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FAMILY HISTORY, CLINICAL MARKER AND READING SKILLS  
IN CHILDREN WITH SPECIFIC LANGUAGE IMPAIRMENT

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**FAMILY HISTORY, CLINICAL MARKER  
AND READING SKILLS IN CHILDREN  
WITH SPECIFIC LANGUAGE IMPAIRMENT**

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*To Thomas, Esther and Doris*

## **ABSTRACT**

This thesis comprises three studies focusing on Swedish school-age children with Specific Language Impairment (SLI). Sixty-one participants with SLI aged 8-12 years were recruited from all existing school language units for children with severe SLI in Stockholm, Sweden. This thesis presents the first Swedish study of family history in SLI and the first empirical study of a clinical marker for Swedish SLI, and contributes to the sparse amount of research studies focusing on reading skills in Swedish children with SLI.

In Study I, the family history of a broad phenotype in SLI was investigated, based on telephone interviews with the parents of the participants with SLI and of 100 matched controls. Significantly higher prevalence rates of language, literacy and social communication problems were found in three generations of SLI relatives as compared to relatives of the controls. In Study II, based on assessments of the participants with SLI and 86 controls, and family history interviews; non-word repetition was reported as a clinical marker for Swedish SLI and was reported to be associated with a family history of language and/or literacy problems. In Study III, reading skills in the participants with SLI were investigated and found to be related to nonword repetition and to a family history of literacy problems in the parents.

The results, based on a large clinical sample with SLI, stress the importance of a family-focused approach to child language pathology. Knowledge about familial aggregation should be considered when making predictions about outcome, prevention and intervention.

## LIST OF SCIENTIFIC PAPERS

The thesis is based on the following papers. They are referred to in the text by their Roman numeral. Reprints were made with permission from the publishers.

- I. **Kalnak, N.**, Peyrard-Janvid, M., Sahlén B. & Forssberg, H. (2012) Family history interview of a broad phenotype in specific language impairment and matched controls. *Genes, Brain and Behavior* 11: 921-927.
- II. **Kalnak, N.**, Peyrard-Janvid, M., Forssberg, H. & Sahlén, B. (2014) Nonword repetition – a clinical marker for specific language impairment associated with parents' language-related problems. *PLoS One*, 9(2), e89544.
- III. **Kalnak, N.**, Peyrard-Janvid, M., Forssberg, H. & Sahlén, B. Reading skills and family history of literacy problems in children with specific language impairment (*submitted*).

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## LIST OF ABBREVIATIONS

CMC	Clinical Marker Controls
FHC	Family History Controls
ICD	International Classification of Diseases
ICF	International Classification of Functioning, Disability and Health
LLP	Language and/or Literacy Problems
LRDPs	Language-Related Diagnoses and Problems
NWR	Nonword Repetition
RI	Reading Impairment
SD	Standard Deviation
SES	Socio-economic Status
SIPS	Sound Information Processing System
SLI	Specific Language Impairment
SLP	Speech Language Pathology/Pathologist
STM	Short Term Memory
SVR	Simple View of Reading



# 1 INTRODUCTION

Specific Language Impairment (SLI) is one of the most common developmental disorders. It is characterized by difficulties in speaking, comprehending and communicating, without obvious causes such as general developmental delay, neurological disorders, sensory disorders (hearing or vision), or social deprivation. SLI does not exclude every other kind of low performance in development (Li & Bartlett, 2012), but problems related to language are the most salient of the developmental challenges these children may face (Bishop, 1997; Leonard, 2000). This thesis is focused on Swedish children attending school language units for children with severe SLI. In the thesis the term SLI is used, which is in agreement with the majority of the scientific literature (Bishop, 2014), but with an awareness that the debate about the concept and label of SLI is ongoing (Ebbles, 2014). SLI is presented more thoroughly in chapter 2.

Children do not refer themselves to speech-language pathologists (SLP) or to anyone else in the healthcare system handling developmental difficulties. Usually it is the parents of the child who seek advice and the child is referred to an SLP by other health professionals (Salameh, Nettelbladt, Håkansson, & Gullberg, 2002). According to my clinical experience as an SLP, parents of children with SLI are not primarily concerned about their child's difficulties in, for example, producing a perfect /s/ sound, or with not having an age-adequate grammar. The most common concern expressed by the parents pertains to their child's current problems, or risk of

future problems, with social interaction due to speech, language and communication difficulties. Moreover, parents of children with SLI often report similar language-related problems in themselves or in their close relatives (Stromswold, 1998), and the more severe the child's SLI, the higher the probability of his or her relatives' having SLI (Conti-Ramsden, Falcaro, Simkin, & Pickles, 2007). Furthermore, in clinical populations, children with more severe language difficulties more often have parents who report distress regarding the language development of their child (Salameh, Nettelbladt, & Gullberg, 2002). The point is that a child with SLI should be treated as an individual, but also as an important part of his or her family history.

Language skills, such as speaking, comprehending, communicating, reading and writing, are highly valued and important prerequisites of participation in a modern society. Poor language and literacy skills are known to be associated with negative effects on academic achievement (Conti-Ramsden, Durkin, Simkin, & Knox, 2009). SLI has been associated with negative effects on social interaction, friendships and quality of life (Conti-Ramsden, Mok, Pickles, & Durkin, 2013; Durkin & Conti-Ramsden, 2007; Fujiki, Brinton, Isaacson, & Summers, 2001); with behavioural problems (Snowling, Bishop, Stothard, Chipchase, & Kaplan, 2006); with poor school achievement (Durkin, Conti-Ramsden, & Simkin, 2012); and with an increased risk of psychiatric disorders (Clegg, Hollis, Mawhood, & Rutter, 2005). SLI is therefore not only of concern for parents and children, but also for society in general.

## 1.1 AIMS

The overall aim of this thesis was to contribute a family-focused approach and to further expand our knowledge regarding the linguistic and cognitive skills of Swedish school-age children with SLI. This thesis aims to contribute to the improvement of clinical diagnoses and prognoses of SLI, which is an important prerequisite for the development of theoretically well-rooted models of prevention and intervention.

The aim in **Study I** was to explore a broader phenotype of family aggregation in SLI by investigating the prevalence rates of language-related diagnoses and problems (LRDPs) in three generations of relatives (grandparents, parents and siblings) of 61 Swedish children with SLI and of 100 typically developing matched controls.

In **Study II**, we aimed to investigate whether nonword repetition (NWR) can serve as a potential clinical marker for Swedish SLI, and whether NWR performance in children with SLI was associated with family history of LRDPs.

The aims of **Study III** were to investigate reading skills in children with SLI attending school language units and to investigate if word/nonword decoding skills and NWR in children were associated with a family history of literacy problems. In addition, we wanted to investigate the link between decoding skills, oral language comprehension and reading comprehension in children with SLI.

## **2 SPECIFIC LANGUAGE IMPAIRMENT**

### **2.1 WHAT IS SLI?**

SLI is a research term for a developmental condition in which children do not acquire language as expected as compared to same-age peers, despite having normal general cognitive abilities, sensory abilities and social opportunities for language development (Leonard, 2014b). This is usually described as an absence of any obvious or known causes of SLI, so that the language impairment is thus ‘unexplained’ (Bishop, 2014).

SLI tends to be persistent (Leonard, 2014b, p. 24; Tomblin & Nippold, 2014, p. 96). Follow-up studies of children identified as having SLI in pre-school or early school years have shown that the language problems in these children tend to persist into school-age and adolescence (Conti-Ramsden, St Clair, Pickles, & Durkin, 2012; Hayiou-Thomas, Harlaar, Dale, & Plomin, 2010). Children with receptive language difficulties are especially likely to have long-term problems as compared to children with speech or expressive language difficulties (Law, Tomblin, & Zhang, 2008).

The nature of the disorder changes over time (Leonard, 2014b; Paul & Norbury, 2012) and varies with the context (Perkins, 2007). This means that the language difficulties found in, for example, a 4-year-old with SLI seldom correspond to the difficulties in the same child at the age of 8 or 12. Context-dependency means that language processing in a child with SLI is influenced by a range of factors in inter-individual interaction, such as the partner, the environment



and the situation (Conti-Ramsden, Hutcheson, & Grove, 1995; Perkins, 2007) as well as in intra-individual cognitive, linguistic and sensory systems (Perkins, 2007). This dynamic picture of language processing is described in the language theories of emergentism (e.g., Perkins, 2007), which stress the complex interaction between nature and nurture and propose that neither is sufficient on its own to support language development.

Traditionally, language impairment has been defined at the lower tail of the normal distribution; that is, the individuals with the poorest performance on language tests. This is sometimes referred to as the naturalist perspective, because it defines an impairment based on deviation from the norm mean performance (Tomblin, 2008). According to the ICD-10 (the International Classification of Diseases and related health problems) the diagnosis of developmental language impairments requires a cut-off at -2 SD on language measures (WHO, 2001). In research, the most commonly applied cut-off values for the classification of SLI usually vary between -1.0 and -1.5 SD (Bishop, 2014).

Another approach to the identification of SLI is based on normative values and considers the negative impact of the child's language difficulties on areas such as school achievement and social interaction (Tomblin, 2008). This normative perspective on functional impairment has been emphasised in the latest version of the Diagnostic and Statistical Manual of Mental Disorders, the DSM-V ([www.dsm5.org](http://www.dsm5.org)), as well as in the upcoming International Classification of Diseases, ICD-11 (Christine Dollaghan, personal communication). In clinical settings psychosocial and educational impacts of SLI are usually assessed by observation and anamnesis. In research they can be described on, for example, the basis of the

biopsychosocial framework of the International Classification of Functioning, Disability and Health, the ICF ([www.who.int/classifications/icf/en](http://www.who.int/classifications/icf/en)).

In Swedish clinical practice, the ICD-10 ([www.socialstyrelsen.se](http://www.socialstyrelsen.se)) is used for diagnosing child language impairment in combination with anamnesis and observation. The diagnoses used are categorical constructs that describe symptoms pertaining to different aspects of language. In addition to the above-mentioned ICD-10 requirement regarding the language measures, non-verbal IQ must be above -2 SD for the child's age and show a discrepancy of at least 1 SD from the language measures. The ICD-10 does not specify the kinds of language measures the diagnostics should be based on, except to say that the measures should be standardised. This is troublesome, as the psychometrics of Swedish language tests vary (Miniscalco, 2003), just as English language tests do (Spaulding, Plante, & Farinella, 2006). It should be noted that a looser clinical definition (Bishop, 2014; Leonard, 2014a) that allow non-verbal IQ to be broadly within norm average is also in use. The current, but ageing, criteria for SLI, which have been highly criticised, are to be reworked and updated (Ebbles, 2014; Reilly, 2014a). Importantly, since there is a pattern of deficits change, and since strengths and weaknesses emerge over time in children with SLI (Leonard, 2014b; Paul & Norbury, 2012), it is crucial to stress that the clinical descriptive diagnoses in child language pathology should be regarded as a current linguistic and communicative profile of the child.

### *Prevalence*

The estimated prevalence of SLI varies between 3–7% (Tomblin, Records, et al., 1997; Weindrich, Jennen-Steinmetz, Laucht, Esser, & Schmidt, 2000) depending on the diagnostic criteria – for example, a narrow or broad phenotype, the method of data collection, the age of the subjects and the cut-offs used. In a systematic review the prevalence of SLI was reported to be about 6% in children up to the age of 16 (Law, Boyle, Harris, Harkness, & Nye, 2000). The prevalence of severe SLI has been found to be 1–2% (Bishop, 1997; Westerlund, 1994; Westerlund & Sundelin, 2000).

### *Degree of severity*

The definition of the severity of SLI is often based on the child's performance in terms of language measures, and not on the impact of the SLI on the child's social interaction and school attainment. In general, -1.5 SD to -2 SD is used as cut-off for moderate to severe performance on language measures and below -2 SD as severe performance (see, e.g., Law, McBean, & Rush, 2011; Locke, Ginsborg, & Peers, 2002). Moreover, severity has also been defined on the basis of a holistic and qualitative clinical picture of the child's language profile (Salameh, Nettelbladt, & Gullberg, 2002). Furthermore, as described above psychosocial aspects should also be included in the definition of severity. Children in need of special education because of persistent and pervasive language problems are often regarded as having a more severe SLI than are those who can manage in the ordinary school system - these children are focused on in the present thesis.

## *Gender*

Population-based studies have found either small or no statistically significant differences in the proportion of males with SLI versus that of females with SLI (Law et al., 2000; Tomblin, Records, et al., 1997). Clinical studies, however, have found higher rates of males with SLI, with proportions ranging from 2:1 to 3:1 (Bishop, 1997, p.38). Several Swedish clinical studies have reported similar findings showing that more boys than girls have been identified as having language impairment (Miniscalco, 2003; Salameh, 2003; Westerlund, 1994). Boys have been found to be more often referred to SLP services (Zhang & Tomblin, 2000).

One possible explanation for this gender discrepancy could involve social norms – for example our expectations of communicative behaviour in girls as opposed to boys. Another possible explanation could be that boys more often present a co-morbid picture including behavioural problems, which increases the likelihood of clinical referral (Zhang & Tomblin, 2000). A third possible explanation comes from reading research. Typically, reading skills do not differ between the genders in terms of mean performance. However, there is a significantly greater variance in the reading skills of males than in those of females, with longer tails at the ends of the normal distribution in males and a gathering around the mean in females (Hawke, Olson, Willcut, Wadsworth, & DeFries, 2009). Just as in reading research, this may explain the lack of significant gender differences in prevalence rates in population-based SLI studies, and possibly also the higher ratio of males with SLI in clinical samples.

### *Theoretical accounts of SLI*

There are two major theoretical accounts of the underlying processes of SLI: a linguistic competence-based account and a cognitive processing account. Models grounded in linguistic nativist theories propose deficits in specific linguistic domains, such as, for example, the rules of grammatical morphology (Rice, 2003; Rice, Wexler, & Cleave, 1995). Models in the domain of cognitive processing theories propose either deficits in specific cognitive processing capacity, as for example with phonological short term memory (STM) (Gathercole & Baddeley, 1990) or in general cognitive processing, as for example with procedural learning (Hedenius et al., 2011; Ullman & Pierpont, 2005).

### *Nature and nurture*

Based on the findings of twin studies, SLI has been suggested to be highly heritable (Bishop, North, & Donlan, 1995), especially regarding clinically identified SLI (Bishop & Hayiou-Thomas, 2008). Moreover, heritability has been found to increase with the severity of the language impairment, suggesting a stronger genetic influence in the extremely low language performers (Viding et al., 2004).

Molecular genetic studies have reported four genetic loci (locations) for SLI in the genome (Newbury, Monaco, & Paracchini, 2014): on chromosome 7q35-q36 (*SLI4*, MIM 612514) (Villanueva et al., 2011), chromosome 13q21 (*SLI3*, MIM 607134) (Bartlett et al., 2002), chromosomes 16 (*SLI1*, MIM 606711) and chromosome 19 (*SLI2*, MIM 606712) (Consortium, 2004; Falcaro et al., 2008). Two

candidate genes have been proposed: *CMIP* and *ATP2C2*, both at the *SLI1* locus (Newbury & Monaco, 2010).

