How Does Fragile X Syndrome Affect Speech and Language Skills?

Children with Fragile X Syndrome (FXS), the most common known inherited cause of intellectual disability, typically experience communication difficulties. Children with other intellectual disabilities such as Down syndrome also experience communication difficulties. Further many boys with FXS (some estimates are as high as 35 percent) also are diagnosed with autism.

Professionals who work with children with language impairments need to tailor language interventions. But what if language difficulties in children with FXS have a different cause than language challenges in children with Down syndrome or autism? Is having an intellectual disability or autism the root cause of the communication challenges in a child with FXS or does having FXS present a unique set of challenges?

New research is beginning to answer these questions. Recent articles by FPG researchers in several journals explore different facets of communication in boys with FXS, boys with FXS and autism, boys with Down syndrome, and boys developing typically. These studies focused on boys because communication and other aspects of development are more severely affected in boys with FXS than girls. The findings will allow professionals to focus interventions efforts to best help children communicate effectively. This work is funded by the National Institute of Child Health and Human Development, National Fragile X Foundation, March of Dimes, and Ireland Family Trust. This snapshot summarizes the findings to date.

Language Use

This study looked at taped conversations between a researcher and child during a 45-minute play session designed to elicit language and social behaviors from children. The study examined the boys’ ability to maintain a topic of conversation, and how often they repeated themselves. Boys with FXS with and without autism repeated themselves more frequently during conversation than boys with Down
syndrome or typically developing boys of the same nonverbal mental age. In addition, boys with FXS with autism had a more difficult time maintaining a topic of conversation than boys with FXS and no autism, boys with Down syndrome, or typically developing boys. These findings suggest that while repetitive speech may be a defining feature of FXS, some of the language characteristics that are typically associated with FXS, such as the ability to stay on topic, may actually be a result of also having autism. Intervention for boys with FXS should focus on decreasing repetitive language and increasing language that maintains the topic of conversation.

**Vocabulary and Syntax**

A first study examined critical aspects of language development, receptive vocabulary (words understood when heard) and expressive vocabulary (spoken words) using standardized tests. Boys with FXS without autism were found to understand and use words just as well as typically developing boys of the same mental age. The boys with FXS with and without autism did not differ in comprehension or production of vocabulary. Both groups scored higher in their comprehension of vocabulary than the boys with Down syndrome. However, a second study based on a standardized test where the vocabulary was more complex, found that boys with FXS regardless of autism diagnosis scored lower than typically developing boys of similar mental age. Thus, when words are conceptually more difficult, the vocabulary comprehension of boys with FXS is more impaired. This study also examined comprehension of syntax and found that boys with FXS scored lower than younger typically developing boys at similar mental ages. The boys with FXS only (not autism) scored higher than the boys with Down syndrome in both comprehension of conceptually more difficult vocabulary and syntactic structures.

A third study examined the number of different words children used in conversation, as well as their syntax. Boys with FXS used less variety of words and shorter, less complex noun and verb phrases and sentence structure in conversation than typically developing boys of the same cognitive level. These findings suggest that boys with FXS are delayed in language comprehension and production of vocabulary and syntax. Vocabulary difficulties appear to be more evident with respect to comprehension of more complex information and in conversation rather than on a standardized test (conversation places a greater demand on language processing and knowledge). Further, the language difficulties of boys with FXS appear to reflect an overall delay in the ability to express themselves, rather than a specific grammatical or vocabulary delay, since both expressive vocabulary and syntax are impaired. Therefore, intervention should focus on both vocabulary and syntax (specifically, more complex structures) to improve the boys’ ability to use language.

**What is Fragile X Syndrome?**

Fragile X Syndrome (FXS) is a single gene disorder on the X chromosome that occurs in both males and females, but males are usually affected more severely. The disorder can be passed down through generations in a carrier status, with increasing chances of the gene expanding into the full disorder. FXS shuts down the gene responsible for producing FMRP, a protein essential for normal brain function.

Testing for FXS is recommended if there is a family history of FXS or intellectual disabilities of unknown causes. In addition, if a pediatrician observes the following characteristics in a child or hears about these behaviors and characteristics from the parents of a young child, fragile X testing should be considered.

