaneously developing systems.

When considering these results and interpretations, it is important to consider the constitution of the SIBS-A group. While a subset of infant siblings will develop typically, approximately 1 out of 5 will receive a later diagnosis of ASD (Ozonoff et al., 2011), and around 1 in 7 will show deficits in language development (Ritvo et al., 1989), with or without a concomitant diagnosis of ASD. It is possible that those infants who receive a later diagnosis or display impairments in language development may be demonstrating atypical attentional biases.

Attentional biases, such as to speech and other salient social information, are an important factor in the development of language. Failing to participate in social exchanges may provide fewer communication opportunities for the infant. Indeed, several studies have found that, as early as 12 months of age, children with ASD are less responsive to their names or to someone speaking compared to other children (Lord, 1995; Osterling & Dawson, 1994), and they are less responsive to the sound of their mother's voice (Klin, 1991). That is, if infants do not respond appropriately to social input, they may have fewer opportunities to learn about language and engage in social interactions. There is evidence that infants with ASD manifest a range of abnormalities suggestive of profound limitations in social engagement (Wimpory, Hobson, Williams, & Nash, 2000). Wimpory and colleagues (2000) noted impairments in both one-on-one nonverbal communication, inclusive of interpersonal contact (e.g., eye contact, babbling with communicative intent, gestures, sociability in play) and in triadic person-person-object interactions (e.g., the referential use of eye contact, offering and giving objects, pointing at objects/following others' points). These findings complement other research demonstrating that infants with a later diagnosis of ASD showed less imitation of others, reduced engagement in social games (e.g., peek-a-boo), less checking for or being comforted by parents, fewer gestures (e.g., waving goodbye, pointing), and less interest in other children (Vostanis et al., 1998). Infants with ASD may also fail to greet their parents, wave to them, or make eye contact (Hobson & Lee, 1998), which reduces the number of opportunities to engage with others in social and communicative exchanges.

Research demonstrates that young children with ASD are generally rated lower on measures of social competence than typically developing peers, and in some cases, children with other disabilities (Lord, 1993). Furthermore, preschool age children with ASD have been observed to spend less time interacting with others as compared with typically developing children, and demonstrate lower-quality interactions during peer play. Interestingly, children with ASD were found to spend more time engaged in purposeless or no activity or nonsocial play, and/or at greater physical distances from peers (Lord & Magill-Evans, 1995; Sigman & Ruskin, 1999). Social interaction deficits for young children with ASD may reflect deficits in essential social skills (McConnell, 2002) that may be observed early in development. Overall, research demonstrates that while children with ASD make and receive fewer social initiations, respond to fewer of the initiations, and engage in shorter lengths of interaction, many of these children do participate in social interaction with peers (Kennedy & Shukla, 1995). However, interactions for many young children with ASD are not preferred activities. Often, children with ASD would rather spend time in isolate play, proximal on-looking, or other ASD related behaviours (e.g., repetitive play and motor movements, sensory-based investigation), which comprise much of their "free play" activities. Conversely, typically developing children are likely, and expected, to engage in social interaction. These responses decrease opportunities for social learning, and thus may affect development of social skills over extended periods of time.

Social interaction is only one way that children learn language. Infants also learn language by taking in and processing the language they hear even before they can speak (Benasich & Tallal, 2002). Initially, ID speech might serve to direct infants' attention and modulate arousal and affect, but by the second year ID speech can fulfil more specific linguistic purposes. The adjustments made to the ID speech signal occur at a range of levels, from the purely acoustic (e.g., higher pitch, exaggerated prosodic contours) to the semantic (e.g., emphasizing words new to infants), and grammatical (e.g., simplified sentence structure) (for a review, see Kuhl, 2004). There is ample evidence that such ID speech modifications benefit infants' ability to discover the underlying structure and meaning within language at all these different levels of analysis (e.g., Kemler-Nelson, Hirsh-Pasek, Jusczyk, & Cassidy, 1989; Liu, Kuhl, & Tsao, 2003; Ma, Golinkoff, Houston, & Hirsh-Pasek, 2011; Seidl & Johnson, 2006; Song, Demuth, & Morgan, 2010; Thiessen, Hill, & Saffran, 2005). However, recent evidence suggests that social factors play a crucial role in the process of learning phonology. For example, Kuhl, Tsao, and Liu (2003) provided 9-month-old English-hearing infants with input containing multiple instances of a sound contrast not relevant to English but relevant to Mandarin Chinese. Only infants who heard the contrast in a live, face-to-face interaction later showed evidence of being able to discriminate between the sound contrasts, whereas infants who had heard the contrasts in an audio-only format or watched a video of a Mandarin-speaking woman were not able to perceive the contrast (Kuhl et al., 2003). This finding demonstrates that speech input alone is not enough. The input must be embedded within a social context in order for infants to make use of it.

