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Language and Communication Development and Early Intervention Practices for Children with Fragile X Syndrome and Down Syndrome

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LANGUAGE AND COMMUNICATION DEVELOPMENT AND EARLY INTERVENTION
PRACTICES FOR CHILDREN WITH FRAGILE X SYNDROME AND DOWN SYNDROME

by

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B.A., Indiana University, 2010

A Research Paper
Submitted in Partial Fulfillment of the Requirements for the
Master of Science degree

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Fragile X syndrome (FXS) is the most common inherited cause of mental retardation (Barnes, Roberts, Mirrett, Sideris, & Misenheirmer, 2006; Barnes, Roberts, Long, Martin, Berni, Mandulak, & Sideris, 2009; Brady, Skinner, Roberts, & Hennon, 2006; Finestack, Richmond, & Abbeduto, 2009; Flenthrope & Brady, 2010; Mirrett, Roberts, & Price, 2003; Price, Roberts, Vandergrift, & Martin, 2007; Roberts, Long, Malkin, Barnes, Skinner, Hennon, & Anderson 2005) caused by an affected X chromosome (Finestack et al., 2009; Flenthrope & Brady, 2010; Mirrett et al., 2003; Price et al., 2007; Roberts et al., 2005) with a prevalence of 1 in 4,000 births (Barnes et al., 2009; Brady et al., 2006; Finestack et al., 2009; Mirrett et al., 2003; Price et al., 2007; Roberts et al., 2005). Down syndrome (DS) is the most common genetic cause of mental retardation (Abbeduto, Warren, & Connors, 2007; Barnes et al., 2006; Barnes et al., 2009; Price et al., 2007; Roberts et al., 2005; Roberts, Price, & Malkin, 2007) caused by the presence of an extra chromosome 21 (Abbeduto et al., 2007; Price et al., 2007; Roberts et al., 2005; Roberts et al., 2007) with a prevalence of 1 in 920 births (Price et al., 2007; Roberts et al., 2005). Individuals with FXS and DS demonstrate delays across a multitude of language and communicative domains. However, these delays, along with many other developmental delays, vary according to each individual. In addition, language

opportunities provided within the environment can have a large impact on the child's development of language (Price et al., 2007). Therefore, parents should be trained on effective strategies to enhance the opportunities for their children to learn language (Brady et al., 2006; Finestack et al., 2009; Mirrett et al., 2003; Roberts et al., 2007; Smith & Oller, 1981; Warren, Brady, Sterling, Fleming, & Marquis, 2010). The objective of this research paper is to review the literature about 0-8 year old children with DS and FXS in order to identify how these populations develop language and how to effectively intervene to increase language development.

Language Development

There are inconsistent reports about strengths and challenges demonstrated by children with FXS and DS when developing language. However, it is known that the multiple developmental delays which are exhibited by these populations will vary according to each individual. It is important to identify each individual's personal strengths and challenges in order to provide individualized intervention (Mirrett et al., 2003).

Prelinguistic Language Development

Fragile X syndrome. Children with FXS and DS have greater delays in language development than typically developing (TD)

children. Young children with FXS typically exhibit comparable delays across communicative areas such as vocal and gestural communicative domains (Finestack et al., 2009). It has also been reported that children under the age of three with FXS produce words in imitative contexts; however, in non-imitative situations the child is typically nonverbal (Finestack et al., 2009). Flenthrope and Brady (2010) added that their research suggested that children with FXS use contact gestures for a longer period of time during prelinguistic development. They defined contact gestures as gestures which include direct contact from the communicator to the conversational partner (Flenthrope & Brady, 2010). It is important to note that there was a small sample used in this research investigation; therefore, additional research is necessary to confirm the validity of the information (Flenthrope & Brady, 2010). Flenthrope and Brady's (2010) findings may imply that the use of contact gestures does not promote positive language growth in this population. Therefore, providing intervention which promotes the use of other social gestures may have a positive impact on language and communication development from individuals with FXS (Flenthrope & Brady, 2010).

