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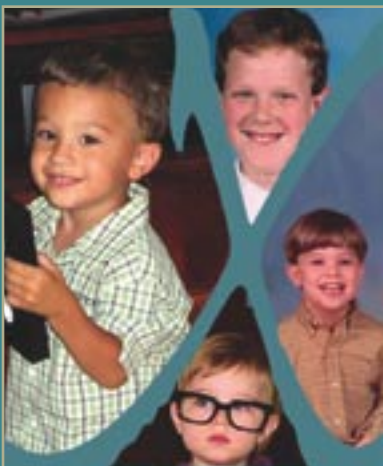
early developments

Speech, Language, and FXS

ASK FIVE-YEAR-OLD DAMON A QUESTION and his response is likely to be difficult to understand. When he meets new people, he becomes very quiet and avoids eye contact. Seven-year-old Tim answers questions in short sentences, but often is not on the topic being discussed. Tim and Damon's speech characteristics are typical of boys with fragile X syndrome (FXS)—girls who carry this genetic mutation are considerably less affected. By identifying these characteristics early in childhood, researchers at FPG hope to aid in the assessment of children with FXS and prescribe intervention strategies that will improve the speech and language of these children as they mature into adults.

Research into speech and language difficulties among young boys with FXS began at FPG in 2001 under the direction of Joanne Roberts, FPG senior scientist and professor of pediatrics and speech and hearing sciences at the University of North Carolina at Chapel Hill. Roberts and co-investigators David Zajac and Jack Roush are assisted by two post-doctoral fellows, Elizabeth Hennon and Beth Barnes, project coordinator Kathleen Anderson, and research staff Anne Edwards, Cheryl Malkin, Julia Jurgens, Lauren Moskowitz, and Siara Cowan.

Funded by the National Institute of Child Health and Human Development (NICHD) and the March of Dimes, the studies are examining the language of young boys with FXS in comparison to boys with Down syndrome and typically developing boys; and whether young boys with FXS have atypical hearing and auditory processing. The studies are also investigating the factors affecting poor speech intelligibility in conversational speech of young boys with FXS.



Roberts says most males with FXS will show moderate-to-severe delays in communication skills. “Phonological difficulties are common, including consonant substitutions and omissions, which is characteristic of developmentally younger children,” she says. “Conversational speech is often unintelligible, although single-word utterances often are understandable.”

Other speech characteristics of some males with FXS include a rapid and fluctuating rate, and repetitive speech. Males may also have trouble repeating multisyllabic sequences and may demonstrate oral motor difficulties.

With respect to language, boys with FXS often have delays in grammatical and vocabulary development. Some boys may have atypical pragmatic language, including frequent repetition of words, sentences, and topics, poor topic maintenance in conversation, difficulty answering direct questions, and gaze aversion.

“The cause for these pragmatic impairments has often been attributed to hyperarousal, although word retrieval difficulties, syntactic difficulties, and executive function deficits also have been cited as possible causes,” Roberts says.

Males with FXS often have prominent ears, although they do not appear to have hearing difficulties, Roberts says.

FPG’s studies are among the first in the nation to examine the communication development of young children with FXS. Roberts hopes that her findings will aid both in the assessment of children with FXS and in developing appropriate intervention strategies. Because physical characteristics associated with FXS are not present in early childhood, language impairments may offer the first sign of the



Photo: Don Trull, FPG

Assessing speech and language skills involves pointing to pictures with verbal directions.

condition. Roberts hopes that her research will lead to increased awareness of these speech and language impairments and, thus, earlier detection.

The cause of an individual’s speech and language difficulties and the particular domains affected will have important implications for intervention, although the specific contributing factors may be difficult to define. For example, if unintelligible speech is due to difficulties with sound patterns,

then intervention should focus on particular sounds or suppression of specific phonological processes.

However, if difficulties relate to such things as rate

of speech, then slowing the rate of speech would be useful. For children whose communication difficulties are particularly evident in conversation, using scripts and routines in more structured environments and transitioning to more naturalistic environments with peers, teachers, and family members whenever possible would be more useful.