Based on the current knowledge, some of the genes associated with SLI seem to be non-specific to SLI, because they have also been associated with other disorders, for example, the *CNTNAP2* gene which is also associated with autism (Vernes et al., 2008), dyslexia (Newbury & Monaco, 2010) and typical language development (Whitehouse, Bishop, Ang, Pennell, & Fisher, 2011). This has been suggested to reflect the high co-occurrence or misdiagnosis of neurodevelopmental conditions, and moreover, the shared underlying biological mechanisms of neurological development and neurodevelopmental conditions (Zhu, Need, Petrovski, & Goldstein, 2014). To conclude, the aetiology of SLI is presently regarded as multifactorial, with several genes of small effect interacting with various environmental factors (Newbury & Monaco, 2010; Plomin, Haworth, & Davis, 2009).

One environmental factor that has been associated with SLI is socio-economic status (SES). SES is usually measured by family income and/or parental level of education. SLI has been reported to be represented in families of all SES levels (Plante, Shenkman, & Clark, 1996), mainly in low SES families (Tomblin, Smith, & Zhang, 1997) and in high SES families (Bishop & McDonald, 2009; Keegstra, Knijff, Post, & Goorhuis-Brouwer, 2007). These results may focus on differences regarding sample characteristics – for example, population versus clinical samples – and reflect the healthcare systems and cultures in which SLP services are managed in the US, UK and the Netherlands, where the above studies were carried out.

## **2.2 FAMILY HISTORY STUDIES OF SLI**

There has been a lack of studies investigating family history of a broader phenotype of SLI. Moreover, there is no previous family history study of SLI based on a Swedish population; all previous studies are from English-speaking populations. In the following, a survey of methods used in family history studies will first be provided. Thereafter previous findings of family history studies in SLI populations will be described.

### *Methodological approaches in family history studies of SLI*

There are two kinds of approaches to estimating family aggregation of language-related problems: family/self-history reports, and direct testing of the relatives. The first approach comprises two methods: questionnaires and interviews. Questionnaires are the most common method. Only a small number of studies have used interviews as a method of data collection (Beitchman, Hood, & Inglis, 1992; Rice, Haney, & Wexler, 1998). The following language-related problems are commonly explored in questionnaires and interviews: difficulties relating to language development (e.g., having received intervention from an SLP); speech deficits (articulation or stuttering); reading acquisition; and school achievement. The other approach, which involves direct testing of relatives to assess affectedness, is based on test results for language measures. The areas examined usually involve measures of expressive and receptive language, and sometimes also reading skills (Conti-Ramsden, Simkin, & Pickles, 2006).

Questionnaires and interviews may be very similar depending on how they are designed and administered. For example, questionnaires can be handed out for the respondent to fill in, or alternatively can be answered in the presence of a researcher who reads the questions and records the responses (Conti-Ramsden et al., 2006). The latter method of administration does not necessarily differ much from some interview methods. One advantage of questionnaires and interviews is that they can be used not only to identify current problems, as in direct testing, but also to investigate a history of language-related problems.

Family history interviews can use, for example, a response-based approach, in which the wording and order of the questions are highly structured and the concepts unstructured, or an investigator-based approach, where the wording of the questions is semi-structured while the concepts and criteria are highly structured (Conti-Ramsden et al., 2006). In this thesis, the investigator-based approach was used in the family history interviews because it provides opportunities for both parties to ask for clarification, and because both verbal and non-verbal aspects of the interaction during the interview can be captured.

#### *Previous findings in family history studies of SLI*

The prevalence of language-related problems in parents and siblings of children with SLI has been found to be significantly higher than in controls, regardless of the method of data collection - that is, family history report or direct testing (Stromswold, 1998). The rates vary (20–78%) with the type of problems being investigated, the method of data collection and the type of relatives included (Barry,



Yasin, & Bishop, 2007; Conti-Ramsden et al., 2006; Stromswold, 1998). Furthermore, the prevalence of language-related problems in relatives has been found to increase with the severity in the SLI probands (Clark et al., 2007; Conti-Ramsden et al., 2007).

There are contradictory findings as to gender differences in relatives' prevalence rates. In a review by Whitehouse (2010), higher rates of language-related problems were found in male relatives than in female relatives of children with SLI in family aggregation studies based on direct testing. However, this gender discrepancy was not found in studies based on family history reports. The opposite pattern was reported in a study by Conti-Ramsden and colleagues (Conti-Ramsden et al., 2006). This study showed that more older brothers than older sisters were reported to have language difficulties when data was based on parental interviews. Direct testing of the same sample showed no gender discrepancy. Importantly, Conti-Ramsden et al. (2006) also showed that parents are more effective identifiers when the language-related problems in their children are severe.

### **2.3 CLINICAL MARKERS IN SLI**

In the following I will first give a definition of clinical markers, followed by an overview of clinical markers proposed for SLI, in particular NWR.

### *What is a clinical marker?*

A clinical marker is a measurable deficit characterising a particular disorder or condition; that is, it distinguishes between individuals with and without a certain disorder (Poll, Betz, & Miller, 2010). Studies of clinical markers for SLI have focused mainly on NWR, (Graf Estes, Evans, & Else-Quest, 2007) but also on verb morphology (Conti-Ramsden, 2003; Moyle, Karasinski, Weismer, & Gorman, 2011) and sentence recall (Hesketh & Conti-Ramsden, 2013). In Study II in this thesis, NWR has been investigated as a clinical marker for Swedish SLI.

### *What is NWR?*

Nonwords are made-up words that do not exist, but could exist because they are usually constructed on the basis of the phonotactic structure of the language in which they are designed to be used. NWR tests are designed to trigger the STM system and to avoid, as far as possible, the involvement of prior knowledge in the long-term memory. Hence, nonwords should not resemble real words – for example by comprising existing words, pre- or suffixes.

NWR has been found to be insensitive to gender and socio-economic status (Chiat & Roy, 2007; Weismer et al., 2000b), to non-verbal IQ (Bishop, North, & Donlan, 1996; Botting & Conti-Ramsden, 2001; Conti-Ramsden, Botting, & Faragher, 2001) and to cultural or ethnic background (Dollaghan & Campbell, 1998; Weismer et al., 2000b). Moreover, based on twin studies, poor NWR has been found to be heritable (Bishop et al., 1996; Falcato et

al., 2008) and has been reported to be linked to chromosome 16q (Consortium, 2002, 2004; Falcato et al., 2008).

### *NWR a clinical marker for SLI*

Previous studies have pointed out that children with SLI have extraordinary difficulties with NWR as compared to same-aged controls and language-matched controls (Dollaghan & Campbell, 1998; Gathercole & Baddeley, 1990; Kamhi & Catts, 1986; Weismer et al., 2000a). Persistent difficulties with NWR have been reported in children with SLI (Gathercole, 2006) and even in children with recovered SLI (Bishop et al., 1996). In general, longer nonwords (more syllables) are more difficult to repeat. The difference between NWR performance in children with SLI and typically developing children increases with the length of the nonwords; that is, the gap widens (Bishop et al., 1996; Dollaghan & Campbell, 1998; Gathercole & Baddeley, 1990).

NWR was first proposed as clinical marker for SLI in English-speaking populations with SLI (Bishop et al., 1996; Gathercole & Baddeley, 1990; Kamhi & Catts, 1986; Kamhi, Catts, Mauer, Apel, & Gentry, 1988) and has during the last 10 years been replicated in the following languages: Spanish (Girbau & Schwartz, 2007), Dutch (De Bree, Rispens, & Gerrits, 2007), French (Thordardottir et al., 2011), Italian (Bortolini et al., 2006; Dispaldro, Leonard, & Deevy, 2012) and Slovak (Kapalková, Polišíenská, & Vicensová, 2013); though not for Cantonese-speaking children with SLI (Stokes, Wong, Fletcher, & Leonard, 2006). Most of these studies of non-English speaking children with SLI are based on rather small sample sizes (about 10–20) of participants with SLI.

### *NWR in Swedish children with SLI*

Studies of NWR in Swedish children with SLI have focused on methodological aspects; for example, the construction, scoring and analysis of a Swedish NWR test (Ibertsson, Willstedt-Svensson, Radeborg, & Sahlén, 2008; Radeborg, Barthelom, Sjöberg, & Sahlén, 2006; Reuterskiöld-Wagner, Sahlén, & Nyman, 2005; Sahlén, Reuterskiöld-Wagner, Nettelblatt, & Radeborg, 1999) and on the relationship of NWR to linguistic and cognitive measures in children with SLI aged 4–7 (Hansson, Forsberg, Löfqvist, Mäki-Torkko, & Sahlén, 2004; Hansson, Sahlén, & Mäki-Torkko, 2007; Sahlén et al., 1999). In a new test, developed to be used for language screening of Swedish four-year-olds in the child healthcare centres, NWR has been found to be the best single predictor of language outcome (Lavesson, 2012; Lavesson, Hansson, & Lövden, 2014).

All above-mentioned Swedish studies report poor NWR in children with SLI, which points to NWR being a potential clinical marker. However, they lack information regarding effect sizes, sensitivity and specificity values for NWR. In addition, there has been a lack of Swedish data regarding NWR in typically developing children older than seven years.

### *Theoretical foundations of NWR*

The most influential theory explaining poor NWR is based on a model of working memory (Baddeley, 2012; Baddeley & Hitch, 1974) that states that NWR is a measure of phonological STM, (Gathercole, 2006). Phonological STM, defined as a temporary storage of verbal information, has been proposed to have a central

role in the learning of new words – in other words, that it is key in the development of vocabulary (Gathercole, Willis, Emslie, & Baddeley, 1992). The role of poor phonological STM (as measured with NWR) in vocabulary development has been disputed (Melby-Lervag & Hulme, 2012).

NWR is regarded as a language processing task, rather than a measure of accumulated language knowledge (Campbell, Dollaghan, Needleman, & Janosky, 1997). Yet, there is no consensus regarding what NWR exactly measures (Coady & Evans, 2008). However, most researchers agree that NWR is a complex task that taps into a range of cognitive and linguistic input and output skills that may be affected in children with SLI (Archibald, 2008; Coady & Evans, 2008; Sahlén et al., 1999).

The ability to repeat nonwords can be influenced by any of the skills involved in NWR, for example, hearing, speech perception and discrimination, STM storage capacity, phonological representations and articulation skills. In addition qualities of the nonwords may also influence the performance of NWR, such as, the number of syllables (i.e., nonword length) or phonological complexity, e.g., consonant clusters, (Coady & Evans, 2008; Graf Estes et al., 2007). Moreover, phonological characteristics of the nonwords, such as, phonotactic probability and neighbourhood density, will also influence NWR performance: nonwords with higher phonotactic probability and denser neighbourhoods are more often accurately repeated – see, for example, Leonard (2014b, p. 62 ). This means that bottom-up and top-down cognitive processes may both influence NWR performance.

## 2.4 READING SKILLS IN CHILDREN WITH SLI

While there are mountains of research focusing on reading and reading impairment (RI) in children, there are a comparatively limited number of research studies focusing on reading in Swedish children with SLI. In this section I will provide some background to Study III. Study III is one of the few studies describing reading skills in a Swedish population with SLI and reporting preliminary rates of prevalence of RI. First, I will give a definition of RI. Next, I will describe findings regarding co-occurrence of RI and SLI in terms of common co-occurring diagnoses, common genetic markers and NWR as a common clinical marker for both conditions. Finally, I will describe the Simple View of Reading model (SVR).

### *What is reading impairment?*

In the thesis, the term RI refers to difficulties with word or nonword decoding – commonly referred to as the core feature of dyslexia – and to poor comprehension of written language (Kamhi & Catts, 2012). Most research of RI is focusing on dyslectic reading difficulties. When referring to other studies I will therefore often use the term dyslexia to indicate the kind of reading difficulties the study has focused on.

Similar to the criteria for SLI, RI is often classified based on exclusion of general cognitive delays, neurological disorders, and sensory (usually visual) impairments that could explain the difficulties with reading skills (Kamhi & Catts, 2012). In addition, RI is often classified based on exclusion of inadequate opportunities to participate in formal teaching (Kamhi & Catts, 2012), and

sometimes on poor responsiveness to intervention (Vellutino et al., 1996; Vellutino, Scanlon, Small, & Fanuele, 2006). The prevalence of RI is about 5–10% in general populations (Hulme & Snowling, 2011; Kamhi & Catts, 2012) depending on the definition criteria.

### *Co-occurrence of RI and SLI*

RI is known to highly co-occur with SLI. This has been shown in family history studies (Flax et al., 2003), twin studies (Dethorne & Watkins, 2006a; Hayiou-Thomas et al., 2010), follow-up studies of school-age children with SLI (Conti-Ramsden, Botting, Simkin, & Knox, 2001; Davison & Howlin, 1997; Stothard, Snowling, Bishop, Chipchase, & Kaplan, 1998), and in a Swedish longitudinal prospective study of 18-year-old adolescents with SLI identified in preschool (Naucler & Magnusson, 1998). Co-occurrence of SLI and RI in school-age children is associated with a significant risk of poor academic achievement (Conti-Ramsden et al., 2009).

The relatively high co-occurrence suggests common underlying mechanisms. Indeed, common genetic risk markers for SLI and RI have been reported - for example the *CYP19A1* gene, (Anthoni et al., 2012), the *DCDC2* and the *KLAA0319* genes from the *DYX2* locus (Eicher et al., 2014; Newbury et al., 2011). However, both SLI and RI are genetically regarded as complex disorders; likely to be associated with multiple genes of small effect interacting with environmental factors (Asbury & Plomin, 2013; Plomin et al., 2009). Complex disorders are a challenge in behavioural genetics, because even though both SLI and RI often aggregate in families, they do not show an obvious pattern of inheritance (Newbury et al., 2014).

In addition to common genetic markers, poor NWR has been suggested as a common cognitive endophenotype - a clinical marker - for SLI and RI (Moll, Loff, & Snowling, 2013). Some of the first studies associating poor NWR with both RI and SLI (Kamhi & Catts, 1986; Kamhi et al., 1988) found that children with SLI performed more poorly on NWR than did children with RI. More recent studies have reported that children with SLI and adequate decoding skills perform significantly better on NWR than do children with SLI and poor decoding skills (Bishop, McDonald, Bird, & Hayiou-Thomas, 2009; Catts, Adlof, Hogan, & Weismer, 2005; Ramus, Marshall, Rosen, & van der Lely, 2013). In addition, Moll et al. (2013) found that poor NWR was associated with family risk of literacy problems. This has also been reported by Carroll, Mundy, and Cunningham (2014), who found that poor readers with family risk of literacy problems show poorer NWR performance than do poor readers without family history of literacy problems.

The relation between SLI and RI has been discussed in several previous studies (e.g., Bishop & Snowling, 2004; Catts et al., 2005; Ramus et al., 2013). Some researchers advocate that SLI and dyslexia are on a dimension of severity; in which children with SLI show poorer phonological deficits than children with dyslexia (Kamhi & Catts, 1986). Others have proposed that the two conditions are qualitatively distinct disorders with the same underlying cause, (i.e., phonological deficits), but with additional factors underlying the language difficulties in SLI (Bishop & Snowling, 2004; Snowling, Bishop, & Stothard, 2000). And, finally, a third model suggests that SLI and dyslexia are distinct disorders and that phonological deficits are more associated with dyslexia than with SLI (Catts et al., 2005).



### *Family risk of literacy problems*

Family risk (often also referred to as family history) of literacy problems is defined as having a first-degree relative with RI, usually with a focus on difficulties in word decoding (i.e., dyslexia). Family risk of literacy problems is associated with a higher risk of having RI – around 40%, according to Carroll et al. (2014) – and an increased risk of early speech and language difficulties (Pennington & Lefly, 2001; Snowling, Muter, & Carroll, 2007). Despite the high co-occurrence of RI and SLI, studies investigating the relation between family risk of literacy problems and the reading skills of children with SLI are lacking.