Research pertaining to language development in individuals with FXS is minimal (Brady et al., 2006; Finestack et al., 2009; Flenthrope et al., 2010; Mirrett et al., 2003). Consequently,

information regarding the onset of canonical babbling in these children was not found. Further research should be conducted in order to obtain information regarding this area of language development in children with FXS.

On average, boys with FXS master most early and middle developing consonants and two-thirds of later developing consonants (Roberts et al., 2005). Mirrett et al. 2003 reported hypotonia of the oral-motor structures and sensory integration issues which could be attributed to unintelligible connected speech in individuals with FXS; however, these individuals often display intelligible speech at the single word level.

Down syndrome. Infants with DS are often delayed in the acquisition of reciprocal eye contact, but demonstrate strengths in imitation and use of gestures (Abbeduto et al., 2007). The preference for gestural communication in individuals with DS is associated with a delay in speech production (Caselli, Vicari, Longobardi, Lami, Pizzoli, & Stella, 1998). Abbeduto et al. (2007) cites inconsistent reports pertaining to pragmatic functions in children with DS: some reports indicate a delay in commenting, but no delay in requesting; yet, it has also been reported that children with DS do demonstrate a delay in requesting (Abbeduto et al., 2007; Roberts et al., 2007). Children with DS are similar to TD children in their use of gestures in early communicative development; however, they

demonstrate fewer requests by use of gestures (Roberts et al., 2007). The use of nonverbal requests is limited in children with DS (Mundy, Kasari, Sigman, & Ruskin, 1995). Their findings also suggested that the weak display of nonverbal requesting could be correlated with the expressive language deficit often observed in children with DS (Mundy et al., 1995). It is important to note that Caselli et al. (1998) reported that children with DS display greater receptive language skills than expressive language skills.

Nonverbal social interactions are associated with expressive and receptive language development (Mundy et al., 1995). For example, use of turn-taking, eye contact, and physical interaction can be predictors of overall language development (Mundy et al., 1995). Therefore, providing intervention to children with DS focusing on turn-taking, eye contact, and physical touch can provide positive results in their overall language development.

Abbeduto et al. (2007) and Roberts et al. (2007) found inconsistent results pertaining to the onset of canonical babbling for children with DS. Some studies report a small delay, whereas other studies report that children with DS begin babbling two months later than TD children. Roberts et al. (2007) reported that infants with DS continue babbling until two years of age which is longer than TD children; consequently,

there is typically a delay in their first spoken words. The findings from Smith and Oller (1981) appears to indicate that the onset of reduplicated babbling for infants with DS is on average 8.4 months old, while the onset of reduplicated babbling for TD infants is 7.9 months old. This suggests that the difference between the groups is non-significant (Smith & Oller, 1981).

Roberts et al. (2005) found that boys with DS express three-fourths of early developing consonants, only half of middle developing consonants, and slightly more than one-third of later developing consonants when compared to TD children. However, it is important to add that Smith and Oller (1981) stated that infants with DS and TD infants are similar in regards to frequency of production of consonants. For example, the production of alveolars is infrequent in the early stages of babbling; however, the production of alveolars increases and becomes much more frequent in the later stages of babbling (Smith & Oller, 1981). Children with DS frequently exhibit difficulty with palatal consonants and frequently demonstrate difficulty with lateralization of sibilants (Roberts et al., 2005).

Intelligible speech is typically mastered at 48 months for TD children, whereas producing intelligible speech is difficult for individuals with DS to master; often, this is a challenge

exhibited throughout their life (Roberts et al., 2007). This could be due to anatomical differences found in individuals with DS (Abbeduto et al., 2007; Roberts et al., 2007).

