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Joint Attention and Early Social Developmental Cascades in Neurogenetic Disorders

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Abstract

This review examines what is known about joint attention and early social development in three neurogenetic syndromes: Down syndrome, Williams syndrome, and fragile X syndrome. In addition, the potential cascading effects of joint attention on subsequent social development, especially social interaction and social cognition are proposed. The potential issues and complexities associated with conducting prospective, longitudinal studies of infant social development in neurogenetic disorders are discussed.

Keywords

Social Development; Joint Attention; Neurogenetic Disorders; Down syndrome; fragile X syndrome; Williams syndrome; Developmental Cascades

The presentation of phenotypic outcomes in neurogenetic disorders has received significant research attention in the last several decades. Behavioral phenotypes are the measureable behavioral outcomes observed in individuals with neurogenetic disorders (Dykens, 1995; O'Brien, 1992). According to Dykens (1995), a behavioral phenotype is conceptualized as “the heightened probability or likelihood that people with a given syndrome will exhibit certain behavioral or developmental sequelae relative to those without the syndrome” (p. 523). Therefore, within a specific neurogenetic disorder, there are behaviors that are more probable, but these behaviors may not emerge for all individuals with that disorder (Dykens, 1995; Hodapp & Desjardin, 2002; Hodapp, 2004). In addition, while certain behaviors may be associated with a specific syndrome, they may not be completely unique to that syndrome (e.g., sociability and friendliness in Down syndrome and Williams syndrome; Dilts, Morris, & Leonard, 1990). As the field has advanced, more nuanced investigations have been conducted that provide deeper insight into and understanding of the behavioral phenotype associated with syndromes such as Down syndrome, Williams syndrome, and fragile X syndrome, as well as the potential implications of these behavioral phenotypes for other aspects of development and on later development.

One domain that has been of great interest to researchers who study neurogenetic disorders is social development, particularly aspects of social cognition. Social cognition is the ability to think and reason about the social world (Tager-Flusberg, Skwerer, & Joseph, 2006;

Trevarthen & Aitken, 2001). Like some other domains of development, social development starts in the very first days of life. Interestingly, only a limited amount of research has been conducted on early social development in neurogenetic disorders, despite recognition from the field of the importance of examining the early developmental profiles associated with neurogenetic disorders or starting states (Karmiloff-Smith & Thomas, 2003; Karmiloff-Smith, 1998). A greater understanding of early social development and social starting states in neurogenetic disorders can shed light on how social phenotypic profiles emerge and develop over time. To conceptualize how such behavioral profiles may emerge over time, Fidler, Lunkenheimer, and Hahn (2011) proposed the notion of using dynamic systems theory and developmental cascades.

Dynamic systems theory provides a useful framework for conceptualizing how development occurs over time (Granic, 2005; Thelen & Smith, 2006). According to this approach, developmental outcomes can be understood as a product of a process called self-organization. Self-organization is the emergence and consolidation of new forms (e.g., behaviors) that comes from interactions between simpler forms (Lewis, 1997, 2000). Within the context of self-organization, emergence is defined as the ongoing process of the “coming-into-existence” of new forms or properties within a system (Lewis, 2000). Therefore, emergence can be used to understand change and novelty in natural systems. The process of self-organization leads to change and growth (Lewis, 2000) and this organization can involve the interaction of genes, brain, behavior, and environment (Howe & Lewis, 2005). This framework aligns with the neuroconstructivist approach that has been used to conceptualize development in neurogenetic disorders as a dynamic, ongoing process of atypical brain development resulting in different developmental patterns and pathways (Karmiloff-Smith & Thomas, 2003; Karmiloff-Smith, 1998, 2006). Based on these perspectives, development occurs when a new form or behavior emerges, and in turn it may cause change in other areas or systems either directly or indirectly (Karmiloff-Smith & Thomas, 2003; Lewis, 1997).

One way to conceptualize these direct and indirect effects that occur in human development is through the analogy of developmental cascade. Developmental cascades are the process by which early functioning in a domain influence, or have a cumulative effect, on the development of other domains of functioning over time (Masten & Cicchetti, 2010). Thus, a self-organization framework may help researchers understand how emerging behavioral phenotypes shape development, and has cascading effects on development in multiple domains throughout the life course in neurogenetic disorders (Fidler et al., 2011).

Early Social Development

The first signs of social interest and understanding develop during infancy. This is also known as intersubjectivity, which is conceptualized as the intuitive recognition and understanding of the impulses and desires of another's mind (Trevarthen & Aitken, 2001). Intersubjectivity is divided into primary intersubjectivity and secondary intersubjectivity. Primary intersubjectivity occurs when infants consolidate the skills of maintaining mutually regulated, dynamic interactions and show an active and responsive appreciation of the communications of their social partners, usually their caregivers (Trevarthen, 1979). Thus

primary intersubjectivity provides infants with the ability to participate in mutually regulated, dynamic interactions. This in turn, supports the ability to incorporate objects and events outside the social interaction into the dynamic interaction, which leads to secondary intersubjectivity. Secondary intersubjectivity occurs when infants show dynamic awareness for both people and objects or person-person object awareness (Trevvarthen, 1998; Trevvarthen & Hubley, 1978). Together, the development of primary and secondary intersubjectivity provides infants and young children with the necessary foundational skills to function in the social world (Meltzoff & Moore, 1994, 1998; Trevvarthen & Aitken, 2001). There are many skills associated with both primary and secondary intersubjectivity that are important for social development, however, one skill associated with secondary intersubjectivity that seems to be particularly critical for social development is joint attention.

