Chapter 5: Intellectual disability

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Abstract:

Pragmatic language skills are often impacted in individuals with intellectual disability, a developmental condition defined by deficits in intellectual and adaptive skills. In this chapter, we review the literature on pragmatic language in three genetically-based causes of intellectual disability – Down syndrome, fragile X syndrome, and Williams syndrome. We focus on group-comparison studies of young verbal individuals and cover a range of critical pragmatic skills (e.g. speech acts, topic initiation and maintenance, management of communication breakdowns, and narrative). We draw special attention to matching strategies utilized in the design of these studies which have critical implications for interpreting existing literature and guiding future studies. We conclude with discussions of theoretical implications, research directions, and clinical applications based on our review.

Key words: communication; Down syndrome; fragile X syndrome; genetic disorder; intellectual disability; language; neurodevelopmental disorder; pragmatics; Williams syndrome

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5.1 Introduction

Intellectual disability (ID), previously referred to as mental retardation, is a developmental condition defined by deficits in intellectual functioning (e.g. an IQ score below 70) and adaptive skills, such as self-management, social behavior, and language and communication (American Psychiatric Association 2013). Historically, little consideration was afforded to understanding the symptom profiles of ID, and few if any intervention efforts existed, with individuals with ID commonly placed in institutions along with patients suffering from a variety of psychiatric conditions (Braddock and Parish 2002). It is now recognized that individuals with ID represent a considerably heterogeneous population, and detailed clinical assessment of skills across different cognitive, social, and linguistic domains is of paramount importance in developing and implementing effective interventions. Because pragmatic competence relies on a complex integration of skills across these domains, pragmatic abilities are frequently impacted in ID (Abbeduto 2003; Abbeduto et al. 2007; Abbeduto and Hesketh 1997; Rice et al. 2005; Roberts et al. 2008). Pragmatic impairment affects communication and social interaction, with potential to impact relationships with family members, peers, and other community members. Thus, the pragmatic skills of individuals with ID warrant special consideration in research and intervention efforts.

In this chapter, we review the research literature on pragmatic language in three genetically-based causes of ID – Down syndrome (DS), fragile X syndrome (FXS), and Williams syndrome (WS). As well as discussing our own findings, we include a few original examples of discourse from our data. We focus on these conditions of genetic origin because of our own expertise and the available research literature. However, we acknowledge that other etiologies, including environmental conditions such as fetal alcohol syndrome, are frequently implicated in ID as well. For each syndrome, we begin with a description of general characteristics. Literature permitting, we then report findings from standardized tests and rating systems, and consider what is known about speech acts (functions), conversational topic initiation and maintenance, communication breakdowns, and pragmatic (macrostructural) aspects of narrative (storytelling). We focus this literature review on verbal children, adolescents, and young adults,

and include only group comparison studies. Many group comparison studies matched research participants on, or controlled for, general cognitive ability to determine whether pragmatic competence is below nonverbal mental age expectations in individuals with ID. According to the autism literature, where studies of pragmatics abound, accounting for structural language (vocabulary and syntax) skills is a more appropriate and relatively more recent approach to matching (Capps et al. 1998; Ozonoff et al. 1990; Tager-Flusberg 2004). Because pragmatics refers to the *use* of language for social interaction, making sure that linguistic 'building blocks' are equated across groups is especially important. Therefore, we draw attention to these details in our review of the literature below, and revisit this issue in considering theoretical implications. The chapter concludes with research directions and some clinical applications.

5.2 Down syndrome

DS occurs in about 1 in 700-800 live births. It has a population prevalence of about 1 in 1,000 for children and adolescents and 1 in 1,200 overall, making it the most common known genetic cause of ID (Centers for Disease Control 2006; Parker et al. 2010; Presson et al. 2013; Shin et al. 2009). The vast majority of cases are caused by an extra copy of chromosome 21, with translocation (when part of this chromosome attaches to another chromosome) and mosaicism (when only some cells include an extra copy of chromosome 21) representing less frequent causes. Intellectual ability in DS varies from average intelligence to severe disability, with most individuals having ID in the moderate range (Pueschel 1995; Roizen 2007). Verbal short-term memory may be particularly impaired (Jarrold and Baddeley 2001; Laws 2002), whereas visuo-spatial processing and perception may represent a relative cognitive strength (Fidler et al. 2006; Jarrold et al. 1999). Individuals with DS have been described as affectionate, very social, and engaging (Moore et al. 2002; Wishart and Johnston 1990). The pragmatic language profile of DS is notable for its blend of strengths and weaknesses, as described below.

Most studies of pragmatic language in DS have focused on discrete pragmatic skills (i.e. specific skills such as signaling noncomprehension of a message or contributing novel information to a topic of conversation). However, one longitudinal study using the Comprehensive Assessment

of Spoken Language (CASL; Carrow-Woolfolk 1999) found that typically developing boys showed more skill at baseline (controlling for structural language and mental age) and developed pragmatic skills more quickly over time than boys with DS (Martin et al. 2013b). Similarly, on the Children's Communication Checklist (CCC; Bishop 1998), a questionnaire for measuring pragmatic skills (along with speech and structural language) that is rated by parents or teachers, individuals with DS showed lower overall pragmatic skills than younger typically developing controls (Laws and Bishop 2004). Using the second edition of the CCC (Bishop 2003), Losh et al. (2012) found that boys with DS performed more poorly overall (and on subscales of initiation, coherence, scripted language, and context in particular) than typically developing boys after controlling for nonverbal mental age as well as structural language skills, but did not differ from boys with FXS.

Except for requesting, children with DS display a similar range of communicative functions (i.e. answers, comments, and protests) as typically developing children matched on language age or developmental level (Beeghly et al. 1990; Coggins et al. 1983). Weakness in requesting may begin early and be less amenable to intervention. This is confirmed in one study of young children with DS (Yoder and Warren 2002), in which parent education and prelinguistic skills training improved prelinguistic commenting and lexical density but not requesting.

Contingent language use, or the ability to stay on topic, appears to be an additional strength. Children with DS appear able to stay on topic for as many turns as mental or developmental age-matched children (Beeghly et al. 1990; Tannock 1988) and for even more turns than children matched on mean length of utterance (MLU, a measure of syntactic complexity) (Beeghly et al. 1990). Moreover, evidence suggests that children with DS are more contingent during conversation than both children with FXS and children with autism (Roberts et al. 2007; Tager-Flusberg and Anderson 1991). However, as Abbeduto and Hesketh (1997) have argued, measuring topic maintenance ability by contingency alone overlooks the quality of topic-maintaining turns. In fact, Roberts et al. (2007) found that boys with DS elaborated on topics less often, and produced more turns that maintained a topic by adding minimal or no new

information (e.g. acknowledgments, simple responses) compared with younger, typically developing boys of similar mental age. Of note, most questions were coded as elaborate topic maintenance, and so findings of reduced requesting in DS referenced previously (Beeghly et al. 1990; Yoder and Warren 2002), along with structural language deficits, may also help to explain these differences. Children with DS also initiate fewer new topics than mental age-matched typically developing children (Tannock 1988).

Pragmatic difficulties continue as children with DS grow older. For instance, when describing novel shapes for a naïve listener during a structured referential communication task, youth with DS expressed messages that were less clear (e.g. more ambiguous) than those of mental age-matched, typically developing children (Abbeduto et al. 2006). Performance on this task was related to the expressive language ability of individuals with DS. In another structured task, Abbeduto et al. (2008) found that young individuals with DS signaled noncomprehension of confusing messages less often than mental age-matched typically developing children. In this same study, individuals with DS did not differ from those with FXS. In work from our own group (Martin et al. 2015), after controlling for mental age and receptive vocabulary skills, we also found that children and adolescents with DS signaled noncomprehension less often than younger, typically developing controls (comparisons with the FXS group are reported in section 5.3). Note that one typical way of signaling noncomprehension is to make a request for clarification, suggesting that requesting in particular may continue to be an area of weakness for children with DS as they become older.

