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# Subtyping Children With Speech Sound Disorders by Endophenotypes

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## Abstract

**Purpose—**The present study examined associations of 5 endophenotypes (i.e., measurable skills that are closely associated with speech sound disorders and are useful in detecting genetic influences on speech sound production), oral motor skills, phonological memory, phonological awareness, vocabulary, and speeded naming, with 3 clinical criteria for classifying speech sound disorders: severity of speech sound disorders, our previously reported clinical subtypes (speech sound disorders alone, speech sound disorders with language impairment, and childhood apraxia of speech), and the comorbid condition of reading disorders.

**Participants and Method—**Children with speech sound disorders and their siblings were assessed at early childhood (ages 4–7 years) on measures of the 5 endophenotypes. Severity of speech sound disorders was determined using the *z* score for Percent Consonants Correct—Revised (developed by Shriberg, Austin, Lewis, McSweeny, & Wilson, 1997). Analyses of variance were employed to determine how these endophenotypes differed among the clinical subtypes of speech sound disorders.

**Results and Conclusions—**Phonological memory was related to all 3 clinical classifications of speech sound disorders. Our previous subtypes of speech sound disorders and comorbid conditions of language impairment and reading disorder were associated with phonological awareness, while severity of speech sound disorders was weakly associated with this endophenotype. Vocabulary was associated with mild versus moderate speech sound disorders, as well as comorbid conditions of language impairment and reading disorder. These 3



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endophenotypes proved useful in differentiating subtypes of speech sound disorders and in validating current clinical classifications of speech sound disorders.

**Keywords**

endophenotypes; oral motor skills; phonological awareness; phonological memory; severity; speech sound disorders; speeded naming; subtypes; vocabulary

Children with early speech sound disorders (SSD) of unknown etiology comprise a heterogenous group with high prevalence rates, especially during the preschool years (Campbell et al., 2003). Subgrouping children into clinically validated subtypes of SSD allows for more precision in the diagnosis, treatment, and prognosis of these disorders. However, to date, there is no single classification system of SSD that is employed uniformly by SLPs (speech-language pathologists). Children with SSD may be grouped differently depending on whether or not the subgroups are based on hypothesized etiology (Shriberg et al., 2005), psycholinguistic models (Stackhouse & Wells, 1997), or behavioral symptoms of SSD (e.g., phonological processes; Dodd, 2005). These classification systems have provided insight into the nature of SSD and have resulted in different diagnostic categories and treatment approaches. However, comorbidity of subtypes (e.g., a combined articulation and phonological disorder), as well as different combinations of related disorders such as language impairment (LI) and reading disorders (RD), result in overlapping diagnostic categories, complicating understanding of causal relationships and choices of treatment approaches.

Some past studies have subtyped children with SSD based on the clinical features of the disorders, such as speech sound errors and comorbid LI. Other studies have examined associations of cognitive–linguistic skills or endophenotypes with SSD. Endopheno-types are objectively measurable cognitive, linguistic, or neuropsychological parameters that are closely associated with a specific behavioral trait and are useful in detecting genetic influences on the behavioral phenotype (Gottesman & Gould, 2003; Inoue & Lupski, 2003), for example, phoneme awareness has been associated with RD. Few studies, however, have investigated the extent to which endophenotypes are related to classifications based on the clinical features of SSD. As endophenotypes relate more directly to underlying genetic influences (Gottesman & Gould, 2003), differential association of these skills with clinical features of SSD may help to elucidate the sources of important clinical variations.

## Subtyping SSD on the Basis of Speech Errors

Previous attempts to subtype children with SSD have relied on various classifications of features of the child's speech. Traditionally, clinical severity ratings of the disorder as mild, moderate, or severe have depended on the SLP's judgment of the child's intelligibility in conversational speech or the number of speech sound errors that the child exhibits on a standardized articulation test. Analyses of speech error types, such as phonological process analyses or classification of errors as omissions, substitutions, or distortions, imply an assumption that children with similar error types share a common etiology and/or may benefit from similar treatment approaches. Children with a large proportion of omission errors, for example, have been considered to have a linguistically based disorder rather than a motor-based disorder and have been viewed as qualitatively different from children with primarily substitution or distortion errors (Panagos, 1974). Preston and Edwards (2010) found that children with atypical speech sound errors had poorer phonological awareness skills and lower receptive vocabularies than children with more typical speech sound errors.

Other classification systems have grouped children with SSD on the basis of a combination of factors that include history, severity, and associated conditions, as well as speech sound error characteristics (Dodd, 2005; Shriberg, 1994; Shriberg & Kwiatowski, 1988; Shriberg, Kwiatkowski, Best, Hengst, & Terselic-Weber, 1986). Such systems have been useful clinically in deriving diagnostic profiles of children with SSD. For example, a system proposed by Dodd (2005) classifies SSD into five subtypes: articulation disorder, delayed phonological acquisition, consistent deviant disorder, inconsistent deviant disorder, and other (including dysfluency, dysarthria, and apraxia of speech). This system does not consider the etiology of the SSD, and, therefore, children within a subtype may have diverse hypothesized causal factors that may influence prognosis and treatment.