Joint Attention

Joint attention is a uniquely human skill (Tomasello, Carpenter, & Hobson, 2005) that involves shared attention between an infant and social partner around an object or event of interest (Mundy et al., 2003; Wetherby & Prizant, 2003). Because of the dynamic nature of joint attention, researchers have found it helpful to define instances of joint attention as either initiating or responding (Bruinsma, Koegel, & Koegel, 2004; Mundy & Newell, 2007). Initiating joint attention involves the infant seeking the attention of a social partner to start the interaction, while responding to joint attention involves responding to a social partner in order to maintain the interaction. In the field of intellectual and developmental disabilities, there has been a great interest in the role that joint attention impacts social development.

Joint engagement is another term that is used to describe periods of joint attention between an infant and a social partner (Adamson, Bakeman, & Deckner, 2004; Adamson & Chance, 1998; Bakeman & Adamson, 1984). Joint engagement adds additional complexity to the study of joint attention by differentiating the emergence of joint attention, referred to as supported joint engagement, and the consolidation of the skills needed to actively use joint attention in interactions with social partners, referred to as coordinated joint engagement (Adamson & Chance, 1998). Specifically, supported joint engagement occurs when an infant and a social partner are engaged with the same object, but the social partner is scaffolding the interaction and the infant is not actively and reciprocally responding to the social partner (Adamson et al., 2004; Bakeman & Adamson, 1984), while coordinated joint engagement is when the infant is actively engaged with the object and the social partner in a dynamic and reciprocal interaction (Adamson et al., 2004; Bakeman & Adamson, 1984). Therefore, during supported joint engagement, the ability to share attention with a social partner is just emerging for the infant and needs support in order for this skill to fully develop into the active, reciprocal process that is characteristic of joint attention. Regardless of the term, both joint attention and joint engagement involve a consolidation of the other skills associated with intersubjectivity (i.e., using eye gaze and affect to share interest with a social partner in a dynamic interaction), making joint attention a critical skill for infants because its emergence and development indicate a shift in the way infants interact with the world, especially the social world (Hubley & Trevvarthen, 1979; Meltzoff & Moore, 1994, 1998; Trevvarthen & Hubley, 1978). Furthermore, joint attention continues to influence social

interactions throughout the life course (Brazelton, Koslowski, & Main, 1974; Stern, 1974; Tronick, 1982).

In order to share attention and communicate interest to social partners, infants use a combination of eye contact, vocalizations, facial expressions, and gestures (Bates, Benigni, Bretherton, Camaioni, & Volterra, 1979). Most of the time these behaviors are combined to provide the most information to the social partner and they can be used either to initiate an interaction with a social partner or respond to a social partner. For example, an infant may use eye gaze and a vocalization to indicate that they see a toy their caregiver is showing them (i.e., responding to joint attention). Or, an infant may look to her caregiver, vocalize to get the caregiver's attention, and then with her eyes direct the caregiver to a toy while smiling and vocalizing (i.e., initiating joint attention). The infant will then look back to the caregiver to see if they see the toy too. However, if there are many toys in the room it may not be clear to the caregiver what the infant is interested in. The addition of a gesture, especially a pointing gesture, will help to clarify this.

Pointing can have two broad functions: to direct attention (i.e., a protodeclarative point), and to control or manipulate the behavior of others (i.e., a protoimperative point; Bates et al., 1979; Bates, Camaioni, & Volterra, 1975). Protodeclaratives usually take the form of a point, or an approximation of a point (i.e., hand or several fingers extended), and can be either proximal (close to the infant) or distal (far away from the infant). For example, an infant may notice leaves moving in a tree and think the movement is interesting leading the infant to point up at the leaves to share her excitement with her caregiver, as if to say, "Hey! Do you see these things moving above us?" An infant may also use a point as a protoimperative to tell the caregiver that she wants more milk by pointing to the milk carton on the counter. Pointing falls within the category of deictic gestures, which are used to direct attention or reference an object or event. Other deictic gestures include reaching, pushing, giving, and showing. All of these gestures can be used to share interests and attention with a social partner. To initiate an interaction an infant may give a caregiver a ball in order to start a back and forth play routine or they may show them an object found in the sandbox that is unfamiliar to the infant. Or if a caregiver notices their infant with a ball, the caregiver may say, "Oh! Do you have a ball?" leading the infant to look at the caregiver and show the ball.

Collectively, joint attention becomes integrated into other skill sets in order to support the development of the more complex social understanding that is needed for functioning in everyday life (i.e., social interaction, communication, learning, etc.) throughout the life course in a variety of contexts (e.g., school, work, home) and in many different social situations (e.g., making friends, dating, interviewing for a job, etc.). For example, we use joint attention skills to coordinate our attention to the topic our teacher is discussing in class (Mundy & Sigman, 2006), or to share in the excitement of a friend receiving a good grade (Adamson, Bakeman, & Deckner, 2004; Tomasello, 1995). Both of these examples also depend on social cognition, which is supported by the development of joint attention. Specifically, through the development of joint attention, young children begin to attribute emotions, desires, and intentions to the actions people produce (Trevvarthen & Aitken, 2001). This in turn leads to the understanding that people are intentional beings that have their own thoughts and minds (Meltzoff & Moore, 1998; Tager-Flusberg, 2005). Therefore, the

behavior of others can be interpreted in terms of mental states (Tager-Flusberg, 2005). This, in turn, leads to the development of theory of mind or the ability to understand, or to theorize about, what another person is thinking and take their perspective—a key component of social cognition (Tager-Flusberg & Skwerer, 2007; Zinck, 2008).