Narrative, or storytelling, abilities appear to represent a relative strength in DS. Children and adolescents with DS have been found to include a similar number of plot elements as mental age-matched, typically developing children (Boudreau and Chapman 2000), and more references to plot and theme than language-matched controls (Boudreau and Chapman 2000; Miles and Chapman 2002). Even when matched on mental age alone, adolescents and young adults with DS used more evaluation (e.g. references to characters' mental states) in their narratives than typically developing controls in another study (Keller-Bell and Abbeduto 2007).

More recently, Finestack et al. (2012) reported that adolescents and young adults with DS performed similarly to younger, MLU-matched typically developing children across all macrostructural elements studied (e.g. character development, references to character's thoughts and feelings, linguistic cohesion through complex syntax). In another recent study, children and adolescents with DS included fewer episodic elements in their narratives than typically developing children matched on nonverbal cognitive skills (Channell et al. 2015). However, MLU accounted for these group differences, suggesting that structural language may be a key limiting factor in narrative skills for children with DS. Of note, less narrative content is recalled when stories are presented in audio only (Kay-Raining Bird et al. 2004). This may be explained by the visual processing strengths and verbal short-term memory deficits described at the beginning of this section.

Together, existing findings suggest that narrative abilities may be a relative strength in the pragmatic profile of DS, at least when visual supports are present. Critical skills such as narrative evaluation, the use of complex syntax to cohere narrative elements, and integration of local episodes within overarching narrative themes are all comparable to comparison groups matched on structural language abilities. To illustrate these crucial skills, the following narrative is produced by a 9.8 year-old boy with DS and is based on a wordless picture book. This boy has a nonverbal mental age of 5.6 years and a nonverbal IQ of 62:

> There was one, um, there was a boy named Bed. Name Christin. She was, he was sleeping in his bed. But he woked up. Because there was one cat on his bed. And he slept and slept and slept. He woked up. He tried to look for his cat. He tried to look under his bed. There's no cat. He was so sad because he doesn't have his cat. A ball. He looked behind the window. She looked behind the plants. He looked in in the toy bag. Toy box. He looked on the tree. He looked on the tree. He looked under here. But there was a spider. And he was crying crying crying. He slept and slept and slept and slept. And he woked up. That's what he looked like. He turned on the lights.

> And there was cats on his bed. That's how the story ended. And he snuggled. They, he kissed them. And they hugged him. He hugged them.

In spite of some grammatical errors (e.g. overregularizations such as 'woked'), this excerpt illustrates a number of strengths in narrative skill. Complex syntax, though not extensively employed, is used to link episodes causally, as with the adverbial clause in 'he was sad *because* he doesn't have his cat'. Protagonists' internal states, goals, and motivations are also described and elaborated in a manner that advances the story. And importantly, the narrative is imbued with an overarching structure with a clear beginning, middle, and end. In line with existing literature on pragmatics in DS more generally, this language sample illustrates how the pragmatic profile of individuals with DS is marked by both strengths and weaknesses. Strengths include contingent language use and picture-supported narrative skills, whereas challenges include requesting, initiation of topics and communicative repairs, and topic elaboration. This profile may be described as somewhat passive in nature, potentially requiring a good amount of scaffolding but lacking in features that would likely frustrate a communication partner (as opposed to noncontingent language and perseveration, as described in section 5.3 below).

Finally, while this review has focused on pragmatic language, children with DS also have poorer speech intelligibility, or understandability, than younger, typically developing children (Barnes et al. 2009; Chapman et al. 1998). Although studies of pragmatic language have typically accounted for these difficulties by evaluating only intelligible utterances from language samples, poor speech intelligibility can clearly impact pragmatic competence by limiting communicative effectiveness.

5.3 Fragile X syndrome

Although less prevalent than DS, FXS is the most common known inherited cause of ID (Dykens et al. 2000; Hagerman and Hagerman 2002), with the full mutation of the Fragile X Mental Retardation-1 gene (*FMR1*) present in approximately 1 in 2,500 to 1 in 5,000 individuals (Coffee et al. 2009; Fernandez-Carvajal et al. 2009; Hagerman 2008; Pesso et al. 2000). In individuals

with the full mutation of this gene, *FMR1* shuts down (becomes methylated). This causes a deficiency in production of the Fragile X Mental Retardation Protein (FMRP), which is thought to be essential for normal cognitive functioning (Devys et al. 1993; Jin and Warren 2003). Because females have two X chromosomes, females affected with FXS still have one functioning copy of *FMR1*. This copy is able to produce FMRP so that females are nearly always less affected than males.

Whereas females tend to exhibit only mild ID or intellectual abilities within the normal range, ID in males with FXS typically ranges in severity from moderate to severe (Hagerman and Hagerman 2002; Loesch et al. 2003; Reiss and Dant 2003). Social anxiety (Bregman et al. 1988; Cordeiro et al. 2011; Hagerman 2002) and deficits in attention (Hooper et al. 2000; Wilding et al. 2002) are also common. FXS is also the leading, identified single-gene condition associated with autism spectrum disorder (ASD), with about 40%-75% of males with FXS meeting criteria for ASD in a research setting (Clifford et al. 2007; Hall et al. 2008; Kaufmann et al. 2004; Klusek et al. 2014a; Philofsky et al. 2004; Rogers et al. 2001). Autism status often affects the severity and quality of language impairments in FXS and is associated with increased likelihood of both males and females receiving speech-language therapy (Martin et al. 2013a).

Because females are generally less affected than males, most research has focused on males with FXS. Accordingly, the following review will focus on males only. That said, results of case studies and a few mixed-age group studies suggest that pragmatic impairment, including difficulties in initiating social interactions, may be present in females as well (Hagerman et al. 1999; Lesniak-Karpiak et al. 2003; Mazzocco et al. 1997; Spinelli et al. 1995).

Several studies of overall pragmatic skills in males with FXS have utilized a standardized measure or comprehensive rating system. In the same longitudinal study reviewed in section 5.2 (Martin et al. 2013b), typically developing boys outperformed boys with FXS (with and without ASD) on the CASL at the first time-point and also developed pragmatic skills more quickly over time. Boys with both FXS and ASD performed more poorly than those with FXS

only. Losh et al. (2012) found that boys with comorbid FXS and ASD, but not those without ASD, performed more poorly than typically developing boys on the CASL after controlling for nonverbal mental age, receptive and expressive lexical skills, and MLU. Regardless of ASD status, boys with FXS performed more poorly overall on the CCC-2 (and on subscales of initiation, coherence, scripted language, context, and nonverbal communication in particular) than controls in this same study. Boys with FXS and ASD also did not differ significantly from boys with FXS only on any subscale, suggesting that the CCC-2 is not sensitive to pragmatic language differences in FXS based on ASD status.

Most recently, Klusek et al. (2014b) applied the Pragmatic Rating Scale-School Age (Landa 2011) to seminaturalistic interactions. They reported that boys with FXS (regardless of ASD status) showed greater impairment than younger typically developing boys after controlling for mental age and structural language. Further, boys with both FXS and ASD showed greater deficits than boys with FXS without ASD and boys with DS. In both of these studies (Klusek et al. 2014b; Losh et al. 2012), boys with comorbid FXS and ASD showed pragmatic impairment that was similar in severity to an additional comparison group of boys with idiopathic ASD.

As is the case for DS, most studies of pragmatic language in males with FXS have focused on discrete pragmatic skills. Males with FXS have been reported to contribute more off-topic or tangential turns (i.e. noncontingent language) to a conversation than males with ID without FXS, including those with DS (Sudhalter and Belser 2001; Wolf-Schein et al. 1987). Of note, autism status of participants with FXS was not specified in these early studies. More recently, Roberts et al. (2007) found this pattern to be specific to boys with FXS who also met criteria for ASD. These boys were additionally found to be more noncontingent than boys with FXS without ASD even after controlling for nonverbal mental age. Roberts and colleagues also found that boys with FXS with and without ASD, like boys with DS, were less likely to add new information in conversational turns (i.e. they were less elaborative) than younger typically developing boys. The following example illustrates the use of noncontingent language during a semistructured

interaction. It is from a 12.2 year-old boy with FXS and ASD who has a nonverbal mental age of 5.3 years and a nonverbal IQ of 42:

Examiner: How do we get in the airplane?