Another framework for subtyping children with SSD that does consider etiology, proposed by Shriberg and colleagues (Shriberg, 1993; Shriberg et al., 2005; Shriberg & Kwiatkowski, 1982, 1994; Shriberg, 2010), is the Speech Disorder Classification System (SDCS). This classification system proposes seven subtypes of SSD: speech delay—genetic, speech delay—otitis media with effusion, speech delay—apraxia, speech delay—dysarthria, speech delay—developmental psychosocial involvement, and two categories of speech errors limited to distortions of speech sounds. Children with speech delay—apraxia, also called childhood apraxia of speech (CAS), are of particular interest because of the severity of the disorder, the prolonged course of treatment, and the distinct speech characteristics (see the American Speech-Language-Hearing Association [ASHA] position statement report for a detailed description; ASHA, 2007). In our previous studies, we have demonstrated that children with CAS represent a distinct subtype of SSD with a long-term impact on school-age academic skills (Lewis, Freebairn, Hansen, Taylor, Iyengar, & Shriberg, 2004; Lewis, Free-bairn, Hansen, Iyengar, & Taylor, 2004).

## Subtyping by Comorbid LI

One factor that appears to distinguish subgroups of SSD is the presence or absence of comorbid LI. Shriberg, Tomblin, and McSweeny (1999) reported 11% to 15% comorbidity of speech delay with LI at 6 years of age, with considerably higher rates of comorbidity for preschool children with speech delay (40%–60%; Shriberg & Kwiatkowski, 1994). A growing body of literature suggests that children with LI in addition to the SSD have poorer outcomes than children with SSD in isolation (Aram & Hall, 1989; Bishop & Adams, 1990; King, Jones, & Laskey, 1982; Lewis, Freebairn, & Taylor, 2000; Nathan, Stackhouse, Goulandris, & Snowling, 2004; Shriberg & Austin, 1998; Young et al., 2002).

In our own studies, children with SSD accompanied by LI performed more poorly than children with isolated SSD at school age on measures of spelling, reading decoding, reading comprehension, and written language, as well as on language measures and phonological awareness. Eighteen percent with isolated SSD and 75% with combined SSD and LI had reading problems at school age (Lewis, Freebairn, & Taylor, 2000; Lewis, O'Donnell, Freebairn, & Taylor, 2002). Despite the poorer academic outcomes, children with combined SSD and LI did not present with more severe SSD at early childhood than children with isolated SSD. This suggests that co-morbid conditions such as LI may not be useful in forming subgroups of children with SSD at early childhood.

## Associations of Endophenotypes with SSD

Recently, genetic studies of SSD have examined endophenotypes as the presumed heritable components that reflect genetic influences leading to SSD. Endophenotypes are thought to influence clinical phenotypes such as SSD, LI, and RD (Bishop & Snowling, 2004; Castellanos & Tannock, 2002; Fisher & DeFries, 2002; Pennington, 1997). The endophenotype is hypothesized to involve fewer genes than the clinical phenotype,

simplifying the genetic analysis (Gottesman & Gould, 2003). Endophenotypes, such as oral motor skills, phonological awareness, phonological memory, speed of processing, and vocabulary, are postulated to influence SSD.

Several studies have demonstrated that children with speech and language disorders present with poorer *motor skills* than normally developing children (Bishop, 2002; Bishop & Adams, 1990; Bishop & Edmundson, 1987; Hill, 2001). Bishop (2002) employed a tapping task and a peg movement task that were timed. She found that the tapping task was related to speech production accuracy and the peg movement task was related to nonword repetition. Hill (2001) reviewed 28 studies assessing children with LI employing motor tasks such as peg moving, bead threading, and finger tapping and found associations of poor motor skills to LI. Timed tasks such as those employed by Bishop (2002) may be confounded with slowed speed of processing in general. Studies specific to oral motor skills have demonstrated that children with primarily articulation disorders perform more poorly on diadochokinetic tasks than their normal peers (Bernthal, Bankson, & Flipson, 2009). Other factors such as tongue strength, fine tongue control, and stability may also play a role.

*Phonological awareness*, also an endophenotype associated with SSD, is assessed by tasks that require children to manipulate phonemes in spoken words, such as by phoneme deletion or reversal. Deficits in phonological awareness have been identified as one of the strongest earliest predictors of RD (e.g., Raitano, Pennington, Tunick, Boada, & Shriberg, 2004). Studies also have demonstrated that children with SSD, both with and without comorbid LI, perform more poorly than controls on phonological awareness measures (Colledge, et al., 2002; Kovas et al., 2005; Nathan, Stackhouse, Goulandris & Snowling, 2004; Preston & Edwards, 2010). Thus, phonological awareness is a useful endophenotype for SSD as well as RD.

*Phonological memory* refers to coding information phonologically for temporary storage in working or short-term memory (Gathercole & Baddeley, 1990; Torgesen & Wagner, 1992). It is usually assessed by asking children to repeat nonwords. Deficits in phonological memory are thought to impair an individual's ability to learn both spoken and written new words. Poor phonological memory may result in weak phonological representations that underlie SSD and RD (Adams & Gathercole, 1995).

*Speeded naming* has been implicated as an endophenotype in studies that have shown that children with LI and/or RD have a generalized slowing of information processing when compared with control children (Windsor & Hwang, 1999). Slower speed of processing may also impact the establishment of phonological representations for words as the phonological information held in memory may decay before the representation is established (Montgomery, Magimairaj, & Finney, 2010).