### *The Simple View of Reading (SVR)*

The SVR is a model stating that reading comprehension is composed of two components: word decoding and language comprehension (Gough & Tunmer, 1986). These two components contribute strongly and independently to reading comprehension (Catts, Hogan, & Fey, 2003). The relative contribution of each component to reading comprehension has been found to vary over development in typically developing children, with decoding skills contributing more in earlier school years and language comprehension being a better predictor in later school years (Gough, Hoover, & Peterson, 1996). This link, between reading comprehension, decoding skills and language comprehension has not yet been explored in a sample with SLI.

## **3 THE PARTICIPANTS**

### **3.1 THE PARTICIPANTS WITH SLI**

The children with SLI were recruited from all fourteen school language units for children with SLI in Stockholm County. The general admission requirement for these language units is SLI as the primary or only diagnosis, and a non-verbal IQ of approximately >80. In other words, it excludes autism spectrum disorders and intellectual disability, but allows inclusion of additional diagnoses such as ADHD or dyslexia, as reported by the teachers in the schools. In Stockholm, these children are usually referred to the school language units by clinical SLPs or psychologists. Regarding admission requirements to the schools, please refer to, e.g., the guidelines for the Sollentuna Municipality school language units ([www.sollentuna.se/uweb/Tal-och-sprakcentrum](http://www.sollentuna.se/uweb/Tal-och-sprakcentrum)). The following criteria were used for the participants in the present thesis:

- SLI at the time of admission to school and at the time of recruitment to our study, that is, we did not invite children regarding whom the SLI diagnosis had been questioned by the school or changed after admission to the school
- Non-verbal IQ within or above average range at the time of school admission
- Normal hearing and vision
- Monolingual Swedish-speaking
- Adoption was an exclusion criterion for our study

### *The recruitment process*

We targeted all 8–12-year-old children with SLI, attending any of the Stockholm school language units. The head of each school was contacted for permission to recruit participants. However, sometimes children are accepted to the school units for reasons other than those described above and we therefore had to consult each unit separately in order to identify the children fitting our criteria.

One hundred and seventy-five children fitted the study's age criterion of being 8–12-years old, but 75 of these children did not fit our remaining study criteria. The majority of the excluded children were bilingual (n=63, 84%). The remaining were excluded due to adoption (n=5, 7%), neurological disability (n=3, 4%), or hearing or vision impairment (n=3, 4%). One child was excluded due to adverse social circumstances (1%).

One hundred children with SLI satisfied the study criteria and were then invited by means of a letter to their parents. Bilingual participants were excluded because of the difficulty of controlling for their exposure to Swedish as first or second language, which is important as it could affect the outcomes of the linguistic/cognitive assessments of the child. In addition, bilingual children are necessarily assessed in all their languages (Salameh, 2003), which was not possible and outside the aim of this thesis. Importantly, this means that we excluded more than a third (36%) of the available SLI population.

Sixty-one children with SLI and their families agreed to take part in a family history interview with the parents, undergo linguistic/cognitive assessment of the child with SLI and donate

saliva for DNA extraction from all members of the nuclear family. Written informed consent was obtained from the parents and oral informed consent was obtained from each child with SLI at the time of their participation in the project. The results from the linguistic/cognitive assessment of the participants with SLI were shared over the phone with each child's parent(s). In this thesis, the results from the family history interviews and from the assessments of the children with SLI are reported. DNA samples were successfully collected from 85% complete nuclear SLI families. However, the genetic analyses are part of ongoing work that is not included in the thesis.

#### *Non-responders and attrition*

Of the 100 children invited to participate in the present project, parents of 39 children did not respond to our invitation. We did not ask the non-responding families for their reasons for not responding. However, according to the schools, the main reasons were that some families had recently participated in another intensive research project and were therefore not ready to take part in a new project. The distribution of genders among the non-responders corresponded to the distribution among the children who did participate.

One family originally agreed to participate in the project, but their child with SLI decided to interrupt the assessment and during the interview the parents decided to withdraw their consent to participate in the project.

### *Characteristics of the participants with SLI*

The participants with SLI had been attending the school language units for at least one year, and in most cases longer. They were all accepted to the language units because of SLI, in most cases at the age of 5–6. According to the schools, the children recruited to our study had language impairment as their main developmental problem; this was confirmed by the parents during the interviews. This means that the parents and the school staff (teachers, SLPs and psychologists) of the participants did not suspect any explanations for the language difficulties in these children other than SLI at the time of their participation in our project.

The mean age of the children was 9.3 years, SD 1.2. The proportion of boys to girls was 3:1, with 46 boys and 15 girls participating. All participants with SLI were monolingual Swedish-speaking, which does not exclude some of the parents having been bilingual, with Swedish as their second language. Swedish was therefore spoken at home and at school.

According to our linguistic/cognitive assessment, each of the participants with SLI performed below -1.5 SD or below the 10<sup>th</sup> percentile on several tests of both expressive and receptive language skills, measuring aspects of phonology, grammar, lexicon, narration and pragmatics. The participants had a mean non-verbal IQ of 99.34, (SD 14.4, min-max 70-120), at the time of their participation in our study, as measured with the Raven's Coloured Matrices (Raven, 1986). According to the parents, four of the 61 children with SLI were also diagnosed with ADHD (6.6%), and three with reading impairment or dyslexia (4.9%).

### *Family background*

Among the participants with SLI, 91% had siblings (64% full siblings only, 17% half and full siblings, 10% half siblings only). Only full siblings are included in the family history analyses. Three of the participants with SLI were full siblings, and their family history data was analysed as one.

In the present study, level of parental education was categorised in three groups based on the highest level of education achieved: elementary schooling (corresponding to nine years of school in Sweden); completion of upper secondary school (in Swedish: *gymnasium*); having pursued at least three years of higher education, that is, university studies. These categories are influenced by the categories of educational attainment used by Statistics Sweden (Statistiska Central Byrån, [www.scb.se](http://www.scb.se)). We have information regarding the educational level of all 61 children's biological parents, except for one father. Level of parental education was distributed in the three categories as follows: elementary schooling, 14%; upper secondary school, 48%; higher education, 38%. The corresponding rates for the Swedish population in the ages of 25–64, were 13%, 45% and 40%, respectively ([http://www.scb.se/en\\_/Finding-statistics/Statistics-by-subject-area/Education-and-research/Education-of-the-population/Educational-attainment-of-the-population/Aktuell-Pong/9575/Behallare-for-Press/372838/](http://www.scb.se/en_/Finding-statistics/Statistics-by-subject-area/Education-and-research/Education-of-the-population/Educational-attainment-of-the-population/Aktuell-Pong/9575/Behallare-for-Press/372838/)).

### *The school language units*

The school language units are affiliated with mainstream schools. These schools provide intensive (daily) intervention for children with persistent and often pervasive SLI. The classes are small and there is a higher ratio of staff-to-pupils than in the ordinary Swedish elementary school system. Some of the schools provide classes for all nine elementary school years, but most only provide up to school year 5 or 6.

All of the included schools, except one, provide exclusive education. This means that the children with SLI attend school full time in a language unit. One of the schools offers semi-inclusive education, in which children spend their school day attending a regular mainstream class in the mornings and a separate class specifically for children with SLI in the afternoons. Two children were recruited from the school with a semi-inclusive education.

While the staff at the language units consists mainly of teachers (e.g., primary school teachers, special education teachers and preschool teachers), several of the schools also have either an SLP or a psychologist providing services to the school.

The schools admit pupils once a year, after the summer holiday. The assessments required for admission are usually performed by a team comprising an SLP, a psychologist and a teacher. The language abilities assessed by the SLP are: language comprehension, grammatical production, lexical abilities, phonological output, oral motor skills and social-communication abilities. General cognitive ability is assessed by a psychologist. A teacher will typically observe the child while in kindergarten together with peers.

## **3.2 THE CONTROL GROUPS**

Two control groups comprising typically developing children were included in this thesis: 100 controls in the family history interviews (Studies I, II, III) and 86 controls in the clinical marker study (Study II).

### *Family history controls, FHC (Studies I, II and III)*

One hundred control children were recruited from the Swedish birth register to participate in our family history data collection and to donate saliva for DNA extraction from all members of the nuclear family. In the thesis, only the control data regarding the family history interviews was used – not the medical, social and genetic data. The controls were matched to each of the children with SLI according to five recruitment criteria in order of mandatory priority. The criteria were the following: (1) year of birth, (2) gender, (3) municipality of birth, (4) country of origin of the mother, (5) district of birth within the municipality. Information regarding the typical development of the control children was based on parental reports given during the interviews. This data was collected about a year after the SLI data collection.

### *Clinical marker controls, CMC (Study II)*

A control group consisting of 86 typically developing children was recruited from mainstream schools within a municipality in central Sweden. They were originally recruited for another project (Wass, 2009), in which the last author of Study II was involved. The control



group children were 8–12-years old (mean age 9.4, SD 1.3), distributed as 43 females and 43 males, and monolingual Swedish-speaking. They had no history of developmental problems according to parents' and teachers' reports. Non-verbal IQs were within normal limits (102.4, SD 21.7) as measured using the Block Design Subtest from the WISC-III battery (Wechsler, 1991). This means that different tests were used for assessment of non-verbal IQ in the CMS controls and in the SLI sample. Raven's Coloured Matrices and the Block Design Subtest were used for estimation of non-verbal IQ. Both tests strongly correlate with full-scale IQ, and with each other (Kluever, 1995). Based on these tests, we found that the SLI and TD groups did not differ significantly regarding non-verbal IQ ( $p = .444$ ).

**Table 1.** Overview of participants. Study I and Study II are case-control studies. Study III is a single group study.

Participants	Study	n=	Gender <sup>1</sup>		Age <sup>2</sup>	
			n=	%	Mean (sd)	Min–max
SLI	I–III	61	46:15	75:25	9:3 (1:2)	8–12
FHC	I	100	78:22	78:22	*	*
CMC	II	86	43:43	50:50	9:4 (1:3)	8–12

SLI= Specific Language Impairment; FHC= Family History Controls; CMC= Clinical Marker Controls; <sup>1</sup> Boys:girls; <sup>2</sup>Years:Months \*This control group was matched for year of birth

### **3.3 ETHICAL APPROVALS**

This work has been approved by the Ethics Committee in Stockholm. The relevant reference numbers are as follows: 2008/543-31/3 – the main application; 2008/1052-32 – the approval of collecting saliva samples instead of blood samples; 2010/1746-32 – approval of collecting control data for the family history interviews and DNA-samples; and 2012/1938-32 – approval to use typically developing control data originally collected for another project.

## 4 DATA COLLECTION AND STATISTICS

Two methods were used for data collection. Family history interview was applied in all three studies. Linguistic/cognitive assessments were applied in studies II and III. Descriptions of the methods of data collection are found in Table 2. At the end of this chapter there is a section summarising the statistical methods used in each study of the thesis.

**Table 2.** Overview of study designs, samples and methods used in each study

Study	Design	Sample (n)	Methods
I	Cross sectional study, case-control design	SLI (61) FHC (100)	Family history interviews
II	Cross sectional study, case-control design	SLI (61) CMC (86)	Family history interviews*, linguistic and cognitive assessments
III	Cross sectional study, single group design	SLI (61)	Family history interviews*, linguistic and cognitive assessments

\*Family history data in Studies II and III concern analysis based on data from the SLI sample, but not from the FHC.

## 4.1 COGNITIVE AND LINGUISTIC ASSESSMENTS

### *Procedure*

The children with SLI were individually assessed with a broad linguistic and cognitive battery of tests, but only the tests or tasks included in the studies in the thesis are described here. The assessments were administered by an SLP (the author of the present thesis) during a single session in a quiet room at each participant's school. The assessments lasted a maximum of three hours per participant, including short breaks, and were audio-video recorded for later analysis.

All of the children were assessed in the early or late morning. There were two lists of test orders, and these were counterbalanced based on gender and on odd and even subject numbers. The test orders and the early or late morning test starts were not statistically significantly correlated with any of the test measures included in this thesis and there was no significant difference between schools.

Fidelity control was performed in a random sample of 10% ( $n=6$ ), based on the recordings, regarding whether the instructions and the test orders were followed. Cases underlying the fidelity control were decided by lottery (two in the beginning, two in the middle and two cases at the end of the data collection). The fidelity control showed that the instructions and test orders were followed.

### *Material*

The following test measures were included: NWR, oral language comprehension (receptive grammar), reading comprehension,

decoding of words and nonwords and non-verbal IQ, and an assessment of speech production status. An overview of these measures is provided in Table 3 and descriptions of each measure follow below.

Inter-rater reliability was performed for assessments of speech production and for the test of NWR, and these procedures are described below in the descriptions of each measure. The responses for the test assessing decoding skills were based on online scoring. Unfortunately, the quality of the recordings of this test was imperfect, and inter-rater reliability control was therefore not possible to carry out. The measures of reading comprehension and non-verbal IQ were paper and pen tests, and the oral language comprehension test was a multiple-choice picture-pointing test. These three tests do not require any analysis for scoring. Therefore, reliability controls were not considered to be necessary.

**Table 3.** Overview of the functions and analysis/scoring of the tests/measures used in the studies

<b>Test/Measure</b>	<b>Function</b>	<b>Analysis/Scoring</b>	<b>Study</b>
<b>NWR</b>	Phonological processing	Binary and PCC	II, III
<b>TROG</b>	Receptive grammar	Accuracy	III
<b>SL40/60</b>	Reading comprehension	Accuracy/time	III
<b>TOWRE</b>	Test of Word Reading Efficiency	Accuracy/time	III
<b>RCM</b>	Non-verbal IQ	Accuracy	I, II, III
<b>SPEECH</b>	Speech production status	Categorical	II

NWR= Nonword repetition; PCC= percent correct consonants; TROG= Test of Reception of Grammar; SL40/60= Sentence comprehension; TOWRE= Test of Word Reading Efficiency; RCM= Raven's Coloured Matrices; SPEECH= speech output status.

### *The NWR test*

The NWR test is part of the computer-based test battery Sound Information Processing System, SIPS, (Wass et al., 2008). It consists of 24 nonwords, based on equal numbers of 3- and 4-syllable nonwords. The nonwords are balanced in terms of stress pattern and number of nonwords with a consonant cluster; half of the clusters follow Swedish phonotactic rules and half violate these rules. The responses – that is, the children’s repetitions of the nonwords – were transcribed online. The responses were scored binary, as either correct or incorrect for each of the nonwords (NWR Binary) and as percent correct consonants (NWR PCC) of a maximum of 120 consonants in the 24 nonwords.

Inter-rater reliability of the NWR scoring was analysed in a random sample of 18% (11/61) of the SLI children based on individual scorings by two SLPs (the first and last authors of Study II). The proportion of inter-rater agreement was 100% for NWR Binary and 96.1% for NWR PCC. All NWR responses in the control group were scored individually by a psychologist and an SLP. The inter-rater agreement was 100% for NWR Binary, and for NWR PCC. There were only a couple of items for which scoring differed between raters and consensus on these was reached by discussion. The findings from the reliability control are in line with previous research, for example, based on binary scoring in a sample of typically developing Swedish children aged 4–6, the inter-rater agreement was 96% (Radeborg et al., 2006), and based on phoneme-by-phoneme scoring in a sample of Italian children with SLI and controls aged 4 years, the inter-rater reliability was 96% (Dispaldro et al., 2012).

