



Heritability and longitudinal outcomes of spelling skills in individuals with histories of early speech and language disorders

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ABSTRACT

This study examined the spelling skills in middle childhood and adolescence in individuals with histories of early childhood speech sound disorders (SSD) with and without language impairment (LI). Youth without such histories were also included (No SSD/LI group). The heritability of spelling skills at each age level was estimated. Children with SSD were classified as SSD-only, SSD with LI but without childhood apraxia of speech (SSD + LI/No CAS), and CAS and LI (CAS + LI). The SSD-only group did not differ in spelling from the No SSD/LI group, suggesting that SSD-only did not increase risk for poor spelling. The SSD + LI/No CAS and CAS + LI groups had poorer spelling skills than the SSD-only and No SSD/LI groups. Spelling was associated with phonological awareness in the middle childhood and adolescent samples and with rapid automatized naming in the adolescent sample. Heritability of spelling skills was stronger in adolescence than in middle childhood. Differences in the correlates of spelling and in heritability at the two ages suggest developmental changes in the factors contributing to spelling.

1. Introduction

Over the past two decades, links between spoken language and literacy skills have been identified and a body of literature has emerged that indicates that children with early spoken language deficits are at risk for later literacy difficulties. Although much of the research has focused on the relationship between language impairment and reading skills (Bishop & Snowling, 2004; Catts, 1996; Catts, Adlof, Hogan, & Weismer, 2005; Catts, Bridges, Little, & Tomblin, 2008; Conti-Ramsden, Durkin, Simkin, & Knox, 2009; Kamhi & Catts, 2012), children with speech sound disorders (SSD), with accompanying language impairment (LI), may be at risk for spelling difficulties (Lewis, Freebairn, & Taylor, 2002). SSD, defined as a delay in acquiring speech sounds, are common communication disorders experienced by young children with an estimated 3.8% of 6-year-olds demonstrating SSD (Shriberg, Tomblin, & McSweeney, 1999). Although most SSD resolve by 8 years of age, more than half of these children continue to present with spelling difficulties (Bird, Bishop, & Freeman, 1995; Lewis et al., 2002).

Studies examining spelling problems in children with SSD have focused on primarily on early spelling difficulties and have not followed students into adolescence to determine if spelling difficulties persist

(Bird et al., 1995; Hayiou-Thomas, Carroll, Leavett, Hulme, & Snowling, 2017; Lewis et al., 2002; Overby, Masterson, & Preston, 2015). Evidence from genetic studies indicates that spelling skills have greater heritability at older ages, and that environmental factors, such as exposure to print and instruction in spelling, are more strongly related to spelling skills at younger ages (DeFries, Stevenson, Gillis, & Wadsworth, 1991; Friend, DeFries, Wadsworth, & Olson, 2007; Stevenson, Graham, Fredman, & McLoughlin, 1987). However, these studies have primarily included typical children or children with reading disabilities and have not focused on individuals with histories of SSD or identified the cognitive-linguistic abilities associated with spelling at younger and older ages. The present study examined spelling skills in middle childhood (7–11.9 years) and adolescence (12–17.9 years) in a large cohort of students with prior histories of SSD. Our primary aims were to determine if spelling difficulties were evident at both ages, to explore potential age differences in the skills associated with spelling, and to identify differences in spelling outcomes among clinical subtypes of SSD. The second aim was to determine if spelling skills had greater heritability in adolescence than in middle childhood, as reported in studies of typically developing students (Friend et al., 2007; Stevenson et al., 1987).

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1.1. SSD and risk for spelling difficulties

Previous studies demonstrated that children with SSD are at risk for spelling difficulties (Bird et al., 1995; Hayiou-Thomas et al., 2017; Lewis et al., 2002; Overby et al., 2015; Peterson, Pennington, Shriberg, & Boada, 2009). These difficulties may result from weaknesses in mapping phonemes to graphemes (Overby et al., 2015), reduced phonological memory and/or perception (Fowler, 1991), and inaccurate or “fuzzy” phonological representations (Lewis et al., 2002). Any of these basic skill deficits in children with SSD may undermine proficiency in spelling and reading (Gillon, 2005; Gillon & Moriarty, 2007).

Our previous studies have indicated that children with histories of SSD with co-morbid LI are more at risk for literacy difficulties than children with histories of SSD-only (Lewis et al., 2002; Lewis et al., 2015; Lewis, O'Donnell, Freebairn, & Taylor, 1998). Children with SSD-only were found to demonstrate a weakness in spelling relative to their language and reading scores (Lewis et al., 2002). Paired *t*-test comparisons revealed that children with SSD-only performed more poorly on the spelling measure than the language or reading decoding measure. One explanation for this finding may be that although word recognition requires only partial identification of orthographic and phonological patterns, spelling requires more complete orthographic and phonological representations (Friend et al., 2007). Another study that employed factor analysis demonstrated that articulation and phonology skills in early childhood are related to later spelling abilities, partially independent of language skills (Lewis et al., 2006). Spelling impairment at school-age was also predicted by early childhood use of phonological processes, word discrimination, and syntax (Lewis et al., 2002). Taken together these findings confirm that children with early childhood SSD and LI are at greater risk for spelling difficulty than children with SSD-only. An SSD may impact spelling skills to a lesser extent than LI possibly due to phonological processing deficits.