Taken together, then, joint attention is hypothesized to have a cascading influence (see Fidler et al., 2011; Karmiloff-Smith, 2011), on later social interactions and social cognition (Bukowski, Newcomb, & Hartup, 1998; Carpendale & Lewis, 2006; Mundy & Newell, 2007). Successfully achieving joint attention can promote development in these domains, while frequent and repeated disruptions in achieving joint attention can lead to adverse effects in these domains (Murphy & Abbeduto, 2005).

Early Social Development in Neurogenetic Disorders

Considering the importance of joint attention on development, the question that arises is, what happens when these skills do not develop in the manner that is expected? Specifically, how does this influence the trajectory of development and what are the implications of typical or atypical joint attention on later development? It is speculated here that disruptions in joint attention can lead to not initiating as many interactions with a social partner and not always responding in a manner that helps to sustain the interaction, which results in fewer opportunities to participate in a social exchange and gain information about social interactions. The cumulative effect of these limited social experiences over time will result in a lack of the social knowledge needed to respond and participated appropriately in social situations throughout the life course. Thus, it is important to conduct nuanced examinations of joint attention to understand the starting state of these skills, followed by examinations of how they develop and change over time. This will allow for unique insight into the cascading effects of joint attention related to the social phenotype of a specific neurogenetic disorder. Additionally, this type of prospective characterization of behavioral phenotypes has the potential to provide meaningful implications for the timing and targets of early interventions.

Three neurogenetic disorders that are particularly relevant for examining early social developmental cascades are Down syndrome (DS), Williams syndrome (WS), and fragile X syndrome (FXS). For all three neurogenetic disorders there is a critical need to examine the emergence of joint attention. The social phenotypes of these three neurogenetic disorders suggests that while there may be areas of overlap, there are also syndrome-specific patterns of social development. In addition, these three syndromes can be viewed on a continuum from developmentally appropriate competence in social skills (DS) to dysregulated social behavior (WS) to impaired social skills (FXS). In the following sections, I provide information on what is known about joint attention and early social development in these three syndrome groups, and identify areas where more research is needed to inform future intervention and treatment work.

Down Syndrome

DS is the most common and recognizable neurogenetic disorder associated with intellectual disability (incidence rate of 1 in 691 live births; Parker et al., 2010). A relative strength in social functioning is a hallmark feature of children, adolescents, and adults with DS (Fidler,

Barrett, & Most, 2005; Fidler, Most, Booth-laforce, & Kelly, 2008; Gilmore, Campbell, & Cuskelly, 2003; Rosner, Hodapp, Fidler, Sagun, & Dykens, 2004). These social strengths appear to emerge in infancy as evidence by the majority of infants with DS achieving the developmental milestones associated with primary intersubjectivity (Fidler, 2006). However, there are noticeable differences in the achievement of these milestones as compared to the typical trajectory. For example, mutual gaze in infants with DS is slow to emerge, but then persists at a high level into the middle of the 1st year at a time when typically developing infants begin to shift their focus from people to the world around them (Berger & Cunningham, 1981; Carvajal & Iglesias, 2000; Slonims & Mcconachie, 2006).

Despite strengths in social functioning, children with DS are at elevated risk for an autism spectrum disorder with 7% to 18% meeting diagnostic criteria (Diguiseppi et al., 2010; Kent et al., 1999; Lowenthal, Paula, Schwartzman, Brunoni, & Mercadante, 2007). Nonetheless, some have argued that social strengths serve as a protective factor for many individuals with DS; others have argued that children with DS tend to overuse their social skills to compensate or to distract others when presented with a challenge (Fidler, 2006; Freeman & Kasari, 2002; Pitcairn & Wishart, 1994; Ruskin, Kasari, Mundy, & Sigman, 1994). Thus, a critical question related to social development in DS that needs to be examined is, what is the impact of early social competencies on development?

Joint Attention in DS—Studies of young children with DS indicate mental-age appropriate levels of initiating joint attention (Fidler, Philofsky, Hepburn, & Rogers, 2005; Mundy, Sigman, Kasari, & Yirmiya, 1988; Mundy et al., 1995) and total joint attention (Kasari, Freeman, Mundy, & Sigman, 1995; Kasari, Mundy, Yirmiya, & Sigman, 1990). All of these studies have used the Early Social Communication Scale (Mundy et al., 2006; Seibert, Hogan, & Mundy, 1982) to examine joint attention and other related early social skills (i.e., social interaction behaviors and requesting), which has a standardized administration and scoring procedure. Similar to the results related to joint attention, behaviors related to social interaction (e.g., turn taking) appear to be used with relative ease with studies reporting either more social interaction behaviors used by children with DS (Mundy et al., 1988) or commensurate levels as compared to mental-age matched peers (Mundy et al., 1995). Specifically, toddlers with DS demonstrate evidence of increased nonverbal social acts including gesturing, pointing, smiling, turn taking, and play acts than typical controls (Franco & Wishart, 1995; Kasari, Mundy, et al., 1990; Mundy, Kasari, Sigman, & Ruskin, 1995; Mundy et al., 1988).