### *Sentence comprehension*

Sentence comprehension (receptive grammar) was assessed with a Swedish version (Holmberg & Lundälv, 1998) of the Test of Reception of Grammar, TROG (Bishop, 1983). TROG comprises 80 multiple choice items. Each item has four pictures of which only one is correct and the other three represent lexical or grammatical distractors. The items are divided into 20 blocks, each testing a specific grammatical structure with increasing complexity. The test administrator reads one sentence at a time and the child is instructed to indicate the picture that corresponds to the sentence. The scoring was based on accuracy of items and blocks.

### *Decoding of words and nonwords*

A Swedish version (Byrne et al., 2009) of the Test of Word Reading Efficiency (TOWRE), (Torgensen, Wagner, & Rashotte, 1999) was used to assess decoding of words and nonwords. TOWRE comprises one subtest assessing word decoding and one subtest assessing nonword decoding. Each subtest consists of two lists of, in total, 208 words and 126 nonwords. Children are instructed to accurately read out loud as many words/nonwords as possible from one list at a time, during 45 seconds per list. The responses are scored binary, and on-line, as either correct or incorrect for each of the word/nonwords, and summed up for each subtest. The two lists from the two subtests significantly correlated for decoding of words ( $r=.967$ ,  $p<.001$ ) and for decoding of nonwords ( $r=.944$ ,  $p<.001$ ). The two subtests of word and nonword decoding also showed statistically significant high correlation ( $r=.908$ ,  $p<.001$ ).

### *Reading comprehension*

A Swedish version of the SL40/60 (Nielsen, Poulsen & Søgaard, 1989; Swedish version by Hellquist, Magnusson, & Naucler, 1997) was used to assess reading comprehension at the sentence level. The SL40/60 consists of two versions: one for children in school years 1-3 and one for years 3-5. Children are asked to silently read one sentence at a time and select (marking with a pen) one of 5 pictures corresponding to the sentence they have just read. There is a time limit of 15 minutes. The scoring was based on accuracy.

### *Speech production*

Speech production was assessed based on two tasks from the comprehensive linguistic/cognitive assessment: a picture-naming task and a narrative task. Speech production was then defined in three broad categories: (1) no speech production deficits; (2) minor speech production deficits – for example, occurrence of substitutions of /r/, or lispings; and (3) major speech production deficits – for example, substitutions of consonants or vowels, reductions of syllable structure, reduplication of syllables and assimilations.

The reliability of the assessments of speech production in the participants with SLI was independently analysed and categorised in a randomly selected sample of 10% (6/61) of the recordings by an SLP research assistant. The inter-rater agreement was 100%.



### *Non-verbal IQ*

Raven's Coloured Matrices (RCM) was used to assess non-verbal IQ in the children with SLI (Raven, 1986). RCM is a multiple-choice test considered to measure non-verbal visual reasoning and is often used as a measure of fluid intelligence (Bölte, Dziobek, & Poustka, 2009). The RCM consists of 36 items. For each item the children were asked to identify a missing element that completes a pattern. There is no time limit. The scoring was based on accuracy.

## **4.2 FAMILY HISTORY INTERVIEWS**

### *Procedure*

The family history interviews were administered over the phone by one of two SLPs. The interviews were carried out with the parents of 61 children with SLI and the parents of 100 matched controls, and took 15–40 minutes (mean 30 minutes).

First, based on the parents' information about the family structure, a pedigree (family tree) was drawn. The pedigree was used for the coding and documentation of responses, that is, the information regarding all relatives (siblings, parents and grandparents). Then, the pre-phrased questions were asked in the same order in all interviews. The order of the questions was only modified if the parent brought up a topic in advance. Notes were taken orthographically by the interviewer. The interviews were based on the aim of mutual understanding and an online confirmation of the answers. This means, for example, that when the responses were not specific enough, the interviewer asked for clarification. This in turn means

that we used an investigator-based interview approach, that is, semi-structured wording of the questions and highly structured criteria.

### *Material*

The interview questions comprised two parts, each targeting a couple of domains. The first part comprised information about two domains: medical and social/genetic issues chiefly regarding the children with SLI and the control children, while the second part comprised information about the two domains ‘prevalence of language-related diagnosis’ and ‘prevalence of language-related problems’ regarding all individuals in the pedigree.

In this second part of the interview, the questions were constructed based on the findings of follow-up studies (Conti-Ramsden et al., 2009; Snowling et al., 2006) and on co-occurring diagnoses and symptoms in neurodevelopmental disorders. The two domains in the second part of the interview are collectively referred to as ‘language-related diagnoses and problems’ (LRDPs). In the three studies comprising the thesis, the analyses from the family history interviews have mainly concerned the LRDPs, as well as the demographic data covered by the questions regarding social issues. The questions in each domain concerned the following information:

**Medical domain:** Information about birth-weight, gestation age, postnatal status, IVF (*in vitro* fertilization), hearing and vision of the SLI and control children. Information regarding abortions and miscarriages of the mothers of the children with SLI and controls.

**Social/Genetic domain:** Information about highest level of parental education and parents' occupation. Information regarding whether any relative was adopted, sibling relationship and information about consanguinity between the parents and the grandparents of the children with SLI and controls. Information as to age and cause of death of all deceased relatives.

**Language-related diagnoses:** The questions targeted having a diagnosis of reading impairment/dyslexia, autism spectrum disorder, Asperger syndrome, stuttering, intellectual disability, ADHD, cleft palate, or congenital impairment of hearing or vision. These questions are based on previous findings of co-occurrence of language-related diagnoses and problems in neurodevelopmental disorders, on longitudinal studies of SLI and on clinical experience in child language pathology.

**Language-related problems:** This domain covered the history of difficulties, or current difficulties, in the following categories: (1) language, (2) reading, (3) school achievement, (4) attention and/or hyperactivity and (5) social communication problems. This domain was included based on the assumption that it is not relevant to ask only about diagnoses in the older generations since the use of neurodevelopmental diagnoses has changed over time. The five categories of questions were the following:

- Difficulty with language acquisition; late talkers (older than 3 years); having received speech/language therapy; or having a diagnosis of language impairment.
- Reading impairment; difficulties in learning to read and write neither due to inadequate schooling nor to bilingualism; adults needing help in reading and/or understanding official correspondence; or having a negative attitude towards reading. Diagnosis of dyslexia was not included.
- Learning difficulties and poor school achievement; having received special education at school; having to retake a school year due to learning difficulties not related to social factors.
- Problems with attention and/or hyperactivity affecting learning or the social aspects of life; but not being diagnosed with ADHD.
- Difficulties in making friends and/or maintaining friendships and relationships; speech anxiety; selective mutism.

#### *Analysis and scoring of interview data*

Information concerning LRDPs in three generations of relatives was coded as '0' for non-affected and '1' for affected. In order to control for agreement between the two interviewers (SLPs), four families were interviewed in a pilot sample that was not included in the study. The responses were scored and analysed independently. The overall proportion of agreement for the first part of the interviews (regarding the domains of medical, social/genetic issues) was 100%, while for the second part of the interviews (the LRDPs) it was 97%.

### 4.3 STATISTICAL ANALYSES

The results in all three studies were analysed using the IBM SPSS versions 20–22. For all analyses the level of statistical significance was set at  $p$ -value  $<.05$ , based on two-tailed analysis. Cohen's  $d$  was used to estimate the effect sizes in Study II and Study III:  $d \geq 0.5$  was regarded as a medium and  $d \geq 0.7$  as a large effect size. An overview is provided in Table 4.

**Table 4.** Summary of statistical tests used in the individual studies

METHODS/ANALYSES	Study I	Study II	Study III
Descriptive statistics	X	X	X
Spearman's correlation		X	
Pearson's correlation		X	X
Chi-square	X	X	
Cohen's D		X	X
Risk ratio/relative risk	X		X
Odds ratio	X		
One-sample t-test			X
One-Way Analysis of variance (ANOVA)		X	X
Mann-Whitney U-test	X		
Receiver-Operating Characteristic Curve		X	
Likelihood ratios		X	
Bivariate logistic regression		X	
Simple linear regression			X

## 5 SUMMARY OF EACH STUDY

In the following sections each study will be briefly summarised. More detailed descriptions, tables and figures are found in the original articles at the end of the thesis.

### 5.1 STUDY I

#### *Family History Interview of a Broad Phenotype in Specific Language Impairment and Matched Controls*

This is the first study investigating family aggregation of a broad phenotype of SLI and also the first to be based on a non-English-speaking population. Data was collected covering more than 400 and 600 relatives of children with SLI and controls, respectively.

In Study I the prevalence and co-occurrence of language-related diagnoses and problems (LRDPs) were explored in the relatives of 61 children with SLI and of 100 controls, 8–12 years of age, based on family history interviews. The children with SLI were recruited from school language units in Stockholm. The controls were attending regular classes in school.

The parents, usually the mothers, were interviewed by phone by one of two SLPs, following a semi-structured interview method with an investigator based approach. The family pedigrees comprised three generations (grandparents, parents and siblings). The broad phenotype was defined based on previous findings of follow-up studies of children with SLI and on known co-occurring diagnoses

and symptoms in neurodevelopmental disorders. The interview questions regarding the prevalence of LRDPs targeted developmental problems related to language, literacy and learning; speech anxiety; cleft palate; diagnoses of ADHD, Autism Spectrum Disorder, Asperger syndrome, Dyslexia or RI, and stuttering.

We found significantly higher prevalence rates of problems related to language, literacy and social communication in all three generations of relatives of the children with SLI as compared to the relatives of the controls (Table 2 in the article). The relative risk of having at least one parent with one or more LRDPs was 85% for the children with SLI, and 13% in the control children (Table 3 in the article). Problems of language and literacy were the most commonly reported in both the SLI and the control relatives, and these two LRDPs highly co-occurred. No significant gender differences regarding prevalence rates of any of the LRDPs were found in any of the generations of relatives of the children with SLI.

Family history from Swedish-speaking families with SLI is unique. This study fills in the gap of knowledge about the familial history environment of Swedish children diagnosed with SLI. Based on the findings of Study I, documentation of family history is strongly advisable in clinical child language pathology, for example, for early identification of siblings who are at a high risk of having LRDPs. An understanding of the prevalence and co-occurrence of LRDPs is important for intervention planning and also for theories of aetiology and background mechanisms.

## 5.2 STUDY II

### *Nonword Repetition – a Clinical Marker for Specific Language Impairment in Swedish Associated with Parents' Language-Related Problems*

Study II is the first study to empirically investigate a clinical marker for Swedish school-age children with SLI. In this study, NWR was investigated as a potential clinical marker for Swedish SLI, and the relation of NWR to family history and the parents' level of education was explored.

Individual assessment using an NWR test was carried out on 61 children with SLI attending school language units and 86 typically developing matched controls attending regular classes. The participants were 8–12-years old. The NWR test (Wass et al., 2008) consists of 24 three-to-four syllable nonwords. Family history of language and/or literacy problems (LLP) was investigated in the sample with SLI based on family history interviews with the parents.

We found 90.2% sensitivity and 97.7% specificity at a cut-off level of -2 SD for binary scoring of nonwords (Table 3 in the article). Differences between the SLI and TD groups showed large effect sizes for the two scoring measures binary ( $d=2.11$ ) and percent correct consonants ( $d=1.79$ ). Performance on NWR was insensitive to age, gender, non-verbal IQ and to level of parental education in the children with SLI. Family history of LLP in the parents was significantly correlated with lower scores on NWR in the children with SLI (Table 5 in the article). The difference on NWR performance between the children with SLI with and without a family history of LLP was large ( $d=0.7$ ).



This study contributes to the development of diagnostics in Swedish SLI by providing support for NWR as a clinical marker in a Swedish-speaking population with SLI. Furthermore, the finding of an association between poor NWR and family history of LLP in children with SLI contributes to a broader understanding and implementation of a family-oriented clinical approach in child language pathology.

### **5.3 STUDY III**

#### *Reading Skills in Relation to Family History of Literacy Problems in Children with Specific Language Impairment*

This study explores reading skills in children with SLI attending school language units. The associations between family history of literacy problems and the participants' reading skills and NWR are investigated. The SVR model (Gough & Tunmer, 1986) was used to investigate the link between decoding skills, oral language comprehension and reading comprehension.

Sixty-one children with SLI aged 8-12 years, attending school language units in Stockholm, in Sweden, were assessed with measures of NWR, oral language comprehension and two aspects of reading skills – reading comprehension and decoding of words and nonwords. Family history of literacy problems was based on family history interviews with the parents.

We found that 97% of the sample showed poor outcomes ( $\leq 10^{\text{th}}$  percentile) in decoding and/or reading comprehension, and the majority performed poorly on both measures (Table 1 in the

manuscript). This finding is discussed in relation to the fact that only 5% (3 of 61) of the sample was previously clinically diagnosed with reading impairment. Co-occurrence of poor reading comprehension and poor oral sentence comprehension was 90%.

Family history of literacy problems was reported in 64% of the participants and was associated with lower performance on decoding of nonwords, but not with decoding of words. Decoding of words and nonwords were both significantly correlated with NWR. No significant relation was found between NWR and categorically defined poor decoding skills.

Based on a regression analysis, we found that decoding is a better predictor of reading comprehension in the lower (1<sup>st</sup>-2<sup>nd</sup>) as compared to the upper (3<sup>rd</sup>-5<sup>th</sup>) school years. We further found that oral language comprehension was a significant predictor for reading comprehension in the upper school years, though not in the lower years (Table 3 in the manuscript).

The findings have important implications for prevention and intervention, emphasising the necessity of assessment of literacy skills in children involved in school language units. A family approach, such as would involve counselling and support to families, is recommended considering the high prevalence of literacy problems in the parents and the association between a positive family history and poor decoding skills.

## 6 GENERAL DISCUSSION

The current on-going debate as to the label and concept of SLI (Ebbles, 2014) – also regarding the concept of dyslexia, see for example Elliott and Grigorenko (2014) – is a sign of a healthy reaction and a sane critical approach to neurodevelopmental diagnoses and definitions, by several of the most influential professionals in the field (clinicians and/or researchers). The debate reflects a frustration about the lack of coherence in the definitions of SLI. Medical, psychological or psychiatric diagnoses are not set in stone. Historically they have followed new trends, shifts in norms and new understandings of, for example, the development and acquisition of diverse human skills. There is no such thing as a constant understanding of these conditions, but rather a dynamic picture over time and in the context of different cultures (as, e.g., in clinical settings versus research, or in Sweden versus let's just say, among the Inuit in North America (Crago, 1990)). Only about a hundred years ago illiteracy was part of the norm in Sweden, and unfortunately it still is in many countries. However, nowadays illiteracy is regarded as a disability in countries like Sweden, because it is not part of the norm anymore.

One problem with the label SLI is the first word, 'specific', since today it has several connotations. The classical definition of the concept of SLI (Stark & Tallal, 1981) has changed over time due to experience and knowledge added to the field. Alternatives to the label have been suggested, for example, Developmental Language Disorder or Language Learning Impairment (Bishop, 2014); Language Impairment (Reilly, 2014b); and some researchers suggest

that the label 'SLI' should be kept (Rice, 2014); but with the broader and looser definition that is already in use by many researchers and clinicians (Leonard, 2014a). In Swedish clinical practice, the label Language Impairment is used, that is, '*språkstörning*' (Hansson, 2014; Nettelbladt, Samuelsson, Sahlén, & Hansson, 2008). There is currently a call for an international consensus conference and an updated definition of SLI that can provide us with an important basis for how we may refer to this condition (Reilly, 2014a). Meanwhile, to enable generalisability of research findings, we need to provide clear descriptions of the samples we include in studies.