Past research reveals an association of speech errors with spelling difficulties at early school age (Gillon, 2002; Hayiou-Thomas et al., 2017; Overby et al., 2015; Raitano, Pennington, Tunick, Boada, & Shriberg, 2004). Children with different clinical subtypes of SSD may also be at differential risk for spelling difficulties. For example, spelling disabilities in children with SSD and comorbid LI, one subtype of SSD, likely reflect problems in phonological representations and other domains, such as semantics, syntax and morphology. Hayiou-Thomas et al. (2017) reported that literacy difficulties at 8 years of age were related to the persistence of the SSD, disordered speech versus delayed speech, co-morbid LI, and a family history of dyslexia, but that these difficulties were not associated with the severity of the SSD at 3 ½ years of age.

A subtype of SSD is childhood apraxia of speech (CAS). CAS is a severe form of SSD with impairment in the planning and/or programming of speech movement sequences (ASHA, 2007). There is disagreement as to the core diagnostic features of CAS. Some view CAS as solely a motor programming deficit that may also present with difficulties in the suprasegmental aspects of speech (Shriberg et al., 2017). Others consider CAS to be a symptom complex that includes linguistic deficits, especially with phonological representations (Aram, 1984; Marion, Sussman, & Marquardt, 1993; Marquardt, Sussman, Snow, & Jacks, 2002; Velleman & Strand, 1994). Froud and Khamis-Dakwar (2012) provided evidence that children with CAS demonstrate phonological over-specification. They concluded that CAS has a core phonological component in addition to a motor planning component. Other studies have examined auditory perceptual deficits in children with CAS (Nijland, 2009) and found children with CAS to show auditory perceptual deficits for both higher and lower order linguistic input. Deficient phonological representations and impaired auditory perceptual deficits may place children with CAS at greater risk for spelling difficulties than children with other types of SSD. Stackhouse and Wells (1997) hypothesized that accurate phonological representations needed for spelling are not formed as the child with CAS is unable to produce

complex words, words are not correctly encoded, and auditory feedback is disrupted. The child with CAS may also have cognitive and phonological deficits, or difficulties in morphology and syntax (Gillon & Moriarty, 2007; Lewis, Freebairn, Hansen, Iyengar, & Taylor, 2004) that impair spelling skills. In our previous work we demonstrated that weakness in phoneme segmentation and phoneme sequencing in children with CAS were associated with poorer speech production than in children with other subtypes of SSD and spelling problems (Lewis et al., 2004). In the current study we focused on spelling skills in a sample of children with early childhood SSD to examine potential differences related to the type of SSD, underlying cognitive skills, and age at assessment.

1.2. Endophenotypes for spelling

Endophenotypes are conceptualized as the basic cognitive-linguistic competencies that contribute to acquisition of more complex traits such as spelling with potential utility in detecting genetic influences (Gottesman & Gould, 2003). Phonological awareness (PA) is an endophenotype hypothesized to underlie speech, language, reading and spelling (Nathan, Stackhouse, Gouldandris, & Snowling, 2004; Raitano et al., 2004; Lewis et al., 2011). Poor PA may result in inaccurate mappings of phonemes onto graphemes. PA is dependent on strong phonological representations that may be deficient in children with SSD, as well as some children with LI (Bird et al., 1995; Preston & Edwards, 2010; Rvachew & Grawberg, 2006) and may also contribute to difficulties in encoding phonemes onto graphemes (Ball & Blachman, 1991; Catts, Kamhi, & Adlof, 2012).

Another endophenotype in common with reading and spelling abilities is speed of name retrieval as assessed by rapid automatized naming (RAN), a skill that has been hypothesized to tap lexical access and phonological processing (Denckla & Rudel, 1976; Norton & Wolf, 2012). RAN requires naming of a small set of randomly presented colors, objects, numbers or letters as quickly as possible. Although most studies have examined the association of RAN performance to reading abilities, cognitive skills that are associated with RAN may also underlie spelling skills (Stainthorpe, Powell, & Stuart, 2013). RAN predicts spelling disability independent of PA in typical children and children with reading disabilities, suggesting separate spelling-related processes (Furnes & Samuelssen, 2010; Norton & Wolf, 2012). These spelling-related processes include the graphotactics, orthography, and phonological representations, rapid access and retrieval of phonological information, and oral motor skills—all skills that may be impaired in children with SSD (Norton & Wolf, 2012; Treiman, 2017). Stainthorpe et al. (2013) demonstrated that children with good PA but poor RAN skills were poorer in spelling skills than matched controls, especially in spelling irregular words such as “yacht”. They concluded that RAN was associated with the establishment of fully specified orthographic representations. PA skills, on the other hand, may be more associated with phonological decoding. The present study examined the contribution of PA and RAN to spelling skills at middle childhood and adolescence in individuals with histories of SSD.

1.3. Genetics of spelling disorders

In our previous work, we noted familial aggregation of poor spelling skills in family pedigrees of children with SSD (Lewis et al., 2002). Although school-age children with combined SSD and LI performed more poorly than children with SSD-only on spelling measures, children with SSD-only exhibited a weakness in spelling skills relative to their reading skills, language abilities and performance IQ. These findings suggest that spelling in particular may be difficult for children with a history of SSD and that these spelling difficulties aggregate within families.

Compared to the many family and genetic studies of reading, few studies have examined spelling skills independently of a reading

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