Speech and Intelligibility Characteristics in Fragile X and Down Syndromes

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Outline

- Characteristics of FXS and DS
- Project Aims and Specific Research Questions
- Previous Findings – Speech Rate
- Current Studies – Segmental Features and Single Word Intelligibility
- Conclusions & Questions
Background

Fragile X Syndrome (FXS)

- Most common cause of *inherited* intellectual disability
- Fragile X Mental Retardation 1 (FMR1) gene does not produce FMR protein
- 1-in-2500 to 1-in-5000 individuals (full mutation)
- Males more severely affected than females
Background

- **Fragile X Syndrome (FXS)**
  - 18 to 74% may have co-diagnosis of autism or autism spectrum disorder (ASD)
    - Difficulty with adaptive & social skills
Background

- **Fragile X Syndrome (FXS)**
  - Moderate to severe delays in speech & language
  - Speech intelligibility affected to various degrees
    - Connected speech more affected than single words
    - Apraxia-like errors: Inconsistent, phonemic transpositions and omissions; prosodic disturbances
Speech characteristics of FXS:

- Frequent articulation errors (omissions, substitutions, distortions)
- Fast & fluctuating rate of speech
- Cluttering (Hanson et al., 1986)
- Unequal rhythm (Brun-Gasca & Antigas-Dallares, 2001)
- Peculiar sounding speech
Background

- **Down Syndrome (DS)**
  - Most common *genetic* cause of intellectual disability
  - 1-in-700 to 1-in-800 live births
  - Caused by a trisomy set of genes located on chromosome 21
Considerable variability in phenotypic features in DS

- Congenital heart defects ≈ 40%
- Eustachian tube dysfunction common due to midface hypoplasia
- Hypotonia (low muscle tone) common
- Cognitive impairment is ubiquitous
Background

Speech Characteristics of DS

- Reduced rate
- Resonance disturbances
- Phonological processes typical of younger-aged children
- Poor intelligibility
Project Aims & Specific Research Questions

1) Identify syndrome-specific differences in speech characteristics (segmental, prosody/voice, and intelligibility) in FXS or DS

2) Determine how segmental and prosody/voice features relate to speech intelligibility in boys with FXS. Are there different predictors of intelligibility among boys with FXS, DS, and typical development?
Previous Findings: Rate and Prosody

Research Question:
Do boys with FXS differ in articulation rate from typically developing boys of similar nonverbal mental age (DA) and typically developing boys of similar chronological age (CA)?

Zajac et al. (2006)
Study Participants

- 38 boys with FXS
- 21 DA boys (similar nonverbal mental age)
- 16 CA (similar chronological age)

Zajac et al. (2006)
Method: Speech Samples

- For FXS and DA boys conversation samples obtained from ADOS (Autism Diagnostic Observation Schedule, Lord, Rutter, DiLavore, & Risi, 2002)

- CA conversation samples taken from 10 minute discussion with examiner

Zajac et al. (2006)
Method: Rate Determination

- **5 INTELLIGIBLE** utterances of at least 3-4 syllables taken from each child

- Duration of utterance (in seconds) & number of syllables per second (Articulation Rate) computed

- Number of Pauses (periods without voicing exceeding .20 seconds) calculated

Zajac et al. (2006)
Results: Means of articulation rate in syllables-per-second (sps)

![Articulation Rate Graph]

- FXS (N=35)
- DA (N=19)
- CA (N=16)
Results: Duration and Pauses

- Mean Duration of Utterance (seconds)
- Mean Number of Pauses Used

Graph showing comparisons for FXS, DA, and CA.
Results: Summary

- DA boys had slower rate than CA boys
- Boys with FXS had a faster rate than DA boys but not CA boys
- Boys with FXS used shorter utterances than both CA and DA boys (cluttering?)
- Boys with FXS tended to pause less often than DA boys and CA boys (cluttering?)

Zajac et al. (2006)
Results: Summary

Why no articulation rate (sps) difference?

