Toward a Phenotype Marker for Genetically Transmitted Speech Delay

Lawrence D. Shriberg

Phonology Project, Waisman Center on Mental Retardation and Human Development
University of Wisconsin-Madison, Madison, Wisconsin
E-mail: Shriberg@waisman.wisc.edu
Web site: http://www.waisman.wisc.edu/phonology/

ABSTRACT

Converging evidence supports the likelihood that some speech-language disorders of currently unknown origin, including a subtype of child speech-sound disorders termed *Speech Delay* (*SD*), are genetically transmitted. Molecular genetics research has not, to date, identified a susceptibility gene for SD, but a susceptibility locus on chromosome 7q31 has recently been reported. As with other complex behavioral traits, methodological challenges in speech-genetics research include heterogeneities in etiologies, genotypes, endophenotypes, and phenotypes. The method and findings summarized in this report address etiological heterogeneity—specifically, the development of methods to differentiate children and family members with genetically based SD from speakers with other possible subtypes of SD (i.e., phenocopies). We report the first data supporting the possibility of an acoustic marker for genetically transmitted SD. The phenotype marker is based on a speaker's production errors on the alveolar sibilants /s/ and /z/.

1. INTRODUCTION

The research context for the present study is the hypothesis that at least one subtype of child speech-sound disorder of currently unknown origin is genetically transmitted [*1*]. Behavioral genetics studies of speech-language disorder provide strong support for this hypothesis [*2, 3*], and molecular genetics findings are beginning to emerge. The Monaco group has identified a susceptibility gene on chromosome 7q31(FOXP2) that co-segregates with an orofacial apraxia and apraxia of speech, and is inherited as an autosomal dominant trait [*4–6*]. Closer to the present concerns, Schick et al. [*7*] report a candidate region near the FOXP2 locus that significantly co-segregates with a speech disorder (optionally comorbid with language disorder) but not with specific language disorder.

As with other complex behavioral traits, methodological constraints on the genetics of speech-language disorders of unknown origin include heterogeneities in putative etiologies, genotypes, endophenotypes, and phenotypes. In speech-genetics research there is a core need for valid, reliable, and readily accessible phenotype markers to classify and quantify the speech status of probands and family members [*8*]. Direct testing is available for young probands, but classification of older siblings and other family members is challenging because they may have normalized prior speech delay and their speech error histories may be unavailable or unreliable by recall report.

Rationale for the present approach was based on findings from an associated research series toward a set of acoustic markers for child speech-sound disorders. We have identified acoustic markers with promising diagnostic accuracy to differentiate children with several etiologic subtypes of speech-sound disorders, including those associated with early recurrent otitis media with effusion [*9, 10*], apraxia of speech [*11, 12*], and phoneme-limited articulatory distortions of /r/ [*13*] and /s/ [*8*]. Crucially, a prior diagnostic marker study [*10*] suggested that when speech-delayed children with positive histories for otitis media with effusion (SD-OME) distort the alveolar sibilants /s/ and /z/, they tend to produce them significantly more often with posterior tongue placement (i.e., *backing*). Such distortions contrast with the dentalized or *fronted* distortions commonly observed in children with speech delay of unknown origin. Findings in an associated concurrent validity study [*14*] have supported the first spectral moment [*15*] as a sufficient metric to discriminate backed alveolar sibilants (lowered first spectral moments) from fronted alveolar sibilants (higher first spectral moments).

This report summarizes the perceptual data from a study of a subtype of child speech-sound disorders presumed to be transmitted as a quantitative trait, termed *Speech Delay-Genetic* (*SD-GEN*). The question for diagnostic marker development is whether the sibilant errors produced by children at risk for SD-GEN differ from the sibilant errors of speech-delayed children meeting classification criteria for SD-OME.

2. METHOD

Participants

Conversational speech samples from 121 preschool children were selected from an audiocassette-recorded archive. The conversational samples had been collected in the course of ongoing etiological studies of child speech-sound disorders. The samples met two criteria.

ISBN 1-876346-48-5

First, children must have been classified as having speech delay of unknown origin (SD), using a set of assessment tools in the PEPPER suite, which was developed for research in speech-sound disorders [*16*]. Second, children were sub-classified by the number of nuclear family members (siblings and/or biological parents) who currently or previously had a speech-language impairment. Children assigned to Group A (n = 24) each had at least two nuclear family members (sibling(s) and/or parent(s)) who reportedly had a speech-language impairment; children assigned to Group B (n = 49) each had one nuclear family member meeting this criterion; and children assigned to Group C (n = 48) had no nuclear family members with prior or current speech-language impairment. Thus, Group A was considered most at risk for genetically transmitted SD, Group B at intermediate risk, and Group C at least risk. Sociodemographic information on participants is provided in the presentation.

