A Diagnostic Marker to Discriminate Childhood Apraxia of Speech From Speech Delay

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Motor Speech Disorders & Speech Motor Control
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Premises

- Both Childhood Apraxia of Speech (CAS) and Speech Delay (SD) are characterized by delays in auditory and somatosensory representational and feedback processes (Shriberg, Lohmeier et al. 2012).

- CAS is characterized by additional deficits in transcoding (planning/programming) and feedforward processes.

- A highly valued diagnostic marker of CAS requires conclusive psychometric support for one cross-linguistic, lifespan sign that identifies and quantifies the transcoding and feedforward deficits.
III. Clinical Typology
(Behavioral Phenotype)

Speech Delay (SD)
- Speech Delay-Genetic (SD-GEN)
- Speech Delay-Otitis Media With Effusion (SD-OME)
- Speech Delay-Developmental Psychosocial Involvement (SD-DPI)

Speech Errors (SE)
- Speech Errors -/s/ (SE-/s/)
- Speech Errors -/r/ (SE-/r/)

Motor Speech Disorder (MSD)
- Motor Speech Disorder-Apraxia Of Speech (MSD-AOS)
- Motor Speech Disorder-Dysarthria (MSD-DYS)
- Motor Speech Disorder-Not Otherwise Specified (MSD-NOS)

IV. Diagnostic Markers
(Criterial Signs of Phenotype)

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Speech Disorders Classification System (SDCS)^a

I. Etiological Processes
(Distal Causes)

- Genomic and Environmental Risk and Protective Factors
- Neurodevelopmental Substrates

II. Speech Processes
(Proximal Causes)

- Representation
  - Auditory
  - Somatosensory
- Transcoding
  - Planning
  - Programming
- Execution
  - Feedforward
  - Feedback

III. Clinical Typology
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(Criterial Signs of Phenotype)

Two Frameworks to Integrate Signs of SD and CAS With Their Genomic and Neurodevelopmental Substrates

- Dual Stream Neurodevelopmental Framework
  - Focus on ventral and dorsal substrates of speech processing in CAS
    (Hickok, Poeppel, & colleagues, others [see References])
# Neurodevelopmental Substrates of CAS Cast Within a Dual Stream Framework

## Ventral Stream
- Earlier Ontogeny
- Auditory Perception
- Phonemic
- Semantic, Syntactic
- Instantiated

## Dorsal Stream
- Later Ontogeny
- Somatosensory Production
- Phonetic
- Articulatory Novel
Speech Disorders Classification System (SDCS)

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Generic Speech Processing Framework
- Seven-element, significantly underspecified framework (Friederici, Guenther, Hickok, Levelt, Maassen, Nijland, Poeppel, Preston, Terband, van de Merwe, Ziegler, others [see References])

II. Speech Processes (Proximal Causes)

<table>
<thead>
<tr>
<th>Framework</th>
<th>Preliminaries</th>
<th>Method</th>
<th>Findings</th>
<th>Conclusions</th>
</tr>
</thead>
</table>

**Framework Method Findings Conclusions**

Preliminaries

II. Speech Processes (Proximal Causes)

1. Representation
   - Auditory
   - Somatosensory

2. Transcoding
   - Planning
   - Programming

3. Execution

4. Feedforward

5. Feedback

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  - Planning
  - Programming

- Execution

SD and CAS

---

Two Frameworks to Integrate Signs of SD and CAS With Their Genomic and Neurodevelopmental Substrates

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**II. Speech Processes (Proximal Causes)**

- **Representation**
  - Auditory
  - Somatosensory

- **Transcoding**
  - Planning
  - Programming

- **Execution**

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SD and CAS

CAS

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**Speculative Integration of Four Candidate Signs of CAS with the Dual Stream and Speech Processes Frameworks**

<table>
<thead>
<tr>
<th>SDCS Level I</th>
<th>SDCS Level II</th>
<th>SDCS Levels III &amp; IV</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dual Stream</td>
<td>Speech</td>
<td>Four Signs of CAS</td>
</tr>
<tr>
<td>Framework</td>
<td>Processes</td>
<td>Framework</td>
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<td></td>
<td>Framework</td>
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<tr>
<td>Ventral</td>
<td>Dorsal</td>
<td>Rate</td>
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<tr>
<td>X</td>
<td>X</td>
<td>Pauses</td>
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<td>X</td>
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<td>Stress</td>
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<td>X</td>
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<td>Transcoding</td>
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<td>X</td>
<td>Representation</td>
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<td>Execution</td>
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<td>X</td>
<td>Feedback</td>
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</table>