Oral Structure and Function

Fragile X syndrome. Barnes et al. (2006) stated that when compared to TD peers, individuals with FXS score lower on speech function tasks and on oral functions of the velopharynx, tongue, and lips. There is evidence that indicates that individuals with FXS have a delay in oral motor functions not accompanied by speech, such as in imitation of oral movements (Barnes et al., 2006).

Down syndrome. Individuals with DS score lower on oral structure than individuals with FXS and TD peers (Barnes et al., 2006). This is consistent with previous findings: oral structures displaying the lowest scores included tongue, lips, and velopharyngeal structure (Barnes et al., 2006). Oral structural differences frequently observed in individuals with DS include large, forward protruding tongue, small oral cavity, missing, additional, or poorly differentiated facial musculature (Roberts et al., 2007), irregular dentition, and narrow, high arched palate (Barnes et al., 2006; Roberts et al., 2007). Although atypical tongue structure was verified in individuals with DS, there was no significant difference in oral function of the tongue and mandible when compared to TD children. This

evidence appears to indicate that atypical structure of an articulator may not affect its' function (Barnes et al., 2006). Findings by Roberts et al. (2007) do not support results by Barnes et al. (2006). Roberts et al. (2007) suggested that difficulty coordinating articulators for speech, as well as limited range of motion and reduced speech are possibly attributed to anatomical differences found in individuals with DS. Barnes et al. (2006) pointed out that unlike individuals with FXS, individuals with DS perform better on oral function tasks than on speech function tasks. This evidence is not surprising due to the frequently observed unintelligible speech in individuals with DS. Barnes et al. (2006) added that, although individuals with DS display more irregular oral structures than individuals with FXS, both populations performed with similar accuracy on speech function tasks; therefore, this evidence suggests that typical oral structure may not contribute to appropriate speech production skills (Barnes et al., 2006).

Phonology

Fragile X syndrome. Boys with FXS demonstrate use of phonological processes similar to TD boys (Roberts et al., 2005; Barnes et al., 2009). Research findings suggest that boys with FXS produce phonological processes similar to younger TD children matched on mental age (Finestack et al., 2009). Roberts et al. (2005) reported different research findings about the use

of phonological processes in boys with FXS. The occurrence of final consonant deletion, unstressed syllable deletion, and gliding are reported findings in other research studies. However, Roberts et al. (2005) found that at the single-word level boys with FXS display consonant substitution and omission errors. Barnes et al. (2009) reported that boys with FXS display liquid simplification and final consonant deletion as commonly used phonological processes at the single-word level and in connected speech.

Down syndrome. Children with DS also display phonological processes similar to TD children; however, the elimination of phonological processes occurs over a longer period of time (Barnes et al., 2009; Roberts et al., 2005). Roberts et al. (2005) cited differences in research pertaining to phonological processes exhibited by boys with DS. It has been reported that individuals with DS exhibit stopping and gliding as commonly used processes; however, Robert et al. (2005) did not find gliding as a frequently used process. In addition, it was found that the use of stopping occurs more frequently in later developing fricatives; however, it is an inconsistent occurrence in individuals with DS. Boys with DS also exhibit greater use of syllable structure processes than substitution processes. However, it is noted that the use of substitution processes are similar to TD boys and boys with FXS while use of syllable

structure processes occurs considerably more (Roberts et al., 2005). The use of syllable structure processes are demonstrated mostly with cluster reductions while final consonant deletion occurs as well (Roberts et al., 2005). Barnes et al. (2009) agreed with Robert et al. (2005) that boys with DS tend to omit a greater number of entire syllables and consonant segments than do boys with FXS and TD boys. Due to the use of many phonological processes, difficulty with later developing sounds, and reduced word shapes, boys with DS display less intelligible speech than boys with FXS and TD peers (Barnes et al., 2009; Roberts et al., 2005).