Child: Through the door. That's really small.

Examiner: Mhm.

Child: And you want gummy bear?

Examiner: Let's play a little more.

Another behavior that can affect the flow of conversation is perseveration, or excessive self-repetition. Boys and adult males with FXS (autism status sometimes not specified) have been found to produce more perseveration than males with DS or typical development of similar cognitive or language level (Levy et al. 2006; Roberts et al. 2007; Sudhalter et al. 1990; Wolf-Schein et al. 1987). In more recent work, boys with comorbid FXS and ASD were found to use more perseveration, controlling for mental age, than those with FXS only, DS, and typical development, whereas the group with FXS without ASD did not differ significantly from the DS or typically developing groups (Martin et al. 2012). The following conversational sample illustrates the tendency of this group to perseverate on both a local, utterance level as well as more globally with repetitive themes across utterances. It is from a 10.4 year-old boy with FXS and ASD who has a nonverbal mental age of 5.8 years and a nonverbal IQ of 63:

Child: What is this? What is it? What is this? What is this?

Examiner: Hmm.

Child: What is it?

Examiner: I think (interrupted)

Child: What is it?

Examiner: It's something that twirls.

…

Child: What is this? What is this guy? What is, what is this?

Examiner: He's a fireman.

Child: No. He's a fireman. What is this guy?

Like young individuals with DS, those with FXS may also have difficulty either expressing comprehensible and unambiguous messages, which could lead to a communication breakdown, or repairing communication breakdowns once they occur. In the same study reviewed earlier on referential communication (Abbeduto et al. 2006), adolescents and young adults with FXS were less successful at describing novel shapes for a listener during a structured task than were mental age-matched controls. In a second study by Abbeduto and colleagues using a structured task (Abbeduto et al. 2008), also reviewed earlier, adolescents and young adults with FXS signaled noncomprehension of unclear messages less often than younger typically developing controls but did not differ from those with DS. Neither of these studies included a separate group of participants with FXS and comorbid ASD. Moreover, investigators excluded from the FXS group only those who met DSM-IV criteria for autistic disorder (American Psychiatric Association 1994), making it likely that those who would meet DSM-5 criteria for ASD (American Psychiatric Association 2013) remained in the sample. Therefore, it is not clear, as in other studies reviewed previously, whether these pragmatic difficulties may be specific to or more pronounced in those with comorbid ASD. Work from our group suggests that this may be the case. We found that children and adolescents with comorbid FXS and ASD were less likely than typically developing controls to signal noncomprehension, whereas youth with FXS without ASD did not differ from controls and signaled noncomprehension more often than those with DS (Martin et al. 2015).

Compared with conversational discourse skills, less research has focused on narration in FXS and findings are mixed. In one study of recalled narratives, after controlling for nonverbal mental age, short-term memory, and expressive syntax, boys with FXS with and without ASD did not differ from boys with DS but included fewer references to a protagonist's goal-motivated actions than younger typically developing boys (Estigarribia et al. 2011). This finding mirrors those in the ASD literature, where causal explanations for protagonist behaviors,

thoughts, and feelings tend to be impaired (Capps et al. 2000; Losh and Capps 2003; Tager-Flusberg and Sullivan 1995). Further, in this study, boys with both FXS and ASD, but not boys with FXS only, also scored lower than the typically developing group in story grammar overall, suggesting that ASD in FXS further undermines narrative ability. Conversely, in another study (Hogan-Brown et al. 2013), no group differences in macrostructural skills (e.g. thematic maintenance) emerged for language age-matched boys with FXS with and without ASD, DS, idiopathic ASD, and typical development. Similarly, no differences were found in the use of evaluation devices between adolescents and young adults with FXS and mental age-matched controls in one other study (Keller-Bell and Abbeduto 2007).

In the study by Finestack et al. (2012) reviewed in section 5.2, adolescents and young adults with FXS without autistic disorder did not differ from those with DS but were more adept than MLU-matched typically developing controls in their use of story introductions (i.e. opening character and setting details). Participants in this study were more verbal than those in other work, with the FXS sample having an average MLU of 6.1 relative to a mental age of just 4.4 years. In sum, the few studies of narrative macrostructure in FXS have resulted in inconsistent findings. Of note, the only study to report impaired performance relative to controls (Estigarribia et al. 2011) used the Bus Story Language Test (Crowley and Glasgow 1994). In this story, the bus is highly anthropomorphized, making relation of character intentions potentially more difficult. Other studies that reported no evidence of narrative macrostructure impairments relied on more basic picture-description story tasks (e.g. Frog Goes to Dinner; Mayer 1977) that may require less proficiency in adopting the perspective of a character.

In summary, pragmatic language is generally impaired in males with FXS. Like males with DS, challenges for males with FXS include initiation of communicative repairs and topic elaboration. Unlike individuals with DS, pragmatic characteristics of males with FXS also include noncontingent language and perseveration. Pragmatic impairment may be pronounced in, and in some cases specific to, boys with comorbid FXS and ASD. More limited research has been conducted on narrative and with females. Finally, as is the case for DS, males with FXS have less

intelligible speech than younger, typically developing controls (Barnes et al. 2009), which can impact pragmatic ability and communicative effectiveness.

5.4 Williams syndrome

WS is caused by a microdeletion of approximately 25 genes on chromosome 7 (region 7q11.23). It affects 1 in 10,000 individuals (Strømme et al. 2002). A prominent characteristic of WS is a hyper-sociable personality, with a strong desire to seek out and initiate conversations with both familiar and unfamiliar individuals (Martens et al. 2008; Riby and Porter 2010). ID in WS is typically mild to moderate, although ability level ranges from severe ID to average intelligence (Donnai and Karmiloff-Smith 2000; Martens et al. 2008; Mervis et al. 2012; Riby and Porter 2010). Of note, individuals with WS demonstrate an uneven cognitive-linguistic profile where verbal skills typically exceed nonverbal abilities. Although this profile and characteristic loquaciousness initially led to hypotheses about the modularity of language and cognitive skills (Bellugi et al. 1990; Donnai and Karmiloff-Smith 2000; Pinker 1994), the advantage in verbal abilities has since been shown to be more complex than initially understood, with selective strengths and weaknesses in language ability relative to typically developing controls (Jones et al. 2000; Karmiloff-Smith 2007; Losh et al. 2001; Reilly et al. 1990; Reilly et al. 2004). Similarly, despite their sociability, more recent research suggests that individuals with WS present with a unique profile of pragmatic challenges, described below.

Two studies have characterized the pragmatic profile of individuals with WS using the CCC, a measure described in section 5.2. Laws and Bishop (2004) found that individuals with WS showed relatively weaker pragmatic skills overall than younger typically developing controls (matching criteria not specified). Also included in this study were DS and specific language impairment (SLI) groups. However, individuals with WS were the only clinical group to differ from controls in inappropriate initiation of conversation. This likely reflects the hypersociability that is characteristic of this group. Individuals with WS also used more stereotyped conversation than those with DS or SLI. Controlling for parent-reported expressive language skills, Philofsky et al. (2007) reported that children with WS showed greater pragmatic skills

overall on the CCC-2 than similarly aged children with ASD, but demonstrated similar rates of impairment as the ASD group on other subscales, including inappropriate initiation.