### (‘Seven Attributes of’) Highly Valued Diagnostic Markers

<table>
<thead>
<tr>
<th>Construct</th>
<th>Premise</th>
<th>Rationale</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Accuracy</strong></td>
<td>The higher the diagnostic accuracy of a diagnostic marker the more highly valued in research and clinical settings.</td>
<td>Diagnostic markers deemed conclusive for a disorder require &gt;90% sensitivity and &gt;90% specificity, yielding positive and negative likelihood ratios of at least 10.0 and at most .10, respectively.</td>
</tr>
<tr>
<td><strong>Reliability</strong></td>
<td>The higher the reliability of a diagnostic marker the more highly valued in research and clinical settings.</td>
<td>Reliable diagnostic markers have robust point-by-point intrajudge and interjudge data reduction agreement and internal and test-retest stability of scores, each estimated across relevant participant heterogeneities.</td>
</tr>
<tr>
<td><strong>Coherence</strong></td>
<td>The greater the theoretical coherence of a diagnostic marker the more highly valued in research and clinical settings.</td>
<td>As portrayed in Figure 1, conclusive diagnostic markers (Level IV) for each of the putative SSD subtypes (Level III) are highly valued for integrative descriptive-explanatory accounts when tied to their genomic, environmental, and developmental neurocognitive and sensorimotor substrates (Levels I and II).</td>
</tr>
<tr>
<td><strong>Discreteness</strong></td>
<td>Diagnostic markers from discrete, on-line events are more highly valued than diagnostic markers derived from off-line tallies of events.</td>
<td>Behavioral signs that that can be spatiotemporally associated with neurological events have the potential to inform explanatory accounts of speech processing deficits and identify biomarkers.</td>
</tr>
<tr>
<td><strong>Parsimony</strong></td>
<td>The fewer the number of signs in a diagnostic marker the greater its theoretical parsimony and psychometric robustness.</td>
<td>Each sign required for a diagnostic marker adds theoretical complexity and requires additional (multiplicative) psychometric stability.</td>
</tr>
<tr>
<td><strong>Generality</strong></td>
<td>The more extensive the generality of a diagnostic marker the more highly valued in research and clinical settings.</td>
<td>Diagnostic markers with the most extensive external validity may be used to identify risk for future expression of disorders, identify active expression of a disorder, and postdict prior disorder.</td>
</tr>
<tr>
<td><strong>Efficiency</strong></td>
<td>The greater the efficiency of a diagnostic marker the more highly valued in research and clinical settings.</td>
<td>More highly valued markers require the fewest tasks, equipment, examiner proficiencies and participant accommodations and the least time and costs to administer, score, and interpret.</td>
</tr>
</tbody>
</table>

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*Shriberg et al. (2014). A pause marker to discriminate Childhood Apraxia of Speech from Speech Delay. Manuscript in preparation. The seven constructs are listed in their estimated rank order of importance.*
## Participants

<table>
<thead>
<tr>
<th>Group</th>
<th>Cohort</th>
<th>Title</th>
<th>n</th>
<th>Age (yrs)</th>
<th>% Males</th>
<th>Percentage of Consonants Correct (PCC)</th>
</tr>
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<tbody>
<tr>
<td>Suspected Childhood Apraxia of Speech (CAS)</td>
<td>Idiopathic CAS</td>
<td>CASI</td>
<td>41</td>
<td>8.7</td>
<td>4.1</td>
<td>65.9</td>
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<tr>
<td></td>
<td>Neurogenetic CAS</td>
<td>CASN</td>
<td>23</td>
<td>10.6</td>
<td>4.8</td>
<td>47.8%</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td></td>
<td>64</td>
<td>9.3</td>
<td>4.4</td>
<td>59.4%</td>
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<tr>
<td>Adult-onset Apraxia of Speech (AAS)</td>
<td>Apraxia of Speech</td>
<td>AOS</td>
<td>14</td>
<td>62.1</td>
<td>10.9</td>
<td>78.6</td>
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<tr>
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<td>Primary Progressive AOS</td>
<td>PPAOS</td>
<td>16</td>
<td>72.4</td>
<td>9.1</td>
<td>56.3</td>
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<td></td>
<td>30</td>
<td>67.6</td>
<td>11.1</td>
<td>66.7</td>
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<tr>
<td>Speech Delay (SD)</td>
<td>Clinical Cohort</td>
<td>SD1</td>
<td>88</td>
<td>4.3</td>
<td>1.3</td>
<td>73.0</td>
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<td>Research Cohort</td>
<td>SD2</td>
<td>23</td>
<td>5.5</td>
<td>0.6</td>
<td>72.7</td>
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<td>Research Cohort</td>
<td>SD3</td>
<td>84</td>
<td>3.9</td>
<td>0.7</td>
<td>71.4</td>
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<td>Research Cohort</td>
<td>SD4</td>
<td>30</td>
<td>4.5</td>
<td>0.9</td>
<td>48.3</td>
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<td>Total</td>
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<td>225</td>
<td>4.3</td>
<td>1.1</td>
<td>69.2</td>
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</tbody>
</table>

*a* Includes participants with copy number variants (n=11) identified in related research, and participants with neurodevelopmental disorders associated with disruptions in FOXP2 (n=4), 4q;16q translocation (n=3), 16p11.2 microdeletion syndrome (n=2), terminal deletion of chromosome 22 (n=1), Joubert syndrome (n=1), and Prader Willi syndrome (n=1).
Madison Speech Assessment Protocol (MSAP)

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)
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