4.2 ASD Behaviour and Communication

Our objective to prospectively observe early developing ASD-like behaviours using a measure of ASD allowed us to determine the extent to which such behaviours were present in an at-risk population within the first year and a half. We found that SIBS-A demonstrated overall higher scores on our measure of early ASD behaviours, as well as elevated number of markers than did SIBS-TD. Furthermore, our measures of language and ASD specific behaviours correlated at both 12 and 18 months, reflecting an important relationship between expressive and receptive language and ASD behaviours. Our study supports evidence found in recent studies demonstrating early ASD related behaviours in an at-risk population. Longitudinal research has shown that by 12 months of age, siblings who are later diagnosed with ASD may be distinguished from siblings who are not later diagnosed and from SIBS-TD on the basis of a number of behavioural markers (e.g., atypical eye contact, visual tracking, disengagement of visual attention, orienting to name), as well as and delayed expressive and receptive language (Zwaigenbaum et al., 2005). The implications of these findings suggest that it may be possible to identify early markers and thus provide an early diagnosis and early intervention. However, findings presented in this research may also help tease apart whether there are core symptoms that contribute to the developmental cascade towards the broader phenotype of autism in infants who do not receive a later diagnosis. Indeed, it is important to monitor the development of these high-risk infants, as there is ample research that shows differences in the early presentation of ASD-like behaviours and language ability in this group.

Another recent study revealed a relationship between infants' preference for speech at 12 months and ASD-like behaviours (as measured by the AOSI; Bryson, Zwaigenbaum, McDermott, Rombough, & Brian, 2008; Zwaigenbaum et al., 2005) at 18 months in SIBS-A (Curtin & Vouloumanos, 2013). For low-risk SIBS-TD, a preference for speech correlated with language abilities but not ASD-like behaviours. Furthermore, when presented with speech and computer generated speech analogues (non-speech), SIBS-TD listened significantly longer to speech than to non-speech at 12 months (as in other studies, e.g., Vouloumanos & Curtin, under review; Vouloumanos & Werker, 2004, 2007). Conversely, SIBS-A did not listen reliably longer to speech. Based on their data, and other research that shows impaired attention to social stimuli in children with ASD (e.g., Bird et al., 2006; Chawarska et al., 2010; Dawson et al., 1998; Osterling, Dawson, & Munson, 2002; Whitehouse & Bishop, 2008), the authors speculate that SIBS-A may be more interested in non-social stimuli (non-speech) rather than being less interested in social stimuli (speech). These atypical species-specific biases for speech may underlie atypical social development in SIBS-A.

Infants' early communication attempts hold multiple functions, many of which are socially based, such as social engagement, requesting, sharing, or informing. These communicative patterns, which emerge at around 10 to12 months, reflect developmental connections between communication and social cognition (Carpenter, Nagell, & Tomasello, Butterworth, & Moore 1998). However, in children with ASD, communication skills are often used in order to gain or relay necessary information for learning about the world. That is, often in children with ASD, communication is primarily used for instrumental purposes (Tager-Flusberg, 1996). That is not to say that children with ASD do not use language to maintain some social interactions (Wetherby & Prutting, 1984), but rather that social communication is used differently and may be more limited. For instance, language may not be used to seek or share attention, comment on past or forthcoming activities, provide new information, or express intentions or mental states (Tager-Flusberg, 1992; 1993; 1997). Indeed, studies examining early aspects of language processing have revealed differences in social communication between preschool age children with ASD and typically developing children (Dawson, et al., 2004). This divergence in social communication skills in the ASD population may in fact emerge prior to preschool age,suggesting that it is imperative to monitor early language and social communication in infants at heightened risk for ASD.

The diagnostic criteria for ASD cluster around communicative, linguistic, and social deficits (DSM-IV; American Psychiatric Association, 1994), which are not only related, but appear to influence on another. A bias or preference for speech and faces is a first step in the development of social communication. By attending to the highly engaging features of ID speech that facilitate language learning, early preferences emerge that draw the infant into social communication, while they are concomitantly learning about other relevant social information conveyed through faces. A relative absence of such biases, or reduction in the time spent attending to it may negatively impact on social communication and language development. Conversely, by attending to non-social information (e.g., checkerboards, non-speech) may result in a relative reduction in amount of time spent attending to social information (e.g., face, speech; Curtin & Vouloumanos, 2013). This may then have cascading effects on the development of language and/or autistic like behaviours. Further to this, our findings revealed that vocabulary development at 12 months is related to the expression of ASD behaviours at 18 months. Thus, it is possible that early developing speech biases may indirectly influence and shape SIBS-A social communication skills through their effects on the development of expressive language. These findings suggest that early attentional biases to speech and faces are crucial for the acquisition of linguistic and social skills.