Thus, we have a label and a concept in change. It is within this changing framework the present research has been performed. The findings in this thesis are based on a sample representing monolingual 8–12-year old children attending special school language units for children with SLI in the Stockholm area. They may be described as having a severe or a persistent and pervasive SLI based on their significantly poor performance on several linguistic measures and on their need for special education because of their significant difficulties in language development. Moreover, the proportions of boys and girls (3:1) in our sample are typical for that of clinical samples of children with SLI (Bishop, 1997; Botting & Conti-Ramsden, 2003). However, the results based on these children are probably not generalisable to, for example, younger children with SLI, or children with less severe SLI or to bilingual children with SLI. Today there is a lack of longitudinal studies of Swedish SLI and of studies involving bilingual school-age children with SLI. Hence, longer and broader perspectives of SLI are highly needed.

The inclusion criteria of the participants with SLI applied in this thesis did not comprise a criterion of discrepancy between linguistic skills and nonverbal IQ at the time for participation in the project. According to the schools, the participants with SLI had a non-verbal IQ within or above norm average at the time for admission to the schools. At the time of this thesis project, when the participants with SLI were 8–12 years old, they performed within norm average on group level (99.34, SD 14.4, n=61), which means that on an individual basis some of the participants (n=2, approx. 3%) performed below -1SD. This may be regarded as fewer than expected based on previous follow-up studies reporting performance-drop of non-verbal IQ in a British population of about 200 children attending school language units (Botting, 2005; Conti-Ramsden, Botting, Simkin, et al., 2001). Interestingly, in a subsequent follow-up study of the same sample as just mentioned, the reported performance-drop of non-verbal IQ was found to be restored to normal level in late adolescence: “...*there is little evidence of slowing or acceleration in growth of nonverbal skills in the group as a whole*” (Conti-Ramsden et al., 2012, p. 1725).

To sum up, despite the fact that we did not apply a non-verbal IQ discrepancy criterion, all participants with SLI (except for the two children with non-verbal IQ scores below -1SD) would fit a definition of SLI including a non-verbal IQ discrepancy criterion. Most researchers seem to agree about the dropping of the IQ discrepancy criteria (Reilly, 2014a) since there is no evidence indicating that the nature of language impairment is qualitatively different depending on non-verbal IQ (Dethorne & Watkins, 2006b) or that the level of non-verbal IQ is of importance for

intervention outcome; given that non-verbal IQ is broadly within norm average (Bishop, 2014).

Still, a criterion of nonverbal IQ broadly within norm average has been important in both research and clinical identification of SLI. In Sweden, this is, perhaps, primarily driven by a structural need for differential diagnosing between SLI and intellectual disability. The reason for this is that children with intellectual disabilities have legal rights to attend special schools and follow a special curriculum throughout elementary (9–10 years) and upper secondary (appr. 3 years) school ([www.skolverket.se/skolformer](http://www.skolverket.se/skolformer)). In the school language units for children with SLI the conventional curriculum is followed, and these units seldom provide schooling throughout all the nine elementary school years; but usually only up to the years 3 or 5. Therefore in the admission requirements to language units in Sweden it is important to tease out if the language difficulties of a child could be part of a general low cognitive ability; to make sure that both children with intellectual disability as well as children with SLI are offered the best option of schooling.

## **6.1 FAMILY HISTORY**

The purpose of family history studies in SLI is to provide estimates of familial aggregation, which is, clustering of, e.g., symptoms or diagnosis within a family. This kind of data has both theoretical and clinical relevance. It has potential impacts on early detection of language related diagnoses and problems, prognosis, intervention and general policy information (Conti-Ramsden et al., 2006). It extends our knowledge of, e.g., co-occurring patterns of symptoms and conditions. Family history studies may also underlie analysis of

genetic risk markers; results from interviews or questionnaires may classify relatives on a qualitative basis as affected or non-affected, and for quantitative classification of affectedness a method of direct testing would be required.

When data from several generations of relatives is included in a family history study, we get insights into the interactions of nature and nurture of language related problems from the perspective of the extended family. Importantly, these two factors (nature and nurture) cannot be disentangled in a family history study because both genetically and environmentally driven aspects in life can aggregate in families. It would have taken a twin study design to explore the unique genetic and environmental influence on, for example, language skills.

#### *Family history of several generations*

The results presented in Study I, showed that there is a significant increased risk in school-age children with severe SLI to grow up in families where the parents and siblings have LRDPs. In addition, it was found that that the parents of the children with SLI also showed an increased risk of having grown up in a similar context themselves. The type of LRDPs being reported in an extended family may indicate the prognosis of the child's language difficulties and point out the direction of focus on prevention work. Information from several generations might also give us an idea of language related experiences and activities in the home environment, for example, regarding the attitude to reading of the children and also of the parents.

### *Family history from the child's perspective*

In Study I, it was found that 85% of the children with SLI had parent(s) with one or several LRDPs, and in Study II, 80% were reported to have parent(s) with LLP and in Study III, 64% of the children with SLI had parents with literacy problems. This way of reporting family history data is novel. Usually the proportion with language problems is reported for each relative type as, for example, for mothers and fathers, or in all first degree relatives. In study I, the usual way of reporting family history data is also provided. In addition, the relative risk of a child with SLI to grow up in a family where the parents have LRDPs is reported. This family approach from the child's perspective is highly relevant in family-oriented clinical settings. Importantly, children are dependent on their parents or caregivers, for example, when it comes to pursuing language intervention.

A previous large Swedish cohort study of over 400 children with SLI (Salameh, Nettelbladt, Håkansson, et al., 2002) found that the more severe SLI a child had, the higher risk of being discharged from SLP services due to non-attendance, that is, the families interrupt the intervention. This could be linked to findings showing that heritability increases with the severity of the child's language difficulties (Viding et al., 2004), and also to the findings of high prevalence of family history of LRDPs in children with severe SLI (Study I in this thesis). My point here is that SLP services might be less attractive to parents of children with severe SLI, because many of the parents have LRDPs themselves.

A better understanding of the child's family context may contribute to the improvement of more effective, acceptable - and therefore



attractive - intervention services. As professionals we need to consider family aggregation of LRDPs when planning prevention and intervention services. Moreover, it is crucial to investigate the children's self-perceptions and attitudes to their own strengths and difficulties, and to psychosocial aspects of their life. This kind of information has already been collected in the project of the present thesis, and is to be analysed in upcoming work.

### *SLI siblings – a high risk group*

Siblings to children with SLI present a high risk group of having LRDPs. The SLI siblings had a five times higher prevalence of language problems and a three times higher prevalence of literacy problems, when compared with general prevalence rates (Study I). About 50% of the siblings were reported with LRDPs. Yet, we may expect an underestimation based on that the youngest siblings are too young to show symptoms or to yet have some of the diagnoses or problems we asked about (e.g., reading impairment). In addition, parents are more effective identifiers the more severe language-related problems their children have, that is, they may not report less severe problems (Conti-Ramsden et al., 2006). Clark et al. (2007) reported high prevalence of SLI in siblings of school-age children with severe SLI, and found that the parental reports regarding the siblings underestimated the prevalence of SLI identified by direct testing.

To conclude, siblings of children with SLI are at high risk of having LRDPs and their difficulties are in danger of being unrecognised. It is therefore important to provide support to the families in identifying LRDPs in the siblings. This is necessary knowledge to

add to the Swedish child health care centres, where practically all families in Sweden have their children regularly screened for physical and cognitive development up to the age of 4–5.

### *Challenges of multi-generational family history studies*

Including several generations of relatives in a family history interview, as in Study I, comprises methodological challenges. First of all, since there have been changes over time regarding the view of neurodevelopmental conditions, the formulation of the questions needs careful consideration. Hence, we did not ask only for the prevalence of language related diagnoses, since the older generations (parental and grandparental) were children in the days when, for example, neurodevelopmental disorders were viewed differently and diagnosed less often than today. Apart from describing the prevalence rate of clinical diagnoses, the aim of Study I was also to identify relatives with severe problems that could possibly have led to diagnoses if these individuals were children today. Accordingly, the interview questions regarding ‘language-related problems’ were therefore incorporated into the interviews.

Another challenge to the reliability of the interviews could be that the relatives’ family history was reported by the parents, usually the mothers. It needs to be acknowledged that this aspect of secondary information might be a limitation that primarily risks underestimating the LRDPs in the relatives. By consequence, the reports of the LRDPs in the grandparents’ generation in both the SLI group and the control group (Study I) risk being underestimated. Although one could also suggest reasons for overestimation (e.g., misinformation, exaggeration of problems) the risk

of under-estimation of the family history reports, especially regarding the grandparent and parent generations, is probably more reasonable to expect.

### *History report versus direct testing*

By including three generations, we have tried to capture a lifetime developmental perspective of family aggregation of a broad phenotype in SLI. Direct testing would not have been adequate or possible as a method for this approach. A few studies have compared family aggregation in SLI obtained from the same sample using both interview or self-reports and direct testing methods (Barry et al., 2007; Clark et al., 2007; Conti-Ramsden et al., 2006; Plante et al., 1996; Tallal et al., 2001). All of these studies, with the exception of Clark et al. (2007) and Plante et al. (1996) found no statistical difference between prevalence rates obtained from self-reports and results from direct testing. Plante et al. (1996) found that self-reports underestimated the number of affected parents as compared to direct testing. Clark et al. (2007) reported underestimation of speech, language and communication impairments in siblings of children with severe SLI based on parental reports as compared to direct testing. However, comparison of these two approaches of data collection is complex and may partly be explained by questionnaires targeting both current problems and a history of problems, while direct testing principally targets current speech and language status.

In summary, the findings from the family history interviews in the thesis contribute to the range of previous family history studies. However, this is the first study exploring a broader phenotype of

SLI than what has previously been published, the first family history study of SLI being based on a non-English-speaking population, and it is based on a large clinical SLI sample and controls. The parents of the children with SLI have probably answered or thought about many of the interview questions before, but for the control parents it was probably for the first time. Moreover, the risk of under-estimation, regarding the prevalence rates, is likely to be higher than that of over-estimation in the SLI group and the control group. It is important to remember that the purpose of family history studies is not to certify absolute prevalence rates, but estimates of the diagnoses and problems investigated.

## **6.2 CLINICAL MARKER**

NWR has been of intense interest to SLI research for several decades, as a language processing measure and a clinical marker. NWR is an important measure, because, as previously mentioned, it is neither associated with SES nor with non-verbal IQ. This is important because a clinical marker for SLI should not be a proxy for general cognitive ability or SES. The non-sensitivity of environmental input on NWR is illustrated in a study of communication skills in 138 children from socially deprived areas in Scotland (Law et al., 2011). These children showed overall low language abilities as compared to standardised test norms, but their performance on NWR and non-verbal IQ did not differ from norm average.

One rationale for NWR to be associated with language development could be that repeating nonwords resembles language learning. When we learn new words and grammatical features we need to be

able to hear what people say, correctly perceive the speech sounds, focus our attention on the speech we want to take in, keep new strings of speech sounds in our phonological STM, and reproduce the word. Already, before the age of 6 months, babies typically try to imitate language input they do not yet understand (Kuhl & Meltzoff, 1996). In western cultures we like to ask toddlers to repeat words and most children seem to like to, and be able to, repeat on request before their first birthday (unless they do not feel like it...). When we ask children to participate in a NWR test, the integration of a range of linguistic and cognitive skills is challenged.

*NWR is a clinical marker for Swedish SLI*

Study II provides support for NWR as a clinical marker for SLI in a Swedish-speaking population aged 8–12 years. With the NWR test from the SIPS test battery (Wass et al., 2008), 90.2% of the children with SLI and 97.7% of the typically developing controls were correctly identified. Moreover, the findings showed that performance on NWR was insensitive to gender and non-verbal IQ in the SLI group and in the control group, as to the level of parental education in the sample of children with SLI (there was no data available as for the level of parental education in the controls). These results are replicating previous studies based on different languages reporting sensitivity and specificity values showing that NWR is a clinical marker for SLI (e.g., Bortolini et al., 2006; Dollaghan & Campbell, 1998; Gray, 2003; Kapalková et al., 2013; Thordardottir et al., 2011). Since Study II is the first empirical study of a clinical marker for Swedish SLI, the results would need to be replicated. In addition, it still remains to be explored if the NWR

test used in Study II is equally accurate as a clinical marker in children with less severe SLI. Furthermore, previous studies suggest a combination of NWR with other linguistic measures to reach high accuracy of identification of SLI based on samples of other ages than in the present work. For example, a combination of NWR and receptive language in children aged 4 (Lavesson, 2012); NWR and a measure of past tense in children aged 5 (Conti-Ramsden, 2003); NWR, sentence repetition and a grammatical judgment task in adults (Poll et al., 2010). These are interesting findings and some of the measures are possible to explore in a future study based on available data from the project of the present thesis. From a clinical perspective, if a screening battery is aiming at incorporating a differential diagnostic approach, it is important to combine NWR with other tasks.

#### *Methodological and statistical considerations in clinical marker studies*

Ideally, a clinical marker shows a bimodal distribution, with no overlap between those with and those without the disorder (Rice, 2003). However, in general this will depend on, for example, whether the clinical marker is a normally distributed phenotype or a more distinct feature of the disorder. A clinical marker is an indication of an increased risk of having the disorder or condition in question. This understanding fits the interpretation of clinical markers and genetic markers of SLI and RI.

Sensitivity and specificity are crucial values to investigate in studies evaluating the accuracy of potential clinical markers. Group differences on a test do not necessarily mean acceptable accuracy of a test for a correct classification of a disorder (Fidler, Plante, &

Vance, 2011). Sensitivity tells us the proportion of true positives, that is, the individuals who have the disorder and specificity tells us the proportion of true negatives, that is, the individuals who do not have the disorder. Therefore, the gold standard of the controls included in clinical marker studies is usually typically developing children; identified in terms of attending regular school and not having any history of developmental problems according to parental and/or teacher reports. However, there is no broadly accepted guideline for interpretation of the clinical importance of sensitivity and specificity values (Lalkhen & McCluskey, 2008). When following a suggested threshold of 90% (Plante & Vance, 1994), the results in Study II can be considered as “good” based on only 9.8% false negatives and 2.3% false positives. The importance of empirically derived cut-off levels, in contrast to commonly used ones, such as, e.g., -1 SD or -1.5 SD, is illustrated in Study II by the inspection of less severe cut-off levels. With the less severe cut-off levels (-1.5 SD and -1 SD, instead of -2SD) no change or small gain was found for the sensitivity, but a loss of up to 10.5% in specificity.

#### *The relation between speech output deficits and NWR*

In studies using NWR tests, children’s speech output deficits have been dealt with in different ways depending on the purpose of the study and the targeted age groups. Developmental speech errors are normally not present in Swedish 8–12 year-old children (Lohmander, Lundeborg Hammarström, & Persson, 2014; Nettelblatt & Salameh, 2007) and since the participants in Study II did not make any systematic speech output errors, only correct repetition of the nonwords was accepted. Another option would

have been to exclude children with speech production deficits, an approach that has been applied in previous studies (Bishop et al., 1995; Gathercole, Willis, Baddeley, & Emslie, 1994; Montgomery & Windsor, 2007). However, excluding one third of our sample of children with SLI, because of speech output deficits, would have affected the representativeness of our sampling. The rate of speech output deficits was higher in the sample of children with SLI in the present thesis, as compared to a population-based study of much younger children, aged 6 years (Shriberg, Tomblin, & McSweeney, 1999), in which the co-occurrence of speech output deficits in children with SLI was found to be 5–8%. This is probably explained by clinical samples being known to more often include speech output deficits than do population based samples (Zhang & Tomblin, 2000). Usually this bias of expressive language problems in clinical samples is explained by, that these characteristics are more readily observable than for example receptive language difficulties (Zhang & Tomblin, 2000). In Sweden, it may probably also be influenced by the current focus of the language screening at the child health care centers.

