Childhood Apraxia of Speech (CAS) in Neurodevelopmental and Idiopathic Contexts

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Abstract

We have proposed that programmatic studies of apraxia of speech as it reportedly occurs in diverse neurodevelopmental and neurological disorders can inform research on the core features and diagnostic markers of idiopathic Childhood Apraxia of Speech, a putative pediatric speech sound disorder [1]. This paper describes the research plan, summarizes primary elements of the speech assessment and analysis methods, and reports summary perceptual and acoustic findings from four initial studies. Assessment of speech, prosody, and voice for signs of apraxia of speech consistent with contemporary diagnostic perspectives was completed on four study groups: a sample of adults with acquired apraxia of speech, a mother and daughter with a chromosome translocation disrupting FOXP2, three siblings with an unbalanced 4;16 chromosome translocation, and eight children and adolescents with classic galactosemia. Positive findings from the four studies are interpreted as support for the research framework. Discussion focuses on the theoretical and clinical implications of a unified perspective on the core features, signs, and diagnostic markers of CAS in neurodevelopmental and idiopathic contexts.

1 Background

Genetic studies of Childhood Apraxia of Speech (CAS) were catalyzed by the widely-cited research series on a London family in which a mutation affecting *FOXP2* was identified in half of the members of the then three-generation family [2]. As indicated in the abstract, we have suggested that research in CAS as it reportedly occurs in a number of genetically diverse neurodevelopmental disorders and as a sequelae of neurological disorders may inform the core features, signs, and diagnostic markers of idiopathic CAS [1]. Research support for the latter form of CAS has been the focus of considerable speculation, with the most recent literature review proposing CAS of unknown origin as a valid nosological classification within pediatric speech sound disorders [3]. This review, however, and anecdotal reports world wide, indicate that a putative idiopathic form of CAS is heavily overdiagnosed, creating significant service delivery needs for public health care systems.

Table 1. *Complex neurodevelopmental disorders reporting significant speech disorder/suspected CAS.*

Autism
Chromosome Translocations
Coffin-Siris syndrome (7q32-34 deletion)
Down syndrome (Trisomy 21)
Rolandic Epilepsy
Fragile X syndrome (*FMR1*)
Joubert syndrome (*CEP290*; *AHI1*)
Galactosemia
Rett syndrome (*MeCP2*)
Russell-Silver syndrome (*FOXP2*)
Velocardiofacial syndrome (22q11.2 deletion
Williams-Beuren locus duplication (7q11.23)

Table 1 is a list of some of the genetic conditions and neurodevelopmental disorders that reportedly include children with significant speech sound problems or specifically, apraxia of speech. Although most reports of suspected apraxia of speech as a secondary sign in complex neurodevelopmental disorders contain limited speech information, findings in this heretofore unexamined literature base provide a rich source of information for genotype-phenotype studies in CAS. Many reports include speakers with sporadic cytogenetic events in which copy number variations (deletions and duplications of genomic material) have resulted from chromosome translocations.