Little is known about early developing behaviours related to ASD and the individual preferences for speech and faces in children with ASD in their infant years that may predict their communication and language outcomes. It is important to understand the range of language abilities among these children and to what extent they may be associated with an underlying attention-to-language deficit or associated with social communication deficits that make it difficult for a child to attend to significant people or to participate in language learning interactions. In the current studies, we found that infants at risk for ASD relied perhaps more heavily on additional social cues that supported language learning due to a weaker reliance on speech specific cues such as ID speech. Further, infants' early ASD-like behaviours were related to their expressive language and vice versa. Early pre-linguistic, psychological, and/or social deficits and the relationships between them appear to contribute to atypical language development (Bruner, 1975; Tomasello, 1992). The research presented here revealed links between early preferences for faces and speech, as well as between expressive language and ASD behaviours in infancy, and has implications for early identification of language disorders and ASD. The current studies provide researchers, clinicians, and educators with longitudinal data obtained from two populations (high- and low-risk infants) on measures of language development and ASD specific behaviours, as well as preferences for socially relevant species-specific information, such as speech and faces.

Based on our longitudinal data, we provide prospective evidence that in the first year and a half, high-risk infants show delayed receptive and expressive language and behaviours consistent with ASD, as indexed by the CDI-II and AOSI. Information obtained from this research may aid in identifying behavioural markers and the developmental sequence to help with early identification. Prospective studies of high-risk infants may also identify novel behavioural markers that may contribute to the development of more effective and inclusive early identification and screening measures (Zwaigenbaum et al., 2005). For instance, we were able to identify atypical social attention in SIBS-A that may reflect deficits in social motivation or cognition. Indeed, a more in-depth investigation of individuals who do go on to receive a later diagnosis will aid in our understanding of ASD expression in those who receive a later diagnosis compared with typically developing relatives. However, these findings may be consistent with studies that show a genetic liability in unaffected relatives of individuals with ASD in the expression of features that are milder, but qualitatively similar, to the defining characteristics of ASD (see Bailey, Le Couteur, Palferman, & Heavey, 1998). Observing and identifying differences in this high-risk population early on will help to further our understanding of the early behavioural manifestations of ASD once a diagnosis has been made, and will help to identify any divergent patterns in development in our SIBS-A. Therefore, it is important to map relationships between skill development and deficits, as they will impact on the expression of associated skills.

4.3 Cognitive Theories of ASD

In conjunction with the growing body of literature focusing on bettering our understanding of ASD, several theories have emerged that attempt to account for the social and communication deficits and repetitive, stereotyped behaviours observed in ASD. These theories are based on behaviours observed in individuals with ASD. The current study examined precursors to these behaviours and the perceptual underpinnings that might influence vocabulary development and a set of behaviours characteristic of ASD. Indeed, development is a continuously unfolding process with later complex linguistic and social capacities building on early visual and auditory processing biases (e.g., Werker & Curtin, 2005). That is, ASD may be characterized by impairments in mechanisms that bias individuals from early infancy to orient towards, attend to, and assign value to socially relevant stimuli (e.g., Johnson, Dziurawiec, Ellis, & Morton, 1991). This early, basic impairment could form the basis of deficits in social communication skills that are characteristic of ASD (Surian & Siegal, 2008; Volkmar, Chawarska, & Klin, 2005). With this in mind, a complete discussion of the ample body of literature discussing theories of ASD is beyond the scope of this thesis; however, it's important to contextualize our findings in the broader theories of ASD.

4.3.1 Theory of Mind

A relatively well-known account describes a core deficit in theory of mind skills. The theory of mind framework attempts to explain pragmatic impairments of language and communication as they relate to social deficits. According to Baron-Cohen (1988), theory of mind rests on the premise that mental states are not directly observable, but have to be inferred, requiring a complex cognitive mechanism. This 'mechanism' allows a person to infer mental states, such as 'beliefs about something' and use them to explain and predict another person's behaviour. In individuals with ASD, the capacity for meta-representation is impaired, as are the pragmatic aspects of language usage (Walenski, Tager-Flusberg, & Ullman, 2006). Indeed, Volden and colleagues argue that a deficit in pragmatic language is integral to ASD (Volden, Coolican, Garon, White, & Bryson, 2009). This research revealed that children with ASD who demonstrated better pragmatic skills had fewer ASD-related communicative symptoms, suggesting a strong association between social communication and communication deficits in ASD. Thus, this cognitive theory proposes that the observed pragmatic deficits in ASD are those that would be expected if children with ASD use language without a theory of mind (Baron-Cohen, 1988).

According to Tager-Flusberg (2005), limitations in functional and social use of language in individuals with ASD are attributable to impaired understanding of other minds, which is arguably foundational for word learning (Baldwin, 1993; 1994; Tomasello & Farrar, 1986). Language acquisition rests on the child's interpretation of the speaker's use of words and gestures as intentional acts. Tager-Flusberg (2005) suggests that early word learning depends on this interpretation. When attaching meaning to novel words, the child must attend to both the speaker and the object in order to connect it to a spoken word – the label. This allows the child to fix the referent to the label. While this social attention (e.g., attention to language and the speaker) supports vocabulary development, it is but the first step. Indeed, semantic processes internal to language must contribute a great deal to word learning, while at the same time the syntactic context of words is also used by young children to demarcate the possible word class in which a new word resides (Golinkoff, Mervis & Hirsch-Pasek, 1994; Markman, 1994; Naigles, 1990). The social context of shared eye gaze and pointing delimit the possible meanings of a new word, suggesting that the theory of mind framework can be applied to the process of simple word learning early in childhood.