Other studies have directly examined conversational skills in individuals with WS. Lacroix et al. (2007) examined parent-child interactions in French-speaking children and adolescents with WS. The WS group spoke less and took fewer conversational turns than typically developing controls, similar to IQ-matched children with DS. However, they used more utterances that express their own mental states than chronological age-matched typically developing peers and individuals with DS, and at a rate similar to mental age-matched typically developing (i.e. chronologically younger) controls. Children and adolescents with WS have been found to have difficulty interpreting questions, as evidenced by noncontingent responding, relative to typically developing chronological age-matched peers (Stojanovik 2006). However, this ability has not been examined relative to mental age- or language-matched typically developing control groups, and thus may be attributed to more general delays in language and cognition. Children with WS also included fewer continuations (i.e. utterances adding new information, similar to what was termed elaborative topic maintenance in the DS and FXS literatures) relative to both typically developing individuals of a similar chronological age and individuals with SLI with similar receptive language abilities (Stojanovik 2006; Stojanovik et al. 2001). It is important to note that these studies are limited by small sample size (n=4-12 individuals with WS).

Communicative repair also represents an area of vulnerability for children with WS. In an experimental task where an examiner incorrectly responded to a child's request for one of two objects, children with WS were less likely than mental age-matched typically developing controls to vary requests or rejections in response to the communication breakdown (Asada et al. 2010). During conversation, children with WS also provided less information in response to an examiner's request for clarification relative to typically developing chronological age-matched peers in the study of French-speaking children with WS by Lacroix and colleagues (2007) described earlier.

The aspect of pragmatic language that has been explored most extensively in WS is narrative ability. Individuals with WS have been found to produce narratives similar in length to typically developing controls (accounting for mental or chronological age) or chronological age-matched children with SLI (Lacroix et al. 2007; Marini et al. 2010; Stojanovik et al. 2004). They include greater rates of key narrative plot points relative to mental age-matched typically developing controls and individuals with DS (Lacroix et al. 2007). However, individuals with WS produce less cohesive narratives than mental age-matched typically developing controls (Marini et al. 2010). Further, Reilly et al. (2004) asked individuals with WS to narrate a wordless picture book and noted a tendency for individuals with WS to describe individual scenes in great detail at the expense of an integrated, thematic whole. Indeed, in a later study, individuals with WS were found to include fewer reiterations of story theme relative to both chronological and mental age-matched controls (although more than individuals with DS) (Lacroix et al. 2007).

Perhaps the most notable feature of narrative in individuals with WS is their frequent employment of narrative evaluation (e.g. mention of characters' thoughts and emotions, explaining causal motivation for protagonist behavior), and more frequent attempts to engage the listener during their narrative relative to typically developing *chronological age-matched* children, as well as clinical groups including DS, traumatic brain injury, and SLI (Lacroix et al. 2007; Losh et al. 2001; Reilly et al. 2004). Evaluation is a critical narrative device for engaging one's interlocutor, for example, through use of engagement devices such as character speech and emphatic statements. Indeed, effective narration hinges on the ability to explain the psychological content of events, such as explaining protagonists' motivations for actions driving the plotline, as well as the ability to infer and express causal relationships across narrated events. Therefore, despite clear structural language and cognitive difficulties in WS, narrative evaluation appears to be a key strength, consistent with the hypersociability noted repeatedly in this population. However, it is important to note that the over-use of this device may ultimately detract from narrative competence in real-world settings, as frequent use of these devices may become distracting or even overwhelming to the listener.

In summary, like individuals with DS and FXS, young individuals with WS may have difficulty elaborating conversational topics (relative to chronological age-matched controls) and repairing communication breakdowns. In addition, they may have difficulty initiating conversation appropriately and telling cohesive narratives. However, individuals with WS also demonstrate a notable strength in the use of evaluation during narration and conversation, even exceeding their chronological age-matched peers.

5.5 Methodological considerations and theoretical implications

The influence of theory on language research in ID and the contribution of this research to theory have been discussed at length by other authors. These authors have argued in support of approaches that consider language problems in the broader framework of genetics, cognition and behavior as defined by a particular syndrome's phenotype and environment (Abbeduto and Boudreau 2004; Abbeduto et al. 2001), as well as the limitations of group-matching designs in developmental disabilities research (Mervis and Klein-Tasman 2004; Mervis and Robinson 2003). We will not repeat these issues here, but will briefly comment on how our review of the literature on pragmatics in DS, FXS, and WS underscores several theoretical and related methodological issues. The vast majority of investigations in this area presume at the outset a strong link between pragmatic language and cognition. However, studies reporting differences between clinical groups and mental age-matched typically developing controls indicate that some pragmatic difficulties (e.g. the ineffective handling of communication breakdowns which was observed across groups) cannot be attributed to cognitive level alone.

One factor that may be critical to pragmatic language development beyond the effects of general cognition is structural language ability. While the strategy of matching on mental age makes much sense for most domains of speech and language, it is not sufficient for studies of pragmatic language. As mentioned in the introduction of this chapter, pragmatics by definition refers to the *use* of language for social interaction. Thus, making sure that this linguistic foundation is similar across groups is key in order to make meaningful conclusions regarding pragmatic competence specifically. In some instances, as indicated in the preceding review,

individuals with ID outperform controls when structural language skills are taken into account. This suggests that studies controlling for mental age alone may be conflating pragmatic and structural language difficulties in these groups and, more central to the discussion of theory, that structural and pragmatic aspects of language are closely related.

Neither general cognition nor structural language, however, sufficiently explains all pragmatic difficulties evident in existing literature. For example, even after accounting for language ability, individuals with DS and FXS were reported to perform more poorly on global measures of pragmatic ability, and males with FXS produced more perseveration than controls. The pragmatic language profile in WS also showed marked divergences from mental-age matched controls, although studies that account for structural language level are largely lacking in the WS literature. Strong links between social cognition, or theory of mind, and pragmatic language have been found for individuals with idiopathic ASD (Capps et al. 1998; Capps et al. 2000; Losh and Capps 2003; Loveland and Tunali 1993; Surian et al. 1996; Tager-Flusberg 2000; Tager-Flusberg and Sullivan 1995), and could inform potential underpinnings of pragmatic language profiles in some instances of ID. Indeed, our review revealed that children with comorbid FXS and ASD showed more pragmatic difficulties than children with only FXS in a few studies that controlled for both nonverbal mental age and structural language skills. Moreover, Losh and colleagues (2012) reported that children with idiopathic and FXS-associated ASD showed similar deficits in theory of mind, and that these skills related to pragmatic ability across ASD, FXS, DS, and typically developing groups.

Surprisingly, in two studies reviewed previously, Abbeduto and colleagues did not find a significant relationship between social cognition and referential communication (Abbeduto et al. 2006) or noncomprehension signaling (Abbeduto et al. 2008) in DS or FXS. In both cases this was unexpected by the authors and was attributed partly to limitations in measurement and sample size as well as the developmental level of participants. Whereas participants' mental ages were comparable across studies, the sample size of the FXS group differed considerably – 57 in Losh et al. (2012) which also included more participants with ASD, versus 18 in the studies

by Abbeduto et al. (2006, 2008). Moreover, Abbeduto and colleagues used a single false belief task to assess theory of mind in both studies, whereas Losh et al. employed a battery of tasks including false belief as well as more basic tests of intentionality and desires. These were designed to decrease verbal and cognitive load and better capture a range of theory of mind abilities in the participants with ID. Social cognition and other influences, such as executive function and environmental factors, may indeed play important roles in the pragmatic competence of individuals with DS, FXS, and WS, although further research is needed.

5.6 Research directions

Review of the literature suggests several important areas for future research. First, studies should continue to elucidate the pragmatic profile of individuals with ID, matching on structural language abilities (for the reasons outlined in section 5.5), and directly compare pragmatic profiles across clinical groups. Second, studies that examine predictors of individual differences in pragmatic skills, beyond general cognition and structural language abilities, are largely lacking in the literature. Although Losh and colleagues (2012) did report links with theory of mind for boys with FXS and DS and with *FMR1*-related genetic variation in boys with FXS, other hypotheses have been proposed. For example, researchers have commonly ascribed pragmatic difficulties in FXS, and perseveration in particular, to excessive arousal and/or anxiety (e.g. Belser and Sudhalter 1995; Cornish et al. 2004; Klusek et al. 2015; Murphy and Abbeduto 2007). Heightened arousal was related to increased perseveration and noncontingent language in a preliminary study of two males with FXS (Belser and Sudhalter 1995). In more recent work, Klusek et al. (2013) reported that arousal dysregulation was related to poorer vocabulary skills, and marginally to poorer pragmatic language, in a larger sample of boys with FXS. In FXS, a recently developed quantitative method for measuring reduced FMRP expression via Luminex technology (LaFauci et al. 2013) presents a valuable opportunity for examining molecular-genetic correlates of pragmatic language in future investigations. Studies focused on these and other potential underlying mechanisms of pragmatic impairment, and whether they differ by syndrome group or from typical development, could inform general knowledge and theory, as well as intervention.