*Vocabulary* knowledge is an endophenotype that plays a critical role in reading acquisition and skill (Wise, Sevcik, Morris, Lovett, & Wolf, 2007). The semantic content of a word has been shown to influence phonological skills as well. For example, vocabulary has been shown to account for 25% to 30% of the variance in phonological awareness in preschool and young school-age children (Bishop & Adams, 1990; Elbro, Borstrom, & Peterson, 1998; Rvachew & Grawberg, 2006). Metsala (1999) hypothesized that children who know more words have phonological representations that are more adult-like in their features and organization.

## Purpose of the Study

The preferred practice patterns for the profession of speech language pathology (ASHA, 2004) indicate that clinicians should document the type and severity of the SSD and its

associated conditions. Yet, as reported by Flipsen, Hammer, and Yost (2005), there is little agreement on ratings of severity of SSD even by experienced SLPs, especially in the middle of the scale range. Past studies have not examined possible endophenotypic differences between clinical subtypes. Discovery of such differences would shed light on the deficits underlying clinical variations in SSD and help guide further studies of underlying genetic mechanisms. To address this gap in knowledge, this study examined associations of endophenotypes of oral motor skills, phonological memory, phonological awareness, vocabulary, and speeded naming with clinical classifications on the basis of severity and comorbidity. These classifications included (1) severity of SSD (moderate SSD, mild-moderate SSD, mild SSD, recovered SSD, and no SSD), (2) comorbidity of SSD with LI and RD (SSD alone, SSD with RD, SSD with LI, SSD with RD, and LI), and (3) our previously defined subtypes of SSD (SSD alone, SSD with LI, and CAS).

## Method

### Participants

Participants were 237 children who were enrolled in a large longitudinal genetic study of SSD, including both the probands with SSD and their nonreferred, typically developing siblings. Probands were identified as having SSD ($n = 168$) at 4 to 7 years of age ($n = 79$ participants aged 5 years and younger) from the clinical caseloads of SLPs working at community speech and hearing centers or in private practice in the greater Cleveland area. All participants demonstrated the following: (1) normal hearing acuity as defined by passing a pure tone audiometric screening test at 25 dB HL ISO for 500, 1000, 2000, and 4000 Hz bilaterally and fewer than six reported episodes of otitis media before the age of 3 years as reported by the parent; (2) absence of a history of neurological disorders or developmental delays other than speech and language as reported by the parent; and (3) *normal intelligence* defined as a performance intelligence quotient (PIQ) of 80 or above on the Wechsler Intelligence Test Scale for Children–Third Edition (WISC-III; Wechsler, 1991) or the Wechsler Preschool and Primary Scales of Intelligence–Revised (WPPSI-R; Wechsler, 1989).

The severity of SSD of the probands was defined in terms of a conversational speech sample of at least 50 utterances, obtained using technical and interlocutor procedures for free speech sampling, as described by Shriberg and Kwiatkowski (1982). The Percentage of Consonants Correct-Revised (PCCR) metric (Shriberg et al., 1997) was calculated. The PCCR is a well-respected and widely used measure, both clinically and in research, to estimate the severity of speech impairment. It is simple to calculate and has demonstrated validity and reliability (Shriberg, Austin, Lewis, McSweeny, & Wilson, 1997; Shriberg & Kwiatkowski, 1982). It is similar to the Percent Consonant Correct (PCC) metric, with the exception that speech sound distortions are not counted as errors. *Z* scores were created on the basis of Shriberg's work to account for age (Shriberg et al., 1997). On the basis of these *z* scores of the PCCR, five groups were created: zPCCR less than −2.0 (moderate SSD), zPCCR less than -1.0 but greater than −2.0 (mild-moderate SSD), zPCCR less than 0 but greater than −1.0 (mild SSD), zPCCR greater than 0 but reported history of therapy for SSD (recovered SSD), and zPCCR greater than 0 with no history of SSD (no SSD).

### Procedures

The participants were tested individually in two sessions to reduce potential effects of fatigue on test results. Testing was carried out in a speech research laboratory at Case Western Reserve University or, at the parent's request, in a quiet and adequately lit room in the family's home. Speech productions were recorded on high-quality audiocassette. Responses were recorded initially online using broad phonetic transcription. Speech sound

samples were later transcribed. All tests were presented in a counter balanced manner, with the more time-consuming measures alternating with less lengthy measures. Parents completed a developmental questionnaire and structured interview on their children to document comorbid conditions of LI, RD, and a diagnosis of CAS.

## Measures of endophenotypes

**Oral motor skills—**Diadochokinetic rates of single and multisyllables on The Oral and Speech Motor Control Protocol (Robbins & Klee, 1987) or the Fletcher-Time-by-Count technique (Fletcher, 1977) were used to assess oral motor skills. The same syllables were employed in both tests to assure that the tasks were comparable; *z* scores were obtained.

**Phonological awareness—**Tests of phonological awareness included the Elision and Sound Blending Subtests of the Comprehensive Test of Phonological Processing (CTOPP; Wagner, Torgesen, & Rashotte, 1999). Standard scores were used in data analysis. The first available measurement for this variable was used for analysis. If children were younger than 5 years at their first visit, the CTOPP assessment from their secondvisit was used in analyses; *z* scores were obtained.