Further research exploring pragmatic language in children with ASD has speculated that theory of mind understanding is necessary for the comprehension of metaphor (Happé, 1993). Children with language impairments have poor vocabulary skills and impoverished semantic and conceptual knowledge necessary for metaphor comprehension. In this study, those individuals who were the most successful on the metaphor task demonstrated better language skills. This suggests that at least some children with ASD may have difficulty with aspects of pragmatic language and that their poor performance with metaphors may be due to problems with language rather than theory of mind. A more recent study examined semantic knowledge, theory of mind ability, and metaphor comprehension across groups of children with developmental language disorder, ASD only, and a combination of the two (Norbury, 2005a). Results showed that only children with language impairment, with or without concurrent ASD symptomotology, were impaired on the metaphor task. Furthermore, theory of mind skills examined in this study, which assessed first order knowledge (e.g., “John thinks X”), did not ensure metaphor comprehension. Instead, semantic ability was a stronger predictor of performance on the metaphor task. Thus, Norbury (2005a) posits that theory of mind alone is not sufficient for metaphor understanding, but rather language ability in general and semantic skills specifically are important for metaphor comprehension.

Most investigation of theory of mind development has focused on 3 to 4 year old children (e.g., Chandler, Fritz, & Hala, 1989; Fodor, 1992; Siegal & Beattie, 1991; Sullivan & Winner, 1993). Three year olds typically fail to recognize their own and other's false-beliefs (holding beliefs that conflict with reality; Wimmer & Perner, 1983). However, antecedents to theory of mind development may be evident in infancy. From 3 months of age, typical children can recognize different facial expressions (e.g., Barrera & Maurer, 1981; Labarbera, Izard, Vietze, & Parisi, 1976; Young-Browne, Rosenfeld, & Horowitz, 1977), but understanding the meaning occurs a few months later (e.g., Nelson, Morse, & Leavitt, 1979). Once young children are able to reliably interpret the facial expressions of others, they begin to use this nonverbal information to guide their behaviour (Campos & Stenberg, 1981; Klinnert, 1984). For example, a toddler may look at his mother's face for cues about whether it is safe to approach an unfamiliar person. Dawson and Osterling (1994) studied videotapes of first birthday parties of typical children and children who later received a diagnosis of ASD and found that the best predictor of future diagnosis was lack of attention to the face of others.

Our findings, presented in Chapters 2 and 3, explored preferences for ID speech and faces, vocabulary, and ASD related behaviours in general. While the age examined in both sets of studies and the tasks used do not speak directly to this theory, they do perhaps suggest that the precursors to theory of mind might be disrupted in the high-risk infants. For instance, data from the first study, discussed in chapter 2, indicates that both low- and high-risk infants look differentially to faces over checkerboards, but this preference was weaker in SIBS-A than in SIBS-TD. Lower looking time to faces my influence the amount of socially relevant information that is gleaned from faces. Thus, early precursors to theory of mind, such as attention to faces (e.g., Dawson & Osterling, 1994) develop differently in high-risk infants and may have down-stream effects the development of theory of mind in SIBS-A.

Other precursors to theory of mind that are impaired in children with ASD include joint attention (Tomasello, 1995), appreciation of intentionality (Tomasello, 1995), recognition that different people have different perspectives (Flavell, Everett, Croft, & Flavell, 1981), use of mental state words (Shatz, Wellman, & Silber, 1983), and pretend play (Astington & Jenkins, 2000). For children with deficits in these precursors and social attention, language learning is also severely compromised (Baron-Cohen, 1988). Of interest, studies have confirmed that language predicts later theory of mind. For instance, parent reports of vocabulary size at 2 years old have been shown to predict later theory of mind at age 4 years, using appearance-reality, unexpected contents, and change of location tasks (Farrar & Maag, 2002). Thus, language and theory of mind performance are related, suggesting that language (particularly vocabulary) is a precursor to theory of mind.

