Diagnostic Signs of Childhood Apraxia of Speech in Idiopathic, Neurogenetic, and Complex Neurodevelopmental Contexts

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Topics

I. Premises

II. Method

III. Results

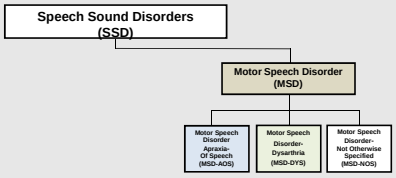
IV. Conclusions

Premise 1
CAS is One of Three Subtypes of MSD

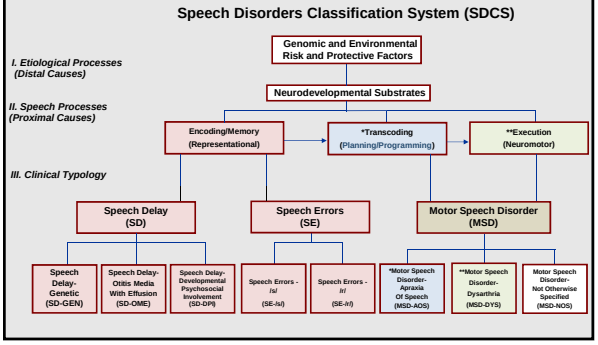
Childhood Apraxia of Speech (CAS) is one of three subtypes of a class of Speech Sound Disorders (SSD) termed Motor Speech Disorders (MSD)

Cover term: Speech Sound Disorders (SSD)
Class term: Motor Speech Disorders (MSD)
Subtype terms:
Motor Speech Disorders-Apraxia of Speech (MSD-AOS)
Motor Speech Disorders-Dysarthria (MSD-DYS)
Motor Speech Disorders-Not Otherwise Specified (MSD-NOS)

Premise 1
CAS is One of Three Subtypes of MSD



Premise 2
A Transcoding Deficit Differentiates CAS from Speech Delay, MSD-DYS and MSD-NOS



Premise 3
Diagnostic "Checklists" for CAS Should Only Include Signs of the Transcoding Deficit

- CAS is not itself a syndrome, although it appears to be a co-occurring speech sign in a number of complex neurodevelopmental disorders
- Diagnostic checklists for CAS should include only signs of the transcoding deficit, not signs of the consequence of a transcoding deficit (e.g., lowered intelligibility) or signs of co-occurring deficits in other domains (e.g., auditory processing deficits)

Method
Pediatric Adaptation of Mayo Clinic System[a]

Classification of a speaker as positive for CAS (CAS+) requires at least 4 of the following 10 signs in at least 3 speech tasks:

- vowel distortions
- difficulty achieving initial articulatory configurations or transitionary movement gestures
- equal stress; lexical or phrasal stress errors
- distorted substitutions
- syllable or word segregation
- groping
- intrusive schwa
- voicing errors
- slow speech rate and/or slow DDK rates
- increased difficulty with multisyllabic words

[a] Shriberg, L.D., Potter, N.L., & Strand, E.A (2011).

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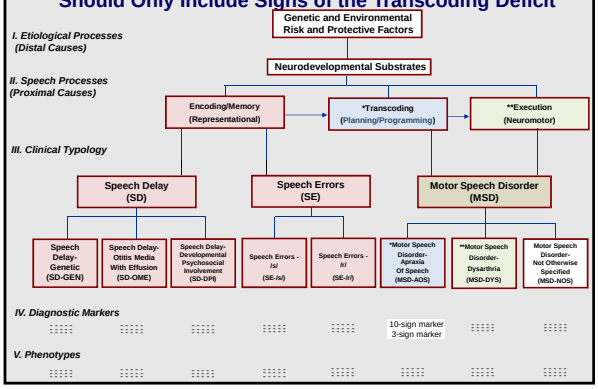

Premise 4
Idiopathic CAS is Informed by MSD-AOS Research in Neurogenetic, Neurodevelopmental, and Neurologic Contexts

- Lack of a validated inclusionary criterion (a gold standard) for Idiopathic CAS has yielded excessive false positives in research and excessive over diagnoses in practice
- Idiopathic CAS is a placeholder for the genomic pathways to CAS emerging from whole exome sequencing (WES) and upcoming whole genome sequencing (WGS)
- Low prevalence of Idiopathic CAS limits research sample sizes[a]

[a] A rare disease in North America = < 1/1500 (Roubertoux & de Vries, 2011).

