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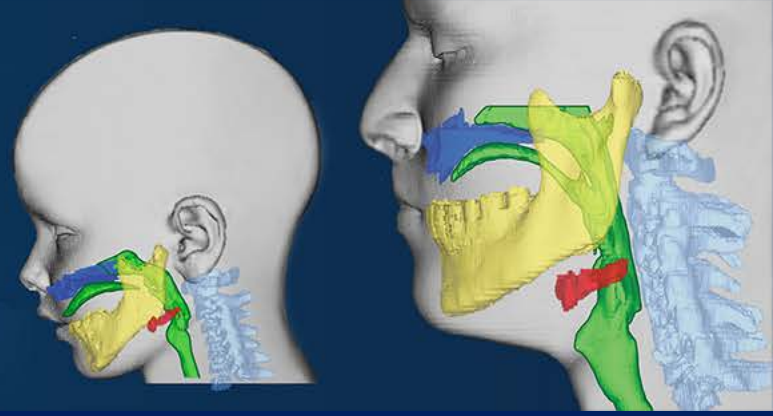

Speech Production in Children and Adults with Down Syndrome: Perceptual and Acoustic Data

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Down syndrome (DS) - Trisomy 21

1. The most common form of intellectual disability (1 in 700-800 births).

2. One of the most complex genetic perturbations compatible with survival.
   - ➢ In only 2 generations, life expectancy increased from 12 to nearly 60 years.

3. Speech intelligibility often is compromised and can be a lifelong problem
   (Kent & Vorperian, 2013; Kumin, 1994; Wild et al., 2018).
   - ➢ Voice enabled technology -- Google initiative: “Project Understood”.

4. The speech impairments relate to multiple factors, such as:
   - motor impairments (hypotonia, dysarthria, apraxia of speech)
   - phonological delay or disorder
   - hearing loss
   - intellectual disability
   - craniofacial and laryngeal dysmorphologies