4.3.2 Central Coherence Theory

Frith (1989) proposed that ASD is characterized by a failure of a central system, which integrates sources of information (i.e., weak central coherence). This theory proposes that individuals with ASD are impaired in their ability to understand context because they attend to small pieces of information rather than large, globally coherent patterns of information. In short, children with ASD pay preferential attention to parts rather than wholes (Frith, 1989). This impairment in central coherence underlies the deficits in ASD and attempts to explain how some people diagnosed with ASD can show remarkable ability or skill in some areas (e.g., music, mathematics, engineering, drawing) yet have trouble with language skills and social interaction. For example, Happé (1997) found that individuals with ASD, including those who passed theory of mind tasks, do not use the preceding sentence context when interpreting the meaning of a word with one spelling but multiple meanings (i.e., homographs). That is, participants with ASD did not seem to integrate the sentence context to arrive at the correct interpretation of the word. Further research has shown that children with ASD display a selection bias towards the most common (context independent) interpretation of homographs (Frith & Snowling, 1983; Jolliffe & Baron-Cohen, 1999a) and ambiguous sentences (Jolliffe & Baron-Cohen, 1999b). This suggests that they are unable to use context to access the less common, but sometimes more appropriate, meaning. Thus, an alternative explanation for pragmatic deficits is that individuals with ASD experience difficulty when comprehending certain linguistic constructions not because of social inference difficulties per se, but because of a more pervasive inability to use context to derive meaning. Taken together, these studies suggest that language may in fact be more difficult for those with ASD due to impairments in integration of context relevant information.

Indeed, central coherence has been linked to difficulties with pragmatic conversational skills that are common in individuals with ASD (Noens & van Berckelaer-Onnes, 2005). Weak central coherence theory suggests that individuals with ASD do not use context to disambiguate linguistic material (Happé, 1997). However, recent research found that individuals with lower language scores demonstrate reduced sensitivity to sentence context, irrespective of a diagnosis of ASD (Brock, Norbury, Einav, & Nation, 2008). Norbury (2005b) explored whether deficits in contextual processing are specific to ASD or are subject to more general linguistic influences. Interestingly, deficits in contextual processing were not observed in individuals with ASD who had core language scores within the normal range. Only those with structural language impairment show evidence of weak central coherence in the verbal domain. These results suggest that language ability, as opposed to symptoms related to ASD, is more closely tied with contextual facilitation and suppression of irrelevant information.

Interestingly, Jarrold, Butler, Cottington, and Jiminez (2000) speculate that central coherence may contribute to the development of theory of mind such that it biases the individual to take on a global view of a situation, and to integrate the perspectives of multiple people within that situation. For instance, Jolliffe and Baron-Cohen (1999a) found that individuals with ASD can answer whether a statement is true or false, but not explain this same statement within the context of a story. The authors speculate that this finding suggests that the theory of mind deficits may result from a difficulty with contextual integration.

While the findings in this dissertation do not directly speak to whether weak central coherence is one of the processing difficulties in individuals with ASD, it is possible that early perceptual atypicalities underlie the overall processing of linguistic information. Indeed, it would be interesting to explore whether discrimination of different prosodic contexts is different in infants at risk for ASD and whether this relates to their later ability to interpret pragmatic cues.

4.3.3 Executive Functioning

Executive function is an umbrella term for functions including initiating, sustaining, shifting and inhibition/stopping (Denkla, 1996). Executive function relies abilities that allow us to plan and problem solve, such as impulse and inhibitory control, set maintenance, organized search, and flexibility of thought and action (Ozonoff, Pennington, & Rogers, 1991). The Executive Function theory posits that individuals with ASD have problems with many aspects of executive function. For example, individuals with ASD may show symptoms such as a need for sameness, a difficulty switching attention, a tendency to perseverate, and a lack of impulse control, which all require executive function abilities (Baddeley & Wilson, 1988). Because the executive function hypothesis is so multifaceted, it can account for many of the non-social aspects of ASD, while also speaking to the cognitive and motor (repetitive hand clapping, rocking) characteristics often observed in ASD.

Findings with individuals with ASD are mixed in terms of their executive functioning abilities. Ozonoff and colleagues (1991) found that neuro-typical controls performed better than a large majority (e.g., 96%) of their ASD group on executive function tests. In contrast, Pellicano, Maybery, Durkin, and Maley (2006) reported executive function deficits in only half of their ASD group. Studies have found planning impairments in children and adults with autism when examining planning abilities (Bennetto, Pennington, & Rogers, 1996; Ozonoff & Jensen, 1999; Ozonoff & McEvoy, 1994; Ozonoff et al., 1991). However, Mari, Castiello, Marks, Marraffa, and Prior (2003) found that planning ability was related to IQ rather than ASD per se in a kinematic reach-to-grasp task. On inhibition tasks, individuals with ASD display similar problems of interference as controls in two studies investigating the Stroop effect (Eskes, Bryson, & McCormick, 1990; Ozonoff & Jensen, 1999). However, on a different inhibition task used in Russell, Mauthner, Sharpe, and Tidswell's (1991) study, individuals with ASD demonstrated problems inhibiting automatic responses. Similar results were observed on self-monitoring tasks, which tap into the ability to monitor one's own thoughts and actions and to adjust one's behaviour accordingly, with no differences shown between individuals with and without ASD (e.g., Hill & Russell, 2002; Russell & Hill, 2001).