Interestingly, despite appropriate use of joint attention and social interaction behaviors, fewer social referencing shifts have been reported in children with DS (Kasari et al., 1995). One possible explanation for this is difficulties with shifting attention. However, if this were the case then difficulties with joint attention would also be observed in DS. The other possibility is that there is a preference for social over nonsocial stimuli (Kasari et al., 1995). In an earlier study, Kasari and colleagues (1990) noted that children with DS look more to their social partner's face, which would suggest a preference for social stimuli. However, children with DS also looked away from the social interaction more than mental age matched typically developing children. In addition, this longer looking time to a social partner's face in children with DS was negatively related to joint attention, such that those

who shared shorter looks with their social partner used more joint attention behaviors (i.e., pointing, showing, etc.). Taken together, perhaps there is a combination of both difficulties shifting attention and a preference for social stimuli in DS. Specifically, for those with great difficulties with shifting attention, they are less able to shift their attention away from a social partner—the preferred stimuli. This preference for social stimuli may also be present in the use of requesting behaviors.

In contrast to these studies indicating strengths in joint attention, there are several studies that suggest initiating joint attention may be particularly difficult for infants with DS between 12 and 26 months (Legerstee & Fisher, 2008; Legerstee & Weintraub, 1997). In these studies, infants with DS were more likely to passively sharing attention to objects, rather than actively initiate an interaction by pointing to objects leading to fewer instances of initiating joint attention (Legerstee & Fisher, 2008; Legerstee & Weintraub, 1997). There are several potential reasons for these different findings. Frist, Legerstee and colleagues (Legerstee & Fisher, 2008; Legerstee & Weintraub, 1997) coded joint attention during naturalistic play observations with the infant's mother. Thus, unlike the studies using the Early Social Communication Scales, there is no standard administration or scoring. In addition, the Early Social Communication Scales provide opportunities for joint attention and other social behaviors that may not always occur during a 20- to 30-minute interaction. There are also differences in the chronological age ranges of these studies. Studies noting mental-age appropriate joint attention in young children with DS have been conducted with children anywhere between 12 months to 55 months. However, the two studies reporting difficulties with joint attention have used a more narrow age range (12 to 26 months). Because these studies are cross-sectional with a large age range, it is difficult to discern when joint attention emerges and perhaps these differences indicate that joint attention is emerging during the 2nd year in DS (i.e., after 12 months). Nonetheless, it has been suggested that the joint attention presentation in DS involves a unique combination of social strengths and cognitive weaknesses (i.e., shifting attention, instrumental thinking; Fidler, Philofsky et al., 2005; Moore, Oates, Hobson, & Goodwin, 2002; Ruskin, Kasari, Mundy, & Sigman, 1994).

Proposed Early Social Developmental Cascade in DS—It could be speculated that the strengths observed in joint attention and related behaviors in DS should support the development of social cognition. However, there is evidence that social cognition in children with DS may be compromised and that joint attention may not serve the same supporting role in the overall development as it does in typically developing children (Cebula et al., 2010; Cebula & Wishart, 2008; Wishart, 2007). The most studied aspect of social cognition in DS is theory of mind. Collectively, these studies suggest difficulties with most standard theory of mind tasks (e.g., appearance reality, false belief, and representational change tasks; Yirmiya, Solomonica-Levi, Shulman, & Pilowsky, 1996; Zelazo, Burack, Benedetto, & Frye, 1996).

To examine the influence of joint attention on early theory of mind performance, Hahn, Fidler, Hepburn and Rogers (2013) examined the performance of children with DS on a failed intentions task (i.e., the behavioral reenactment procedure; Meltzoff, 1995), and how this performance related to joint attention and affect sharing. Results indicated that the

ability to view a social partner's actions as intentional is not an area of particular strength or weakness in children with DS. Additionally, although high rates of both joint attention and affect sharing were observed in this study, they did not appear to be supporting the development of the understanding of intentionality in the manner that would be expected. Specifically, neither joint attention nor affect sharing were significant predictors of children's ability to accurately interpret their social partner's failed intentional action. Also, higher levels of affect sharing in children with DS did predict more imitations of the social partner's failed intentional action, which suggests that high affect sharing may be a barrier to the understanding of intentionality. Together, these findings indicate that intersubjective skills may influence the development of more complex social cognitive skills via two pathways. First, children with DS who experience deficits in joint attention (Cebula & Wishart, 2008; Legerstee & Weintraub, 1997) may miss the intentional information needed to interpret the intentions of others. Second, high affect sharing, or an overuse of social skills, in children with DS may interfere with joint attention (Fidler, Philofsky, et al., 2005; Kasari et al., 1995; Mundy et al., 1988; Sigman & Ruskin, 1999), which in turn may negatively impact the development of more complex social cognitive skills, such as the understanding of intentionality, in the manner that is expected based on the typical trajectory (Cebula & Wishart, 2008; Wishart, 2007). Collectively, the early phenotypic characteristics of children with DS may have a cascading effect on the understanding of intentionality, which may continue to influence the development of later social cognitive skills and social interactions (i.e., ability to understand the perspectives of others; Fidler et al., 2011).

In sum, there are many questions and avenues for future research on social phenotype associated with DS. Specifically, there is a need to examine the emergence and trajectory of early social development starting from infancy. These studies will allow for a better understanding of whether these skills, especially joint attention, are intact, delayed, or different from the typical trajectory. This will also support the examination of how these early social skills may have a cascading effect on social interactions for individuals with DS.