“Our ultimate goal is to identify the types of intervention that are most appropriate for children with FXS,” Roberts says. “We want to know if there are specific strategies, different from those used with individuals with other forms of mental retardation, that we can recommend to help a child with FXS.” | [ed](#) |

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Finding Clues in Family Videos

Home videos are a great tool for preserving a family record, and especially for capturing those phases of early childhood that seem to pass so quickly. Now, FPG researchers are using retrospective analysis of home videos as a way of detecting previously unnoticed signs of fragile X syndrome among children aged 9 to 12 months.

Signs of FXS are often very subtle among infants and toddlers and may not become obvious for months or even years. Even if parents suspect something may be wrong with their child, physicians may downplay their fears, believing that the child is at the late end of normal development, and defer developmental evaluations until later in the second year of life. The result is that FXS is typically not diagnosed (in children without a family history of developmental delays) until children reach two years of age. Valuable time is lost during which intervention strategies could have been used. Clearly, there is a need for better methods of early detection. But since children with FXS are rarely identified during the first year of life, studying their earliest development has been virtually impossible.

Grace Baranek, FPG researcher and associate professor in UNC's Department of Allied Health Sciences, has used an innovative strategy—retrospective video analysis—to address this issue. This approach involves collecting home videos of children later diagnosed with developmental problems and analyzing them to see if unusual behaviors or delays can be detected. Raters are trained to view the videos and code a variety of social and sensory-motor behaviors in the context of natural daily activities. Her first study using this technique showed that by 9 to 12 months of age, the behaviors of children later diagnosed with autism were markedly different from those of children with delayed or typical development.

Baranek has just completed video analysis research on FXS conducted in a similar fashion to her autism research. She coded the same behavioral markers at 9 to 12 months, plus a few additional features such as repetitive movements that may reflect hyperarousal common to children with FXS. Findings so far confirm that even at this young age, children with FXS do exhibit markedly different behaviors than their normally developing peers, and can also be differentiated from children with autism.



Photo: Don Trull, FPG

Grace Baranek (upper left) examines home videos of infants at her laboratory with team members Fabian David and Cassandra Danko.

“Infants with FXS were showing significant delays in the way they played with toys,” Baranek says. “We also observed a high level of unusual postures, reflecting motor difficulties.”

Baranek says her findings may not be practical yet. Although she has proved that early signs of developmental problems are evident during the first year of life, they may not be obvious to pediatricians or other professionals who don't have the advantage of watching, rewatching, and coding videotapes. However, these findings provide important clues about how FXS is expressed in infancy and hopefully will stimulate other research on early development.

Furthermore, “awareness of these early features of FXS may help a clinician to select evaluation methods and interventions that target the family's specific needs,” Baranek says. “For example, activities designed to promote play skills, or environmental modifications that support engagement in daily activities would be recommended.” ■

PARENTS AND PROFESSIONALS SEEKING MORE INFORMATION ON FRAGILE X will want to access FPG's new website for the Fragile X Information Center (www.fpg.unc.edu/~fxic). The website provides a wealth of information, including the effects of FXS on young children, summaries of research findings, family experiences and intervention strategies, services available for children with disabilities, and how to advocate for those services. Lists of publications, personnel, and resources related to FXS are also included.

Jane Roberts, manager of the information center, says FPG's website is geared toward those who want detailed and objective information on FXS and related disabilities with references to specific studies. The website content is written by a team of authors and reviewed by a panel of parents and professionals.

Parents who have previewed the site have given it positive reviews. "The fragile X website is an essential tool for educators and parents alike," says Nichole Tooz, parent and educator. "Its friendly format provides relevant information that is not overwhelming or foreign, even to those new to fragile X. It addresses the most pertinent topics with a simple, yet thorough style. I personally have used information from this website in public presentations and with my son's teachers and therapists. It has become an on-going resource, as the site is frequently updated with contributions from parents and professionals."

In the future, FPG plans to add content in the areas of temperament, academic achievement, educational transitions, educational services, toilet training, social skills, sibling issues, disability laws, alternative therapies, effects in permutation carriers, mental health issues, and communication skills.

FPG's Fragile X Information Center has been funded by a grant from the Ronald McDonald House Charities. Other funding sources include the Office of Special Education programs of the US Department of Education, the FRAXA Research Foundation, and the National Fragile X Foundation. Other websites available include those of the National Fragile X Foundation (www.fragilex.org) and the FRAXA Foundation (www.fraxa.org). **|ed|**

Jane Roberts, manager of the Fragile X Info Center, monitors the website.



Photo: John Cotter, FPG

Fragile X Info Center