Procedures

Narrow phonetic transcription of each of the 121 conversational samples had been completed by research transcribers using diacritic symbols and transcription conventions developed for research in typical and atypical speech development [*17*]. Quantitative analyses from the PEPPER package included descriptive metrics and inferential statistical tests at the level of sounds, features, error types, and diacritics. For the present purposes, a participant was considered to have dentalized /s/ or backed /s/ (palatalized, retroflexed, or lateralized) if at least 15% of their distortions on /s/ met criteria, respectively, for each of the two error classes. Moments analyses [*15*] were completed on selected /s/ tokens from each speaker. These acoustic data are presented elsewhere.

3. RESULTS

Figure 1 is a panel from the PEPPER output that provides both numeric (upper section) and graphic (lower section) information on the omission errors of the participants in groups A, B, and C. As shown in the upper section, titled *Percentage of Absolute Omissions*, participants in Group A had significantly more omission errors on fricatives. As shown in the lower section, titled *Percent Relative Omissions*, participants also had significantly more fricative omissions when calculated as a proportion of all errors (i.e., relative errors normalize for absolute differences in severity of involvement). Group A also tended to have more relative omission errors as cross-tabulated by Sound Class (Sonorant, Obstruent), Voicing Feature (Voiced, Voiceless), and three of the five manner classes.

As described in the introduction, we were particularly interested in comparing the types of errors observed on sibilant fricatives in the three groups. As shown in Figure 2, there were no statistically significant differences in the percentage of participants meeting criteria for dentalized /s/ productions (i.e., fronting). There was, however, a statistically significant difference in the percentage of Group A participants who backed /s/ (72%) compared to the percentage of Group A participants (55%) who backed /s/.

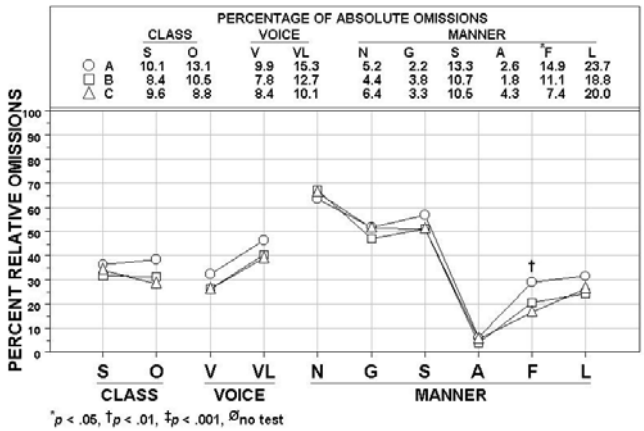

Figure 1. Comparison of omission errors.

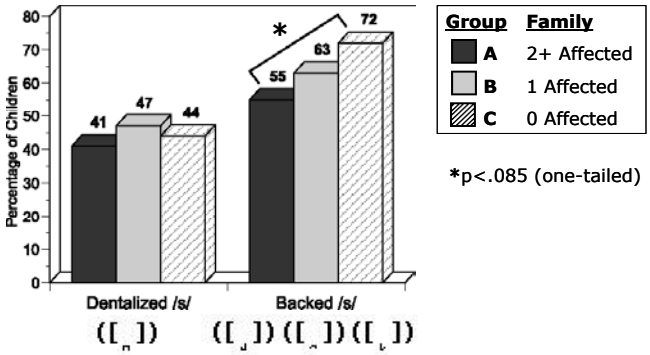

Figure 2. Comparison of distortion errors.

4. DISCUSSION AND CONCLUSION

These auditory-perceptual findings provide the first reported data on the hypothesis that there may be speech markers of a subtype of SD that is genetically transmitted (i.e., SD-GEN). As developed at the outset of this paper, prior research suggests that such phenotype markers could be available and persistent in family members studied in speech-genetics research. The diagnostic accuracy of this potential marker using acoustic methods is currently being assessed.

Theoretical implications of these findings and the associated acoustic data relate to possible proximal causes for speech delay. Neurolinguistic and psycholinguistic substrates of speech delay include possible constraints in auditory-perceptual, cognitive-linguistic, and speech-motor processes [*10*]. Increased omission errors in children at greatest risk for SD-GEN (see Figure 1) are consistent with the array of cognitive-linguistic constraints implicated in verbal trait disorders (e.g., phonological awareness, phonological working memory, speed of processing) that may be genetically transmitted endophenotypes. In contrast, as reviewed in the introduction, the atypical or "non-natural" distortion errors aggregated as backing are consistent with the fluctuant conductive hearing loss (i.e., auditory-perceptual constraints) associated with early recurrent otitis media with effusion (SD-OME).

ISBN 1-876346-48-5

ACKNOWLEDGMENTS

This research is supported by the National Institute on Deafness and Other Communication Disorders, NIDCD DC00496.

