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Heterogeneity in speech sound disorders: New findings in multigenerational families

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Abstract

Speech sound disorder (SSD) is a disorder affecting a child's ability to produce speech that is easily understood by others. Speech sound errors include distortions, substitutions, omissions, and insertions. SSD can run in families but causal genes have not yet been validated, although several candidate regions and genes have been proposed. In the past, genetic studies of SSD were based on samples of children from many different families ascertained with a broad SSD definition. There are reasons, however, to suspect that SSD has various phenotypically distinct subtypes and genetic etiologies. Traditionally, difficulty with sequentially integrating motor commands across articulatory subsystems has been associated with the term childhood apraxia of speech (CAS). Our recent work shows that in children with CAS, sequential processing deficits can be observed beyond speech production, affecting nonword imitation, reading, and spelling, and that this global deficit can even be observed in adults with a CAS history whose conversational speech has long normalized. Genetic etiologies may differ from family to family, even among families with familial CAS. In the project described here, SSD is studied in individual multigenerational families with familial SSD to reduce the effects of heterogeneity in each sample. Complementary genomic methods include linkage analysis, IBD screens, and exome sequencing. This approach is leading to the discovery of new genomic candidate regions and genes. Future work will elucidate the role of sequential processing deficits as a shared endophenotype of genetic origin in CAS and other communication disorders characterized by deficits in sequential processing.

Biography

Beate Peter is Research Assistant Professor in the Department of Speech and Hearing Sciences at the University of Washington in Seattle. In addition to a clinical master's degree in Speech-Language Pathology and a Ph.D. in Speech and Hearing Sciences, she holds the Graduate Certificate in Statistical Genetics, earned during her three years as postdoctoral trainee in Medical Genetics at the University of Washington. She is first editor of *Comprehensive Perspectives on Speech Sound Development and Disorders*. Her research focuses on the genetics of speech and reading disorders.