Williams Syndrome

WS is one of the most widely researched neurogenetic disorders (Bellugi, Lichtenberger, Jones, Lai, & St George, 2000), with an incidence rate of 1 in 7,500 (Strømme, Bjørnstad, & Ramstad, 2002). There is a distinct social profile associated within WS that emerges in infancy (for a review see Järvinen-Pasley et al., 2008; Järvinen, Korenberg, & Bellugi, 2013; Thurman & Fisher, 2015), characterized by an overly friendly, highly sociable (termed hypersociability; Gosch & Pankau, 1997), highly approachable, highly gregarious, and people oriented personality style (Mervis & Klein-Tasman, 2000). Children with WS are described as good social partners who are very friendly, highly empathic, and able to tune into other people's mental states because they seem to have special sensitivity to the emotional states of others (Dykens & Rosner, 1999; Gosch & Pankau, 1997; Hodapp & Desjardin, 2002; Jones et al., 2001; Plesa-Skwerer, Faja, Schofield, Verbalis, & Tager-Flusberg, 2006). A combination of relative strengths in interpersonal skills and the distinct social profile associated with WS drives individuals with WS to socially engage with others (Hodapp & Desjardin, 2002; Jones et al., 2001; Mervis & Klein-Tasman, 2000).

Indications of the distinct social phenotype associated with WS is apparent from infancy. This includes increased frequency of smiling behavior, increased frequency of attending to the faces of others (Järvinen-Pasley et al., 2008), a strong interest in people (Tager-Flusberg et al., 2006), and a preference for social stimuli over nonsocial stimuli (Järvinen-Pasley et al., 2008). Also, infants and toddlers with WS spend more time looking at their mother and at strangers than typically developing infants and toddlers of the same chronological or developmental age (Mervis et al., 2003). However, despite these strengths, it seems that children with WS have poorer than expected joint attention. In addition, it is possible that there are two related, but different processes influencing social development in WS: social-perceptual skills (i.e., skills related to the affective system that involve making judgments based on the perceptual, social, and emotional information available during a social interaction) and social-cognitive skills (i.e., the higher order thinking abilities that make up the traditional conceptualization of theory of mind; Hepburn, Fidler, Hahn, & Philofsky, 2011; Tager-Flusberg, 2005; Tager-Flusberg & Sullivan, 2000).

Joint Attention in WS—Young children with WS appear to have difficulties with joint attention in comparison to both mental-age and chronological age matched peers (Laing et al., 2002; Mervis & Bertrand, 1993, 1997; Mervis et al., 2003). Specifically, fewer instances of initiating joint attention and initiating requests, including protodeclarative and protoimperative pointing, have been reported in comparison to mental age-matched, typically developing peers (Laing et al., 2002). Considering the strong social drive and orientation of infants and young children with WS, it would not be surprising if they showed mental-age appropriate, or even strengths, in joint attention. However, it has been noted that during a triadic social interaction (i.e., between the infant, a social partner, and a toy) children with WS quickly turn this interaction into a dyadic interaction (i.e., a face-to-face interaction with the social partner that does not include the toy), especially when a novel person is present (Doyle, Bellugi, Korenberg, & Graham, 2004). There are two possible explanations for this shift in the interaction. First, this may be related to poor joint attention skills (Laing et al., 2002; Mervis & Bertrand, 1993, 1997; Mervis et al., 2003). On the other hand, this pattern may also be related to an overall preference for dyadic interaction (Järvinen-Pasley et al., 2008). This preference for dyadic interactions, or dyadic strength (Laing et al., 2002), may reduce the opportunity for children with WS to engage in joint attention and, in turn, lead to delays in this skill. Specifically, there appears to be a split in abilities in young children with WS, such that initiating joint attention skills are delayed, while responding to joint attention develops similar to the typical trajectory (Hepburn et al., 2011). This split in joint attention abilities could disrupt, and influence, later social development (Hepburn et al., 2011).

Proposed Early Social Developmental Cascade in WS—Given their high levels of empathy, emotional responsivity, and attunement to others, it would be plausible to speculate that children with WS might show relative sparing or even strength in social cognition. However, research on social cognition in WS indicates a combination of strengths and weaknesses in this population that likely stem from difficulties with joint attention (Gagliardi et al., 2003; Plesa-Skwerer, Faja, Schofield, Verbalis, & Tager-Flusberg, 2006; Riby & Back, 2010; Tager-Flusberg, Boshart, & Baron-Cohen, 1998; Tager-Flusberg,

Skwerer, & Joseph, 2006; Tager-Flusberg & Skwerer, 2007). These studies combined with the research on the social profile associated with WS, lead to the hypothesis that there are two processes that underlie the development of social cognition, especially theory of mind: social-perceptual skills and social-cognitive skills (Tager-Flusberg & Sullivan, 2000; Tager-Flusberg, 2005). This split in social cognitive abilities may emerge from early difficulties with joint attention.

In order to make the types of social judgments associated with theory of mind, individuals use skills that are supported by joint attention. That is, instead of sharing attention about an object or event with a social partner they need to consider the mind of that individual based on the linguistic and emotional information provided as the third variable in social interaction. The difficulties reported in joint attention may make it challenging for individuals with WS to obtain this type of information, especially when there is little or conflicting emotional information (e.g., your friend says, "I'm fine," but they are showing facial indicators of sadness). When clear emotional cues are present it is possible that individuals with WS are more successful at understanding their social partner's perspective due to their strong interest in the emotions of others (Dykens & Rosner, 1999; Gosch & Pankau, 1997; Hodapp & Desjardin, 2002; Jones et al., 2001; Plesa-Skwerer et al., 2006). This could explain why stronger social-perceptual skills and weaker social-cognitive skills are reported in WS (Tager-Flusberg & Sullivan, 2000). It is also likely that this dissociation is also observed in early social cognitive prior to the development of theory of mind (Fidler et al., 2007; Laing et al., 2002).