Overall, there is variability in findings of executive function studies with an ASD population. In fact, researchers have sometimes failed to replicate results of others even when using identical tasks and methodologies (see Pennington & Ozonoff, 1996, for a review). Not all individuals with ASD show executive problems and those who do may have differing profiles of executive function abilities. Moreover, executive difficulties are not unique to ASD and are seen in other disorders (e.g., Attention Deficit/Hyperactivity Disorder; Ozonoff & Jensen, 1999). Some researchers have even argued that executive abilities may even be related to theory of mind (e.g., Hala & Russell, 2001; Russell, Hala, & Hill, 2003; Russell et al., 1991). The data presented in this dissertation does not directly address this theory of ASD, as executive function abilities were not assessed. It may be of interest to examine social attention as it relates to later executive function tasks, given findings that there is a relationship between social behaviours and executive function (McEvoy, Rogers, & Pennington, 1993). It is possible that factors associated with perceptual abilities and language skills specifically affect the development of some regions of the brain associated with the development of executive function.

4.3.4 Social Motivation

A recently presented theory of ASD describes underlying deficits in social motivation (Chevallier, Kohls, Troiani, Brodkin, & Schultz, 2012). The authors propose that motivational deficits have cascading effects on the development of social cognition, and deficits in social cognition are therefore a result, rather than a cause, of disrupted social interest. The premise rests on observations that social interactions are intrinsically reinforcing; objects with social importance are prioritized by attention; and there is a biologically driven desire to maintain and enhance relationships that significantly influence interpersonal behaviours. Individuals with ASD, however, demonstrate atypical social behaviours that reflect a notable lack of attentional weight allocated to social information (Chevallier et al., 2012).

Highly relevant social signals, such as faces and eye contact, are effective in securing attention in typically developing individuals (Senju & Johnson, 2009). These signals help in distinguishing gender, facial affect, and identity, as well as gleaning meaning from social cues

(Cohen & Tronick, 1983; Corkum & Moore, 1998; Pascalis, De Haan, Nelson, & De Schonen, 1998; Scaife & Bruner, 1975; Tronick, 1989). Moreover, typically developing infants will often smile in response to social smiles, suggesting an internally reinforcing mechanism that also sustains social interactions (Berger, 2006). Sustained social interactions often lead to language exchanges, regardless of the infants' language level. Caregivers will talk to their infants and elicit verbal responses in return as a part of social exchanges that also promote language (Bruner, 1975; Tomasello, 1992; Trevarthen, 1979). Reciprocal social smiling, for example, entails social attention, internal reward, and social maintenance (Chevallier et al., 2012), which in turn leads to language exchanges (Berger, 2006), all of which appear to be lacking in ASD (Dawson et al., 1998; Mundy & Neal, 2001).

Within this framework, one could argue that our high-risk infants do not demonstrate the same motivational factors necessary to direct attention to social information, as seen in measures of decreased attention to faces and ID speech. Diminished social orienting and engagement are characteristic of children with ASD (Dawson et al., 2004) and have been observed in infant siblings children with ASD (Zwaigenbaum et al., 2005). However, it is difficult to gauge motivation in infants using our measures, so while our findings might be consistent with this theory, they cannot explicitly speak to whether motivation or interest underlie their performance on these tasks.

No single theory can integrate the socio-linguistic, perceptual, and sensorimotor aspects of the disorder. For one theory to account for all the symptom domains that are characteristic of ASD, it would have to apply from birth, through infancy, to adulthood and across a range of abilities on the spectrum. Although the data presented throughout this dissertation do not directly speak to these theoretical accounts of ASD, it is possible that early developing atypicalities underlie deficits in social communication skills that are characteristic of ASD (Surian & Siegal, 2008; Volkmar et al., 2005). In turn, data on early developing perceptual abilities, communication, and ASD-related behaviours lends itself to research with a focus on identifying early markers of ASD for the purpose of developing early diagnostic measures.

4.4 Clinical and Practical Implications

Given the range of language abilities in individuals with ASD and their younger counterparts, it is imperative that we attempt to understand the source of this difficulty. Deficits may be associated with an underlying perceptual deficit or with social motivation or interest, the presence of either or both making it difficult to form connections with people and also to one's surroundings. Early language and verbal ability are strong predictors of later language among children with ASD (Smith, Mirenda, & Zaidman-Zait, 2007). Children who are able to use language by age 5 to 6 years have a much greater chance of achieving levels of functionality that allow for greater development than those who do not possess this skill (Howlin, Goode, Hutton, & Rutter, 2004). Thus, understanding and exploring the nature of the underlying problem will help researchers develop appropriate tools for facilitating and screening of language development.

There is increasing evidence that early intervention improves the outcome of children with ASD (Alberta Children's Services, Expert Panel Report, 2002). The earlier a diagnosis can be made the sooner a child can receive appropriate intervention. Given the increased risk of sub-clinical ASD symptoms expressed in SIBS-A and/or of a later ASD diagnosis, it is important for families to have information about normal development as early as possible. The identification of early markers that help to predict outcome will assist infants and their parents in obtaining appropriate services so that screening and support can begin.