Early Intervention

Evidence suggests that providing early intervention in the first few months of life will increase language development (Roberts et al., 2007). Interventionist should train parents on how to respond to children's communication attempts. Parents' implementation of intervention goals during the child's daily life will promote their development (Abbeduto et al., 2007; Roberts et al., 2007; Ronski & Sevcik, 2005).

Prelinguistic Language Development

Fragile X syndrome. It is important to note that there are currently no known studies that have examined the effectiveness of intervention for the FXS population; consequently, this impacts evidenced-based practice (EBP) for these individuals.

However, researchers provide suggestions which appear effective intervention strategies for working with individuals with FXS. Intervention strategies are derived from behavioral and developmental characteristics frequently portrayed by these individuals.

According to Finestack et al. (2009), in order for children to learn how to get their needs met it is imperative to target prelinguistic communication skills such as coordinated eye gaze, gestures, and vocalization. These prelinguistic skills should be targeted in isolation and simultaneously at the earliest age possible. Intervention should focus on family priorities, concerns and routines (Brady et al., 2006). When planning intervention for individuals with FXS, the child's communication priorities, developmental level, and interests should be considered in order to create an individualized program that fits each child's specific needs (Mirrett et al., 2003). In order to ensure individualized intervention, it is critical to assess all language domains to determine the individual's personal strengths and challenges (Finestack et al., 2009).

Brady et al. (2006) and Finestack et al. (2009) agreed that although research justifies an obvious need for speech and language intervention, there are currently no known studies which provide exact intervention techniques for individuals with FXS. However, it is noted that intervention programs targeting

language learning deficits appear appropriate; for example, programs could include Picture Exchange Communication System (PECS) (Brady et al., 2006; Finestack et al., 2009) and Responsivity Education/Prelinguistic Milieu Teaching (RE/PMT) (Finestack et al., 2009).

Parent responsivity is important for language development. Training parents to be responsive to their child's behaviors can provide positive influences on social and communication development due to correlations in high levels of mother responsivity and receptive and expressive language growth (Brady et al., 2006). Training parents to implement strategies focusing on prelinguistic development such as prompting for gestures and vocalizations, requesting for clarification, and responding to nonverbal and verbal communication attempts may be beneficial for communication growth in children with FXS (Brady et al., 2006).

Individuals with FXS demonstrate strengths in simultaneous processing (Finestack et al., 2009; Mirrett et al., 2003). Due to this strength, simultaneously providing visual and auditory input will aid in learning new concepts and enhance successful communication (Mirrett et al., 2003). Depending on the child's age and severity, it may be beneficial to use more concrete visual cues such as toys (Mirrett et al., 2003). The use of these visual cues will promote speech, language, attention,

comprehension, and allow for greater ease when transitioning topics and activities (Mirrett et al., 2003).

The environment plays an important role in language acquisition. Individuals with FXS demonstrate challenges in inhibitory control and sustained attention (Finestack et al., 2009). Due to these challenges, intervention should include consistent routines in an environment that is highly structured in order to eliminate distractions (Finestack et al., 2009; Mirrett et al., 2003).

Down syndrome. Roberts et al. (2007) highlighted that there is a lack of empirical support pertaining to speech intervention effectiveness for children with DS. On the other hand, evidence does indicate successful outcomes for communication and language development with prelinguistic intervention (Roberts et al., 2007). Evidence shows the importance of early intervention during the first few months of life for individuals with DS (Roberts et al., 2007). Higher communication scores were obtained from infants who received intervention initially after birth, rather than 3–6 months later (Roberts et al., 2007).

Price et al. (2007) discussed that language development in TD children can be greatly impacted by their environment. This also holds true for children with DS and FXS (Price et al., 2007). Parent-oriented intervention provides the child with gains in development of prelinguistic skills (Roberts et al.,

2007). In order for any intervention approach to be truly successful and for maximum gains to be achieved, the parents of the child must not only receive education about the intervention approach, but also utilize the target intervention practice in the child's daily life (Ronski & Sevcik, 2005). In other words, parents should play a primary role in early intervention practices (Ronski & Sevcik, 2005).