**Phonological short-term memory—**Phonological short-term memory was assessed by the Digit Span subtest of the WISC III, the Sentence Imitation Subtest of the Test of Language Development–Primary 3rd Edition (TOLDP:3; Newcomer & Hammill, 1997), and the repetition of multisyllabic real (Catts, 1986) and nonsense words (Catts, 1986). The rationale for using nonsense words to assess short-term phonological memory is that because the word is novel, the participant does not have it coded or stored in his lexicon. Again, if children were younger than 5 years at their first visit, the Digit Span sub-test from their second visit was used for analysis. Raw scores were age standardized according to the age of the child at assessment and the age-standardized scores were used in data analysis.

**Vocabulary—**The Expressive One Word Picture Vocabulary Test, Revised (EOWPVT-R; Gardner, 1990) and the Peabody Picture Vocabulary Test, 3rd Edition (PPVT-III; Dunn & Dunn, 1997) assessed receptive and expressive vocabulary skills. Standard scores were used in data analyses.

**Speeded naming—**Rapid Naming of Colors (Denckla & Rudel, 1976) assessed speeded naming, a measure of speed of processing. Rapid naming requires efficient retrieval of phonological information. *z* scores derived from the normative data by Denckla and Rudel (1976) were used in data analysis.

## Data analyses

Age-standardized *z* scores were created for each endophenotype by adjusting the individual measures for age and age squared. Our previous work has shown that, although performance on these measures improves with age, it does not do so in a straight line (Stein et al, 2011). Adjustment for age and age squared approximates this relationship so that all scores are on the same scale. A domain or factor score was derived for each endophenotype's *z* scores for the measures comprising that factor. The measures included were the diado-chokinetic rates of single and multisyllables on either The Oral and Speech Motor Control Protocol (Robbins & Klee, 1987) or the Fletcher-Time-by-Count (Fletcher, 1977) for the oral motor factor, the CTOPP subtests Elision and Sound Blending for the phonological awareness factor, the real-word and nonsense-word repetition tasks and the TOLD-P-3 Sentence Imitation for the phonological memory factor, the PPVT and EOW-PVT for the vocabulary factor, and the Rapid Naming of Colors for the speeded naming factor. The assignment of tests to domains was based on results from our previous factor analyses of the test battery (Lewis et al., 2006;

Stein et al., 2004; Sices, Taylor, Freebairn, Hansen, & Lewis, 2007). As shown in Table 1, significant correlations ($p < .05$) were observed among the phonological memory factor, the speeded naming factor, the phonological awareness factor, and the vocabulary factor.

Analysis of variance was employed to compare groups of individuals classified according to each of the three clinical dimensions on the factor scores. These classifications included severity (moderate SSD, mild-moderate SSD, mild SSD, recovered SSD, and no SSD) described earlier, comorbidity of RD (SSD alone without RD, SSD + LI without RD, SSD + RD, and SSD + LI + RD), and clinical subtype (no SSD, SSD only, SSD + LI, and CAS). The 168 children with SSD were grouped differently in each of the three clinical dimensions. When examining the comorbidity of SSD with RD, the SSD alone group ($n = 74$) was split into SSD alone without RD ($n = 67$) and SSD with RD ($n = 7$). The SSD + LI group ($n = 94$) was split into the SSD + LI without RD ($n = 54$) and the SSD + LI + RD ($n = 40$). When grouping the participants by clinical subtypes, children with a diagnosis of CAS ($n = 41$) formed a subgroup. All children with CAS except one presented with comorbid LI. Thus, in the clinical subtype analyses, 73 children remained in the SSD alone group and 53 children remained in the SSD + LI group. Pairwise differences between groupings were examined using Tukey's test with $\alpha = .05$.

## Results

### Severity groupings and associations with endophenotypes

As shown in Table 2, the five severity groups (moderate SSD, mild-moderate SSD, mild SSD, recovered SSD, and no SSD) differed significantly in gender, $\chi^2 = 12.3$, $p = .002$, and age, $F = 21.14$, $p < .0001$. The SSD groups comprised more males than females, with the moderate group 72% male, the mild-moderate group 50% male, and the mild group 65% male; whereas the typical group was 48% male. The recovered SSD group was older (mean age in years = 6.31, $SD = 0.97$) than the other SSD groups (mild group mean = 5.35, $SD = 1.31$; mild-moderate mean =.13, $SD = 1.31$ years; and moderate mean = 4.86, $SD = 1.36$). The groups also differed on the composite language scores of the TOLD-P:3 or the Clinical Evaluation of Language Fundamentals 3rd Edition (CELF-3; Semel, Wiig, & Secord, 1995), with the moderate SSD group performing more poorly (mean = 86.98 $SD = 16.47$) than the recovered group (mean = 101.26, $SD = 14.99$), the mild group (mean = 105.18, $SD = 14.16$), and the typically developing group (mean = 106.52, $SD = 15.65$). No group differences were observed on PIQ.

Comparisons of severity groups on the factor scores revealed that the groups differed on the phonological memory factor, with the moderate SSD presenting with lower scores on phonological memory, $F(4, 209) = 12.05$, $p < .0001$, than the recovered SSD and mild SSD groups, and lower scores on vocabulary, $F(4, 212) = 4.55$, $p < .0001$, than the recovered SSD group (Table 3). The no SSD group differed from the recovered SSD group, the mild-moderate SSD group, and the moderate SSD group on phonological memory, and from the moderate group on vocabulary. Phonological awareness showed a tentative difference among severity groups, $F(4, 172) = 2.10$, $p = .083$, though none of the pairwise comparisons was statistically significant at $\alpha = .05$.