Evidence that signs of social attention, communication, and language delays can be present as early as 12 months in high-risk infants has important clinical implications. Due to the wide range of language abilities in typical development, clinicians may not be immediately concerned when a child presents with delays in language at 12 to 18 months of age. Our findings suggest that it may be prudent to monitor infants who are at elevated risk for developing ASD more closely. Consistent with our findings, high-risk infants present with lower vocabulary, divergent social attention, and atypical behaviours associated with ASD. Regardless of whether infants are at heightened risk for ASD, this research provides evidence that early language abilities, specifically vocabulary size, are related to later expression of ASD behaviours, and conversely, that ASD behaviours that may interfere with social learning opportunities earlier in development are related to later deficits in language development. These findings suggest that monitoring the progress of high-risk infants may be valuable as soon as concerns arise within the developmental domains associated with ASD.

Several symptom areas were examined in this dissertation that have direct implications for studies seeking to identify early markers of ASD. Findings from both studies above suggest that early attention to socially relevant information is related to vocabulary size, which in turn is related to ASD behaviours. These results are consistent with the prospect that perceptual and language atypicalities in a high-risk population may have cascading effects on the development of behaviours related to ASD, and these effects may be observable prior to 2 years of age. Such findings contribute to a broader understanding of the onset and developmental progression of specific symptom domains of ASD. Given the increased risk of sub-clinical ASD symptoms expressed in SIBS-A and/or of those for whom a later ASD diagnosis, it is important for families and practitioners to have information about atypical development as early as possible. Data from retrospective sources have provided insights into early behavioural manifestations of ASD (e.g., Werner, Dawson, Osterling, & Nuhad, 2000; Baranek, 1999; Osterling & Dawson, 1994). Indeed, findings from these studies have formulated current practice in early identification (Filipek et al., 2000). However, there are some limitations to retrospective parent reports, as discussed in Chapter 3. Thus, prospective studies, such as those presented in this paper, have implications for research into diagnostic tools for earlier identification.

There is increasing evidence that early intervention improves the outcome of children with ASD (Alberta Children's Services, Expert Panel Report, 2002). The identification of early markers that help to predict outcome will assist infants and their parents in obtaining appropriate services so that screening and support can begin. The earlier a diagnosis can be made the sooner a child can receive appropriate intervention. However, clinical detection of ASD is still difficult prior to 24 months. At present, there are very few diagnostic instruments for children younger than 24 months (Luyster et al, 2009; Zwaigenbaum, et al., 2005), and those that do assess early ASD in very young children are still in development. Currently, the clinical implications of this dissertation research are more directly relevant to the research community than in clinical assessment, and will likely be more beneficial to the practitioner once early diagnostic instruments can be developed based on findings from studies such as these.

While we acknowledge that this high-risk group is a heterogeneous population, our results are striking in that we were able to detect perceptual preferences, language skills, and ASD-related behaviours that differed significantly from our low-risk controls. Attempting to understand the source of this variability by identifying which group of infants may be driving the effects (i.e., SIBS-A who do not receive a later diagnosis of ASD or language impairment; SIBS-A who receive a later diagnosis of ASD; SIBS-A who are later identified with a language impairment; SIBS-A who receive a later diagnosis of ASD with language impairment without ASD symptoms) will help inform our understanding of possible underlying mechanisms that contribute to ASD and the overt behaviours observed in this population. For instance, one might further explore possible differences in the kinds of words that low- and high-risk infants first produce and/or attend to, their early speech preferences in the absence of a visual display that may modulate possible biases (and vice versa), or behaviours that are specific to a later diagnosis through an item analysis on the behavioural measure of ASD. This body of longitudinal research is the first step toward understanding early behaviours related to ASD, their developmental sequence, and the relationships between impairments in early presentation and later behavioural manifestations.

Once we have a clearer picture of the early behavioural markers in a high-risk population, parents and clinicians will be better able to identify those specific behaviours that arise early in childhood and are indicative of ASD. This may be particularly useful since parents of children with ASD may be highly attuned to early developmental signs that were present in their older child prior to their diagnosis, and/or may overlook certain early markers if they are in denial about the possibility of their younger child presenting with ASD symptoms. Finally, there is a range of what constitutes typical behaviours in infancy, which may make it difficult to identify what may be typical or atypical development. Thus, by examining low- and high-risk infants, information related to the range of variability exhibited in the typically developing population will help us to further hone in those atypical patterns of behaviour specific to ASD. This information will help parents and clinicians consider and monitor more reliably those behaviours related to ASD.