Third, girls with FXS should be the focus of future investigations, and girls and boys should be examined separately across syndrome groups to determine whether any sex differences exist which could inform understanding of underlying physiological processes or social influences, as well as clinical approaches. Fourth, future studies should continue to examine the impact of ASD status on pragmatic language in individuals with FXS, using valid and well-characterized groups (with and without comorbid ASD) so that findings are more comparable across studies. Future investigations should also include an idiopathic ASD group in order to better understand the overlap in FXS-associated and idiopathic cases of ASD, which could help to identify specific ASD traits that are linked to the *FMR1* gene involved in FXS (see Chapter 3, this volume for a review of pragmatic language in idiopathic ASD). Similarly, ASD is more common in DS and WS than in the general population (Hepburn et al. 2008; Richards et al. 2015), making the co-occurrence of ASD and its impact on pragmatic language an area of future research for all groups. Fifth, many of the studies reviewed here utilized mixed-age groups, spanning from childhood through adulthood, with relatively small sample sizes. Future studies should focus on discrete age groups and also examine pragmatic language longitudinally in order to determine changes over time as well as predictors of change. Finally, intervention research that targets the phenotypic characteristics described above for children with DS, FXS, and WS is critically needed. These studies should measure outcomes in pragmatic language specifically and related social development, such as peer relationships, more generally.

5.7 Clinical applications

A few clinical implications of the preceding review of pragmatics in ID for assessment and intervention are worth mentioning (for more detailed discussion, readers are referred to Chapter 21, this volume). Although individualized assessment and intervention that takes into account the developmental level and needs of a particular child and family is recommended, knowledge of phenotypic characteristics common to each syndrome could also help a clinician to focus or tailor assessment and intervention. Assessment approaches may also be informed by the research literature. For instance, in the Klusek et al. (2014b) study reviewed in section

5.3, group differences for the seminaturalistic context were more robust than those based on a standardized measure of pragmatics. Thus, clinical assessment should utilize a multi-method approach, including results of standardized assessments but also direct observation of more naturalistic interaction in multiple contexts and with various communication partners.

Ultimately, the goals of language intervention for individuals with ID should include improved functioning in communicative, social, academic and vocational domains (American Speech-Language-Hearing Association 2005). While this chapter has necessarily focused on pragmatic language, there is a vast literature documenting relative strengths and weaknesses of all three groups for speech and language more broadly that should be considered (Abbeduto et al. 2007; Mervis and Becerra 2007; Rice et al. 2005; Roberts et al. 2008). Assessment and intervention for children with ID should of course also focus on speech intelligibility and structural language to ensure that children with ID have the necessary tools for pragmatic language. Finally, intervention studies and research that uncovers the underlying mechanisms of pragmatic difficulties in each group will clearly have important implications for clinical management.

5.8 Summary

Pragmatic competence is frequently impacted, to varying degrees, in young individuals with DS, FXS, and WS. Future studies should continue to compare syndrome groups to each other and to typically developing controls appropriately matched on structural language ability. Knowledge of the phenotypic characteristics of each syndrome group may inform clinical efforts to some extent, though well-designed intervention studies are critically needed for all three groups. These studies, and intervention in general, will be guided by research that further elucidates the pragmatic language profile of each group, as well as the underlying mechanisms of pragmatic impairment in ID and whether they differ by etiological category.

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REFERENCES

Abbeduto, L. (Ed.) (2003). *Language and communication in mental retardation*. New York: Academic Press.

Abbeduto, L., & Boudreau, D. (2004). Theoretical influences on research on language development and intervention in individuals with mental retardation. *Mental Retardation and Developmental Disabilities Research Reviews,* 10(3), 184-192.

Abbeduto, L., Brady, N., & Kover, S. T. (2007). Language development and fragile X syndrome: Profiles, syndrome-specificity, and within-syndrome differences. *Mental Retardation and Developmental Disabilities Research Reviews,* 13(1), 36-46.

Abbeduto, L., Evans, J., & Dolan, T. (2001). Theoretical perspectives on language and communication problems in mental retardation and developmental disabilities. *Mental Retardation and Developmental Disabilities Research Reviews,* 7(1), 45-55.

Abbeduto, L., & Hesketh, L. J. (1997). Pragmatic development in individuals with mental retardation: Learning to use language in social interactions. *Mental Retardation and Developmental Disabilities Research Reviews,* 3(4), 323-333.

Abbeduto, L., Murphy, M. M., Kover, S. T., Karadottir, S., Amman, A., & Bruno, L. (2008). Signaling noncomprehension of language: A comparison of fragile X syndrome and Down syndrome. *American Journal on Mental Retardation,* 113(3), 214-230.

Abbeduto, L., Murphy, M. M., Richmond, E. K., Amman, A., Beth, P., Weissman, M. D., Kim, J. S., Cawthon, S. W., & Karadottir, S. (2006). Collaboration in referential communication: Comparison of youth with Down syndrome or fragile X syndrome. *American Journal of Mental Retardation,* 111(3), 170-183.

American Psychiatric Association (1994). *Diagnostic and statistical manual of mental disorders: DSM-IV*. Fourth edition. Washington, DC: American Psychiatric Association.

American Psychiatric Association (2013). *Diagnostic and statistical manual of mental disorders: DSM-5*. Fifth edition. Arlington, VA: American Psychiatric Association.

American Speech-Language-Hearing Association (2005). Roles and responsibilities of speech-language pathologists serving persons with mental retardation/developmental disabilities. http://www.asha.org/policy/GL2005-00061.htm. Accessed 19 January 2016.

Asada, K., Tomiwa, K., Okada, M., & Itakura, S. (2010). Fluent language with impaired pragmatics in children with Williams syndrome. *Journal of Neurolinguistics,* 23(6), 540-552.

Barnes, E., Roberts, J. E., Long, S. H., Martin, G. E., Berni, M. C., & Mandulak, K. C. (2009). Phonological accuracy and intelligibility in connected speech of boys with fragile X syndrome or Down syndrome. *Journal of Speech, Language, and Hearing Research,* 52(4), 1048-1061.

Beeghly, M., Weiss-Perry, B., & Cicchetti, D. (1990). Beyond sensorimotor functioning: Early communicative and play development of children with Down syndrome. In D. Cicchetti & M. Beeghly (Eds.), *Children with Down syndrome: A developmental perspective* (pp. 329-368). New York: Cambridge University Press.

Bellugi, U., Bihrle, A., Jernigan, T., Trauner, D., & Doherty, S. (1990). Neuropsychological, neurological, and neuroanatomical profile of Williams syndrome. *American Journal of Medical Genetics Supplement,* 6, 115-125.

Belser, R. C., & Sudhalter, V. (1995). Arousal difficulties in males with fragile X syndrome: A preliminary report. *Developmental Brain Dysfunction,* 8(4), 270-279.

Bishop, D. (1998). Development of the Children's Communication Checklist (CCC): A method for assessing qualitative aspects of communicative impairment in children. *Journal of Child Psychology and Psychiatry,* 39(6), 879-891.

Bishop, D. (2003). *The children's communication checklist-2*. London: Psychological Corporation.

Boudreau, D. M., & Chapman, R. S. (2000). The relationship between event representation and linguistic skill in narratives of children and adolescents with Down syndrome. *Journal of Speech, Language, and Hearing Research,* 43(5), 1146-1159.

