Speech & Language in Fragile X & Down Syndrome

Joanne Roberts, Ph.D.
Gary E. Martin, M.A.
FPG Child Development Institute (FPG)
University of North Carolina at Chapel Hill (UNC)

Elizabeth Barnes, Ph.D.
North Carolina State University

Johanna Price, Ph.D.
Mississippi University for Women

David J. Zajac, Ph.D.
School of Dentistry, UNC

Kerry Callahan-Mandulak, M.A.
FPG, UNC

American Speech-Language-Hearing Association Convention
Boston, MA November, 2007

Research supported by:

National Institute of Health - NICHD (R01 HD38819, R01 HD 44935, & R03 HD40640), National Fragile X Foundation, March of Dimes, & Ireland Family Trust
Outline

- Genetics, Diagnosis, & Behavioral Characteristics
- Language Findings
  - Vocabulary & Syntax
  - Pragmatics
- Assessment & Intervention: Vocabulary, Syntax, & Pragmatics
- Speech Findings
  - Speech Production
  - Oral Motor Skills
- Assessment & Intervention: Speech
- Directions & Questions
ACKNOWLEDGEMENTS

- Children & Families
- Carolina Communication Project

- Other Colleagues: Peg Burchinal, John Sideris, Weejy Neebe, Jan Misenheimer, Steve Hooper, & Dave Zajac

- Funding Agencies: National Institute of Child Health and Human Development, March of Dimes, National Fragile X Foundation, & Ireland Family Trust

Kristin Cooley
Anne Edwards
Bruno Estigarribia
Anne Harris
Gary Martin
Amy Spencer
Anne Taylor
Cheryl Malkin
Sabrina Smiley
Joanne Roberts
General Considerations for Working with Children with FXS

- Develop consistent routine with structured activities & structured environment.
- Monitor frustration levels; allow breaks to minimize anxiety.
- Accommodate arousal & sensory needs.
- Be sensitive to gaze aversion, proximity to child.
- Consider potential impact of medication.
General Considerations for Working with Children with Down Syndrome

- Consider individual profile of strengths and weaknesses
- Monitor frustration and attention levels; allow breaks
- Consider possible vision difficulties
- Recognize low tone and provide adequate support
- Manage OME and associated hearing loss
Carolina Communication Project
Carolina Communication Project

- Speech & language characteristics of boys with fragile X syndrome (FXS) & Down syndrome (DS)
  - FXS with and without autism spectrum disorders (ASD)
- Factors affecting poor speech intelligibility in young males with FXS & DS
- Role of child & family characteristics on speech & language in FXS & DS
Model of Speech & Language Production in Males with FXS & DS

**INPUT**
- Hearing & Perception
- Attention
- Selective Listening

**ORGANIZATION**
- Language
- Memory
- Cognition

**OUTPUT**
- Retrieval
- Sequencing & Coordination

Speech Production
Expressive Language
Participants

- Fragile X (FXS) - 86 boys, 2-15 years
  - FXS ONLY (FXS-O) - 38
  - Autism Spectrum Disorder (FXS-ASD) - 48
- Down Syndrome (DS) - 52 boys, 3-16
- Typically Developing (TD)
  - Developmental Age Matches (DA), 49 boys, 2-6 years
- FXS, DS, & DA similar Leiter Nonverbal mental age
Receptive & Expressive Language
Language in FXS & DS

- Language delays common; considerable variability
- Expressive more delayed than receptive in DS, unclear in FXS
- Inconsistent findings regarding whether language skills of boys with FXS are similar to those of typically developing boys at similar developmental levels
Research Questions

- Do language skills of boys with FXS (with/without ASD), boys with DS, & TD boys differ?
  - Receptive
  - Expressive
Method

- Test of Auditory Comprehension of Language-3 (TACL-3)
  - Vocabulary
  - Grammatical Morphemes
  - Elaborated Phrases & Sentences

- Language sample - Autism Diagnostic Observation Schedule
  - 100 utterances transcribed using CHILDES
  - Syntax complexity – Index of Productive Syntax (IPSyn)
TACL-3 Age Equivalents for Boys with FXS-Only, FXS-Spectrum, FXS-Autism, DS, & TD

Price et al., 2007
Receptive/Expressive Language: Summary

- Both boys with FXS & DS scored lower in receptive & expressive language than TD boys
  - Boys with FXS & DS have expressive & receptive language delays
- Boys with FXS with & without autism did not differ in measures of receptive or expressive language
- Boys with DS scored lower than all boys with FXS on measures of overall expressive syntax & expressive questions/negations
Language: Pragmatics
Pragmatics: Background

- Perseveration, poor topic maintenance, tangential language, inappropriate turn-taking, and gaze aversion reported in studies of children, adolescents, & adult males with FXS
Pragmatics: Method

- Coded 100 complete & intelligible turns during ADOS
  - Examiner-child interaction of structured & semi-structured activities
- Topic Continuity - maintenance or change
- Topic Quality
  - Maintenance - elaborate, adequate, or noncontingent
  - Change - appropriate or noncontingent
- Perseveration
  - Repetition of words, phrases, sentences & topics
Examples of Noncontingent Language

- Noncontingent Maintenance
  - E: Have you ever gone to a volcano before?
  - C: Lava.