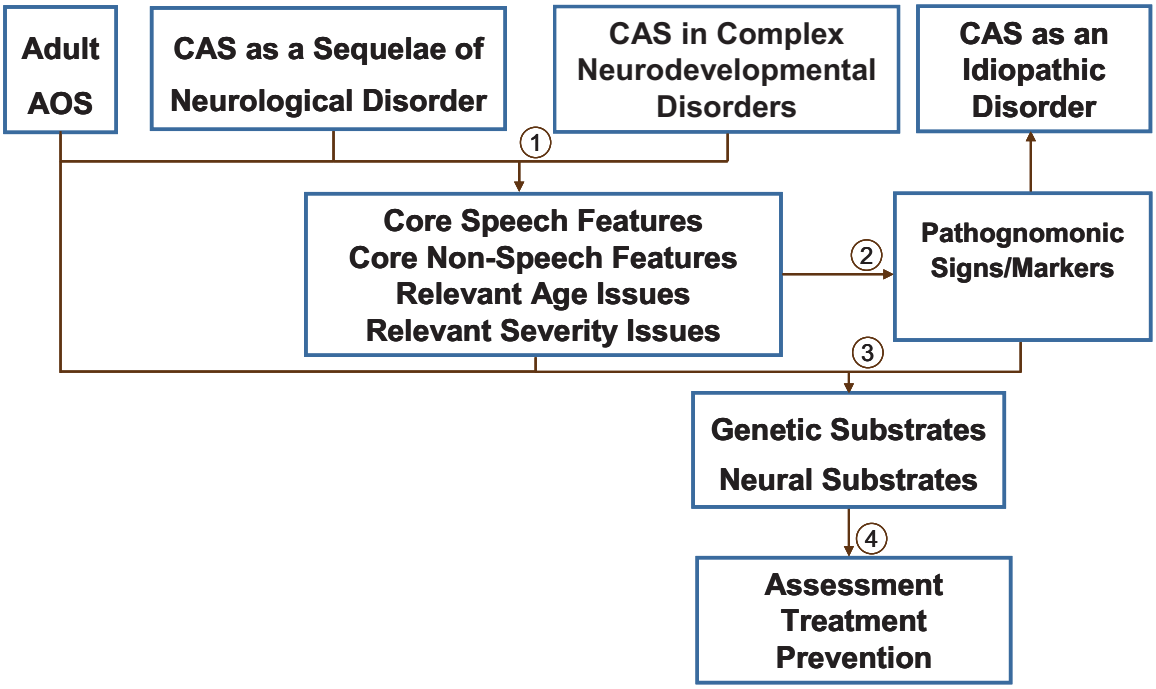

Figure 1: *A neurodevelopmental framework for research in CAS.*

There are a number of rationales for expecting differences between apraxia in idiopathic versus neurodevelopmental and neurologic forms [4], but validation of idiopathic CAS requires at least one praxic feature common to its expression in neurodevelopmental and acquired neurologic forms. Identification of one or more core features and the development of efficient clinical-research methods to quantify their signs and diagnostic markers is the primary goal of the research reported in this presentation.

2 Method

Figure 1 illustrates a four-phase framework for research in CAS. The first of the four phases depicted in Figure 1 is to conduct studies to identify core features of CAS as they occur in adult AOS, in children following neurological disorder (e.g., infection, trauma), and in complex neurodevelopmental disorders. In the second phase, findings from prior studies can be used to inform the inclusional/exclusional criteria to classify participants as positive for idiopathic CAS. Findings from these four forms of CAS can provide the information base needed for third-phase studies of the genetic and neural substrates underlying the pathophysiology of apraxia. Last, in the fourth phase, investigators from a number of disciplines can use aggregate findings to develop optimum methods for assessment, treatment, and ultimately, prevention.

Table 2 is an overview of the assessment and analysis framework. As shown in the two lefthand columns, speech targets are organized by three analytic domains (spatial, temporal, prosody-voice), each subordinated under three analytic constructs reflecting a participant's speech competence, precision, and stability. For each of these 9 domains, prior studies in our laboratory and elsewhere have suggested segmental and suprasegmental indices and variables that may be sensitive and specific diagnostic markers for either apraxia, one of several subtypes of dysarthria, or for a classification termed motor speech disorder-not otherwise specified (i.e. a cover term nonspecific for apraxia or dysarthria). As used in areas such as personality disorders (PD-NOS) and pervasive developmental disorders on the autism spectrum disorders (PDD-NOS), we propose MSD-NOS as a useful classification term for children whose motor speech characteristics differentiate them from phonological-based speech delay, but do not meet our emerging diagnostic markers for apraxia or dysarthria.

Potential diagnostic markers for apraxia (e.g., *unstable planar area, unstable vowel duration, overstressed lexical stress*) are interrogated using a

high-throughput computer platform for acoustic-aided transcription and transcription-aided acoustics. Multiple data sources are used to examine contextual influences on each index/variable obtained from a one-hour assessment protocol that includes 15 speech tasks (e.g., vowel repetition tasks, challenging word tasks, conversational sample). Comparison databases are used to derive age x sex z-scores for each index/variable from each speech task. As shown in Table 2, clinical classification information on the resulting profiles of z-scores is consolidated in a 27-cell matrix.

Studies undertaken for the first phase of the research framework shown in Figure 1 include data from assessments of participants from four clinical populations: (a) a group of 10 adults with acquired apraxia of speech [5], (b) a mother and daughter with a chromosome translocation disrupting *FOXP2* [6], (c) three siblings with an unbalanced 4;16 chromosome translocation [7], and (d) eight children and adolescents with classic galactosemia [8]. All participants reportedly have apraxia of speech, based on clinical or clinical-research findings. For the present studies, participants were group matched to typically-speaking, same sex controls of approximately the same age. Raw score cutoff points from prior studies and z-score based performance of the appropriate comparison groups were used in all quantitative analyses.

Conservative rules to minimize false positives were developed to assign participants to the three clinical classifications (apraxia, dysarthria, motor speech disorder-not otherwise specified) using the speech data alone, that is, without support from nonspeech data and data on other factors that confer risk for motor speech disorder. Participant findings on 28 speech, prosody, and voice markers were aggregated to support individual classification assignments, with each positive marker supported from z-score data from over 150 indices and variables obtained from the multiple speech sample tasks in the assessment protocol.

3 Results and Discussion

At the time this summary report was completed findings were not fully available, pending supplementary cross validation analyses. Results to this date provide strong support for several common speech, prosody, and voice signs and markers among participants with acquired and neurodevelopmental forms of apraxia of speech and among and between participants with different complex neurodevelopmental disorders. Individual and group findings will be summarized using the analytic matrix in Table 2. Discussion will focus on the theoretical and clinical implications of a unified perspective on the core features, signs, and diagnostic markers of CAS in neurodevelopmental and idiopathic contexts.

Table 2. *Assessment and analysis framework*.

Analytic		Clinical Classification		
Construct	Domain	Apraxia	Dysarthria	Motor Speech Disorder – Not Otherwise Specified
Competence	Spatial			
	Temporal			
	Prosody-Voice			
Precision	Spatial			
	Temporal			
	Prosody-Voice			
Stability	Spatial			
	Temporal			
	Prosody-Voice			

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