- Zajac et al. (2006) suggested that other factors may contribute to perceived rate
  - Utterance duration
  - Length & number of pauses
  - Prosodic factors
    - Unequal rhythm?
Zajac et al. (2009)

1) Do listeners perceive boys with FXS as speaking faster than CA controls even when utterances are matched on overall acoustically-determined articulation rate?

2) If so, what factors within the utterances account for perceived articulation rate?
Study Participants

- 12 boys with FXS (10-15 yrs old)
  - 5 co-diagnosed with ASD
  - Subset from Zajac et al. (2006)
- 12 typically developing boys of similar chronological age (CA)
- 10 adult listeners (undergrads) unfamiliar with speech of children with FXS

Zajac et al. (2009)
Sentence Imitation Task
Method:

- All boys were matched on articulation rate based upon the sentence imitation task
  - All used an articulation rate between 3.5 and 4.5 sps for at least three sentences
  - The sentences contained no pauses > 200 ms

Zajac et al. (2009)
Direct Magnitude Estimation (DME)

- 10 adult listeners judged 72 sentences (3 sentences from each of the 24 boys with FXS and CA)

- Listeners rated sentences for how fast they sounded compared to a standard sentence assigned a modulus value of 100

Zajac et al. (2009)
Predictor Variables

- **Voice and Prosody**
  - Mean Fo level
  - Final Fo drop
  - Articulation rate in sps minus the final word (Rate_MFW)

- **Segmental**
  - Percent consonants correct (PCC)

Zajac et al. (2009)
Results: Question 1

GM = 114

GM = 94

FXS (N=12)  CA (N=12)
Results: Question 1

- **DME Articulation Rate**
  - **CA**
    - N=12
    - GM=94
  - **FXS**
    - N=7
    - GM=107
  - **FXS-ASD**
    - N=5
    - GM=125
Results: Question 2

Multiple linear regression to predict perceived rate

In:
- Rate_MFW \( p < .01 \) \( r^2 = .75 \)
- Fo drop \( p < .01 \) \( r^2 = .91 \) \( \Delta r^2 = .16 \)

Out:
- Diagnostic group (CA, FXS, FXS-ASD)
- Fo
- PCC

Zajac et al. (2009)
Conclusions

- Boys with FXS-ASD were judged to articulate faster than CA boys.
- When the final word of the sentence was excluded, *actual* articulation rate (sps) was faster for boys with FXS-ASD.
- Atypical prosody relative to final Fo appears to contribute to perceived rate.

Zajac et al. (2009)
Current Studies

1) To determine if **segmental** differences exist between FXS, DS, and typically-developing boys matched on chronological age (TD-CA).

2) To determine if **single-word intelligibility** differences exist between FXS, DS, TD-CA, and typically-developing boys matched on language age (TD-LA).
Segmental Study – Aim 1: Participants

- **29 Boys with FXS**
  - 12 FXS Only
  - 17 FXS-Autism Spectrum Disorder
  - Ages 6:4 – 14:2
- **9 Boys with DS**
  - Ages 8:0 – 14:6
- **6 TD CA boys (chronological age match)**
  - Ages 8:4 – 11:6
Segmental Study – Aim 1: Method

- Four phonetic contrasts
  - Duration of stress/lax vowels (i/I)
  - Sibilant duration of /s/ and /z/
  - VOT of /t/ and /d/
  - First linear spectral moment of /s/ and /ʃ/
Segmental Study – Aim 1: Method

- Recordings made in sound-booth
- Five productions each of
  - Pete/pit
  - sack/Zack
  - tot/dot
  - sack/shack
Segmental Study – Aim 1: Method

- TF32
  - Duration of i/l and s/z
  - VOT of t/d
  - 1st spectral moment of s/sh
Results: Duration of Tense/Lax Vowels

Duration (msec.)