**Developmental Features**
- Mental retardation
- Developmental delays
- Learning disabilities
- Pervasive developmental delays or autism
- Communication/language delays
- Perseverative speech

**Physical Features**
- Large, prominent ears
- Long, narrow face
- Flat feet
- Single crease that extends straight across the hand
- Double joints or loose joints
- Large testicles (seen in few young males but in the majority of pubescent males)

**Behavioral Characteristics**
- Handflapping and/or handbiting
- Inattentiveness or hyperactivity
- Easily overstimulated by touch
- Low frustration tolerance that may lead to tantrums or aggression
- Autistic-like features such as fascination with an unusual object or event like spinning fans or running water
- Avoidance of eye contact with others
- Attention deficits
- Hyperactivity

What is Fragile X Syndrome?
Speech
In the first study, speech production (the ability to pronounce words) was assessed using a single word standardized test. Boys with FXS pronounced single words as accurately as younger, typically developing boys of the same mental age. In addition, the boys with FXS and the typically developing boys pronounced words more accurately than the boys with Down syndrome.

In the second study, speech production of boys with FXS with and without autism were compared to the other groups on the single word standardized test. The speech of boys with FXS did not differ according to whether they had autism nor did it differ from typically developing boys. Boys with FXS and TD boys had higher speech production scores than boys with Down syndrome.

In the third, ongoing study of speech during conversation, boys with FXS were less understandable in conversation but did not differ in their accuracy of sounds during conversation as compared to typically developing boys of the same mental age.

In the fourth study, we found that boys with FXS did not speak faster, but paused less often, than typically developing boys of the same chronological age during conversation. However, boys with FXS spoke faster than younger typically developing boys of the same mental age.

These findings suggest that although boys with FXS are delayed in their speech production skills, their speech accuracy is similar to that of younger children at a similar mental age. However, the decreased intelligibility in the conversational speech of boys with FXS compared to the younger typically developing children suggests that other factors such as rate, intonation, and fluency may be affecting understandability. Intervention should focus on improving speech accuracy, with an emphasis on approaches that increase speech intelligibility in conversational speech.

Oral Motor
This study looked at the structure and function of features in the mouth including the lips, jaw, teeth, tongue, velopharynx (the area behind the uvula that directs air through the nose or mouth) and larynx (where sound is generated). In addition to having a physical examination of these features, children completed a series of oral motor exercises with speech (e.g., repeating words) and without speech (e.g., imitating mouth movements without using voice). Researchers found that boys with FXS and boys with Down syndrome had some structural differences compared to the typically developing boys.

The study also compared the boys’ oral function to speech function (the ability to say words). Boys with FXS like the typically developing boys did better at speech function tasks than oral function tasks. For boys with Down syndrome, however, speech function was lower than oral function. Further, both the boys with FXS and Down syndrome repeated single syllable words with greater accuracy than multiple syllable words, while the typically developing boys produced both types of words with equal accuracy. These findings suggest that boys with FXS and Down syndrome both have some atypical oral structure and oral motor function, yet differ in certain oral-motor patterns. The effects of these oral motor differences on speech production in boys with FXS are not clear.

Conclusion
These studies identify some of the communication difficulties of boys with FXS. Future research is needed to:

- Examine children’s use of language, including types of repetitive speech, narration (retelling of an event), and repair strategies (reformulation of unclear messages)
- Determine the role of rate, intonation, and fluency in the speech intelligibility in of boys with FXS in conversation
- Conduct longitudinal studies to determine how language patterns change over time
- Compare the language of boys with FXS to boys with autism who do not have FXS to determine the role of autism
- Examine the language characteristics of girls with FXS to determine if they have a similar language profile as boys with FXS
- Investigate mechanisms such as anxiety and memory and the role of the environment (such as responsiveness of the mother) that might explain some of these language characteristics in boys with FXS.
To Learn More


**Carolina Fragile X Project**  
[www.fpg.unc.edu/~fx/](http://www.fpg.unc.edu/~fx/)

**Carolina Communication Project**  
[www.fpg.unc.edu/~carolinacommunicationproject/](http://www.fpg.unc.edu/~carolinacommunicationproject/)

**Fragile X Information Center**  
[www.fpg.unc.edu/~fxic/](http://www.fpg.unc.edu/~fxic/)