- Noncontingent Change
  - E: You know what? It’s time to have some dinner!
  - C: I get my ball. I get my ball. It worked!
Noncontingent Topic Maintenance:
% Turns Noncontingent Maintenance for FXS (with & without ASD), DS, & TD DA Groups After Controlling for Nonverbal Mental Level

FXS ASD more noncontingent topic maintenance than FXS Only, DS, & TD

Roberts et al., 2007
Noncontingent Topic Change:
% Turns Noncontingent Change for FXS (with & without ASD), DS, & TD DA Groups After Controlling for Nonverbal Mental Level

FXS ASD more noncontingent topic changes than FXS Only, DS, & TD

Roberts et al., 2007
Perseveration:

% Turns Perseveration for FXS (with & without ASD), DS, & TD DA Groups After Controlling for Nonverbal Mental Level

Roberts et al., 2007

FXS ONLY & FXS ASD higher on perseverance than TD & DS, but equal to each other.
Summary

- Boys with FXS and ASD produced more noncontingent discourse than boys with FXS only, boys with DS, and TD boys
  - Autism status in FXS affected contingent discourse
- Boys with FXS (regardless of autism status) used more perseveration than boys with DS and TD boys
- Noncontingent language in FXS may be a function of the high rate of autism in FXS, whereas perseveration may be characteristic of FXS
Assessment & Intervention: Vocabulary, Syntax, & Pragmatics
Manage Otitis Media with Effusion & Hearing Loss

- Routinely screen for OME and associated hearing loss
- Use low gain hearing aids/other amplification devices when there is a hearing loss
- Use environmental strategies when there is a hearing loss
  - Reduce background noise
  - Provide close access to the speaker

Roberts, Chapman, Martin, & Moskowitz, 2008
Assess Language in a Variety of Contexts

- Different contexts may elicit different levels of language complexity
  - Standardized Tests
  - Conversation
  - Narration

- Assess language during contexts with differing social demands
  - Large groups
  - One-to-one
  - Familiar vs. unfamiliar persons

Roberts et al., 2008
Consider Cognitive Profile

- **Down Syndrome**
  - Focus on relative weaknesses in auditory-verbal working memory and phonological working memory
    - Provide repeated opportunities to hear words and sentences
    - Provide simple and repeated instructions
  - Build upon relative strength of visual-spatial short-term memory
    - Use visually based materials

- **Fragile X Syndrome**
  - Build upon relative strengths in visual memory, verbal imitation, simultaneous processing, and long-term memory for meaningful information
    - Use picture cues and modeling
    - Associate vocabulary words with a meaningful context

Roberts et al., 2008
Consider Anxiety & Approaches for Children with Autism

- Anxiety/Hyperarousal
  - Do not demand eye contact
  - Keep comfortable distance
  - Use less directive, more incidental approach

- Autism
  - Use structured behavioral approaches to teach vocabulary and syntax
  - Use naturalistic approaches to teach pragmatics (incidental teaching)

Roberts et al., 2008
Vocabulary & Syntax

- **Vocabulary**
  - Create opportunities to learn to understand and use more complex vocabulary
  - Provide repeated opportunities to hear and produce new words in meaningful contexts

- **Syntax**
  - Target specific syntactic forms
  - Use conversational recasts to develop complex syntax (expand on a child’s utterance with semantic or grammatical information)

Roberts et al., 2008
Pragmatics: Perseveration

- Consider possible underlying causes
  - Anxiety, receptive/expressive language, attention-seeking
- Monitor anxiety levels
  - Establish predictable routines
  - Help transition with visual display of what is going to happen
- Provide increased processing time
- Reduce complexity of utterances directed at the child
- Verbally redirect

Roberts et al., 2008
Pragmatics: Noncontingent Language

- Use games with familiar routines or those that require contingent responding (e.g., collaborating with a group to make a story)
- Allow child to select the topic of conversation
- Use video modeling
- Use visual cues
- Consider peer-mediated interventions
  - Teach TD peers to initiate and answer questions

Roberts et al., 2008
Pragmatics: Topic Elaboration

- Consider possible underlying causes
  - receptive/expressive language, cognitive deficits, anxiety
- Have child give directions or plan an event and direct the child to give further information
- Support child-initiated topics with specific questions (open-ended) and comments
- Pause to allow child sufficient time to respond
- Focus on activities, topics, and materials of interest to the child

Roberts et al., 2008
Promote Generalization of Language Targets

- Use naturalistic language methods (e.g., milieu language teaching)
- Provide multiple exemplars of targets skills
- Integrate materials from the child’s home or classroom into intervention (e.g., storybooks, textbooks)
- Work on language in a variety of settings (e.g., classroom, home, community)
- Work on language with a variety of communication partners (e.g., teachers, parents, siblings, classmates)
Speech
Phonology Skills of Boys with FXS or DS

- Boys with FXS & boys with DS are less intelligible than TD boys in conversation.