4.5 Limitations of the Current Studies

The longitudinal, rather than cross-sectional, design of both studies has limitations as well as strengths. The population we examined is specific, thus making it difficult to enrol a large sample size, which resulted in small subgroups. We enrolled participants at 6 months of age and visits occurred at 8, 12, and 18 months of age in the first study, while the second study required parents to the visit the lab twice and complete parent questionnaires at both 12 and 18 month visits. As a result, infants did not always complete all the tasks and, at times, parents failed to complete or return the CDI-II forms. This resulted in missing data and a relatively small final sample. Due to this, protocols in the Speech Development Lab have changed so that parents fill out the CDI-II forms on site before they leave to ensure more complete CDI-II data sets.

The use of a parental report of language development may have been problematic in this specific population. The reports of parents of children with ASD regarding their unaffected sibling may be biased to either maximize or minimize any potential language difficulties, because of having a child with ASD as a reference. This bias is unlikely to be manifested in the SIBS-TD group introducing potential differences in reliability between groups. Future studies may benefit from the use of other standardized assessments (e.g., Peabody Picture Vocabulary Test, Expressive Language Test, Preschool Language Scale) to support data collected from parent reports. However, many of these formal assessments are limited by the age at which they assess language skills. Further examination of the Expressive and Receptive Language scales from the Mullen Scales of Early Learning (MSEL; Mullen, 1995) may support data from parent reports.

The studies also lacked additional comparison groups, such as a group of siblings of children with learning disorders or language delays. Such a comparison group would allow us to examine whether the potential effects of heightened caregiving demands of first-born siblings impacts the needs of later-born siblings. Additionally, a supplemental clinical population would have contributed to a broader understanding of potential early speech biases specific to ASD, which may differ from language and learning disorders. Observing other high risk populations, such as infants born prematurely (Johnson et al., 2010; Juul-Dam, Townsend, & Courchesne, 2001; Kolevzon, Gross, & Reichenberg, 2007; Limperopoulos et al., 2008; Schendel & Bhasin, 2008) may allow for a larger sample size of high risk infants, as well as observations of perceptual biases and language development in a population at risk for other health and developmental issues. That is, developmental trajectories may differ in children who go on to receive a diagnosis of ASD from those who present with language impairments, learning disorders, or other developmental problems.

Finally, our participants were not yet of an age where ASD diagnosis could be made. However, the likelihood of infants in our high-risk group receiving an ASD diagnosis is approximate 19% (Ozonoff et al., 2011), suggesting that just less than 20% of the infants in our study will go on to receive a diagnosis of ASD. A small proportion may be misidentified as having ASD. Conversely, we anticipate that some of the children who do not meet criteria on the ADOS at 24 months will be diagnosed subsequently with ASD. Furthermore, some children will present with a language impairment unrelated to ASD. Following a larger sample until 3 years of age when diagnoses could typically be made would allow us to look at the relationships between preferences for speech and/or faces, language, and ASD behaviours once a diagnosis was made (Mandell, Novake, & Zubritsky, 2005; Ozonoff et al., 2009). The small sample size and young age of the participants makes it difficult to draw any conclusions about how the factors observed (e.g., preferences for face/speech, expressive and receptive vocabulary, ASD-like behaviour) may predict ASD diagnosis. Future research with the ASD population in our lab will continue to assess language development, speech biases, and potential early markers of ASD as standardized measures of ASD severity and later language development are completed, and data from experimental tasks examining early speech preferences is gathered. As infants develop, we will continue to investigate whether SIBS-A who go on to receive an ASD diagnosis differ from those who show language impairments (but not a diagnosis of ASD). Furthermore, data from SIBS-A who go on to receive a diagnosis will be examined as it relates to data from SIBS-A who fail to develop these symptoms and appear to be typically developing. Future research is warranted to document more systematically the early developmental trajectories of children at high risk for either ASD or language impairment to draw more definitive conclusions about the communication and language profiles that distinguish them from both typical children and those with other developmental disorders

4.6 Conclusions

Our findings lend support to the notion that as infants begin to integrate speech signals with other sensory input, information conveyed through the face becomes more salient (Fernald, 1992). Specifically, our TD infants appear to preferentially attend to faces as they learn about socioemotional meaning, while at the same time developing a robust preference for the emotionally laden, highly prosodic ID speech. This research provides support for the role of early perceptual biases in directing attention to linguistic and socially relevant information. Attention to such species-specific information may underlie some of the social communicative deficits observed in children with ASD. Further to this, we found that high-risk infants demonstrate difference in their expressive and receptive vocabulary, as well as significantly more ASD-like behaviours that were also more severe in nature. When we investigated whether relationships existed between ASD behaviours and vocabulary, we found that expressive and receptive vocabulary is significantly correlated to ASD behaviours. These results are consistent with the premise that early perceptual and language atypicalities in a high-risk population may have cascading effects on the development of behaviours related to ASD that may be observable prior to two years of age.

The current studies highlight the importance of language skills and social attention as they relate to the development of ASD behaviours. Thus, in conjunction with data accrued across the infant sibling literature, our study lends further support to the exploration of early atypical perceptual preferences and patterns of behaviour in order to understand the progression of this disorder.

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