Programs such as the Hanen Program for Parents, PMT, a combination of the two, and PECS are appropriate for children with DS who are making the transition from preintentional to intentional prelinguistic communication (Abbeduto et al., 2007). To achieve greater gains in prelinguistic development, intervention approaches should be functional rather than structured (Abbeduto et al., 2007) and involve free-play (Roberts et al. 2007).

Abbeduto et al. (2007) continued to reason that the Hanen Program for Parents can be implemented alone or simultaneously with direct child intervention. This program should enhance parents' awareness of and responses to communication opportunities for their children (Abbeduto et al., 2007). Additionally, the Hanen Program for Parents also teaches parents to model words and other language skills, promote turn-taking, and create communication opportunities for their child (Abbeduto et al., 2007). Consequently, this program should promote turn-

taking during conversation and expand vocabulary (Abbeduto et al., 2007).

Abbeduto et al. (2007) and Roberts et al. (2007) suggested that RE/PMT is an effective prelinguistic intervention approach for children with DS. However, it was noted that greater gains are achieved in intentional communication when parents and interventionists respond to nonverbal communication attempts made to indicate an object or event, and not persisting when child is unresponsive to prompts for requests (Abbeduto et al., 2007; Roberts et al., 2007).

It was previously stated that the use of PECS can enhance language development in children with DS (Abbeduto et al., 2007). In accordance with Abbeduto et al. (2007) idea, Roberts et al. (2007) and Ronski and Sevcik (2005) indicated that AAC can be used to enhance language development while also supplementing verbal communication. Ronski and Sevcik (2005) encouraged interventionists to utilize AAC devices with nonverbal children to center the parent's focus on the intervention goals rather than on the child using verbal communication (Ronski & Sevcik, 2005). If spoken communication is a goal for the future, parents may require reinforcement or counseling that the intervention goals are steps leading to verbal communication.

AAC devices can provide multiple gains in communicative and cognitive development for children at a young age (Ronski & Sevcik, 2005). For optimum gains in communication development to occur, AAC should be introduced to children prior to communication failure (Ronski & Sevcik, 2005). Moreover, research suggests that imitation of words occurs more frequently when a sign is used simultaneously with verbal production of a word (Roberts et al., 2007). This multimodal input "increases the variety of communication options" (Roberts et al., 2007, p. 33).

In addition, past research demonstrated that the use of AAC devices does not hinder production or development of verbal communication (Roberts et al., 2007; Ronski & Sevcik, 2005). The use of AAC may actually reinforce development of verbal communication as well as language (Ronski & Sevcik, 2005). Parents often report using AAC as a means of communication for their child until the child's speech becomes more intelligible or until the child begins to communicate verbally (Ronski & Sevcik, 2005).

When selecting goals for treatment, the interventionist should target items which have the greatest impact on the individual's current communication needs (Roberts et al., 2007). In addition, the goals should be targeted in a way which facilitates generalization. However, generalization of

communication skills is often difficult for individuals with DS. Therefore, it is fundamental to incorporate generalization into intervention (Roberts et al., 2007). The knowledge from Roberts et al. (2007) and Ronski and Sevcik (2005) suggests that including parents in intervention and training parents how to implement their child's intervention in to their everyday life will aid in providing maximum opportunities for generalization to occur. Roberts et al. (2007) emphasized that the best opportunities for generalization arise when intervention is provided in the child's natural environment while also incorporating materials from the child's natural environment. Furthermore, modeling, prompting, and arranging the environment in a manner which promotes opportunities for the child to request items are also effective strategies for promoting generalization (Roberts et al., 2007).