### Comorbid conditions and associations with endophenotypes

Table 4 lists the comorbid conditions reported by the parents for participants in each group. LI ($n = 100$) was the most frequently reported comorbid condition, followed by RD ($n = 53$). Forty-two children (38 in the moderate group, 2 in the mild-moderate group, and 2 in the recovered group) were reported to have been diagnosed with CAS. The moderate group had the highest rates of comorbid disorders.

Group comparisons according to comorbid condition revealed differences in phonological awareness, $F = 15.19$, $p < .0001$; phonological memory, $F = 17.53$, $p < .0001$; and vocabulary, $F = 13.89$, $p < 0.0001$ (Table 5). For phonological awareness, the children with all three comorbid conditions had significantly lower scores than the children with SSD only, and the children with only SSD + LI or SSD + RI. In addition, children with SSD + LI had significantly lower scores on these measures than children with SSD + RD. For phonological memory, children with all three disorders again had significantly lower scores than children with SSD alone or SSD + RD and SSD + LI; and, in this case, children with SSD + LI had significantly lower scores than children with SSD alone. Finally, for vocabulary, children with all three disorders scored significantly lower than children with SSD only.

### Clinical subtypes and associations with endophenotypes

Comparison of the clinical subtypes on factors scores revealed differences in phonological awareness, $F = 11.084$, $p < .0001$; phonological memory, $F = 35.443$, $p < .0001$; and vocabulary, $F = 18.344$, $p < .0001$ (Table 6). For phonological awareness, the CAS subtype had significantly lower scores than both children with no SSD and the SSD only subtype, and the SSD + LI subtype had lower scores than the SSD alone subtype ($p < .05$). For phonological memory, children with no SSD had significantly higher scores than the SSD + LI or CAS subtypes, the SSD only subtype had significantly higher scores than the SSD + LI and CAS subtypes, and the SSD + LI subtype had significantly higher scores than CAS subtype ($p < .05$). Thus, all four clinical subtypes were distinguishable from each other using the phonological memory factor. Finally, for vocabulary, children with no SSD had significantly higher scores than the SSD + LI or CAS subtypes, and the SSD subtypes had significantly higher scores than the SSD + LI or CAS subtypes ($p < .05$).

## Discussion

The goal of the present study was to determine whether endophenotypes of oral motor skills, phonological awareness, phonological memory, speeded naming, and vocabulary distinguish children grouped according to common clinical classifications or dimensions (severity, comorbid conditions, clinical subtype) and thus provide information on underlying basis of these clinical distinctions. Three endophenotypes, phonological memory, phonological awareness, and vocabulary, were associated with severity of SSD, the presence of the comorbid conditions of LI and RD, and our previously reported clinical subtypes of SSD alone, SSD + LI, and CAS. Findings supported a Multiple Deficit Model proposed by Pennington (2006) and others (McGrath et al., 2007 in press; Pennington & Bishop, 2009; Peterson, Pennington, Shriberg, & Boada, 2009).

These findings provide insights into the core deficits associated with SSD and comorbid conditions of LI and RD, as well as support for current clinical diagnostic categories. The endophenotypes of phonological memory, phonological awareness, and vocabulary best distinguished subtypes based on sever ity of SSD, comorbid conditions of LI and RD and clinical diagnostic categories of SSD alone, SSD + LI, and CAS. All three clinical classifications were associated with differences in phonological memory. The results indicate that difficulty in holding onto speech sounds in memory long enough to form robust phonological representations is an important aspect of SSD in young children (Baddeley, 1998; Preston & Edwards, 2010). Kenney, Barac-Cikoja, Finnegan, Jeffries, and Ludlow (2006) suggest that this memory deficit may persist into adulthood for individuals with histories of SSD. Deficits in phonological short-term memory also have been linked to RD and LI (Kamhi & Catts, 1986; Kamhi, Catts, Mauer, Apel, & Gentry, 1988).

A second endophenotype, phonological awareness, has been proposed as a skill that underlies SSD, LI, and RD (Peterson, Pennington, Shriberg, & Boada, 2009; McGrath et al., in press). Phonological awareness distinguished groups basedonboth the presence of comorbid LI or RD and our previous clinical classifications of children into subgroups with SSD only, SSD + LI, and CAS. Children with SSD + LI and those with CAS had lower scores on our measure of phonological awareness than children with SSD alone or no SSD. This is contrary to the findings of Bird, Bishop, and Freeman (1995) who reported that children with expressive phonology disorders scored poorly on phonological awareness whether or not they had additional LI. One possible explanation for this discrepancy is the age of the cohort that we examined. Phonological awareness is a skill that emerges from 3 to 4 years of age with some skills such as phoneme segmentation not established until 6 to 7 years of age (McLeod, 2009). Specific phonological awareness tasks might be difficult for all children with SSD in the age range of our study (4–7 years) but may be more discriminating at older ages when most typically developing children have mastered phonological awareness. Comparisons of children exhibiting different patterns of comorbidity revealed that children with co-occurring LI or RD had poorer phonological awareness skills than those with SSD alone. Interestingly, in this study, children with SSD + LI performed more poorly on tests of phonological awareness than children with SSD + RD. This may be due to the small number of children in the SSD + RD group ($n = 6$), suggesting that it is unusual to find children with SSD + RD without comorbid LI. In addition, children with SSD + RD and no LI may differ in the underlying etiology of the RD. For example, Rvachew& Grawburg (2006) reported that both speech perception and vocabulary predict phonological awareness skills. Children with SSD + RD may have deficits in speech perception that were not examined in this study.

