Motor Speech Deficits in 22q11.2 Deletion Syndrome

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Disclosures

I have no relevant financial or nonfinancial relationships in the products or services described, reviewed, evaluated or compared in this presentation.
Background

Motor Speech Disorders (MSD), including both apraxia of speech and dysarthria, have been reported in children and adults with 22q11.2DS.

Clinical debate on the potential overdiagnosis of Childhood Apraxia of Speech (CAS) in 22q11.2DS is, in part, due to possible overlap in the signs of each disorder.

- Severe articulation disorder with limited speech intelligibility
- Lack of oral movement during compensatory misarticulations

(Persson et al., 2003; Carneol et al., 1999; D’Antonio et al., 2001; Solot et al., 2000; Golding-Kushner, 1985; Kummer et al., 2007; Mills et al., 2006; Zaleski et al., 2009; Baylis 2011).
Research in MSD in 22q11.2DS

• Retrospective studies using chart reviews have reported that 36-85% of children with 22q11.2DS have features consistent with or a clinical diagnosis of CAS (Mills et al., 2006; Kummer et al., 2007).

• Prospective studies using instruments that purport to assess CAS (e.g., Hickman Apraxia Profile, Nuffield Dyspraxia Assessment) have reported that 52% of participants with 22q11.2DS have features consistent with CAS (Mills et al., 2006; Kummer et al., 2007).
Limitations of Research in MSD in 22q11.2DS

• Lack of an explanatory account of CAS at neurologic and speech processing levels;

• Lack of a standardized assessment procedure and diagnostic criteria to classify a child as positive for CAS (ASHA, 2007; McCauley and Strand, 2008);

• Lack of studies comparing speech findings in 22q11.2DS with speech findings in children with other subtypes of speech sound disorders, including Speech Delay and subtypes of dysarthria.
Significance

Research in children with 22q11.2DS using contemporary methods for classification of CAS and other forms of MSD has the potential to refine our understanding of the speech phenotype of children with 22q11.2DS, as well as inform clinical management.

• This study provides the first preliminary estimate of the clinical prevalence of CAS in children with 22q11.2DS using a standardized diagnostic marker of CAS.
Research Questions

1. Do children with 22q11.2DS meet a standardized research criterion for CAS?

2. Do any of the sample meet emerging research criteria for a subtype of Motor Speech Disorders termed “Motor Speech Disorder-Not Otherwise Specified” (MSD-NOS)?
Method

Participants
- Children recruited by mailings from two major Midwest pediatric medical centers
- 18 children with 22q11.2DS
- Ages 6-18 years
- No history of permanent hearing loss
- Native English speakers
Method

- All assessment tasks administered by the same SLP
- **Madison Speech Assessment Protocol** (MSAP, Shriberg et al., 2010)
  - 15 spontaneous and imitated speech tasks in simple to challenging contexts
  - Kaufman Brief Intelligence Testing-2 (K-BIT2)
  - Oral and Written Language Subscales (OWLS)
  - Questionnaires on participant’s medical, speech, and educational histories.
- All MSAP speech tasks audio recorded
Method

• MSAP speech data were reduced independently by trained speech research assistants using the Speech Disorders Classification System (Shriberg et al., 2010), including narrow phonetic transcription, prosody-voice coding, and acoustic analyses.

• Classification as CAS requires participants to meet percentage cutoff criteria on inappropriate pauses and inappropriate stress, using an acoustic-aided motor-speech typology (Shriberg et al., in preparation).

• Classification as having an MSD other than CAS requires criterion z-scores on an emerging set of perceptual and acoustic signs of dysarthria or “MSD-NOS” using data from a reference sample of 150 typical speakers (Potter et al., 2012).
A Diagnostic Phenotype for Childhood Apraxia of Speech: *The Pause-Stress Marker* (a)

- **Pauses (P+):** Inappropriate pauses in continuous speech, coded using a 10-category linguistic typology (e.g., sharp, grope, etc.)

- **Stress (S+):** Excessive and equal stress on words and phrases in continuous speech (sentential level).

- Based on % of opportunities within 24 utterances → If cutoff threshold exceeded, then are coded as (+) for that feature (e.g., P+S+, P+S-, P-S+).

  **P+S+** has been shown to be the main diagnostic feature that separates children with CAS from age/gender matched peers with Speech Delay.

  - Specificity for a sample of 224 children with Speech Delay was > 98% (Shriberg et al., in preparation).

Results

• 3 of the 18 participants (16.7%) with 22q were positive for the diagnostic marker (P+S+) for childhood apraxia of speech.

• Several of the remaining 22q participants were positive on an emerging set of speech, prosody, and voice signs consistent with MSD-NOS (e.g., P-S+).
Conclusions

• Findings support the perspective that approximately 1 in 6 youth with 22q11DS meet contemporary criteria for Childhood Apraxia of Speech. They fail to support prior reports that CAS is a highly prevalent type of speech sound disorder.

• Findings for this sample of children with 22q11DS support the perspective that in addition to their speech delay, some may have additional motor speech involvement.

• Given the critical role of differential diagnosis in treatment planning, findings support the need for larger scale research on the prevalence and genetic and neural substrates of CAS and other types of motor speech disorders in youth with 22q11DS.
Acknowledgments

• National Institute on Deafness and Other Communication Disorders [DC00496] and a core grant to the Waisman Center from the National Institute of Child Health and Development [HD03352], PI: Lawrence Shriberg, PhD.
• UL1RR025755 from the National Center For Research Resources (National Center for Advancing Translational Sciences)
• Phonology Project, Waisman Center, University of Wisconsin-Madison
Key References