- Does phonological accuracy and speech differ for boys with FXS (with and without autism) from boys with DS and TD boys at similar mental ages in:
  - single words?
  - conversational speech?
Method

- Single Words
  - Goldman-Fristoe Test of Articulation Sounds in Words Subtest

- Conversational Speech
  - 100 first occurrence words during structured play

- Computerized Profiling Profile of Phonology (PROPH) analyzed single words and conversational speech

- Computed % Consonants Correct, Phonological Process Occurrence and % Intelligible Words (conversational speech only)
PCC for Boys with FXS, DS, & TD – Single Words

Roberts et al., 2005
Speech Production in Single Words On GFTA Over Time

<table>
<thead>
<tr>
<th>Age Equivalent Score (in Months)</th>
<th>0</th>
<th>20</th>
<th>40</th>
<th>60</th>
<th>80</th>
<th>100</th>
</tr>
</thead>
<tbody>
<tr>
<td>TD</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>FXS Only</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>FXS ASD</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>DS</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Time

Roberts et al., 2007
Phonological Processes for Males with FXS, DS, & DA – Single Words

Roberts et al., 2006
Oral Motor Function: Comparison of Alternating Speech Movements

- FXS (n=53)
- DS (n=34)
- TD (n=37)

* indicates statistical significance.
Oral Motor Function: Accuracy on Single vs. Multi-syllable Words

Performance on Single vs. Multi-syllable Words

FXS (n=53)  DS (n=34)  TD (n=37)

Monosyllable  Multisyllable

*
Results: FXS

- Boys with FXS have similar speech production accuracy as TD boys in single words & conversation
- Boys with FXS produced more unintelligible words than TD boys
- Autism status did not affect phonological accuracy in single words or conversation
- Coordinated Speech Motor function reduced compared to TD boys
Results: DS

- Boys with DS performed lower on speech production accuracy, used more phonological processes than boys with FXS and TD boys.
- Boys with DS produced more unintelligible words than TD boys.
- Coordinated Speech Oral Motor function reduced compared to TD boys.
Consider Role of Factors Contributing to Speech Disorders

- Otitis media & hearing loss
- Oral motor skills
- Cognition and language skills
- Sensory Deficits / Arousal (especially in FXS)
- Assess single words and connected speech
  - Syllable/word shapes, phonological process occurrence
- Prosody
- Communication environment

Roberts, Stoel-Gammon, & Barnes, 2008
Provide Intervention to Improve Accuracy & Increase Intelligibility

- Naturalistic, phonological approaches adapted for children with MR
  - Cycles (double length of duration)
  - Target primary patterns first (impact intelligibility)
    - Syllableness
    - Anterior/posterior contrasts
    - /S/ clusters
  - Address “syllableness”
    - children using syllable structure processes

Kent & Price, 2008; Roberts, Stoel-Gammon, & Barnes, 2008
Provide Intervention to Improve Accuracy & Increase Intelligibility

- Improve articulatory accuracy
  - Complexity Approach
- Normalize prosody to increase intelligibility
- Adapt approaches for Treatment of Apraxia –
  - increase motor command
  - Address prosody (rate, intonation, rhythm, stress)
  - Multimodal cues
  - Vary social and linguistic context of speech practice

Kent & Price, 2008; Roberts, Stoel-Gammon, & Barnes, 2008
Promote Generalization of Speech

- Teach overlearned patterns
- Multiple trials, exemplars
- Select relevant targets
  - family members, ADLs
- Practice in multiple natural communicative contexts

Roberts, Stoel-Gammon, & Barnes, 2008
Directions & Questions
Directions

- Samples: FXS with and without ASD and ASD only comparison; Girls with FXS and DS
- Speech & language characteristics specific to FXS & DS
  - Pragmatics
  - Speech intelligibility
- Growth of boys’ speech/language
- Child & family predictors of speech & language
For more information...

- Contact Joanne Roberts --- joanne_roberts@unc.edu
- Research opportunities
  - NIH Minority Fellowship
    - All academic levels
  - Post-doctoral fellows
- We are recruiting girls & boys with FXS; girls & boys with DS, and boys with autism
Thank You