Another endophenotype that distinguished clinical dimensions of SSD was vocabulary. As expected, children with moderate SSD performed more poorly than children with mild or mild to moderate SSD on vocabulary. Children with CAS and those with SSD + LI also performed more poorly than children with SSD alone. Children with all three disorders (SSD + LI + RD) had poorer vocabulary skills than the other three groups. Vocabulary may be related to phonological memory. The learning of new words requires that the child hold the phonological forms for the new word in working memory while a phonological template for the word is created and stored (Baddeley, Gathercole, & Papagno, 1998). Poor phonological memory and vocabulary may result in poor language abilities that impact reading and spelling skills.

The two other endophenotypes, oral motor skills and speeded naming, were not related to severity of SSD, patterns of comorbidity, or clinical subtypes. Speech sound disorder of unknown origin has historically been viewed as a problem in the motor execution of articulatory movements (functional articulation disorders). General deficits in motor performance may be an indicator of an underlying neurodevelopmental immaturity rather than a true motor disorder. This neurodevelopmental immaturity may explain the high rates of comorbidity of SSD, LI, RD, with motor incoordination (Bishop, 2002; Hill, 2001, Visser, 2003). More recently poor oral motor skills have been considered characteristic of CAS or childhood dysarthria with most children with speech delay demonstrating normal motor skills (Shriberg, 2010). It is possible that oral motor skills may not vary systematically with the clinical dimensions examined here, though may be useful in characterizing other dimensions of SSD, such as the age of resolution of speech disorder or the type of speech therapy that is most effective in treating the disorder.

Rapid Naming of Colors (also known as rapid serial naming) draws on articulatory, vocabulary, and cognitive skills and has been related to RD, LI and attention-deficit/ hyperactivity disorder (Peterson et al., 2009). In this study, however, speeded naming was

not associated with differences in severity, patterns of comorbidity, or clinical subtypes. Lahey, Edwards, and Munson (2001) also failed to find a relation of processing speed to the severity of SLI. Similar results are reported by Raitano et al. (2004) in children with SSD and by Young et al. (2002) in adults with histories of SSD. In contrast, Peterson et al. (2009) found that children with SSD with poor speeded naming skills were more likely to have RD. Thus, reports of deficits in speeded naming in children with SSD are mixed, potentially because of the multiple motor and cognitive skills involved in this task.

In summary, the results suggest that clinical dimensions of SSD are related to a constellation of deficits rather than to singular impairments and that comorbidities of SSD are best explained in terms of the joint contribution of multiple endophenotypes to the child's disorder. These findings support the Multiple Deficit Model proposed by Pennington (2006) that hypothesizes a multifactorial etiology for complex developmental disorders such as SSD. In this model, whether or not an individual presents with a disorder and comorbid clinical impairments depends on the risk and protective factors shared among disorders.

### Limitations and future directions

Study limitations include participant and measurement issues. The participants represented an age range (4–7 years old) during which rapid development of speech and language takes place. Significant differences were observed in age between the recovered SSD group and the other SSD groups. Because clinical classifications may change with age, longitudinal follow-up would be useful in determining the extent to which the endophenotypic differences observed in this study vary across development. As children in the no SSD group were typically developing siblings of the participants with SSD, with some sharing of genetic risks for SSD, it may also be useful in future studies to include an unrelated control group. A further limitation is that the study did not include a group of children with LI alone. Such a group might help to disentangle the relationships of the endophenotypes to SSD versus LI.

Other limitations of the study are related to measurement issues. Comorbid conditions of LI, RD, and CAS were determined by parent report. Future studies may directly test for these conditions to verify the accuracy of the parent's report. Some participants had missing data because they were either too young for the test battery or unable to do the test, as in the case of the phonological awareness measures.

## Conclusions

Despite these limitations, this first study of the association of endophenotypes and subtypes of SSD provides insight as to individual differences in children with SSD, suggests approaches to refine assessment of these disorders, and helps to guide the development of interventions that target core areas of deficiency. Furthermore, common underlying endophenotypes for SSD, LI, and RD may have common underlying genetic influences. Identifying these genes and their role in neurological development will help us better understand the etiological bases of these disorders.