In order to explore the emergence of this dissociation in 2-to-5-year-olds with WS, Hepburn and colleagues (2011) used the Early Social Communication Scale (Mundy et al., 2006; Seibert et al., 1982) and the Autism Diagnostic Observation Schedule (Lord et al., 2012). Examples of social-perceptual skills in this study included directing facial expressions, shared enjoyment, and smiling, while social-cognitive skills included initiating and responding to joint attention and integrating eye gaze. Results indicated that young children with WS demonstrated intact social-perceptual skills and emerging deficits in social-cognitive skills (Hepburn et al., 2011). The conceptualization of social-cognitive skills in this study included both initiating and responding to joint attention, therefore, it is possible that early difficulties in joint attention, especially initiating joint attention, lead to later weaknesses in the social-cognitive skills associated with theory of mind. What is still unclear from this study is how these two processes are manifesting in infancy (i.e., prior to 2 years), and how they are developing over time, which may help to clarify the social phenotype associated with WS. Taken together, despite the social strengths in WS, difficulties with joint attention may help to clarify why they struggle with some areas of social development, especially those related to social cognition (e.g., social reciprocity, making and maintaining friendships) throughout the life course (Jawaid et al., 2012; Riby et al., 2012; Van der Fluit et al., 2012). These areas of strength and weakness in social development continue to provide interesting research avenues within this population, and opportunities to explore the nuances and complexity of these skills on other domains of development.

Fragile X Syndrome

FXS is the most common inherited form of intellectual disability with an incidence rate of 1 in 4,000 males and 1 in 6,000 females (CDC, 2011; Hagerman, 2007). FXS is also the most common single-gene cause of autism spectrum disorders with 30–74% of individuals with FXS meeting diagnostic criteria (Auerbach, Osterweil, & Bear, 2011; Bailey, Raspa, Olmsted, & Holiday, 2008; Clifford et al., 2007; Darnell et al., 2011; Hall, Lightbody, Hirt, Rezvani, & Reiss, 2010; Harris et al., 2008). Individuals with FXS show impairments in social interactions across the life span (Bailey, Hatton, & Skinner, 1998; Cohen et al., 1988; Murphy, Abbeduto, Schroeder, & Serlin, 2007; Wolff, Gardner, Paccia, & Lappen, 1989). Specifically starting in early childhood, individuals with FXS tend to show social withdrawal and anxiety during social interactions, especially those with strangers or in new settings (Bailey, Mesibov, et al., 1998; Cohen et al., 1988; Hall, Lightbody, Huffman, Lazzeroni, & Reiss, 2009; Hessel et al., 2006; Roberts, Weisenfeld, Hatton, Heath, & Kaufmann, 2007; Roberts, Clarke, et al., 2009). Thus, it can be hypothesized that these early impairments in social interaction in FXS may stem from even earlier atypical development of joint attention and joint attention related behaviors that has a cascading effect on social development throughout the life course.

Joint attention in FXS—Research on joint attention in FXS is still in the preliminary stages. However, while there is initial evidence that some aspects of joint attention appear to be mental-age appropriate in FXS (Hahn et al., 2016; Marschik et al., 2014; Roberts et al., 2002), there is also evidence that disruptions in gesture use exist (Flenthrope & Brady, 2010; Roberts et al., 2002). While more research is needed, the available research on joint attention and joint attention related behaviors provides insight into early social development in FXS.

Based on a retrospective video analysis, there is evidence that 9 and 12 month old infants with FXS direct attention to themselves during social interactions (e.g., getting attention, seeking comfort) and respond to utterances directed toward them (Marschik et al., 2014). Similarly, observational studies of young children with FXS suggest that joint attention and joint engagement are commensurate with developmental level (Hahn et al., 2016; Roberts et al., 2002). Also during social interactions, it seems that young children with FXS are particularly skilled at using eye gaze shifts for the purpose of sharing interests, making requests, and commenting (Roberts et al., 2002), despite the extant research on children, adolescents, and adults with FXS that reports impairments in eye gaze, especially for social purposes (Cohen et al., 1988; Hall et al., 2015; Hessel, Glaser, Dyer-Friedman, & Reiss, 2006; Murphy et al., 2007; Wolff et al., 1989). Thus, it is possible that difficulties with eye gaze have not yet emerged as a phenotypic behavior in very young children with FXS (Marschik et al., 2014; Wolff et al., 1989) and may actually be a key behavior that supports joint attention in early development.

However, eye gaze alone is not enough to support the increasing complexity of social interactions, which is where the addition of gestures is needed—an area of impairment in FXS. Specifically, several studies reported limited gesture use during social interactions in both infants and young children with FXS (Flenthrope & Brady, 2010; Marschik et al., 2014; Roberts et al., 2002). Examinations of the types of gestures used by young children with

FXS indicate they use less complex gestures, such as contact gestures, with more ease and frequency than advanced gestures, such as conventional and distal gestures (i.e., gestures where there is not contact between the child and the referent, such as pointing to something that is at a distance; Flenthrope & Brady, 2010; Roberts et al., 2002). Although contact gestures can support joint attention, the support they provide is limited to objects and events that the child can touch, and, therefore, do not support bringing in objects and events from outside the interaction (i.e., such as directing attention to a picture on the wall). Also, early impairments in gesture use may be a contributing factor for why no infants in Marschik and colleagues (2014) study were observed using requesting actions, requesting information, making choices (e.g., between objects), or imitating their social partners. All of these early social interactions may include a gestural component to clarify an infant's intent, such as reaching to request a toy or pointing to make a choice. Thus, this limited gesture use may disrupt the development of joint attention. Coupled with impairments in expressive language (Abbeduto, McDuffie, Brady, & Kover, 2011) and later difficulties with eye gaze, impairments in gestures use may result in very little support for interacting with others later in development.