Braddock, D., & Parish, S. L. (2002). An institutional history of disability. In D. Braddock (Ed.), *Disability at the dawn of the 21st century and the state of the states* (pp. 11-68). Washington, DC: American Association of Mental Retardation.

Bregman, J. D., Leckman, J. F., & Ort, S. I. (1988). Fragile X syndrome: Genetic predisposition of psychopathy. *Journal of Autism and Developmental Disorders,* 18(3), 343-354.

Capps, L., Kehres, J., & Sigman, M. (1998). Conversational abilities among children with autism and developmental delay. *Autism,* 2(4), 325-344.

Capps, L., Losh, M., & Thurber, C. (2000). The frog ate a bug and made his mouth sad: Narrative competence in children with autism. *Journal of Abnormal Child Psychology,* 28(2), 193-204.

Carrow-Woolfolk, E. (1999). *Comprehensive assessment of spoken language*. Circle Pines, MN: American Guidance Services.

Centers for Disease Control (2006). Improved national prevalence estimates for 18 selected major birth defects – United States, 1999-2001. *Morbidity and Mortality Weekly Report,* 54(51 & 52), 1301-1305.

Channell, M. M., McDuffie, A. S., Bullard, L. M., & Abbeduto, L. (2015). Narrative language competence in children and adolescents with Down syndrome. *Frontiers in Behavioral Neuroscience,* 9. doi:10.3389/fnbeh.2015.00283.

Chapman, R. S., Seung, H., Schwartz, S. E., & Kay-Raining Bird, E. (1998). Language skills of children and adolescents with Down syndrome: II. Production deficits. *Journal of Speech, Language, and Hearing Research,* 41(4), 861-873.

Clifford, S., Dissanayake, C., Bui, Q. M., Huggins, R., Taylor, A. K., & Loesch, D. Z. (2007). Autism spectrum phenotype in males and females with fragile X full mutation and premutation. *Journal of Autism and Developmental Disorders,* 37(4), 738-747.

Coffee, B., Keith, K., Albizua, I., Malone, T., Mowrey, J., Sherman, S. L., & Warren, S. T. (2009). Incidence of fragile X syndrome by newborn screening for methylated FMR1 DNA. *American Journal of Human Genetics,* 85(4), 503-514.

Coggins, T. E., Carpenter, R. L., & Owings, N. O. (1983). Examining early intentional communication in Down's syndrome and nonretarded children. *British Journal of Disorders of Communication,* 18(2), 98-106.

Cordeiro, L., Ballinger, E., Hagerman, R., & Hessl, D. (2011). Clinical assessment of DSM-IV anxiety disorders in fragile X syndrome: Prevalence and characterization. *Journal of Neurodevelopmental Disorders,* 3(1), 57-67.

Cornish, K., Sudhalter, V., & Turk, J. (2004). Attention and language in fragile X. *Mental Retardation & Developmental Disabilities,* 10(1), 11-16.

Crowley, J., & Glasgow, C. (1994). *The Renfrew bus story*. Centreville, DE: The Centreville School.

Devys, D., Lutz, Y., Rouyer, N., Bellocq, J. P., & Mandel, J. L. (1993). The FMR-1 protein is cytoplasmic, most abundant in neurons and appears normal in carriers of a fragile X premutation. *Nature Genetics,* 4(4), 335-340.

Donnai, D., & Karmiloff-Smith, A. (2000). Williams syndrome: From genotype through to the cognitive phenotype. *American Journal of Medical Genetics,* 97(2), 164-171.

Dykens, E., Hodapp, R. M., & Finucane, B. M. (2000). *Genetics and mental retardation syndromes: A new look at behavior and interventions*. Baltimore, MD: Paul H. Brookes.

Estigarribia, B., Martin, G. E., Roberts, J. E., Spencer, A., Gucwa, A., & Sideris, J. (2011). Narrative skill in boys with fragile X syndrome with and without autism spectrum disorder. *Applied Psycholinguistics*, 32(2), 359-388.

Fernandez-Carvajal, I., Walichiewicz, P., Xiaosen, X., Pan, R., Hagerman, P. J., & Tassone, F. (2009). Screening for expanded alleles of the FMR1 gene in blood spots from newborn males in a Spanish population. *Journal of Molecular Diagnostics,* 11(4), 324-329.

Fidler, D. J., Hepburn, S., & Rogers, S. (2006). Early learning and adaptive behaviour in toddlers with Down syndrome: Evidence for an emerging behavioural phenotype? *Down's Syndrome Research and Practice,* 9(3), 37-44.

Finestack, L. H., Palmer, M., & Abbeduto, L. (2012). Macrostructural narrative language of adolescents and young adults with Down syndrome or fragile X syndrome. *American Journal of Speech-Language Pathology,* 21(1), 29-46.

Hagerman, P. (2008). The fragile X prevalence paradox. *Journal of Medical Genetics,* 45(8), 498-499.

Hagerman, R. (2002). The physical and behavioral phenotype. In R. J. Hagerman & P. J. Hagerman (Eds.), *Fragile X syndrome: Diagnosis, treatment, and research* (pp. 3-87). Baltimore, MD: Johns Hopkins University Press.

Hagerman, R., & Hagerman, P. (Eds.). (2002). *Fragile X syndrome: Diagnosis, treatment, and research*. Third edition. Baltimore, MD: Johns Hopkins University Press.

Hagerman, R., Hills, J., Scharfenaker, S., & Lewis, H. (1999). Fragile X syndrome and selective mutism. *American Journal of Medical Genetics,* 83(4), 313-317.

Hall, S. S., Lightbody, A. A., & Reiss, A. L. (2008). Compulsive, self-injurious, and autistic behavior in children and adolescents with fragile X syndrome. *American Journal of Mental Retardation,* 113(1), 44-53.

Hepburn, S., Philofsky, A., Fidler, D. J., & Rogers, S. (2008). Autism symptoms in toddlers with Down syndrome: A descriptive study. *Journal of Applied Research in Intellectual Disabilities,* 21(1), 48-57.

Hogan-Brown, A. L., Losh, M., Martin, G. E., & Mueffelmann, D. J. (2013). An investigation of narrative ability in boys with autism and fragile X syndrome. *American Journal on Intellectual and Developmental Disabilities,* 118(2), 77-94.

Hooper, S. R., Hatton, D. D., Baranek, G. T., Roberts, J. P., & Bailey, D. B. (2000). Nonverbal assessment of IQ, attention, and memory abilities in children with fragile-X syndrome using the Leiter-R. *Journal of Psychoeducational Assessment,* 18(3), 255-267.

Jarrold, C., & Baddeley, A. D. (2001). Short-term memory in Down syndrome: Applying the working memory model. *Down Syndrome Research and Practice*, 7(1), 17-23.

Jarrold, C., Baddeley, A. D., & Hewes, A. K. (1999). Genetically dissociated components of working memory: Evidence from Downs and Williams syndrome. *Neuropsychologia,* 37(6), 637-651.

Jin, P., & Warren, S. T. (2003). New insights into fragile X syndrome: From molecules to neurobehaviors. *Trends in Biochemical Sciences,* 28(3), 152-158.

Jones, W., Bellugi, U., Lai, Z., Chiles, M., Reilly, J., Lincoln, A., & Adolphs, R. (2000). II. Hypersociability in Williams syndrome. *Journal of Cognitive Neuroscience,* 12(Suppl 1), 30-46.

Karmiloff-Smith, A. (2007). Williams syndrome. *Current Biology,* 17, R1035-R1036.

Kaufmann, W. E., Cortell, R., Kau, A. S., Bukelis, I., Tierney, E., Gray, R. M., Cox, C., Capone, G. T., & Stanard, P. (2004). Autism spectrum disorder in fragile X syndrome: Communication, social interaction, and specific behaviors. *American Journal of Medical Genetics,* 129(3), 225-234.

Kay-Raining Bird, E., Chapman, R., & Schwartz, S. (2004). Fast-mapping of words and story recall by children with Down syndrome. *Journal of Speech, Language, and Hearing Research,* 47(6), 1286-1300.