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**Table 1**

**Correlation Matrix for Factor Scores of Endophenotypes**

| | Oral motor skills | Speeded naming | Phonological awareness | Phonological memory | Vocabulary |
|---|---|---|---|---|---|
| Oral motor skills | 1.00 | .116 | −.077 | −.097 | −.142 |
| Speeded naming | | 1.000 | −.114 | −.188 * | −.163 * |
| Phonological awareness | | | 1.000 | .489 ** | .353 ** |
| Phonological memory | | | | 1.000 | .440 ** |
| Vocabulary | | | | | 1.000 |

* $p < .05$;

** $p < .001$

**Table 2**

**Comparison of SSD Severity Groups on Demographics and Speech, Language, and PIQ Measures**

| | No SSD | Recovered SSD | Mild SSD | Mild-Moderate SSD | Moderate SSD | Total | Eta$^2$ | F | *p* value |
|---|---|---|---|---|---|---|---|---|---|
| *n* | 45 | 54 | 23 | 16 | 75 | 237 | | | |
| Male/female | 22/23 | 38/16 | 15/8 | 8/8 | 54/21 | 146/91 | | 15.19 | 0.0096 |
| Age mean years[efg] *(SD)* | 5.54(1.76) | 6.31 (0.97) | 5.35(1.31) | 5.13(1.61) | 4.86(1.36) | 5.52 (1.52) | 0.15 | 8.85 | <.0001 |
| GFTA percentile[abcdfghij] | 80.17(20.60) | 59.25 (26.74) | 48.43 (25.01) | 26.87 (20.78) | 10.61 (12.34) | 45.80(34.19) | 0.64 | 89.28 | <.0001 |
| PCC[bcdefgij] | 94.53 (5.63) | 94.37(3.27) | 88.76 (4.69) | 83.08 (6.23) | 71.76(8.34) | 85.81(11.55) | 0.73 | 133.41 | <.0001 |
| PCC-R[bcdefgij] | 97.08 (4.24) | 97.40(1.81) | 91.96(2.51) | 89.34 (3.32) | 75.95 (8.43) | 89.31 (10.60) | 0.75 | 147.72 | <.0001 |
| PIQ standard score | 104.25(15.34) | 102.63 (14.40) | 100.60(17.19) | 105.25(15.73) | 98.23 (12.46) | 102.40 (14.47) | 0.03 | 1.00 | 0.4076 |
| Language standard Score[dgi] | 107.22 (14.86) | 101.26(14.99) | 105.18(14.16) | 100.33 (15.64) | 86.98 (16.47) | 98.42 (17.39) | 0.22 | 10.62 | <.0001 |

*Note.* GFTA = Goldman-Fristoe Test of Articulation (Goldman & Fristoe, 1986); PCC-R = Percent Consonant Correct-Revised; PIQ = performance IQ intelligence quotient; SSD = speech sound disorder.

[a] No SSD differs from recovered SSD.

[b] No SSD differs from mild SSD.

[c] No SSD differs from mild-moderate SSD.

[d] No SSD differs from moderate SSD.

[e] Recovered SSD differs from mild SSD.

[f] Recovered SSD differs from mild-moderate SSD.

[g] Recovered SSD differs from moderate SSD.

[h] Mild SSD differs from mild-moderate SSD.

[i] Mild SSD differs from moderate SSD.

[j] Mild-moderate SSD differs from moderate SSD.

**Table 3**

**Comparison of SSD Severity Groups' Performance on Endophenotype Factor Scores Mean (*SD*)**

| | No SSD | Recovered SSD | Mild SSD | Mild-moderate SSD | Moderate SSD | Eta$^2$ | *F* | *p* |
|---|---|---|---|---|---|---|---|---|
| Oral motor skills | *n* = 38 | *n* = 47 | *n* = 17 | *n* = 12 | *n* = 53 | 0.03 | 1.17 | .325 |
| *N* = 167 | −1.29 (9.9) | −0.99 (6.8) | 0.60 (4.9) | −2.62 (8.16) | 1.34 (7.5) | | | |
| Speeded naming | *n* = 35 | *n* = 45 | *n* = 17 | *n* = 11 | *n* = 50 | 0.00 | 0.12 | .975 |
| *N* = 158 | −2.31 (31.7) | 1.36 (27.3) | 1.04 (36.8) | −2.90 (31.7) | 1.93 (38.9) | | | |
| Phonological awareness | *n* = 42 | *n* = 49 | *n* = 16 | *n* = 14 | *n* = 52 | 0.05 | 2.10 | .084 |
| *N* = 173 | 0.35 (1.7) | 0.35 (1.8) | 0.02 (1.6) | −0.57 (2.7) | −0.58 (2.2) | | | |
| Phonological memory[acdgi] | *n* = 44 | *n* = 54 | *n* = 23 | *n* = 16 | *n* = 73 | 0.19 | 12.05 | <.001 |
| *N* = 210 | 3.00 (4.3) | 0.16 (5.0) | 0.58 (3.4) | −1.87 (5.0) | −2.65 (4.0) | | | |
| Vocabulary[dg] | *n* = 45 | *n* = 54 | *n* = 23 | *n* = 16 | *n* = 75 | 0.08 | 4.55 | .002 |
| *N* = 213 | 1.40 (5.3) | 0.49 (5.1) | 0.85 (4.7) | −0.99 (5.8) | −2.37 (5.7) | | | |

*Note*. Cells contain number of observations and mean (standard deviation) for each factor score. SSD = speech sound disorders.

[a] No SSD differs from recovered SSD.

[b] No SSD differs from mild SSD.

[c] No SSD differ from mild-moderate SSD.

[d] No SSD differ from moderate SSD.

[e] Recovered SSD differ from mild SSD.

[f] Recovered SSD differ from mild–moderate SSD.

[g] Recovered SSD differ from moderate SSD.

[h] Mild SSD differ from mild–moderate SSD.

[i] Mild SSD differ from moderate SSD.