Proposed Early Social Developmental Cascade in FXS—Gestures serve as a way to initiate an interaction, share interest, and imitate a social partner. Without the support of gestures, joint attention may not develop in the manner that is expected and together this may lead to difficulties interpreting information during a social interaction that is usually acquired by using theory of mind. The ability to understand the minds of others involves the accumulation of information from multiple proximal factors (i.e., gestures, facial expressions, vocalizations, context, etc.). Thus, one hypothetical link as to why impairments in theory of mind have been reported in adolescents and adults with FXS (Cornish et al., 2005; Garner, Callias, & Turk, 1999; Grant, Apperly, & Oliver, 2007; Mazzocco, Pennington, & Hagerman, 1994) is a cascading effect of limited gesture use that disrupts the development of joint attention. Specifically, using gestures during a social interaction allows social partners to share thoughts that may not be easily represented by speech alone (e.g., to show how big a spider was that scared the social partner telling the story; Goldin-Meadow & McNeill, 1999). Also, using gestures to support a social interaction is particularly important when an individual does not have the words to share their thoughts clearly to a social partner, like individuals with FXS (Goldin-Meadow, 1999). In addition, difficulties interpreting mental state information by individuals with FXS may lead them to not know how to appropriately respond in social situations. That is, they do not know how to interpret the mental state information provided by their social partner and, in turn, do not always provide a socially appropriate response. This may contribute to social avoidance and anxiety that is observed in FXS.

While more research is needed on joint attention, and early social developmental more broadly, there is initial evidence of unique profile of early social development in FXS. This leads to a preliminary hypothesis that the impairments in social development observed in older children, adolescents, and adults with FXS are the result of a cascading effect of early impairments in gesture use. However, more research is needed to examine this hypothesis. To date, there have been no investigations of less complex social cognitive skills like

understanding desires and intentions, or studies linking early social skills to later social cognitive skills. Also, future studies examining the emergence of joint attention in FXS, prospectively from infancy, have the potential to identify the starting state of joint attention and explore the if gesture impairments in FXS disrupt the development of joint attention and later social development, providing insight into the social phenotype associated with FXS.

Conclusions and Future Directions

Although advances have been made in characterizing the social phenotypes associated with DS, WS, and FXS there are still many aspects of social development that are unclear, especially in regards to how the social phenotype is emerging in infancy—what the starting state is—and the cascading effects on later development. For all three neurogenetic disorders there is a need to examine the emergence of joint attention. Based on the available evidence it seems that similar to the typical trajectory, joint attention is a critical and foundational skill that has a cascading influence on social development. In addition to examining joint attention there are several important questions for future research that are unique to each disorder. Specifically in DS, while there is initial evidence that social strengths can have both positive and negative effects on development, there is a need for more nuanced investigations of social skills. These studies should seek to help clarify the divide within different social skills and the interaction of these skills with other contexts and developmental domains. For example, why is pointing used effectively in social sharing contexts, but not in requesting contexts? In WS, the suggestion that there is a dissociation in social-perceptual and social-cognitive skills warrants further investigation in the area of social cognition, as well as, how this dissociation impacts other domains of development, such as language. Also, examination of this dissociation has yet to be explicitly examined in infancy. Finally in FXS, there is a need for more research on joint attention and other related skills, especially gesture use, in order to elucidate the development of these skills and their connection to later social development. This will help to clarifying the available research suggesting a mixed profile of strengths and weaknesses in joint attention despite many researchers hypothesizing deficits in this skill set. Thus, there are many potential research avenues to explore.

However, the future of research on early social development in infants and young children with neurogenetic disorders will need to overcome primary challenges in recruiting these infants, continuing to refine techniques for examining development, and the rise of comorbid conditions, such as autism spectrum disorders. Although this work is challenging, it is feasible, especially as the field continues to work to identify better solutions to these challenges. The most obvious issue associated with characterizing development in infancy is the need to identify infants with neurogenetic disorders within the first few months of life in order to examine starting states. While it is common for an infant with DS to be diagnosed prenatally or at birth (Bull, 2011), early identification is more difficult for neurogenetic disorders, like WS and FXS, that are not commonly tested for at birth. Unfortunately, it is still common for WS to not be diagnosed until between 5 and 10 years, unless heart problems are present—which can instigate genetic testing—and then a diagnosis may occur between 12 to 16 months (Carrasco, Castillo, Aravena, Rothhammer, & Aboitiz, 2005; Ferrero et al., 2007). The average age of diagnosis for males with FXS is 35 to 37 months

and 41 months for females (Bailey et al., 2009) however, there has been a recent increase in infant studies associated with FXS. This is due in part to increased awareness and understanding of FXS, which leads families who already have a family member with FXS to test their newborns. At the same time, this increase in studies is also due to the tenacity and connectedness of the researchers. Specifically, researchers have fostered strong relationships with the families of children with FXS from prior research studies and are now assessing their infant siblings, cousins, and other family members. In addition, these researchers are connected with one another and are developing ways to refer or “share” participants. Thus, the feasibility of recruiting infants with FXS is becoming more attainable, by collaborating with those who have been successful, participating in the FXS community, fostering rapport with the families, and being tenacious in recruitment efforts. This list applies to all families with infants, and can serve to foster in more studies of infants with neurogenetic disorders. This will allow for a better understanding of early development and how behavioral phenotypes are emerging.