Premise 5:
Genetic and Behavioral Findings in CAS are Consistent With a Multiple Domain Disorder

- Findings in *FOXP2*-CAS studies
  - Bilateral, widespread *FOXP2* expression, including gene regulation in pathways for vision, audition, speech, and other domains (e.g., Horng et al., 2009)
  - Histories of cognitive, auditory-perceptual, language, motor, psychosocial deficits (Rice et al., 2011; Shriberg et al., 2006; Tomblin et al., 2009)
- Findings in Idiopathic and Neurogenetic CAS studies
  - Behavioral findings and developmental histories indicate persistent deficits in multiple domains (e.g., encoding and memory deficits: Shriberg et al., in press)

Topics

Method
Participants

Group	*n*	Age (yrs)			% Males	Percentage of Consonants Correct (PCC)	
		M	SD	Range		*M*	SD
Speech Delay	98	4.0	0.8	3-7	67.3	69.5	9.7
Suspected CAS							
Idiopathic	19	9.2	4.2	4-19	52.6	73.3	11.0
Neurogenetic[a]	12	16.3	11.8	8-50	41.7	80.8	7.8
Neurodevelopmental							
Galactosemia	29	9.0	2.9	5-16	62.1	85.7	11.6
Down syndrome	13	12.2	2.7	8-17	23.1	76.8	9.3
22q deletion	14	10.6	3.2	7-18	71.4	81.0	14.2
Autism	40	6.1	1.2	4-8	82.5	92.0	5.9

[a] Subtypes include *FOXP2* (3), chromosome translocation (3), Joubert syndrome (1), 16p11.2 microdeletion syndrome (2), Prader Willi syndrome (1), terminal deletion of chromosome 22 (1), 16p13.2 copy number loss (1).

Method
Madison Speech Assessment Protocol

Four age-based protocols:

Preschool, school-aged, adolescent, adult

Each protocol includes 15 speech tasks

- Articulation Task
- Challenging Word Tasks (2)
- Challenging Phrase Task
- Consonants Task
- Conversational Sample
- DDK Task
- Phonation Task
- Syllable Repetition Tasks (2)
- Stress Tasks (2)
- Vowel Tasks (3)

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[a] Shriberg, L.D., Potter, N.L., & Strand, E.A (2011)

Method
Speech Disorders Classification System (SDCS)

Candidate Diagnostic Signs for Eight Subtypes of SSD[a]	Components[b]
Organized	Ten Linguistic Domains Analytics (TLDA) Competence, Precision, Stability Analytics (CPSA)
Operationalized	Madison Speech Assessment Protocol (MSAP) Procedures for transcription, prosody-voice analyses, and acoustic analyses
Standardized	Reference Database (200 typical speakers, 3 to 80 years old)

[a] Candidate signs selected from pediatric and adult literature in speech sound disorders of known and unknown origin.

[b] Data reduction and analyses in the PEPPER software environment.

Method
A Three-Sign Diagnostic Marker to Discriminate CAS from Speech Delay

Classification Criterion for CAS+:

Positive Finding on each of three signs

Sign	Finding
Low Accurate Transcoding (AT)	**+**
Low Appropriate Pauses (AP)	**+**
Low Articulatory Rate (AR)	**+**
	= CAS+

Method
Two of the Three Diagnostic Signs Are Obtained from a Conversational Sample

Sign: Low Appropriate Pauses (AP)[a]

- 12 utterances eligible for coding
- Coding scheme includes 15 codes
- $AP = \frac{\text{Appropriate Pauses}}{\text{Appropriate + Inappropriate Pauses}} \times 100$

Sign: Low Appropriate Rate (AR)[a]

- Same 12 utterances as above
- AR = number of syllables per second with pause time removed

[a] Standardized (z-scores) by age and gender

Method:
The Third Diagnostic Sign is Obtained from the Syllable Repetition Task (SRT)[a]

Sign: Low Accurate Transcoding (AT)

1. bada	10. dabama
2. dama	11. madaba
3. bama	12. nabada
4. mada	13. banada
5. naba	14. manaba
6. daba	15. bamadana
7. nada	16. danabama
8. maba	17. manabada
9. bamana	18. nadamaba

[a] Shriberg & Lohmeier (2008); Shriberg et al. (2009; in press); Lohmeier & Shriberg (2011)

Method
Low Accurate Transcoding[a]

