INTRODUCTION

Gene x Environment Interactions

- Gene x environment (g x e) interactions have been relatively neglected in speech, language, and reading disorders.
- Two models make competing predictions about the direction of predicted g x e interactions.
  - Diathesis-stress model (Rende & Plomin, 1992)
    - Effects of genotype are larger in risk environments.
  - Biological model (Bronfenbrenner & Ceci, 1994)
    - Effects of genotype are smaller in risk environments.

METHODS

Participants
- 60 children with SSD (5-7 years) and their biological siblings (5-9 years) = 79 sib-pairs.
- Children with SSD had a history of speech therapy and/or scored below the 30th percentile on the Sounds-in-Words subset of the Goldman Fristoe Test of Articulation.

Procedure
- Composite phenotypes were created based on the results of a confirmatory factor analysis: Articulation, Oral-Motor skills, Semantics, Syntax, Phonological Awareness, Phonological Memory, Letter-Naming, and Rapid Naming.
- Environmental measures were controlled for those that had an impact on the phenotypes. Those that passed the screen were: parent education, shared reading, and home literacy environment.
- Environmental measures that showed g-e correlations were excluded from the analyses.
- DNA obtained from buccal brushes.
- Markers from RD candidate regions on chromosomes 1p36, 6p22, and 15q21 were typed and ibd estimates calculated using Merlin.

Analyses
- Regression-based approaches appropriate for selected samples:
  - DeFries-Fuker multipoint linkage
  - Merlin-Regress multipoint linkage

RESULTS

Localizing the Linkage Peaks

- Analyses first focused on localizing the linkage peaks described by Smith et al. (2005) using this new set of composite phenotypes.
- Results showed fairly good convergence between the DeFries-Fuker and Merlin-Regress methods.
- Evidence for linkage at 6p22 and 15q21, but not 1p36.

DISCUSSION

- Phenotypes showing converging evidence of linkage: Phonological Awareness and Letter Knowledge.
- The graphs were complicated and showed 2 separate linkage peaks.
  - One of the linkage peaks was very close to the 2 proposed RD candidate genes KIAA0319 and (Cope et al., 2005; Francks et al., 2004; Meng et al., 2005; Schuchardt et al., 2006).
  - The other linkage peak is more distal to these genes although significant linkage peaks have also been reported in this region (Grigorenko, 1997).

Gene x Environment Interactions

- Test for g x e interactions in SSD using a sib-pair linkage design.
- Use molecular genetic measures of “g” and previously identified SSD/RD loci in this sample: 1p36, 6p22, and 15q21 (Smith et al., 2005).
- Use psychosocial measures of “e” related to speech, language, or literacy development.

FUTURE DIRECTIONS

- Final confirmation of these results will await identification and replication of candidate genes for SSD and RD.
- In the meantime, these linkage-based methods could be used in larger samples and different developmental disorders to inform theory about g x e models.
- Specifically, more research is needed to determine what factors are important for determining the direction of a g x e interaction:
  - Type of disorder (psychopathology vs. cognitive)?
  - Type of environmental factor (risk vs. protective)?
  - Type of genetic factor (risk vs. protective)?