FXS (n=29)

DS (n=9)

TD-CA (n=6)
Results: Duration of Tense/Lax Vowels

Duration (msec.)

FXS-O (n=12)

FXS-ASD (n=17)
Results: Duration of /s/-/z/
Results: Duration of /s/-/z/

Duration (msec.)

- sack
- Zack

FXS-O (n=10)
FXS-ASD (n=16)
Results: VOT of /t/-/d/

<table>
<thead>
<tr>
<th></th>
<th>Duration (msec.)</th>
</tr>
</thead>
<tbody>
<tr>
<td>FXS (n=29)</td>
<td>75</td>
</tr>
<tr>
<td>DS (n=5)</td>
<td>75</td>
</tr>
<tr>
<td>TD-CA (n=4)</td>
<td>150</td>
</tr>
</tbody>
</table>

* Indicates significant difference.
Results: VOT of /t/-/d/

- Duration (msec.)
  - 0
  - 25
  - 50
  - 75
  - 100
  - 125
  - 150
  - 175
  - 200

- (n=12) (n=17)

- FXS-O
  - (n=12)

- FXS-ASD
  - (n=17)
Results: 1\textsuperscript{st} Spectral Moment of /s/-/sh/

![Bar chart showing 1\textsuperscript{st} Spectral Moment (kHz) for different groups.](chart.png)

- **FXS** (n=23)
  - sack
  - shack
- **DS** (n=4)
  - sack
  - shack
- **TD-CA** (n=6)
  - sack
  - shack

The chart illustrates the 1\textsuperscript{st} Spectral Moment for the sounds /s/ and /sh/ across different groups. The circles highlight specific comparisons between the groups.
Results: 1st Spectral Moment of /s/-/sh/
Summary: Aim 1 Segmental Results

- Boys with DS tended to produce
  - Shortest tense/lax vowel difference
  - Shortest s/z difference
  - Smallest 1st spectral moment difference
- No differences between boys with FXS-O and FXS-ASD
Intelligibility Study – Aim 2: Participants

- **12 Boys with FXS**
  - Ages 6:4 – 14:2

- **12 Boys with DS**
  - Ages 8:0 – 14:6

- **8 TD CA boys (chronological age match)**
  - Ages 8:4 – 11:6

- **8 TD LA boys (language age match)**
  - Ages 4:0 – 6:3
Intelligibility Study – Aim 2: Method

- Single-word intelligibility test
  - 50 unique words randomly selected from 510 words
  - Based on Zajac et al. (2011)
Single Word Intelligibility Test
Intelligibility Study – Aim 2: Method

- 20 adult listeners
  - Normal speech and hearing
  - No experience with FXS or DS speech
- 4 groups of 5 different listeners
  - Orthographically transcribed 10 children
    - 3 FXS, 3 DS, 2 TD-CA, 2 TD-LA
Intelligibility Study – Aim 2: Results

Single Word Intelligibility Scores

<table>
<thead>
<tr>
<th>Condition</th>
<th>Percentage Words Correct</th>
</tr>
</thead>
<tbody>
<tr>
<td>TD-CA</td>
<td>83%</td>
</tr>
<tr>
<td>TD-LA</td>
<td>68%</td>
</tr>
<tr>
<td>FXS-A</td>
<td>58%</td>
</tr>
<tr>
<td>FXS-O</td>
<td>57%</td>
</tr>
<tr>
<td>DS</td>
<td>33%</td>
</tr>
</tbody>
</table>
Conclusions

- There appears to be syndrome specific differences in single word intelligibility between FXS and DS
  - Related to reduced segmental contrasts by DS speakers
- While FXS boys were less intelligible than younger language-age matched boys, DS boys were even less intelligible
Clinical Implications

- DS boys may benefit most from therapy directed to improve phonetic contrast distinctions at the single word level.
- FXS boys – especially those with ASD – may benefit most from therapy directed to reduce rate and normalize prosody at the sentence level.
Thank You!

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Questions