Dysmorphologies and Dysfunctions

Small midface

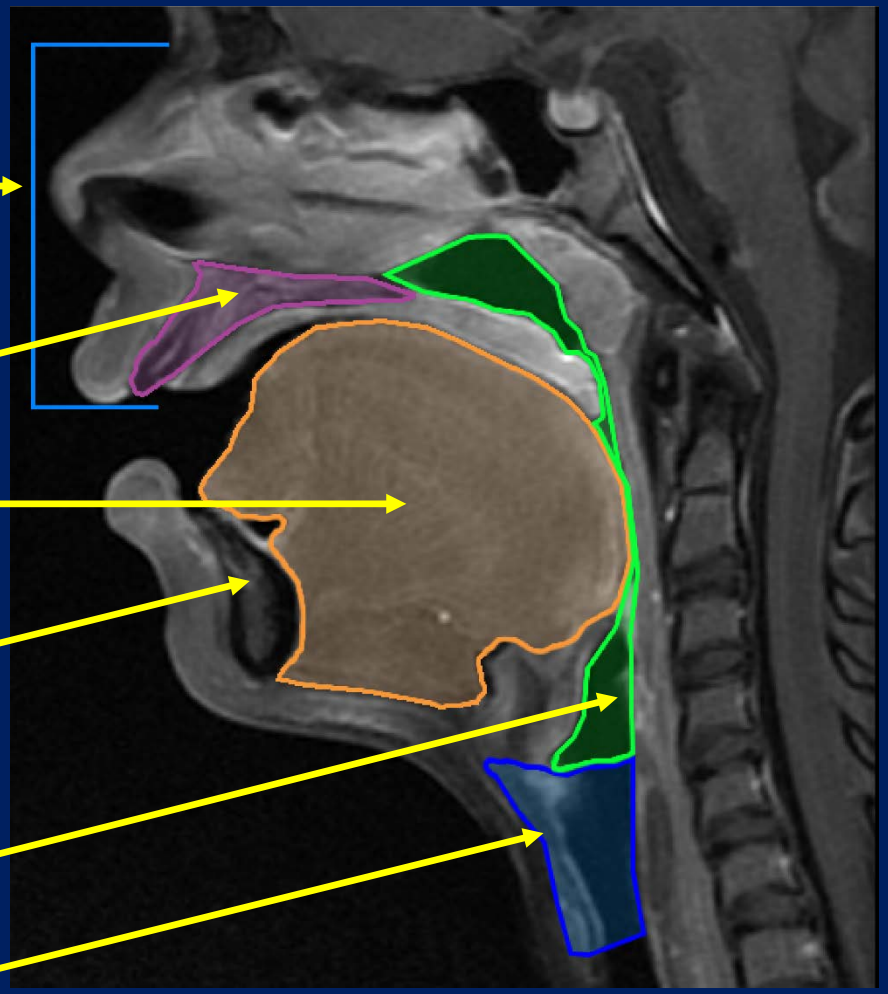
Short and narrow palate

Pseudomacroglossia

Stage III malocclusion and anterior open bite

Constricted airway

Laryngomalacia



Hypotonia*

- Labial
- Lingual
- Laryngeal

*Chu & Barlow (2016) *Advances in Communication Disorder*, 2-40.

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Conflicting Reports on Speech Disorder in DS-- A Few Examples…

1. Articulatory working space (e.g. vowel space area):
   - **Reduced** (Abolhasanizadeh & Olyiaiee, 2018; Bunton & Leddy, 2010; Moura et al. 2008)
   - **Increased** (Rochet-Capellan & Dohen, 2015)
2. Phonatory dysfunction:
   - **Vocal hyperfunction** (Pebbili et al., in press)
   - **Vocal hypofunction** (Wold & Montague, 1979, Moran & Gilbert, 1982)
   - **Nonmodal phonation** (Jeffery, Cunningham, & Whiteside, 2018)
3. Oral-nasal resonance:
   - **Hypernasality** (Montague & Hollien, 1973; Rolfe, Montague, Tirman, & Vandergrift, 1979)
   - **Hyponasality** (Jones et al., 2019)
   - **Other atypical resonance** (Fourakis, Karlsson, Tilkens, & Shriberg, 2010; Jones et al., 2019)

Present Study – Research Questions

Overarching Question:

What is the speech subsystem profile in Down syndrome?

1. *What are the dominant perceptual features for sustained vowels and short sentences?*
2. *What are the acoustic characteristics of vowels and fricatives?*

How can we distinguish functional from structural aspects of the speech disorder?

Methods: Participants and Speech Samples

Down syndrome

- 82 children and adults
- Ages 3 to 53 years
- 40 females
- 42 males

Neurotypical

- 407 children and adults
- Ages 4 to 92 years
- 212 females
- 195 males

Speech sample: Sustained vowels, monosyllabic words, short sentences

Appropriate for individuals with limited cognitive and language abilities over the age range

Speech Samples for this Report

Five words for each corner vowel*

/i/- *bead, bee, eat, sheep, feet* /u/- *boo, boot, zoo, hoot, shoe*

/æ/- *bath, bat, cat, hat, sad* /ɑ/- *dot, hop, pot, top, hot*

**Also used in a single-word intelligibility study (Wild et al., AJSLP, 2018)*

Three short phrases or sentences

The blue duck quacks, Pop the bubble, Cars go beep beep

Sustained vowel /ɑ/

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Perceptual Ratings – 22 Features in 4 Subsystems

1) Phonation/respiration:

Roughness
Breathiness
Strain
Pitch variability
Pitch break
Loudness variability
Dysphonia severity

2) Articulation and resonance:

Imprecise consonant articulation
Distorted vowels
Irregular articulatory breakdown
Atypical resonance

- *Hyponasality*
- *Hypernasality*
- *Cul de sac resonance*
- *Other atypical resonance*

3) Suprasegmental:

Disturbance of speech rhythm
Atypical intonation

4) Overall communication effectiveness:

Reduced intelligibility
Atypical overall quality of speech
Dysfluency

Perceptual ratings

- Perceptual ratings were completed by 3 listeners who were highly familiar with speech production in DS and who participated in consensus training.

