

A neurodevelopmental framework for research in childhood apraxia of speech

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Background

Rationale for a neurodevelopmental framework for research in childhood apraxia of speech (CAS) follows from the central methodological problem in programmatic research in this area—lack of consensus on the behavioral features and signs that define this putative clinical entity. The following widely-cited quote from McNeil and colleagues (McNeil, Robin, & Schmidt, 1997), which over 10 years ago was targeted at research in acquired apraxia of speech in adults (AOS), continues to set the problem for contemporary research in CAS:

. . . On what bases do you select subjects for study when trying to identify and characterize a new clinical entity? Without established inclusional and exclusional criteria, derived from careful experimentation, usually accumulated over a long period of time, and without models that specify the levels of breakdown and the potential mechanisms responsible for the phenomena . . . it is difficult or impossible to have confidence that the individuals and groups actually represent the subject of interest. In other words, it is difficult or impossible to avoid experimental tautologies. (p. 315)

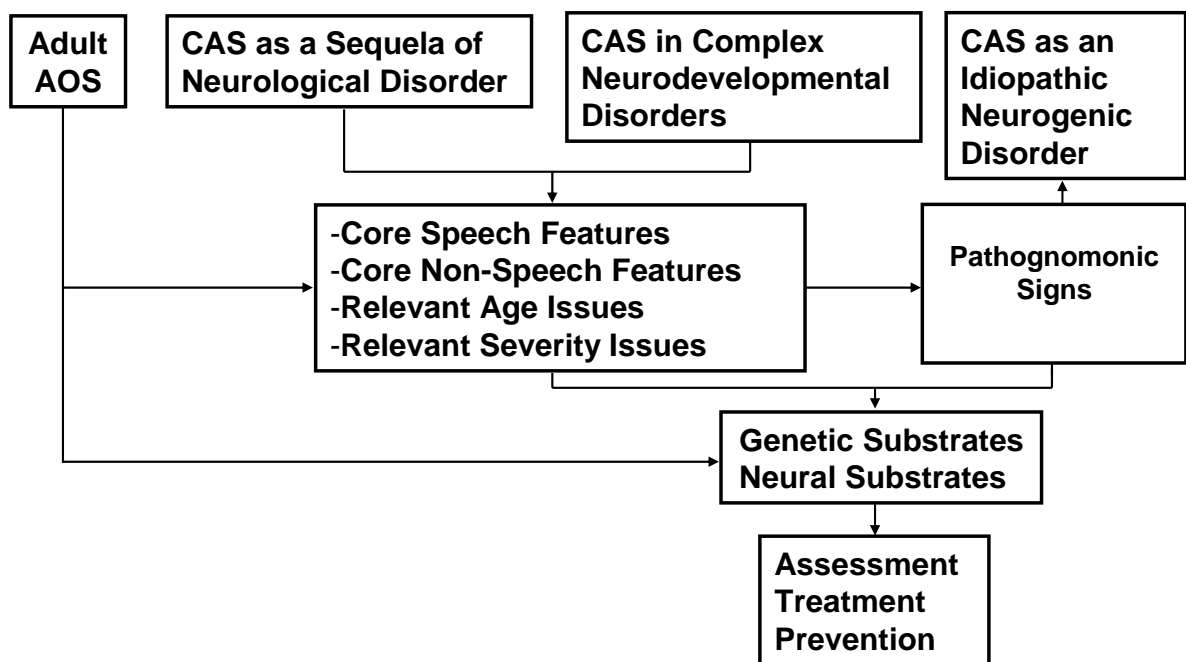
Our solution to this dilemma in CAS research includes a two-part methodological approach. First, we take the position that the only way to avoid the “experimental tautologies” that characterize continuing research in idiopathic CAS is to inform this clinical entity from studies of CAS as it occurs in association with known neurologic and neurodevelopmental disorders. Figure 1 is a graphic description of this approach. [Figure 1 here] The framework is a four stage research sequence in which findings from each stage inform successive stages. Thus, study of the core features of adult AOS in Stage 1 is viewed as requisite for study of CAS as it occurs in neurologic and neurodevelopmental disorders in Stage 2. The thesis is that a disorder of speech *praxis* should have features that are common to both acquired and developmental subtypes, with developmental issues likely moderating or mediating expression of some core features. Findings from studies of individuals within these classifications should yield the pathognomonic signs needed to meet inclusional/exclusional criteria to qualify subjects as having idiopathic CAS (Stage 3). Finally, information from each stage should provide the bases for studies of the genetic and neural substrates that eventually lead to methods for assessment, treatment, and the several forms of prevention.

The second methodological element of the framework for research in CAS is to base measurements at the first three stages on one common well-developed protocol. For this purpose we have developed a suite of high-throughput measures that use acoustic-aided transcription and transcription-aided acoustics. This life-span protocol provides extensive information on speech production, including segmental and especially suprasegmental processes. The goal is to assemble the array of clinical signs that differentiate CAS from other subtypes of speech sound disorders of currently unknown origin, including idiopathic dysarthria.

Findings to Date

As indicated in the abstract, the paper will provide a series of slides reviewing the molecular genetic research in CAS, followed by emerging findings from several study series in children suspected to have CAS in the context of neurodevelopmental disorders. Emphasis will be on findings from two recent studies, including those from a mother and daughter with CAS and a balanced 7;13 translocation affecting *FOXP2* and findings from three siblings with CAS and an unbalanced 4;16 translocation.

Figure 1. A neurodevelopmental framework for research in Childhood Apraxia of Speech (CAS).



References

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