\(^a\)Dr. Strand provided written anecdotal comments on the sources and rationale for each classification.
Pause Marker (PM) Method

1. Transcribe and prosody-voice code 24 utterances from a conversational speech sample.

2. Complete acoustics-aided procedures to identify occurrences of eight types of inappropriate between-word pauses in each utterance:
   - Type I pauses: abrupt, change, grope, other
   - Type II pauses: addition, repetition/revision, long, breath

3. Calculate PM percentage:
   \[ 100 \times (1 - \text{No. Type I Pauses/No. Pause Opportunities}) \]
   where No. Pause Opportunities = No. words - No. utterances

4. Criterion for CAS+: PM < 95\%

CAS+ classification for marginal PM scores (94.5% – 95.5%) requires positive findings on at least two of three supplementary standardized signs of CAS (Slow Articulatory Rate, Inappropriate Sentential Stress, Transcoding Errors).
And <uh> when we take a vacation we go <uh> one or two weeks, maybe two or three times a year. [PART] a year.
Procedures to Resolve MCS-PM Classification Disagreements

1. Assembled best estimates of ‘true positive’ and ‘true negative’ CAS groups:
   - Consensus CAS+ Group (n = 35):
     participants classified CAS+ by both diagnostic markers
   - Consensus CAS- Group (n = 15):
     participants classified CAS- by both diagnostic markers

2. Computed descriptive and inferential statistics for relevant demographic and speech variables for and between the two CAS consensus groups; compared findings for each disagreement to findings for the two CAS consensus groups
3. Determined **case-by-case support** for resolving each MCS-PM classification disagreement as either due to conceptual differences in MCS vs. PM criteria for CAS+, or as ‘questionable’ due to either **method constraints** (e.g., insufficient MSAP data) and/or **statistical support** consistent with the alternative Consensus CAS group.

4. Recalculated the estimated diagnostic accuracy of the PM with all ‘questionable’ disagreements excluded.
MCS-PM Classification Agreement Findings: 64 Participants Suspected Positive for CAS

Includes 7 Participants with 'Questionable' MCS+PM- Classifications

Excludes 7 Participants with 'Questionable' MCS+PM- Classifications

Sensitivity 83.3% Specificity 68.2%
MCS-PM Classification Agreement Findings: 30 Participants with AAS (AOS and PPAOS)

- Includes 11 Participants termed "Voicers"
  - Sensitivity 63.3%
  - Percent Agreement:
    - MCS+ PM+ (8/14) AOS: 57.1%
    - MCS+ PM+ (11/16) PPAOS: 68.8%
    - MCS+ PM+ (19/30) AAS: 63.3%

- Excludes 11 Participants termed "Voicers"
  - Sensitivity 100.0%
  - Percent Agreement:
    - MCS+ PM+ (8/8) AOS: 100.0%
    - MCS+ PM+ (11/11) PPAOS: 100.0%
    - MCS+ PM+ (19/19) AAS: 100.0%
SDCS-PM Classification Agreement Findings: 225 Participants with Speech Delay

**Specificity 98.2%**

<table>
<thead>
<tr>
<th>SD</th>
<th>Agreement (%)</th>
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</thead>
<tbody>
<tr>
<td>SD4</td>
<td>100.0%</td>
<td>30/30</td>
</tr>
<tr>
<td>SD1</td>
<td>98.9%</td>
<td>87/88</td>
</tr>
<tr>
<td>SD3</td>
<td>98.8%</td>
<td>83/84</td>
</tr>
<tr>
<td>SD2</td>
<td>91.3%</td>
<td>21/23</td>
</tr>
<tr>
<td>SD ALL</td>
<td>98.2%</td>
<td>221/225</td>
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</tbody>
</table>
Conclusions

- The PM provides a single-sign marker that likely can be used cross-linguistically to discriminate CAS from SD, and to scale the severity of CAS.

- The Type I pauses identified and quantified by the PM have theoretical ‘Coherence.’ The claim is that these atypical cessations of continuous speech are consequent to deficits in planning, programming, and/or feedforward processes.

- PM findings are interpreted to meet six of the seven proposed criteria for a highly valued diagnostic marker of CAS, requiring additional research to improve ‘Efficiency.’
Research Directions

Methodological

- Cross-validate the current, estimates of intrajudge and interjudge reliability of the PM (low-to mid 80%)

- Cross-validate the current acoustic correlate (steep amplitude rise time) of the most frequent type of inappropriate pause (Type I: ‘abrupt’) and explore automated detection of ‘abrupt’ pauses

- Develop alternatives to continuous speech samples for speakers suspected positive for CAS who have limited verbal output

- Assess the specificity of the PM for speakers with different types of dysarthria
Research Directions

Substantive

- Assess the informativeness of the PM in collaborative neuroscience studies to explicate the genomic and neural correlates of planning, programming, and feedforward deficits in CAS and AAS toward a biomarker of apraxia of speech.

- Assess the utility of the PM in collaborative studies to characterize normalization processes in CAS and to quantify treatment efficacy in studies of CAS and AAS.
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David Wilson

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References


References