Phonology

Fragile X syndrome. Roberts et al. (2005) and Roberts et al. (2007) stated that various factors such as cognitive skills and language deficits could inhibit children with FXS and DS ability to perform certain phonology related tasks. Therefore, a full examination of the individual is necessary. The clinician should assess auditory perceptual skills, verbal memory, oral motor functioning, and any other factors that may be compromising the child's speech and language acquisition. When

assessing children with FXS, phonological processes and word shapes should be assessed at the single word level (Roberts et al., 2005).

Down syndrome. Roberts et al. (2007) revealed that the occurrence of apraxia of speech and dysarthria is occasionally observed in individuals with DS. Therefore, assessment of muscle tone and speech motor coordination is critical for intervention planning. In addition, Roberts et al. (2005) and Roberts et al. (2007) stated that, due to the high frequency of syllable structure processes characterized by individuals with DS such as cluster reduction, deletion of final consonants, and syllable deletion, initial assessment should be done at the single-word level. Multisyllabic words with varying stress patterns and words that contain consonant clusters should be included (Roberts et al., 2005; Roberts et al., 2007). Also, due to high frequency of word-shape reductions, intervention should focus on production of all syllables within words (Roberts et al., 2005).

Conclusion

Overall, children with FXS and DS have a delay in language and communication development. Children with FXS exhibit delays in vocal and gestural communicative domains with strengths in imitation. On the other hand, children with DS demonstrate challenges in reciprocal eye gaze and commenting with strengths in imitation and use of gestures. Due to anatomical differences

found in children with FXS and DS, there is often a delay in speech acquisition which results in a delay in expressive language. Research shows inconsistent findings pertaining to phonological processes used by both populations. Furthermore, when developing an intervention plan for individuals with FXS and DS, the interventionist must provide an individualized plan by incorporating strengths and challenges of the child with inclusion of the family's priorities.

Obtaining an optimal understanding of genetic disorders such as DS would enhance the understanding and function of chromosome 21 (Abbeduto et al., 2007). Therefore, further research comparing DS to other specific disorders would be valuable in adapting intervention strategies for individuals with DS. Although DS is often compared to FXS and Williams syndrome (WS) in research investigations, a comparison of a larger number of genetic syndromes would provide a greater perspective of the function of chromosome 21, that would help in providing individuals with DS a syndrome specific intervention (Abbeduto et al., 2007). In addition, it would be beneficial to compare FXS to additional genetic syndromes, in order to obtain a greater understanding of the function of the X chromosome.

Although there are not many studies pertaining to prelinguistic language skills with the DS population, the existing literature does provide helpful information. However,

more research in this area would provide a variety of intervention techniques that can be provided to the DS population. Also, knowing the impact of variation in the frequency of intervention and intensity of intervention could result in additional gains upon intervention.

There is also a lack of research concerning prelinguistic language skills with the FXS population. Obtaining accurate information is of great importance, as well as a helpful tool when planning intervention for individuals with FXS.

It is evident that further research pertaining to intervention with children who have FXS is needed. The lack of evidence-based research with this population limits speech-language pathologists' (SLP) ability to provide EBP. Although the strategies provided in the literature provide a standard guide for therapist who work with the FXS population, the lack of evidence-based research reduced the chances of achieving the further progress.

Speech and language development differ between typically developing males and females. There has been little research conducted involving females with FXS and their speech and language development. The majority of research obtained about FXS has been conducted on males. Although FXS is more common in males than in females (Brady et al., 2006; Mirrett et al., 2003), further research should be conducted on females in order

to obtain knowledge pertaining to the developmental differences and severity between genders with FXS. This knowledge may provide SLPs with greater direction when providing intervention to females with FXS.

Further research is also needed to determine causes of speech unintelligibility frequently observed in children with DS. It is important to identify if unintelligible speech is truly due to anatomical differences found in these individuals or if it is due to phonological processes often displayed by the DS population. Determining an exact cause of unintelligible speech will provide SLPs with more accurate intervention approaches when targeting unintelligible speech in persons with DS during intervention.

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