Keller-Bell, Y. D., & Abbeduto, L. (2007). Narrative development in adolescents and young adults with fragile X syndrome. *American Journal on Mental Retardation,* 112(4), 289-299.

Klusek, J., Losh, M., & Martin, G. E. (2014a). Consistency between research and clinical diagnoses of autism among boys and girls with fragile X syndrome. *Journal of Intellectual Disabilities Research,* 58(10), 940-952.

Klusek, J., Losh, M., & Martin, G. E. (2014b). A comparison of pragmatic language in boys with autism and fragile X syndrome. *Journal of Speech, Language, and Hearing Research,* 57(5), 1692-1707.

Klusek, J., Martin, G. E., & Losh, M. (2013). Physiological arousal in autism and fragile X syndrome: Group comparisons and links with pragmatic language. *American Journal on Intellectual and Developmental Disabilities,* 118(6), 475-495.

Klusek, J., Roberts, J. E., & Losh, M. (2015). Cardiac autonomic regulation in autism and fragile X syndrome: A review. *Psychological Bulletin,* 141(1), 141-175.

Lacroix, A., Bernicot, J., & Reilly, J. (2007). Narration and collaborative conversation in French-speaking children with Williams syndrome. *Journal of Neurolinguistics,* 20(6), 445-461.

LaFauci, G., Adayev, T., Kascsak, R., Nolin, S., Mehta, P., Brown, W. T., & Dobkin, C. (2013). Fragile X screening by quantification of FMRP in dried blood spots by a Luminex immunoassay. *Journal of Molecular Diagnostics,* 15(4), 508-517.

Landa, R. (2011). *The pragmatic rating scale for school-age children (PRS-SA)*. Unpublished manual.

Laws, G. (2002). Working memory in children and adolescents with Down syndrome: Evidence from a colour memory experiment. *Journal of Child Psychology and Psychiatry,* 43(3), 353-364.

Laws, G., & Bishop, D. (2004). Pragmatic language impairment and social deficits in Williams syndrome: A comparison with Down's syndrome and specific language impairment. *International Journal of Language & Communication Disorders,* 39(1), 45-64.

Lesniak-Karpiak, K., Mazzocco, M. M., & Ross, J. L. (2003). Behavioral assessment of social anxiety in females with Turner or fragile X syndrome. *Journal of Autism and Developmental Disorders,* 33(1), 55-67.

Levy, Y., Gottesman, R., Borochowitz, Z., Frydman, M., & Sagi, M. (2006). Language in boys with fragile X syndrome. *Journal of Child Language,* 33(1), 125-144.

Loesch, D. Z., Huggins, R. M., Bui, Q. M., Taylor, A. K., Pratt, C., Epstein, J., & Hagerman, R. J. (2003). Effect of the fragile X status categories and the fragile X mental retardation protein levels on executive functioning in males and females with fragile X. *Neuropsychology*, 17(4), 646-657.

Losh, M., Bellugi, U., & Anderson, J. D. (2001). Narrative as a social engagement tool: The excessive use of evaluation in narratives from children with Williams syndrome. *Narrative Inquiry,* 10(2), 265-290.

Losh, M., & Capps, L. (2003). Narrative ability in high-functioning children with autism or Asperger's syndrome. *Journal of Autism and Developmental Disorders,* 33(3), 239-251.

Losh, M., Martin, G. E., Klusek, J., Hogan-Brown, A. L., & Sideris, J. (2012). Social communication and theory of mind in boys with autism and fragile X syndrome. *Frontiers in Psychology,* 3. doi:10.3389/fpsyg.2012.00266.

Loveland, K. A., & Tunali, B. (1993). Narrative language in autism and the theory of mind hypothesis: A wider perspective. In S. Baron-Cohen, H. Tager-Flusberg & D. J. Cohen (Eds.),

Understanding other minds: Perspectives from autism (pp. 247-266). Oxford: Oxford University Press.

Marini, A., Martelli, S., Gagliardi, C., Fabbro, F., & Borgatti, R. (2010). Narrative language in Williams syndrome and its neuropsychological correlates. *Journal of Neurolinguistics,* 23(2), 97-111.

Martens, M. A., Wilson, S. J., & Reutens, D. C. (2008). Research review: Williams syndrome: A critical review of the cognitive, behavioral, and neuroanatomical phenotype. *Journal of Child Psychology and Psychiatry,* 49(6), 576-608.

Martin, G. E., Roberts, J. E., Helm-Estabrooks, N., Sideris, J., & Assal, J. (2012). Perseveration in the connected speech of boys with fragile X syndrome with and without autism spectrum disorder. *American Journal on Intellectual and Developmental Disabilities,* 117(5), 384-399.

Martin, G. E., Ausderau, K. K., Raspa, M., Bishop, E., Mallya, U., & Bailey Jr, D. B. (2013a). Therapy service use among individuals with fragile X syndrome: Findings from a US parent survey. *Journal of Intellectual Disability Research,* 57(9), 837–849.

Martin, G. E., Losh, M., Estigarribia, B., Sideris, J., & Roberts, J. (2013b). Longitudinal profiles of expressive vocabulary, syntax, and pragmatic language in boys with fragile X syndrome or Down syndrome. *International Journal of Language & Communication Disorders,* 48(4), 432-443.

Martin, G. E., Hornickel, J., McGrath, S., Barstein, J., Durante, G., & Losh, M. (2015). Noncomprehension signaling following communication breakdown in children with fragile X syndrome, Down syndrome, and autism. Paper presented at the American Speech-Language-Hearing Association Convention, Denver, Colorado, 12-14 November 2015.

Mayer, M. (1977). *Frog goes to dinner*. New York: Puffin Books.

Mazzocco, M. M., Kates, W. R., Baumgardner, T. L., Freund, L. S., & Reiss, A. L. (1997). Autistic behaviors among girls with fragile X syndrome. *Journal of Autism and Developmental Disorders,* 27(4), 415-435.

Mervis, C. B., & Becerra, A. M. (2007). Language and communicative development in Williams syndrome. *Mental Retardation and Developmental Disabilities Research Reviews,* 13(1), 3-15.

Mervis, C. B., Kistler, D. J., John, A. E., & Morris, C. A. (2012). Longitudinal assessment of intellectual abilities of children with Williams syndrome: Multilevel modeling of performance on the Kaufman Brief Intelligence Test–Second Edition. *American Journal on Intellectual and Developmental Disabilities,* 117(2), 134-155.

Mervis, C. B., & Klein-Tasman, B. P. (2004). Methodological issues in group-matching designs: Alpha levels for control variable comparisons and measurement characteristics of control and target variables. *Journal of Autism and Developmental Disorders,* 34(1), 7-17.

Mervis, C. B., & Robinson, B. F. (2003). Methodological issues in cross-group comparisons of language and/or cognitive development. In Y. Levy & J. Schaeffer (Eds.), *Language competence across populations: Toward a definition of specific language impairment* (pp. 233-258). Mahwah, NJ: Erlbaum.

Miles, S., & Chapman, R. S. (2002). Narrative content as described by individuals with Down syndrome and typically developing children. *Journal of Speech, Language, and Hearing Research,* 45(1), 175-189.

Moore, D. G., Oates, J. M., Hobson, P. R., & Goodwin, J. E. (2002). Cognitive and social factors in the development of infants with Down syndrome. *Down Syndrome Research and Practice,* 8(2), 43-52.

Murphy, M., & Abbeduto, L. (2007). Gender differences in repetitive language in fragile X syndrome. *Journal of Intellectual Disability Research,* 51(5), 387-400.

Ozonoff, S., Pennington, B. F., & Rogers, S. J. (1990). Are there emotion perception deficits in young autistic children? *Journal of Child Psychology and Psychiatry,* 31(3), 343-361.