- Features were rated on a monopolar 5-point equal-appearing interval scale Ratings were made for both sustained vowels and sentences.

Vowel Rating Scale

F 8;01

Vowel /a/	Not Present	Mild	Moderate	Severe	Very Severe
Roughness					
Breathiness					
Strain					
Pitch variability					
Pitch breaks					
Loudness variability					
Dysphonia severity					
Atypical resonance					
Hyponasal					
Hypernasal					
Cul de sac					
Other atypical resonance					

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Ratings of Vowels – Principal Components Analysis

Principal Component 1

High ratings of:

Roughness

Dysphonia severity

Strain

Pitch variability

Loudness variability

PC1 = vocal hyperfunction

Principal Component 2

High ratings of:

Breathiness

Atypical resonance

and low rating of:

Strain

PC2 = vocal hypofunction

Principal Component 3

High rating of *Pitch breaks* and

low rating of *Loudness variability*

PC3 = pitch & loudness control

Ratings of Sentences – Principal Components Analysis

Principal Component 1

High ratings of:

Imprecise consonants
Distorted vowels
Disturbance of speech rhythm
Atypical intonation
Breathiness
Atypical resonance
Dysphonia severity

PC1 = severity across systems

Principal Component 2

High ratings of:

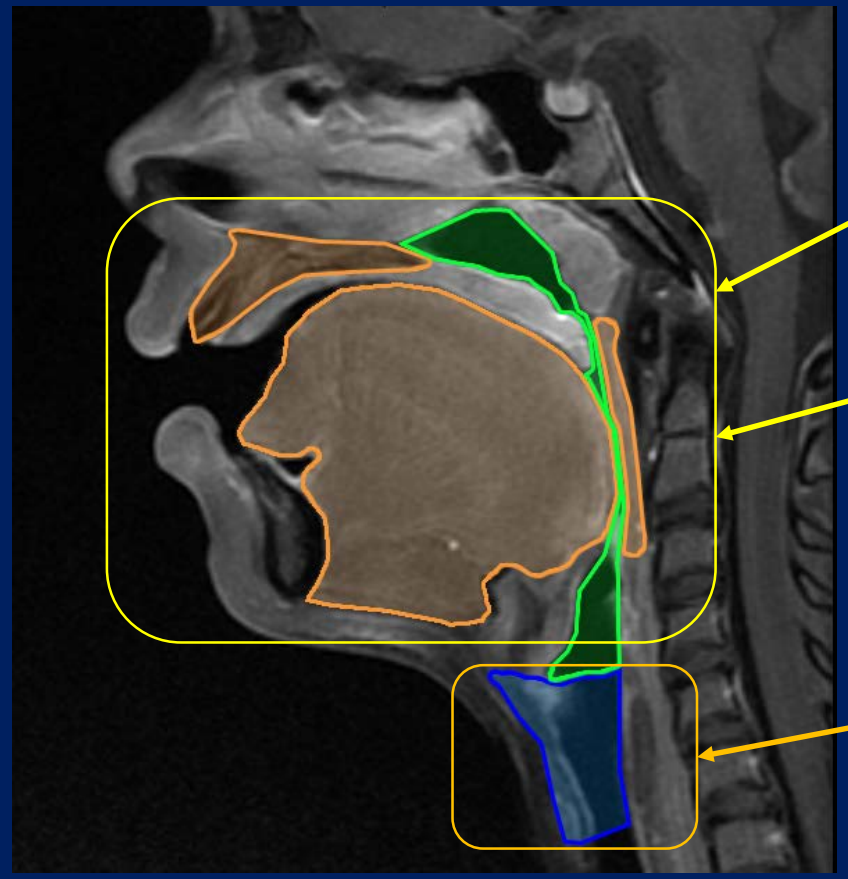
Roughness
Dysfluency
Dysphonia severity

Principal Component 3

High ratings of:

Irregular articulatory breakdown
Strain

Acoustic Measures



Formant frequencies (F1-F4) of corner vowels

Spectral moments (M1-M4) of the fricatives / s / and / ʃ /

Articulation

Multidimensional Voice Program (MDVP)

Cepstral Spectral Index of Dysphonia (CSID)

Phonation

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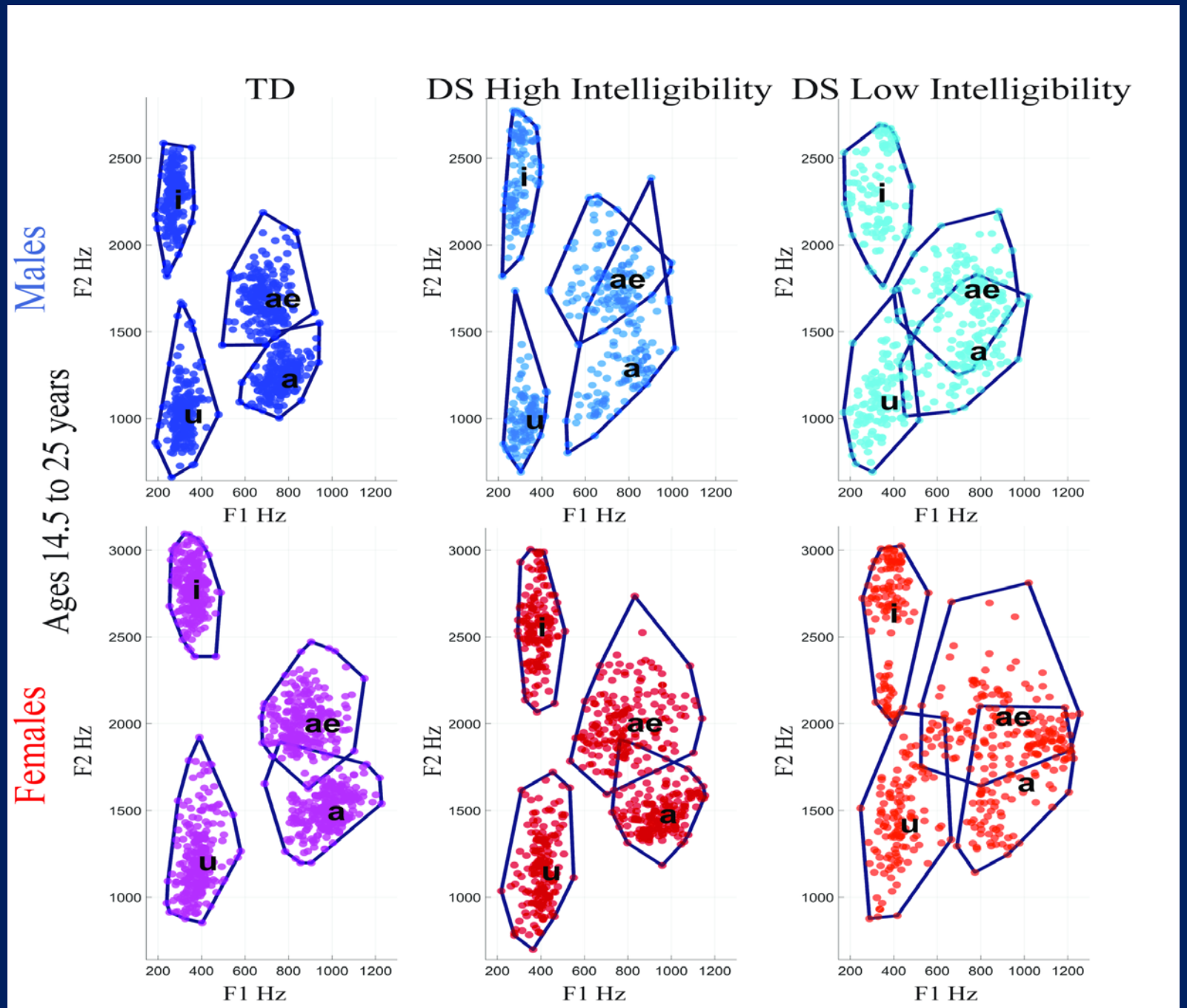