At the same time, it is important to be respectful of the families who have infants with neurogenetic disorders and take caution not to intrude. Thus, it is key to find the appropriate way to approach families about research opportunities for their infants. As more networks, registries, and parent groups are formed, more avenues are available for working relationships to be established with these groups. The strongest supporter of this endeavor (based on personal experiences and observations) is that many parents of infants with neurogenetic disorders appear to reach a stage where they want more information about their child, leading them to want to participate in research opportunities. This leads to a need for researchers to consider our responsibilities in disseminating knowledge gained from our research in family-friendly ways, and to pursue translational research opportunities with clinicians.

Other issues with identifying potential participants in early infancy have to do with measurement and study design. Even with recruitment becoming more feasible, it may not be viable to conduct a prospective longitudinal study for 5 or more years, which would be needed to examine the potential cascading effects of joint attention on social development. However, there are statistical models that can help to overcome these issues. First, when a longitudinal study is not feasible, one approach that can be used is the cross-sectional developmental trajectory (Thomas et al., 2009). This approach was developed to examine research questions intended to determine if an atypical group shows a pattern of development different from the typical developmental pattern (Thomas et al., 2009). Also, this approach is ideal for studying the developmental trajectory of low incidence disorders because it uses a cross-sectional design instead of a longitudinal design, which is useful for small sample studies prior to longitudinal designs, albeit not as rigorous as a longitudinal design (Thomas et al., 2009). In this approach, the group of interest is compared to a chronological age-matched and developmental age-matched group of typically developing peers. Then the trajectories can be plotted together to examine if the trajectory of the group of interest resembles either of the comparison trajectories (Thomas et al., 2009). This type of trajectory method is particularly useful when there is a wide age range in the atypical group, which is an especially important feature for studying low incidence disorders. Finally, this approach can be particularly helpful as a starting point when it is not yet clear if a skill is

simply delayed due to the presence of intellectual disability or if a skill has a different development pattern.

Still, a necessary follow-up to a cross-sectional developmental trajectory, or any cross-sectional design, is a longitudinal study. By using multilevel modeling/longitudinal data analysis, it is possible to examine longitudinal trajectories even when all participants do not have the same number of data points (Singer & Willett, 2003). This approach allows for as much data to be used as possible to estimate the trajectory. One particularly useful longitudinal data analysis approach for studying development in neurogenetic disorders is a descriptive analysis of change over time (Singer & Willett, 2003), which can be used to examine individual change over time.

A final important factor to consider when examining early social development in neurogenetic disorders is the presence of co-morbid autism spectrum disorder. In FXS, the prevalence rate of autism spectrum disorders is 30–74% (for a review see McCary & Roberts, 2013) and in DS the prevalence rate is 7% to 18% (for a review see Diguiseppi et al., 2010; Reilly, 2009). While no prevalence studies have been conducted in WS, it has been suggested that 10% meet diagnostic criteria for autism spectrum disorder (Lincoln, Searcy, Jones, & Lord, 2007). Deficits in joint attention, theory of mind, and language, especially social communication, are hallmark features of autism spectrum disorder (American Psychological Association, 2013; Bruinsma, Koegel, & Koegel, 2004). Therefore, the presence of autism spectrum disorder will also have a cascading effect on early social development in neurogenetic disorders. For example, higher levels of autism spectrum disorder symptomatology have been consistently related to more impaired functioning in FXS (Bailey, Hatton, Skinner, & Mesibov, 2001; Hahn, Brady, Warren, & Fleming, 2015; Roberts, Mankowski, et al., 2009), and this pattern has also been observed in early social development (Flenthrope & Brady, 2010; Hahn et al., 2016). Similarly, a study examining the presence of autism spectrum disorder in DS indicates that potential early indicators include impairments in social and communication skills (Hepburn, Philofsky, Fidler, & Rogers, 2008). Thus, in addition to general within-syndrome variability in early social development, it is also possible that there are sub-phenotypes of children with co-morbid autism spectrum disorder who will have a slightly different trajectory of development or show a greater level of impairment in these skills.

Studying development, especially in low incidence disorders, is always accompanied by challenges. Despite these challenges, continuing to examine early social development starting in infancy has the potential to have a tremendous influence on our understanding of the development of individuals with these neurogenetic disorders. While there have been numerous calls for translational research to promote positive development and well-being in neurogenetic syndromes, it is also important to ensure that we have identified appropriate targets and supports for intervention. Considering the importance of early intervention for promoting positive development, continuing to characterize the infant phenotype associated with a given disorder should provide insight into the optimal targets and supports. Specifically, if deficits in joint attention, are identified in infants with neurogenetic disorders then early interventions can be developed, or current interventions can be modified, to support positive development (Jones, Carr, & Feeley, 2006; Kasari, Freeman, & Paparella,

2006; Martins & Harris, 2006; Rocha, Schreibman, & Stahmer, 2007). At the same time, if areas of strength are identified, it is possible that these skills could be used as leverage points and serve as a bridge to support the development of more complex skills. Therefore, it is necessary to continue to use innovative methodologies to gain deeper understanding of early social development in neurogenetic disorders, especially the potential cascading effects of gene-brain-behavior pathways, to inform intervention and treatment work.

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