Parker, S. E., Mai, C. T., Canfield, M. A., Rickard, R., Wang, Y., Meyer, R. E., Anderson, P., Mason, C. A., Collins, J. S., Kirby, R. S., Correa, A. and the National Birth Defects Prevention Network (2010). Updated national birth prevalence estimates for selected birth defects in the United States, 2004-2006. *Birth Defects Research Part A: Clinical and Molecular Teratology,* 88(12), 1008-1016.

Pesso, R., Berkenstadt, M., Cuckle, H., Gak, E., Peleg, L., Frydman, M., & Barkai, G. (2000). Screening for fragile X syndrome in women of reproductive age. *Prenatal Diagnosis,* 20(8), 611-614.

Philofsky, A., Fidler, D. J., & Hepburn, S. (2007). Pragmatic language profiles of school-age children with autism spectrum disorders and Williams syndrome. *American Journal of Speech-Language Pathology,* 16(4), 368-380.

Philofsky, A., Hepburn, S. L., Hayes, A., Hagerman, R., & Rogers, S. J. (2004). Linguistic and cognitive functioning and autism symptoms in young children with fragile X syndrome. *American Journal of Mental Retardation,* 109(3), 208-218.

Pinker, S. (1994). *The language instinct*. London: Penguin.

Presson, A. P., Partyka, G., Jensen, K. M., Devine, O. J., Rasmussen, S. A., McCabe, L. L., & McCabe, E. R. (2013). Current estimate of Down syndrome population prevalence in the United States. *Journal of Pediatrics,* 163(4), 1163-1168.

Pueschel, S. M. (1995). Down syndrome. In B. Zuckerman & S. Parker (Eds.), *Behavioral and developmental pediatrics: A handbook for primary care* (pp. 116-119). New York: Little Brown.

Reilly, J., Klima, E. S., & Bellugi, U. (1990). Once more with feeling: Affect and language in atypical populations. *Development and Psychopathology,* 2(4), 367-391.

Reilly, J., Losh, M., Bellugi, U., & Wulfeck, B. (2004). 'Frog, where are you?' Narratives in children with specific language impairment, early focal brain injury, and Williams syndrome. *Brain and Language,* 88(2), 229-247.

Reiss, A. L., & Dant, C. C. (2003). The behavioral neurogenetics of fragile X syndrome: Analyzing gene-brain-behavior relationships in child developmental psychopathologies. *Development and Psychopathology,* 15(4), 927-968.

Riby, D. M., & Porter, M. A. (2010). Williams syndrome. In P. Bauer (Ed.), *Advances of child development and behavior, volume 39* (pp. 163-209). UK: Elsevier Inc.

Rice, M. L., Warren, S. F., & Betz, S. K. (2005). Language symptoms of developmental language disorders: An overview of autism, Down syndrome, fragile X, specific language impairment, and Williams syndrome. *Applied Psycholinguistics,* 26(1), 7-27.

Richards, C., Jones, C., Groves, L., Moss, J., & Oliver, C. (2015). Prevalence of autism spectrum disorder phenomenology in genetic disorders: A systematic review and meta-analysis. *Lancet Psychiatry,* 2(10), 909-916.

Roberts, J. E., Chapman, R., & Warren, S. (Eds.) (2008). *Speech and language development and intervention in Down syndrome and fragile X syndrome*. Baltimore: Paul H. Brookes Publishing Co.

Roberts, J. E., Martin, G. E., Moskowitz, L., Harris, A. A., Foreman, J., & Nelson, L. (2007). Discourse skills of boys with fragile X syndrome in comparison to boys with Down syndrome. *Journal of Speech, Language, and Hearing Research,* 50(2), 475-492.

Rogers, S. J., Wehner, D. E., & Hagerman, R. (2001). The behavioral phenotype in fragile X: Symptoms of autism in very young children with fragile X syndrome, idiopathic autism, and other developmental disorders. *Journal of Developmental Behavioral Pediatrics,* 22(6), 409-417.

Roizen, N. J. (2007). Down syndrome. In M. L. Batshaw, L. Pellegrino & N. J. Roizen (Eds.), *Children with disabilities*. Sixth edition (pp. 263-273). Baltimore: Paul H. Brookes.

Shin, M., Besser, L. M., Kucik, J. E., Lu, C., Siffel, C., Correa, A., & Congenital Anomaly Multistate Prevalence and Survival Collaborative (2009). Prevalence of Down syndrome among children and adolescents in 10 regions of the United States. *Pediatrics,* 124(6), 1565-1571.

Spinelli, M., Rocha, A. C., Giacheti, C. M., & Richieri-Costa, A. (1995). Word-finding difficulties, verbal paraphasias, and verbal dyspraxia in ten individuals with fragile X syndrome. *American Journal of Medical Genetics,* 60(1), 39-43.

Stojanovik, V. (2006). Social interaction deficits and conversational inadequacy in Williams syndrome. *Journal of Neurolinguistics,* 19(2), 157-173.

Stojanovik, V., Perkins, M., & Howard, S. (2001). Language and conversational abilities in Williams syndrome: How good is good? *International Journal of Language & Communication Disorders,* 36(S1), 234-239.

Stojanovik, V., Perkins, M., & Howard, S. (2004). Willliams syndrome and specific language impairment do not support claims for developmental double dissociations and innate modularity. *Journal of Neurolinguistics,* 17(6), 403-424.

Strømme, P., Bjørnstad, P. G., & Ramstad, K. (2002). Prevalence estimation of Williams syndrome. *Journal of Child Neurology,* 17(4), 269-271.

Sudhalter, V., & Belser, R. C. (2001). Conversational characteristics of children with fragile X syndrome: Tangential language. *American Journal on Mental Retardation,* 106(5), 389-400.

Sudhalter, V., Cohen, I. L., Silverman, W., & Wolf-Schein, E. G. (1990). Conversational analyses of males with fragile X, Down syndrome, and autism: Comparison of the emergence of deviant language. *American Journal on Mental Retardation,* 94(4), 431-441.

Surian, L., Baron-Cohen, S., & van der Lely, H. K. J. (1996). Are children with autism deaf to Gricean maxims? *Cognitive Neuropsychiatry,* 1(1), 55-71.

Tager-Flusberg, H. (2000). Language and understanding minds: Connections in autism. In S. Baron-Cohen, H. Tager-Flusberg & D. J. Cohen (Eds.), *Understanding other minds: Perspectives from developmental cognitive neuroscience* (pp. 124-149). Oxford: Oxford University Press.

Tager-Flusberg, H. (2004). Strategies for conducting research on language in autism. *Journal of Autism and Developmental Disorders,* 34(1), 75-80.

Tager-Flusberg, H., & Anderson, M. (1991). The development of contingent discourse ability in autistic children. *Journal of Child Psychology and Psychiatry,* 32(7), 1123-1134.

Tager-Flusberg, H., & Sullivan, K. (1995). Attributing mental states to story characters: A comparison of narratives produced by autistic and mentally retarded individuals. *Applied Psycholinguistics,* 16(3), 241-256.

Tannock, R. (1988). Mothers' directiveness in their interactions with their children with and without Down syndrome. *American Journal of Mental Retardation,* 93(2), 154-165.

Wilding, J., Cornish, K., & Munir, F. (2002). Further delineation of the executive deficit in males with fragile-X syndrome. *Neurospsychologia,* 40(8), 1343-1349.

Wishart, J. G., & Johnston, F. H. (1990). The effects of experience on attribution of a stereotyped personality to children with Down syndrome. *Journal on Mental Defiency Research,* 34(5), 409-420.

Wolf-Schein, E. G., Sudhalter, V., Cohen, I. L., Fisch, G. S., Hanson, D., Pfadt, A. G., Hagerman, R., Jenkins, E., & Brown, W. T. (1987). Speech-language and the fragile X syndrome: Initial findings. *ASHA,* 29(7), 35-38.

Yoder, P., & Warren, S. (2002). Effects of prelingustic milieu teaching and parent responsivity education on dyads involving children with intellectual disabilities. *Journal of Speech, Language, and Hearing Research,* 45(6), 1158-1175.