[j] Mild-moderate SSD differ from moderate SSD.

**Table 4**

**Percentage of Individuals by SSD Severity Group who Reported a Comorbid Disorder**

| | Severity group | | | | |
|---|---|---|---|---|---|
| **Comorbid disorder** | **Controls count (%)** | **Recovered SSD count (%)** | **Mild SSD count (%)** | **Mild–moderate SSD count (%)** | **Moderate SSD count (%)** |
| LI *n* = 100 | 6 (6.0) | 21 (21.0) | 9 (9.0) | 9 (9.0) | 55 (55.0) |
| RD *n* = 53 | 6 (11.3) | 16 (30.2) | 5 (9.4) | 6 (11.3) | 20 (37.7) |
| CAS *n* = 42 | 0 (0.0) | 2 (4.8) | 0 (0.0) | 2 (4.8) | 38 (90.5) |

*Note.* CAS = childhood apraxia of speech; LI = language impairment; RD = reading disorder; SSD = speech sound disorder.

**Table 5**

**Comparison of Groups Based on Comorbid Conditions of LI and RD and Performance on Endophenotype Factor Score Mean (*SD*)**

| | Comorbid conditions | | | | | |
|---|---|---|---|---|---|---|
| | **SSD alone No RD *N* = 67** | **SSD + LI No RD *N* = 54** | **SSD + RD *N* = 7** | **SSD + LI + RD *N* = 40** | ***F*** | ***p*** |
| Oral motor skills | *n* = 53 −.11 (5.7) | *n* = 33 −.14 (8.8) | *n*=6 0.68 (3.5) | *n* = 37 0.26 (7.5) | 0.04 | .988 |
| Speeded naming | *n* = 53 −0.22 (31.1) | *n* = 31 −2.61 (31.6) | *n*=5 0.28 (20.9) | *n* = 34 6.91 (40.8) | 0.48 | .695 |
| Phonological awareness[ab] | *n* = 54 0.66 (1.4) | *n* = 37 −0.20 (1.6) | *n*=6 1.88 (3.3) | *n* = 34 −1.77 (2.2) | 15.19 | <.0001 |
| Phonological memory[ac] | *n* = 67 1.23 (4.0) | *n* = 52 −2.49 (3.9) | *n*=7 1.56 (2.8) | *n* = 40 −4.13 (4.7) | 17.53 | <.0001 |
| Vocabulary[d] | *n* = 67 2.14 (4.9) | *n* = 54 −2.90 (5.0) | *n*=7 −1.58 (4.3) | *n* = 40 −3.08 (5.1) | 13.89 | <.0001 |

*Note.* LI = language impairment; RD = reading disorder; SSD = speech sound disorder.

[a]SSD + LI + RD differs from SSD alone, SSD + LI, and SSD +RD.

[b]SSD + LI differs from SSD +RD.

[c]SSD + LI differs from SSD alone.

[d]SSD + LI + RD differs from SSD alone.

Cells contain number of observations and mean (standard deviation) for the factor score. In Table 5, we distinguish subtypes of SSD alone and SSD + LI as to whether or not they have comorbid RD.

**Table 6**

**Comparison of Clinical Subtypes of SSD and Their Performance on Endophenotype Factor Score Mean (*SD*)**

| | Clinical subtypes | | | | | |
|---|---|---|---|---|---|---|
| **Endophenotype measures** | **No SSD *N* = 69** | **SSD alone *N* = 73** | **SSD + LI *N* = 53** | **CAS *N* = 42** | ***F*** | ***p*** |
| Oral motor skills *N* = 187 | *n* = 58 −1.05 (8.6) | *n* = 58 0.23 (5.2) | *n* = 42 −1.12 (7.36) | *n* = 29 1.26 (9.3) | 0.864 | .461 |
| Speeded naming *N* = 175 | *n* = 52 −2.57 (27.9) | *n* = 57 −0.23 (30.5) | *n* = 39 −2.66 (32.2) | *n* = 27 9.64 (41.4) | 1.015 | .387 |
| Phonological awareness[ab] *N* = 192 | *n* = 61 0.25 (2.0) | *n* = 59 0.80 (1.7) | *n* = 45 −0.58 (2.0) | *n* = 27 −1.56 (−.02) | 11.34 | <.0001 |
| Phonological memory[cd] *N* = 234 | *n* = 68 2.93 (4.2) | *n* = 73 1.29 (3.9) | *n* = 52 −2.18 (4.7) | *n* = 41 −4.44 (3.23) | 35.44 | <.0001 |
| Vocabulary[c] *N* = 237 | *n* = 69 1.46 (4.7) | *n* = 73 1.83 (5.0) | *n* = 53 −2.05 (5.2) | *n* = 42 −4.11 (4.5) | 18.34 | <.0001 |

*Note.* CAS = childhood apraxia of speech; LI = language impairment; SSD = speech sound disorders.

[a]CAS differs from no SSD and SSD alone.

[b]SSD + LI differs from SSD alone.

[c]CAS and SSD + LI differs from no SSD and SSD alone.

[d]CAS differs from SSD + LI.

Cells contain number of observations and mean (standard deviation) for the factor score. Table 6 represents clinical subtypes that we have previously reported. In this table we distinguish children with CAS from children with SSD alone or SSD + LI. All children with CAS except for one, have comorbid LI.