In study II, sensitivity was investigated based on the speech output status in the children with SLI. For example, the sensitivity was found to be 75% in children with currently normal speech status. This is not surprising since the scoring is based on the child's speech output performance. Is NWR more of a marker for speech sounds deficits than for SLI-only? NWR is a complex measure. Repetition of nonwords depends on a range of interacting skills. A deficit or a difficulty in any of the skills risk influencing the output. The output constitutes articulation of what one has heard, perceived and memorised, and is the emergent surface



manifestation of the nonword. In subsequent analysis, not included in Study II, the six children who scored above the cut-off level on NWR based on the binary scoring, scored below cut-off level when a more detailed scoring method was used (i.e., percent correct consonants). This means, that the method of scoring is also a factor that may influence the sensitivity of a test.

To conclude, speech output status was found to influence the sensitivity of the binary scoring on the NWR test. Finally, it was not in the scope of the thesis to explore mechanisms behind poor NWR. However, different factors that may underlie poor NWR are possible to study more thoroughly in a forthcoming study.

#### *Association between family history and NWR*

In study II, it was found that children with SLI that grow up with parents with LLP have poorer NWR performance than those who do not have parents with LLP, and the difference between these two groups on NWR was large. This finding may be an indication of an association between family history and NWR. As already mentioned in a previous section, family history data cannot disentangle environmental factors from genetic factors. However, the evidence for genetic influence on SLI is very strong (Bishop, Adams, & Norbury, 2006) and deficits in NWR have been found to be highly heritable with little evidence of environmental influence (Bishop et al., 1999).

It is highly plausible that linguistic input for children with SLI differs depending on whether the parents have LRDPs or not. Yet, it is also important to remember that a child with SLI will contribute to, and

shape the interaction within a family, because of the child's own limitations in speech and language abilities (Fujiki et al., 2001). It is well-known that there is a mutual adjustment of communicative behaviours in parent-child interactions where the child has language impairment, so that the interaction pattern is regulated according to the linguistic ability of the child (Conti-Ramsden & Friel-Patti, 1984; Conti-Ramsden et al., 1995; Lasky & Klopp, 1982). It still remains to be explored how this mutual adjustment is affected in families where parents themselves are struggling with language-related problems.

#### *Level of parental education and LRDPs*

Level of education is one aspect of SES and is often used as an estimation of SES. SES has been found to be important for home language input (Bradley & Corwyn, 2002; Craig, 2006). However, even more important than parents' formal level of education, is the quality of the linguistic, cognitive, social, and emotional stimulation parents are able to provide for their children (Christian, Morrison, & Bryant, 1998; Taylor, Clayton, & Rowley, 2004).

The influences of nature and nurture on SLI can be difficult to disentangle. Low parental education levels may reflect LRDPs in the parents and may therefore also be genetically influenced (Paul & Norbury, 2012). SES may also have an impact on the home language environment (Bradley & Corwyn, 2002; Davis-Kean, 2005), influencing activities and language input for the child. Thus, a low level of parental education may influence language acquisition in children with SLI, but may not be a direct cause of SLI. In addition, it should be mentioned that education level also reflects the

educational system and culture of a country (e.g., education for all or for an exclusive proportion of citizens), and the time of schooling (e.g., being a child today or 60 years ago).

Swedish studies of SES influence on children's language and cognitive environment is sparse. Updated research regarding SES influence on children's development, based on today's generations of families in Sweden, is needed, including focus on cultural diversity and bilingualism. It could help family-oriented clinical child language pathology by adding new knowledge and giving a better understanding of the issues involved.

To conclude, I would first like to stress that NWR is often reported to be poor in children with speech and language deficits that are present in a range of neurodevelopmental disorders (Miniscalco & Gillberg, 2009), for example in children with autism spectrum disorder (Kjelgaard & Tager-Flusberg, 2001), Down's syndrome (Costanzo et al., 2013), and in children with RI (Catts et al., 2005; Ramus et al., 2013) and children with cochlear implants or hearing impairment (Nakeva von Mentzer, 2014). Though, none of these studies has reported the sensitivity and specificity of NWR.

Secondly, I would like to highlight that SLI in school-age children can be difficult to identify because behavioural problems related to, e.g., academic achievement and psychosocial functions, risk hiding the language impairment. Currently, the established routines for language screenings at the child health care centres in Sweden are in the process of being improved. In most cases, children with SLI are identified in their preschool-age. However, it is very important to also identify SLI in school-age children because of these children's risk of having associated problems (Law et al., 2000). To be able to

identify SLI in school-age children, professionals in clinical and educational settings need to have knowledge about child language impairment and a good understanding of the functional impacts of poor language skills.

### **6.3 READING**

In Study III, a high proportion of the children with SLI were found to have poor reading skills: 79% of the children showed poor decoding skills, 85% showed poor reading comprehension skills, and 97% showed difficulties in either or both of the reading measures.

The findings in Study III of a high co-occurrence of SLI and RI are in line with previous studies based on clinical populations. For example, Davison and Howlin (1997) reported poor reading skills – mainly poor reading comprehension – in 89% of a sample of children with SLI attending school language units. Stothard et al. (1998) found 93% RI in a longitudinal follow-up of 15-year-olds with SLI diagnosed at age 5. In a study of 11-year-old children with SLI, Conti-Ramsden, Botting, Simkin, et al. (2001) found poor decoding skills in 69% and poor reading comprehension in 78% of their sample. The findings of RI in these previous reports are very similar to the findings presented in Study III. However, in a longitudinal and population-based study of children identified with SLI at age 5, Catts et al. (2005) found poor decoding skills in around 20% when followed-up in years 2, 4 and 8. This is a lower proportion than has been reported for clinical populations. In addition, the stability of the SLI classification in the study by Catts and colleagues, was low over time (Tomblin, 2008; Tomblin, Zhang,

Buckwalter, & O'Brien, 2003) and only less than half of the children identified with SLI at age 5 were clinically referred because of language difficulties (Paul & Norbury, 2012).

According to Ramus et al. (2013) findings of overlap of SLI and RI in clinical samples risk being biased and over-estimating the “true overlap” regarding the relation of SLI and dyslexia, as compared to representative population samples. On the other hand, one may suggest that population-based studies under-estimate the co-occurrence of RI and SLI in clinical samples. Indeed, population based studies can be too general in that they are not specific in explaining clinical phenomena, while clinical studies can be too narrow in the sense of their generalisability often being limited to the most severe populations.

To conclude, studies investigating the overlap between SLI and RI are in part reflecting differences in sample characteristics. It is more likely to find high rates of RI in clinical samples of school-age children with SLI, than in population based samples or in samples with early identified SLI participating in follow-up studies. Differences between clinical and population based samples regarding the proportions of RI found in children with SLI, may be due to these two types of samples representing phenotypically and ethiologically different groups of SLI (Bishop & Hayiou-Thomas, 2008). For example, in clinical populations it is more likely to find speech deficits (Zhang & Tomblin, 2000) and when SLI is defined in terms of clinical referral to SLP services, and not on the basis of a naturalist perspective (Tomblin, 2008), the genetic influence on SLI is considerably greater (Bishop & Hayiou-Thomas, 2008).

*Discrepancy between direct testing of reading skills and the family history reports of literacy problems in the children with SLI*

According to the parental reports in the family history interviews, only three of the 61 participants were previously clinically diagnosed with RI. There are many possible explanations for the discrepancy between the parental reports and the direct assessments of the children with SLI. One explanation could be that the school language units might be aware of the poor reading skills in these children and are offering them appropriate reading intervention without a need of a formal clinical diagnosis of RI. Moreover, the schools might think that a clinical diagnosis of RI in children with SLI is unnecessary or redundant; because reading difficulties are expected in these children. The latter would suggest that lack of clinical diagnosis does not necessarily imply a disadvantage to the individual child. Furthermore, it also needs to be acknowledged that the measures underlying classification of RI in research studies are often limited as compared to the more comprehensive assessments of reading and language skills in clinical diagnostics. Nevertheless, the discrepancy is still great between the parental reports and the direct assessments and it is crucial to discover the reasons for this discrepancy.

*A shared underlying mechanism - NWR*

Studies investigating if SLI and RI are two distinct disorders are usually based on models illustrating the relationship between SLI, RI and phonological skills. However, in general, boundaries between SLI and RI are hard to draw because of the overlap at the behavioural level and probably also at the etiological level.

The most influential theory of the underlying causes of poor decoding skills in alphabetic languages is the phonological deficit hypothesis (Vellutino, Fletcher, Snowling, & Scanlon, 2004), which states specific deficits in phonological skills (e.g., phonological STM). The phonological deficits in these two conditions have been suggested to be qualitatively different (Ramus et al., 2013), and in addition, found to be more severe in children who have both SLI and dyslexia as compared to children with SLI-only or dyslexia-only (Bishop et al., 2009; Ramus et al., 2013). NWR is commonly used in reading research as a measure of phonological skills. Interestingly, one of the first studies of NWR found that school-age children with SLI performed significantly poorer on NWR than children with RI-only (dyslexic type of RI) (Kamhi & Catts, 1986).

In Study III, the children with SLI perform poorer on NWR than the control children, irrespective of having poor decoding skills or not. These findings corroborate previous reports (Bishop et al., 2009; Catts et al., 2005; Ramus et al., 2013) and might be expected in a clinical sample with SLI, considering that NWR is a clinical marker for SLI (Study II). NWR has also been suggested as a clinical marker for dyslexia (Moll et al., 2013). This means that these two conditions share an endophenotype characterised by (the same or different) difficulties in processing phonological information and therefore respond to the same clinical marker.

Yet, and importantly, cognitive endophenotypes of RI have also been suggested to be based on a broad language phenotype (Bishop & Snowling, 2004; Elwér, 2014), a perspective that corresponds particularly well with RI in children with SLI. For example, the construct of comprehension has been described as an integration of structural language knowledge, broad and context-specific world

knowledge, motivation and processing capacity (Elwér, 2014; Kamhi & Catts, 2012; Kintsch & Rawson, 2005).

### *Family history and reading skills in SLI*

The most common LRDP reported for the parents was related to literacy problems and was found in 37.3% of the parents, a significantly higher prevalence rate than the about 5-10% reported for general populations (Hulme & Snowling, 2011; Kamhi & Catts, 2012). Moreover, the co-occurrence of language and literacy problems was high in the parents: 55% of the parents with language problems also had literacy problems, and 44% of the parents with literacy problems also had language problems. Taken together, these findings based on the family history interviews made it highly important to investigate the reading skills in the sample of children with SLI – in which only a small number was reported with a clinical diagnosis of RI.

Importantly, not only children with a family history of literacy problems, but also most of the children without a family history showed RI. However, the findings of associations between a positive family history of literacy problems and poor decoding of nonwords in Study III, and between family history of LLP and poor NWR in Study II, are in line with previous research (Carroll et al., 2014; DeThorne et al., 2006; Moll et al., 2013). Our data points to the complexity in family aggregation of LRDPs in studies investigating family history, and is stressing a broader phenotype in family history of literacy problems. For example, parents with and without literacy problems were reported as also having other LRDPs, for example in addition to their literacy problems (Study II).



Possible genetic influence from the extended family must also be considered, for example, LRDPs in grandparents and other close relatives (Study I).

*Predictions based on the SVR model*

The SVR model (Gough & Tunmer, 1986; Hoover & Gough, 1990) predicts that the difficulty poor comprehenders have in understanding written text is the result of deficits in either language comprehension or poor decoding skills, or in both.

The framework of the SVR was used in study III to investigate the distribution of our sample of children with SLI into subgroups based on the two SVR components, that is, decoding skills and oral language comprehension. In the same study it was explored if the two components vary in their relative contributions to reading comprehension in the lower and upper school years. In typically developing children decoding is a better predictor of reading comprehension in lower school years and listening comprehension a better predictor in upper years (Gough et al., 1996). This change in the link between reading comprehension, oral language comprehension and decoding skills follows the development of reading – after a couple of years in school children typically become skilled in decoding words and a shift of factors driving the focus in their reading development takes place. Decoding becomes more automatic and less challenging to, for example, the memory system. This opens up for new factors (e.g., language comprehension, world knowledge and cognition in general) to come into the picture with a mission to support reading comprehension (Catts & Kamhi, 2005).

Why is this link interesting to explore in a SLI-sample? Should we expect any other findings than what has been reported for typically developing readers? Yes, because a large majority of the participants in Study III have difficulties in accurate and fluent decoding of words and nonwords, in addition to their SLI. Therefore, decoding skills could be expected to be a better predictor in the lower as well as in the upper school years. Importantly, predictions at group level do not necessarily explain individual differences, thus in intervention it is necessary to take the individual perspective. For every school year the participants in Study III do improve the number of correctly decoded words and nonwords (i.e., even children with SLI can be expected to score higher for every school year). However, and importantly, despite lack of age reference data for children in years 4 and 5, the data clearly suggests that the participants do not reach age adequate norms. It remains for future studies to investigate which other factors contribute to reading comprehension of children with SLI in the upper school years.

In summary, a large overlap of SLI and RI in clinical populations should be expected. The discrepancy between the low number of children being previously clinically diagnosed with RI and the high number of children identified with poor reading comprehension and/or poor decoding skills on the basis of direct assessment needs to be further investigated. When a child has a family history for LLP, and/or has poor NWR skills, or difficulties in decoding of words/nonwords, or a deficient oral language comprehension; professionals involved in intervention and education should be concerned about the child's language and reading development and take necessary steps for prevention and intervention.

## 6.4 CONCLUSIONS

The work presented in the thesis is the first, more comprehensive, step towards a better understanding of Swedish school-age children with SLI. The participants with SLI represent monolingual children who require special education because of persistent and pervasive difficulties in language development.

This thesis contributes to the development of diagnostics in Swedish SLI by providing support for NWR as a clinical marker. The findings of associations between family history, NWR and decoding skills contribute to a broader understanding and implementation of a family-oriented clinical approach in child language pathology. Counselling and support for the families is recommended, taking into account the high prevalence of LRDPs in the parents and siblings, and considering the association between positive family history and, for example, poor decoding skills in the children with SLI. In the efforts to enhance language learning in children with SLI, we need to acknowledge linguistic, social and cultural challenges to the family network of the child.

- Knowledge about familial aggregation should be considered when making predictions about outcome, prevention and intervention.
- Documentation of family history is strongly advisable in clinical child language pathology, for example, for early identification of siblings who are at a high risk of having LRDPs.
- Poor NWR is a clinical marker for Swedish school-age children with SLI. NWR is insensitive to age, gender, non-

verbal IQ and to level of parental education. NWR should be combined with other linguistic markers in screening for SLI.

- The finding of RI in 97% of children with SLI, most showing both poor decoding and poor reading comprehension, has important implications for prevention and intervention, and emphasises the necessity of continual assessments of the literacy skills in children involved in school language units.