SRT Item	Examples of Inaccurate Transcoding: Homorganic Nasal	Heterorganic Nasal	Non-Nasal
bada	banda	bamda	
mada			marda
nabada			nabavda

$$\text{AT Percentage} = 1 - \frac{\text{No. of Additions}}{\text{No. of Eligible Stop Consonants}} \times 100$$

AT positive = < 80%

[a] Addition of a preceding nasal/nasal was the most common addition (92% of all additions) in Group 4; Shriberg, Lohmeier, Strand, & Jakielski [in press]

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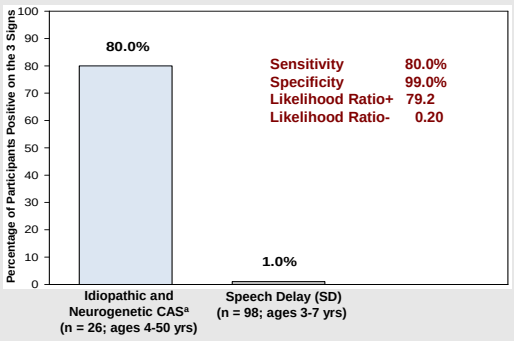
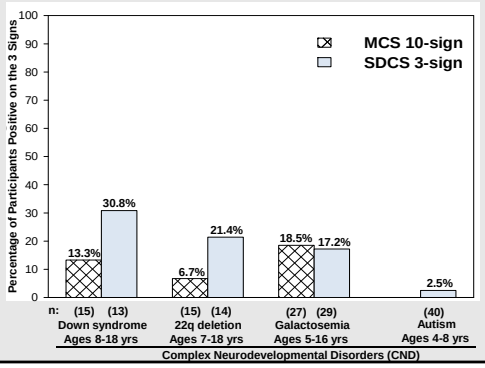
Results
Diagnostic Accuracy Findings for a Standardized Three-Sign Diagnostic Marker of CAS[a]

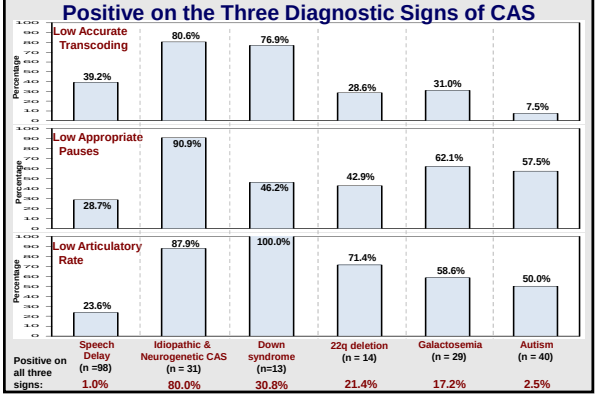


[a] Using a pediatric adaptation of the Mayo Clinic System as the gold standard

Results
CND Findings for a Standardized Three-Sign Diagnostic Marker of CAS



Results
Percentage of Participants with CND Classified as Positive on the Three Diagnostic Signs of CAS



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Conclusions
Take Away

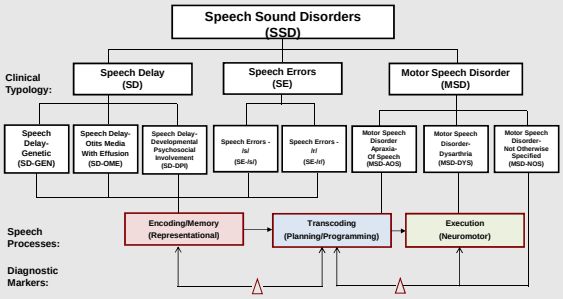
Findings are interpreted as support for

- the premise of standardized diagnostic markers for subtypes of pediatric motor speech disorders—proof of concept
- a three-sign standardized diagnostic marker that discriminates both early and persistent CAS from Speech Delay
- a provisional classification, MSD-NOS, for motor speech disorders that do not meet diagnostic criteria for either apraxia of speech or the conventional dysarthrias

Conclusions
Research Needs

- Construct validity research to develop the more highly valued account of pediatric motor speech disorders—categorical versus dimensional?
- Standardized diagnostic markers to discriminate among MSD-AOS, MSD-DYS, and MSD-NOS
- Population prevalence estimates for idiopathic CAS to complete risk statistics for genomic and other research and public health needs (i.e., the pre-test odds of CAS)

Conclusions
Research Needs



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