REFERENCES

[1] Shriberg, L. D. (1993). Four new speech and prosody-voice measures for genetics research and other studies in developmental phonological disorders. *Journal of Speech and Hearing Research, 36,* 105–140.

[2] Stromswold, K. (1998). Genetics of spoken language disorders. *Human Biology, 70,* 297–324.

[3] Stromswold, K. (2001). The heritability of language: A review and metaanalysis of twin, adoption and linkage studies. *Language, 77,* 647–723.

[4] Fisher, S. E., Vargha-Khadem, F., Watkins, K. E., Monaco, A. P., & Pembrey, M. E. (1998). Localisation of a gene implicated in a severe speech and language disorder. *Nature Genetics, 18,* 168–170.

[5] Lai, C. S. L., Fisher, S. E., Hurst, J. A., Levy, E. R., Hodgson, S., Fox, M., Jeremiah, S., Povey, S., Jamison, D. C., Green, E. D., Vargha-Khadem, F., & Monaco, A. P. (2000). The SPCH1 region on human 7q31: Genomic characterization of the critical interval and localization of translocations associated with speech and language disorder. *American Journal of Human Genetics, 67,* 357–368.

[6] Lai, C. S. L., Fisher, S. E., Hurst, J. A., Vargha-Khadem, F., & Monaco, A. P. (2001). A forkhead-domain gene is mutated in a severe speech and language disorder. *Nature, 413,* 519–523.

[7] Schick, J. H., Kundtz-Kluge, A. M., Tiwari, H. K., Taylor, H. G., Shriberg, L. D., Hansen, A., Freebairn, L. A., Lewis, B. A., & Iyengar, S. K. (2003). *Linkage and association analysis of families with speech-sound disorder: Evidence for a susceptibility locus on 7q31.* Manuscript submitted for publication.

[8] Karlsson, H. B., Shriberg, L. D., Flipsen, P., Jr., & McSweeny, J. L. (2002). Acoustic phenotypes for speech-genetics studies: Toward an acoustic marker for residual /s/ distortions. *Clinical Linguistics and Phonetics, 16,* 403–424.

[9] Shriberg, L. D., Flipsen, P., Jr., Kwiatkowski, J., & McSweeny, J. L. (in press). A diagnostic marker for speech delay associated with otitis media with effusion: The intelligibility-speech gap. *Special Issue: Diagnostic Markers for Child Speech-Sound Disorders, Clinical Linguistics & Phonetics.*

[10] Shriberg, L. D., Kent, R. D., Karlsson, H. B., McSweeny, J. L., Nadler, C. J., & Brown, R. L. (in press). A diagnostic marker for speech delay associated with otitis media with effusion: Backing of obstruents. *Special Issue: Diagnostic Markers for Child Speech-Sound Disorders, Clinical Linguistics & Phonetics.*

[11] Shriberg, L. D., Campbell, T. F., Karlsson, H. B., Brown, R. L., McSweeny, J. L., & Nadler, C. J. (in press). A diagnostic marker for childhood apraxia of speech: The lexical stress ratio. *Special Issue: Diagnostic Markers for Child Speech-Sound Disorders, Clinical Linguistics & Phonetics.*

[12] Shriberg, L. D., Green, J. R., Campbell, T. F., McSweeny, J. L., & Scheer, A. (in press). A diagnostic marker for childhood apraxia of speech: The coefficient of variation ratio. *Special Issue: Diagnostic Markers for Child Speech-Sound Disorders, Clinical Linguistics & Phonetics.*

[13] Shriberg, L. D., Flipsen, P., Jr., Karlsson, H. B., & McSweeny, J. L. (2001). Acoustic phenotypes for speech-genetics studies: An acoustic marker for residual /ɝ/ distortions. *Clinical Linguistics and Phonetics, 15,* 631–650.

[14] Karlsson, H. B., Shriberg, L. D., Scheer, A., & Nadler, C. J. (2003). *Acoustic moments data for palatalized and dentalized sibilant productions from speech delayed children with and without histories of otitis media with effusion.* (Tech. Rep. No. 12). Phonology Project, Waisman Center, University of Wisconsin-Madison.

[15] Milenkovic, P. (2000). Time-frequency analysis for 32-bit Windows [Computer software]. Madison, WI: Department of Electrical Engineering, University of Wisconsin-Madison.

[16] Shriberg, L. D., Allen, C. T., McSweeny, J. L., & Wilson, D. L. (2001). PEPPER: Programs to examine phonetic and phonologic evaluation records [Computer software]. Madison, WI: Waisman Center Research Computing Facility, University of Wisconsin-Madison.

[17] Shriberg, L. D., & Kent, R. D. (2003). *Clinical phonetics* (3rd ed.). Boston: Allyn & Bacon.

 ISBN 1-876346-48-5

ISBN 1-876346-48-5