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## REFERENCES

- Anthoni, H., Sucheston, L. E., Lewis, B. A., Tapia-Páez, I., Fan, X., Zucchelli, M., . . . Castrén, E. (2012). The aromatase gene CYP19A1: several genetic and functional lines of evidence supporting a role in reading, speech and language. *Behav Genet*, *42*(4), 509-527.
- Archibald, L. M. (2008). The promise of nonword repetition as a clinical tool. *Canadian Journal of Speech-Language Pathology and Audiology*, *32*, 21-28.
- Asbury, K., & Plomin, R. (2013). *G is for genes: The impact of genetics on education and achievement* (Vol. 14): John Wiley & Sons.
- Baddeley, A. (2012). Working memory: theories, models, and controversies. [Review]. *Annual Review of Psychology*, *63*, 1-29. doi: 10.1146/annurev-psych-120710-100422
- Baddeley, A., & Hitch, G. (1974). Working memory The psychology of learning and motivation: advances in research and theory. *GH. Bower*.
- Barry, J. G., Yasin, I., & Bishop, D. V. (2007). Heritable risk factors associated with language impairments. [Evaluation Studies Research Support, Non-U.S. Gov't]. *Genes Brain Behav*, *6*(1), 66-76. doi: 10.1111/j.1601-183X.2006.00232.x
- Bartlett, C. W., Flax, J. F., Logue, M. W., Vieland, V. J., Bassett, A. S., Tallal, P., & Brzustowicz, L. M. (2002). A major susceptibility locus for specific language impairment is located on 13q21. *The American Journal of Human Genetics*, *71*(1), 45-55.

- Beitchman, J. H., Hood, J., & Inglis, A. (1992). Familial transmission of speech and language impairment: a preliminary investigation. [Research Support, Non-U.S. Gov't]. *Can J Psychiatry*, 37(3), 151-156.
- Bishop, D. (1997). *Uncommon understanding: Development and disorders of language comprehension in children*: Psychology Press/Erlbaum (UK) Taylor & Francis.
- Bishop, D., & Hayiou-Thomas, M. (2008). Heritability of specific language impairment depends on diagnostic criteria. *Genes, Brain and Behavior*, 7(3), 365-372.
- Bishop, D. V. (1983). TROG-Test for Reception of Grammar: Medical Research Council: Chapel Press.
- Bishop, D. V., Adams, C. V., & Norbury, C. F. (2006). Distinct genetic influences on grammar and phonological short-term memory deficits: evidence from 6-year-old twins. *Genes, Brain and Behavior*, 5(2), 158-169.
- Bishop, D. V., Bishop, S. J., Bright, P., James, C., Delaney, T., & Tallal, P. (1999). Different Origin of Auditory and Phonological Processing Problems in Children With Language Impairment: Evidence From a Twin Study. *Journal of Speech, Language, and Hearing Research*, 42(1), 155-168.
- Bishop, D. V., & McDonald, D. (2009). Identifying language impairment in children: combining language test scores with parental report. *International Journal of Language & Communication Disorders*, 44(5), 600-615.
- Bishop, D. V., McDonald, D., Bird, S., & Hayiou-Thomas, M. E. (2009). Children who read words accurately despite language impairment: who are they and how do they do it? *Child Dev*, 80(2), 593-605.
- Bishop, D. V., North, T., & Donlan, C. (1995). Genetic basis of specific language impairment: evidence from a twin

- study. [Comparative Study Research Support, Non-U.S. Gov't Twin Study]. *Dev Med Child Neurol*, 37(1), 56-71.
- Bishop, D. V., North, T., & Donlan, C. (1996). Nonword repetition as a behavioural marker for inherited language impairment: Evidence from a twin study. *Journal of Child Psychology and Psychiatry*, 37(4), 391-403.
- Bishop, D. V., & Snowling, M. J. (2004). Developmental dyslexia and specific language impairment: Same or different? *Psychol Bull*, 130(6), 858.
- Bishop, D. V. M. (2014). Ten questions about terminology for children with unexplained language problems. *International Journal of Language & Communication Disorders*, 49(4), 381-415. doi: 10.1111/1460-6984.12101
- Bortolini, U., Arfé, B., Caselli, C. M., Degasperi, L., Deevy, P., & Leonard, L. B. (2006). Clinical markers for specific language impairment in Italian: The contribution of clitics and non-word repetition. *International Journal of Language & Communication Disorders*, 41(6), 695-712.
- Botting, N. (2005). Non-verbal cognitive development and language impairment. *Journal of Child Psychology and Psychiatry*, 46(3), 317-326.
- Botting, N., & Conti-Ramsden, G. (2001). Non-word repetition and language development in children with specific language impairment (SLI). *International Journal of Language & Communication Disorders*, 36(4), 421-432.
- Botting, N., & Conti-Ramsden, G. (2003). Autism, primary pragmatic difficulties, and specific language impairment: can we distinguish them using psycholinguistic markers? *Developmental Medicine & Child Neurology*, 45(8), 515-524.

- Bradley, R. H., & Corwyn, R. F. (2002). Socioeconomic status and child development. *Annual Review of Psychology*, 53(1), 371-399.
- Byrne, B., Coventry, W. L., Olson, R. K., Samuelsson, S., Corley, R., Willcutt, E. G., . . . DeFries, J. C. (2009). Genetic and environmental influences on aspects of literacy and language in early childhood: Continuity and change from preschool to Grade 2. *Journal of Neurolinguistics*, 22(3), 219-236.
- Bölte, S., Dziobek, I., & Poustka, F. (2009). Brief report: The level and nature of autistic intelligence revisited. *J Autism Dev Disord*, 39(4), 678-682.
- Campbell, T., Dollaghan, C., Needleman, H., & Janosky, J. (1997). Reducing bias in language assessment: Processing-dependent measures. *Journal of Speech, Language and Hearing Research*, 40(3), 519.
- Carroll, J. M., Mundy, I. R., & Cunningham, A. J. (2014). The roles of family history of dyslexia, language, speech production and phonological processing in predicting literacy progress. *Developmental Science*.
- Catts, H., Adlof, S., Hogan, T., & Weismer, S. E. (2005). Are specific language impairment and dyslexia distinct disorders? *Journal of speech, language, and hearing research: JSLHR*, 48(6), 1378.
- Catts, H. W., Hogan, T. P., & Fey, M. E. (2003). Subgrouping poor readers on the basis of individual differences in reading-related abilities. *J Learn Disabil*, 36(2), 151-164.
- Catts, H. W., & Kamhi, A. G. (2005). *The connections between language and reading disabilities*: Psychology Press.
- Chiat, S., & Roy, P. (2007). The Preschool Repetition Test: An evaluation of performance in typically developing and

clinically referred children. *Journal of Speech, Language, and Hearing Research*, 50(2), 429-443.

- Christian, K., Morrison, F. J., & Bryant, F. B. (1998). Predicting kindergarten academic skills: Interactions among child care, maternal education, and family literacy environments. *Early Childhood Research Quarterly*, 13(3), 501-521. doi: 10.1016/s0885-2006(99)80054-4
- Clark, A., O'Hare, A., Watson, J., Cohen, W., Cowie, H., Elton, R., . . . Seckl, J. (2007). Severe receptive language disorder in childhood—familial aspects and long-term outcomes: results from a Scottish study. *Arch Dis Child*, 92(7), 614-619.
- Clegg, J., Hollis, C., Mawhood, L., & Rutter, M. (2005). Developmental language disorders—a follow-up in later adult life. Cognitive, language and psychosocial outcomes. *Journal of Child Psychology and Psychiatry*, 46(2), 128-149.
- Coady, J. A., & Evans, J. L. (2008). Uses and interpretations of non-word repetition tasks in children with and without specific language impairments (SLI). *International Journal of Language & Communication Disorders*, 43(1), 1-40.
- Consortium, S. (2002). A genomewide scan identifies two novel loci involved in specific language impairment. *The American Journal of Human Genetics*, 70(2), 384-398.
- Consortium, S. (2004). Highly significant linkage to the SLI1 locus in an expanded sample of individuals affected by specific language impairment. *Am J Hum Genet*, 74(6), 1225.
- Conti-Ramsden, G. (2003). Processing and linguistic markers in young children with specific language impairment (SLI).

*Journal of Speech, Language and Hearing Research*, 46(5), 1029.

- Conti-Ramsden, G., Durkin, K., Simkin, Z., & Knox, E. (2009). Specific language impairment and school outcomes. I: Identifying and explaining variability at the end of compulsory education. *International Journal of Language & Communication Disorders*, 44(1), 15-35.
- Conti-Ramsden, G., Falcaro, M., Simkin, Z., & Pickles, A. (2007). Familial loading in specific language impairment: patterns of differences across proband characteristics, gender and relative type. [Research Support, Non-U.S. Gov't]. *Genes Brain Behav*, 6(3), 216-228. doi: 10.1111/j.1601-183X.2006.00250.x
- Conti-Ramsden, G., & Friel-Patti, S. (1984). Mother-child dialogues: a comparison of normal and language impaired children. *J Commun Disord*, 17(1), 19-35.
- Conti-Ramsden, G., Hutcheson, G. D., & Grove, J. (1995). Contingency and breakdown: Children with SLI and their conversations with mothers and fathers. *Journal of Speech, Language and Hearing Research*, 38(6), 1290.
- Conti-Ramsden, G., Mok, P. L., Pickles, A., & Durkin, K. (2013). Adolescents with a history of specific language impairment (SLI): Strengths and difficulties in social, emotional and behavioral functioning. *Res Dev Disabil*, 34(11), 4161-4169.
- Conti-Ramsden, G., Simkin, Z., & Pickles, A. (2006). Estimating familial loading in SLI: a comparison of direct assessment versus parental interview. [Comparative Study Research Support, Non-U.S. Gov't]. *J Speech Lang Hear Res*, 49(1), 88-101. doi: 10.1044/1092-4388(2006/007)
- Conti-Ramsden, G., St Clair, M., Pickles, A., & Durkin, K. (2012). Developmental Trajectories of Verbal and

Nonverbal Skills in Individuals with a History of SLI: From Childhood to Adolescence. *Journal of speech, language, and hearing research: JSLHR*.

- Conti-Ramsden, G., Botting, N., & Faragher, B. (2001). Psycholinguistic markers for specific language impairment (SLI). *Journal of Child Psychology and Psychiatry*, 42(6), 741-748.
- Conti-Ramsden, G., Botting, N., Simkin, Z., & Knox, E. (2001). Follow-up of children attending infant language units: outcomes at 11 years of age. *International Journal of Language & Communication Disorders*, 36(2), 207-219.
- Costanzo, F., Varuzza, C., Menghini, D., Addona, F., Giancesini, T., & Vicari, S. (2013). Executive functions in intellectual disabilities: A comparison between Williams syndrome and Down syndrome. *Res Dev Disabil*, 34(5), 1770-1780.
- Crago, M. B. (1990). Development of communicative competence in Inuit children: Implications for speech-language pathology. *Communication Disorders Quarterly*, 13(1), 73-83.
- Craig, L. (2006). Parental education, time in paid work and time with children: an Australian time-diary analysis. *The British journal of sociology*, 57(4), 553-575.
- Davis-Kean, P. E. (2005). The influence of parent education and family income on child achievement: the indirect role of parental expectations and the home environment. *Journal of Family Psychology*, 19(2), 294.
- Davison, F. M., & Howlin, P. (1997). A follow-up study of children attending a primary-age language unit. *International Journal of Language & Communication Disorders*, 32(1), 19-36.
- De Bree, E., Rispen, J., & Gerrits, E. (2007). Non-word repetition in Dutch children with (a risk of) dyslexia and

SLI. *Clinical Linguistics & Phonetics*, 21(11-12), 935-944.

DeThorne, L. S., Hart, S. A., Petrill, S. A., Deater-Deckard, K., Thompson, L. A., Schatschneider, C., & Davison, M. D. (2006). Children's history of speech-language difficulties: Genetic influences and associations with reading-related measures. *Journal of Speech, Language, and Hearing Research*, 49(6), 1280-1293.

Dethorne, L. S., & Watkins, R. V. (2006a). Language abilities and nonverbal IQ in children with language impairment: inconsistency across measures. *Clin Linguist Phon*, 20(9), 641-658.

Dethorne, L. S., & Watkins, R. V. (2006b). Language abilities and nonverbal IQ in children with language impairment: Inconsistency across measures. *Clinical Linguistics & Phonetics*, 20(9), 641-658.

Dispaldro, M., Leonard, L. B., & Deevy, P. (2012). Real-Word and Nonword Repetition in Italian-Speaking Children with Specific Language Impairment: A Study of Diagnostic Accuracy. *J Speech Lang Hear Res*. doi: 10.1044/1092-4388(2012/11-0304)

Dollaghan, C., & Campbell, T. F. (1998). Nonword repetition and child language impairment. *Journal of Speech, Language and Hearing Research*, 41(5), 1136.

Durkin, K., Conti-Ramsden, G., & Simkin, Z. (2012). Functional outcomes of adolescents with a history of specific language impairment (SLI) with and without autistic symptomatology. [Research Support, Non-U.S. Gov't]. *J Autism Dev Disord*, 42(1), 123-138. doi: 10.1007/s10803-011-1224-y

Durkin, K., & Conti-Ramsden, G. (2007). Language, social behavior, and the quality of friendships in adolescents



- with and without a history of specific language impairment. *Child Dev*, 78(5), 1441-1457.
- Ebbles, S. (2014). Introducing the SLI debate. *International Journal of Language & Communication Disorders*, 49(4), 377–380. doi: 10.1111/1460-6984.12119
- Eicher, J. D., Powers, N. R., Miller, L. L., Mueller, K. L., Mascheretti, S., Marino, C., . . . Smith, S. D. (2014). Characterization of the DYX2 locus on chromosome 6p22 with reading disability, language impairment, and IQ. *Hum Genet*, 1-13.
- Elliott, J. G., & Grigorenko, E. L. (2014). *The dyslexia debate* (Vol. 14): Cambridge University Press.
- Elwér, Å. (2014). *Early Predictors of Reading Comprehension Difficulties*. PhD thesis, Linköping University, Sweden.
- Falcaro, M., Pickles, A., Newbury, D. F., Addis, L., Banfield, E., Fisher, S. E., . . . Conti-Ramsden, G. (2008). Genetic and phenotypic effects of phonological short-term memory and grammatical morphology in specific language impairment. [Research Support, Non-U.S. Gov't]. *Genes Brain Behav*, 7(4), 393-402. doi: 10.1111/j.1601-183X.2007.00364.x
- Fidler, L. J., Plante, E., & Vance, R. (2011). Identification of Adults With Developmental Language Impairments. *Am J Speech Lang Pathol*, 20(1), 2-13. doi: 10.1044/1058-0360(2010/09-0096)
- Flax, J. F., Realpe-Bonilla, T., Hirsch, L. S., Brzustowicz, L. M., Bartlett, C. W., & Tallal, P. (2003). Specific language impairment in families: evidence for co-occurrence with reading impairments. *J Speech Lang Hear Res*, 46(3), 530-543.
- Fujiki, M., Brinton, B., Isaacson, T., & Summers, C. (2001). Social behaviors of children with language impairment on