Differences in:

- Dispersion of F1 and F2
- Overlap of vowels, esp. low vowels /æ/ & /ɑ/

Interpretation:

Anatomic restriction of tongue position in DS

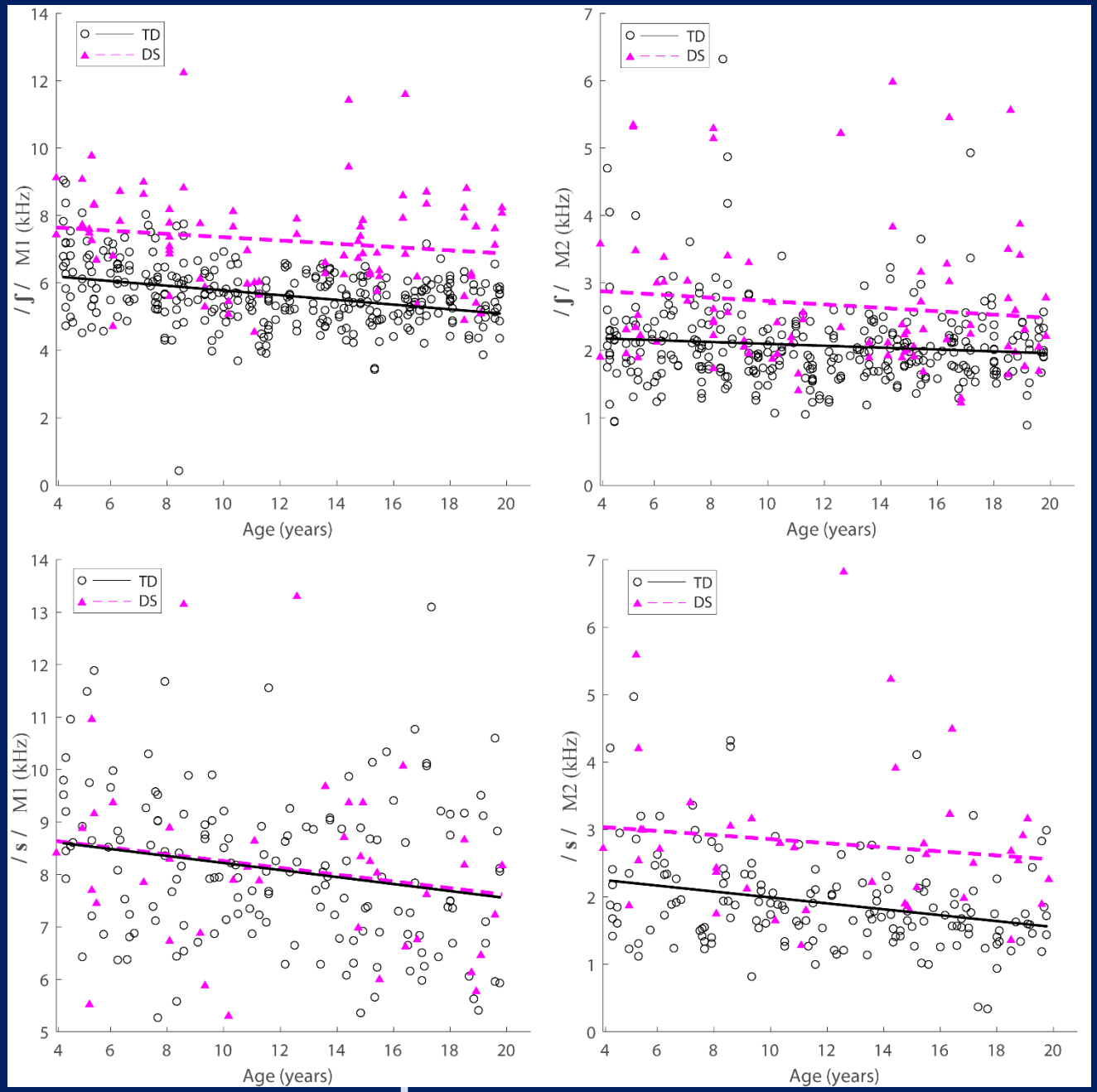
/æ/- /ɑ/ difficulty:

- Perceptual study by Wild et al (*AJSLP*, 2018)
- Acoustic study by Carl (*JSLHR*, in press)

M1 M2

/ʃ/

/s/



Spectral Moments (middle)

For /ʃ/ both M1 and M2 are larger for DS than TD

For /s/ M1 is equal for DS and TD, but M2 is larger for DS

Inference:
Palatal dysmorphology affects the palatal fricative /ʃ/

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Acoustic Analyses of Phonation

- Background studies
  - SLPs can identify DS from sustained vowels (Moran, *J. Com. Dis.*, 1986).
  - No single set of acoustic variables defines voice in DS (Albertini et al., *Res. Dev. Dis.*, 2010; Moran, *J. Com. Dis.*, 1986; Moran & Gilbert, *Am. J. Ment. Def.*, 1982).
- Current study assessed phonation using MDVP & CSID.
  - ➢ Dysphonia severity score correlated mildly but significantly with:
    - MDVP Fundamental Frequency Variation (r=.373, p=.006) &
    - CSID (r= .392; p =.001).
  - ➢ MDVP measures significantly different between DS and TD:
    - Fundamental Frequency Variation (t-test p= .000)
    - Peak Amplitude Variation (t-test p= .000)
  - ➢ CSID also significantly different between DS and TD (t-test p= .000).

There may be different acoustic signatures of the voice disorder in DS

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Conclusions

The Speech Disorder in Down Syndrome:

- Reflects the phenotypic heterogeneity in the syndrome.
- Results from impairments distributed across the systems of speech production.
- Is rooted in both dysmorphology and disordered motor control.
  - Hypothesis: Structure-function interaction through the lifespan.
- Can be better understood through a combination of methods (anatomic, physiologic, acoustic, and perceptual)
  - More interdisciplinary research!

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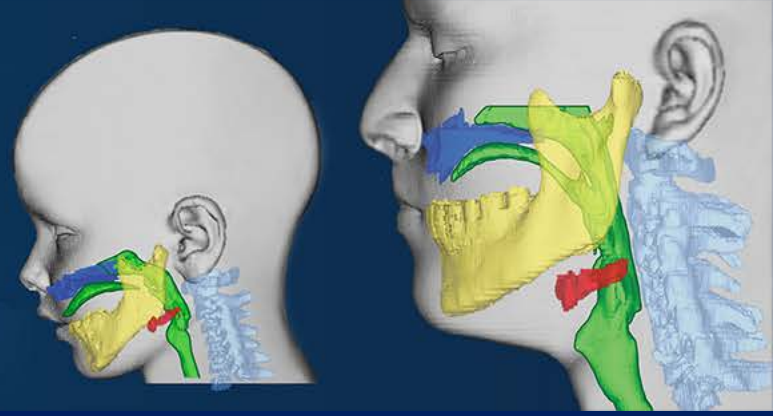

Acknowledgments

Research made possible by funding support from NIH:

NIDCD R01 DC006282; 3R01DC006282-15S1 & NICHD U54 HD090256

We thank all our research participants and their caregivers.
We also thank all the lab members who assisted with the acoustic measurements and data analysis.

Thank you for your attention!

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