- the playground: A pilot study. *Lang Speech Hear Serv Sch*, 32(2), 101.
- Gathercole, S. E. (2006). Nonword repetition and word learning: The nature of the relationship. *Applied Psycholinguistics*, 27(04), 513-543.
- Gathercole, S. E., & Baddeley, A. D. (1990). Phonological memory deficits in language disordered children: Is there a causal connection? *Journal of Memory and Language*, 29(3), 336-360.
- Gathercole, S. E., Willis, C. S., Baddeley, A. D., & Emslie, H. (1994). The Children's Test of Nonword Repetition: a test of phonological working memory. [Research Support, Non-U.S. Gov't]. *Memory*, 2(2), 103-127. doi: 10.1080/09658219408258940
- Gathercole, S. E., Willis, C. S., Emslie, H., & Baddeley, A. D. (1992). Phonological memory and vocabulary development during the early school years: A longitudinal study. *Dev Psychol*, 28(5), 887.
- Girbau, D., & Schwartz, R. G. (2007). Non-word repetition in Spanish-speaking children with Specific Language Impairment (SLI). [Research Support, N.I.H., Extramural Research Support, Non-U.S. Gov't]. *Int J Lang Commun Disord*, 42(1), 59-75. doi: 10.1080/13682820600783210
- Gough, P. B., Hoover, W. A., & Peterson, C. L. (1996). Some observations on a simple view of reading. *Reading comprehension difficulties: Processes and intervention*, 1-13.
- Gough, P. B., & Tunmer, W. E. (1986). Decoding, reading, and reading disability. *Remedial and special education*, 7(1), 6-10.
- Graf Estes, K., Evans, J. L., & Else-Quest, N. M. (2007). Differences in the nonword repetition performance of

- children with and without specific language impairment: a meta-analysis. [Meta-Analysis Research Support, N.I.H., Extramural]. *J Speech Lang Hear Res*, 50(1), 177-195. doi: 10.1044/1092-4388(2007/015)
- Gray, S. (2003). Diagnostic accuracy and test-retest reliability of nonword repetition and digit span tasks administered to preschool children with specific language impairment. *J Commun Disord*, 36(2), 129-151.
- Hansson, K., Forsberg, J., Löfqvist, A., MäkiTorkko, E., & Sahlén, B. (2004). Working memory and novel word learning in children with hearing impairment and children with specific language impairment. *International Journal of Language & Communication Disorders*, 39(3), 401-422.
- Hansson, K., Sahlén, B., & Mäki-Torkko, E. (2007). Can a single hit cause limitations in language development? A comparative study of Swedish children with hearing impairment and children with specific language impairment. *International Journal of Language & Communication Disorders*, 42(3), 307-323.
- Hansson, K., Sandgren, O. and Sahlén, B. (2014). Changing labels for a concept in change. Commentary on Bishop, D. V. M., Ten questions about terminology for children with unexplained language problems. *International Journal of Language and Communication Disorders*, 49(4), 381-415 doi: 10.1111/1460-6984.12101
- Hawke, J. L., Olson, R. K., Willcutt, E. G., Wadsworth, S. J., & DeFries, J. C. (2009). Gender ratios for reading difficulties. *Dyslexia*, 15(3), 239-242.
- Hayiou-Thomas, M. E., Harlaar, N., Dale, P. S., & Plomin, R. (2010). Preschool speech, language skills, and reading at 7, 9, and 10 years: Etiology of the relationship. *Journal of*

*Speech, Language, and Hearing Research*, 53(2), 311-332.

Hedenius, M., Persson, J., Tremblay, A., Adi-Japha, E., Veríssimo, J., Dye, C. D., . . . Ullman, M. T. (2011). Grammar predicts procedural learning and consolidation deficits in children with specific language impairment. *Res Dev Disabil*, 32(6), 2362-2375.

Hellquist, B., Magnusson, E., & Naucler, K. (1997). Meningsläsningsprov SL 60 och SL 40 *Lund Pedagogisk Design*.

Hesketh, A., & Conti-Ramsden, G. (2013). Memory and Language in Middle Childhood in Individuals with a History of Specific Language Impairment. *PLoS One*, 8(2), e56314.

Holmberg, E., & Lundälv, E. (1998). TROG. *SIH Läromedel, Gothenburg*.

Hulme, C., & Snowling, M. J. (2011). Children's Reading Comprehension Difficulties Nature, Causes, and Treatments. *Current Directions in Psychological Science*, 20(3), 139-142.

Ibertsson, T., Willstedt-Svensson, U., Radeborg, K., & Sahlén, B. (2008). A methodological contribution to the assessment of nonword repetition—a comparison between children with specific language impairment and hearing-impaired children with hearing aids or cochlear implants. [Comparative Study Research Support, Non-U.S. Gov't]. *Logoped Phoniatr Vocol*, 33(4), 168-178. doi: 10.1080/14015430801945299

Kamhi, A. G., & Catts, H. W. (1986). Toward an understanding of developmental language and reading disorders. *Journal of Speech and Hearing Disorders*, 51(4), 337.

- Kamhi, A. G., & Catts, H. W. (2012). *Language and reading disabilities*: Pearson.
- Kamhi, A. G., Catts, H. W., Mauer, D., Apel, K., & Gentry, B. F. (1988). Phonological and spatial processing abilities in language-and reading-impaired children. *Journal of Speech and Hearing Disorders*, 53(3), 316.
- Kapalková, S., Polišíenská, K., & Vicenová, Z. (2013). Non-word repetition performance in Slovak-speaking children with and without SLI: novel scoring methods. *International Journal of Language & Communication Disorders*, 48(1), 78-89.
- Keegstra, A. L., Knijff, W. A., Post, W. J., & Goorhuis-Brouwer, S. M. (2007). Children with language problems in a speech and hearing clinic: background variables and extent of language problems. *Int J Pediatr Otorhinolaryngol*, 71(5), 815-821. doi: 10.1016/j.ijporl.2007.02.001
- Kintsch, W., & Rawson, K. (2005). Comprehension. *The Science of Reading: A Handbook*, 209-226.
- Kjelgaard, M. M., & Tager-Flusberg, H. (2001). An investigation of language impairment in autism: Implications for genetic subgroups. *Language and Cognitive Processes*, 16(2-3), 287-308.
- Kluever, R. C. (1995). The WISC-III and Raven Coloured Progressive Matrices Test: A Pilot Study of Relationships.
- Kuhl, P. K., & Meltzoff, A. N. (1996). Infant vocalizations in response to speech: Vocal imitation and developmental change. *J Acoust Soc Am*, 100(4), 2425-2438.
- Lalkhen, A. G., & McCluskey, A. (2008). Clinical tests: sensitivity and specificity. *Continuing Education in Anaesthesia, Critical Care & Pain*, 8(6), 221-223.

- Lasky, E. Z., & Klopp, K. (1982). Parent-child interactions in normal and language-disordered children. *Journal of Speech and Hearing Disorders*, 47(1), 7.
- Law, J., Boyle, J., Harris, F., Harkness, A., & Nye, C. (2000). Prevalence and natural history of primary speech and language delay: findings from a systematic review of the literature. [Review]. *Int J Lang Commun Disord*, 35(2), 165-188.
- Law, J., McBean, K., & Rush, R. (2011). Communication skills in a population of primary school-aged children raised in an area of pronounced social disadvantage. *International Journal of Language & Communication Disorders*, 46(6), 657-664.
- Law, J., Tomblin, J. B., & Zhang, X. (2008). Characterizing the growth trajectories of language-impaired children between 7 and 11 years of age. *Journal of Speech, Language, and Hearing Research*, 51(3), 739-749.
- Lavesson, A. (2012). Språkscreening i fyraårsåldern: Nonordsrepetition och andra kliniska markörer. *Master thesis Lund University*.
- Lavesson, A., Hansson, K., & Lövdén, M. (2014). Language screening at age 4. Construction and standardization of a new test to be used at Health Centers in Sweden. *Poster presentation at International Clinical Phonetics and Linguistic Association, Stockholm Sweden*.
- Leonard, L. (2014a). Replacing one imperfect term with another. Commentary on Reilly, S., Tomblin, B., Law, J., McKean, C., Mensah, F. K., Morgan, A., Goldfeld, S., Nicholson, J. M. and Wake, M., 2014, Specific language impairment: a convenient label for whom. *International Journal of Language and Communication Disorders* 49(4), 416–451. doi: 10.1111/1460-6984.12102

- Leonard, L. B. (2000). *Children with specific language impairment*: MIT Press (MA).
- Leonard, L. B. (2014b). *Children with specific language impairment*: MIT press.
- Li, N., & Bartlett, C. W. (2012). Defining the genetic architecture of human developmental language impairment. *Life Sci*, 90(13-14), 469-475. doi: 10.1016/j.lfs.2012.01.016
- Locke, A., Ginsborg, J., & Peers, I. (2002). Development and disadvantage: implications for the early years and beyond. *International Journal of Language & Communication Disorders*, 37(1), 3-15.
- Lohmander, A., Lundeberg Hammarström, I., & Persson, C. (2014). *Articulation, nasality and phonology in typically developing Swedish individuals: normative data from the SVANTE-test at 3 to 19 years of age*. Poster presentation. International Clinical Phonetics and Linguistics Association Conference. .
- Melby-Lervag, M., & Hulme, C. (2012). Is Working Memory Training Effective? A Meta-Analytic Review. *Dev Psychol*. doi: 10.1037/a0028228
- Miniscalco, C. (2003). Language impairment in Swedish children-A survey of 6-year-olds screened for developmental language disability at 2.5 and 4 years of age. *Licentiat avhandling, Göteborgs Universitet*.
- Miniscalco, C., & Gillberg, C. (2009). Non-word repetition in young school-age children with language impairment and/or neuropsychiatric disorder. [Research Support, Non-U.S. Gov't]. *Res Dev Disabil*, 30(6), 1145-1154. doi: 10.1016/j.ridd.2009.03.002
- Moll, K., Loff, A., & Snowling, M. J. (2013). Cognitive endophenotypes of dyslexia. *Scientific Studies of Reading*, 17(6), 385-397.

- Montgomery, J. W., & Windsor, J. (2007). Examining the language performances of children with and without specific language impairment: Contributions of phonological short-term memory and speed of processing. *Journal of Speech, Language and Hearing Research, 50*(3), 778.
- Moyle, M. J., Karasinski, C., Weismer, S. E., & Gorman, B. K. (2011). Grammatical Morphology in School-Age Children With and Without Language Impairment: A Discriminant Function Analysis. *Lang Speech Hear Serv Sch, 42*(4), 550-560. doi: 10.1044/0161-1461(2011/10-0029)
- Nakeva von Mentzer, C. (2014). *Rethinking Sound. Computer-assisted reading intervention with a phonics approach for deaf and hard of hearing children using cochlear implants or hearing aids*. PhD thesis, Linköping University, Sweden.
- Naucler, K., & Magnusson, E. (1998). Reading and writing development: Report from an ongoing longitudinal study of language-disordered and normal groups from pre-school to adolescence. *Folia phoniatica et Logopaedica, 50*(5), 271-282.
- Nettelblatt, U., & Salameh, E. K. (2007). *Språkstörning hos barn*: Studentlitteratur.
- Nettelblatt, U., Samuelsson, C., Sahlén, B., & Hansson, K. (2008). Språkstörning hos barn utan andra funktionshinder. *Hartelius, L., Nettelblatt, U. & Hammarberg, B (Red.), Logopedi*, 139-146.
- Newbury, D. F., & Monaco, A. P. (2010). Genetic advances in the study of speech and language disorders. *Neuron, 68*(2), 309-320.



- Newbury, D. F., Monaco, A. P., & Paracchini, S. (2014). Reading and Language Disorders: The Importance of Both Quantity and Quality. *Genes*, 5(2), 285-309.
- Newbury, D. F., Paracchini, S., Scerri, T. S., Winchester, L., Addis, L., Richardson, A. J., . . . Monaco, A. P. (2011). Investigation of dyslexia and SLI risk variants in reading- and language-impaired subjects. [Research Support, Non-U.S. Gov't]. *Behav Genet*, 41(1), 90-104. doi: 10.1007/s10519-010-9424-3
- Paul, R., & Norbury, C. (2012). *Language disorders from infancy through adolescence: listening, speaking, reading, writing, and communicating*: Elsevier Health Sciences.
- Pennington, B. F., & Lefly, D. L. (2001). Early reading development in children at family risk for dyslexia. *Child Dev*, 72(3), 816-833.
- Perkins, M. R. (2007). *Pragmatic impairment*. Cambridge: Cambridge University Press.
- Plante, E., Shenkman, K., & Clark, M. M. (1996). Classification of adults for family studies of developmental language disorders. [Comparative Study Research Support, Non-U.S. Gov't Research Support, U.S. Gov't, P.H.S.]. *J Speech Hear Res*, 39(3), 661-667.
- Plante, E., & Vance, R. (1994). Selection of preschool language tests: A data-based approach. *Lang Speech Hear Serv Sch*, 25(1), 15.
- Plomin, R., Haworth, C. M., & Davis, O. S. (2009). Common disorders are quantitative traits. *Nature Reviews Genetics*, 10(12), 872-878.
- Poll, G. H., Betz, S. K., & Miller, C. A. (2010). Identification of clinical markers of specific language impairment in adults. [Research Support, Non-U.S. Gov't]. *J Speech*

*Lang Hear Res*, 53(2), 414-429. doi: 10.1044/1092-4388(2009/08-0016)

- Radeborg, K., Barthelom, E., Sjöberg, M., & Sahlén, B. (2006). A Swedish nonword repetition test for preschool children. *Scand J Psychol*, 47(3), 187-192.
- Ramus, F., Marshall, C. R., Rosen, S., & van der Lely, H. K. (2013). Phonological deficits in specific language impairment and developmental dyslexia: towards a multidimensional model. *Brain*, 136(2), 630-645.
- Raven, J. (1986). Court, JH, & Raven, J.(1986). *Raven's progressive matrices and Raven's coloured matrices*.
- Reilly, S., Bishop, D. V. M. and Tomblin, B. . (2014a). Terminological debate over language impairment in children: forward movement and sticking points. *International Journal of Language & Communication Disorders*, 49(4), 452-462. doi: 10.1111/1460-6984.12111
- Reilly, S., Tomblin, B., Law, J., McKean, C., Mensah, F. K., Morgan, A., Goldfeld, S., Nicholson, J. M. and Wake, M. . (2014b). Specific language impairment: a convenient label for whom? *International Journal of Language & Communication Disorders*,, 49(4), 416-451. doi: 10.1111/1460-6984.12102
- Reuterskiöld-Wagner, C., Sahlén, B., & Nyman, A. (2005). Non-word repetition and non-word discrimination in Swedish preschool children. *Clinical Linguistics & Phonetics*, 19(8), 681-699.
- Rice, M. L. (2003). A unified model of specific and general language delay: Grammatical tense as a clinical marker of unexpected variation. *Language competence across populations: Toward a definition of specific language impairment*, 63-95.

- Rice, M. L. (2014). Advocating for SLI. Commentary on Bishop, D. V. M., Ten questions about terminology for children with unexplained language problems. *International Journal of Language and Communication Disorders* 49(4). doi: 10.1111/1460-6984.12101
- Rice, M. L., Haney, K. R., & Wexler, K. (1998). Family histories of children with SLI who show extended optional infinitives. [Research Support, U.S. Gov't, P.H.S.]. *J Speech Lang Hear Res*, 41(2), 419-432.
- Rice, M. L., Wexler, K., & Cleave, P. L. (1995). Specific language impairment as a period of extended optional infinitive. *J Speech Hear Res*, 38(4), 850-863.
- Sahlén, B., Reuterskiold-Wagner, C., Nettelblatt, U., & Radeborg, K. (1999). Language comprehension and non-word repetition in children with language impairment. *Clinical Linguistics & Phonetics*, 13(5), 369-380.
- Salameh, E. (2003). *Language impairment in Swedish bilingual children-epidemiological and linguistic studies*: Lund University.
- Salameh, E., Nettelblatt, U., & Gullberg, B. (2002). Risk factors for language impairment in Swedish bilingual and monolingual children relative to severity. [Research Support, Non-U.S. Gov't]. *Acta Paediatr*, 91(12), 1379-1384.
- Salameh, E., Nettelblatt, U., Håkansson, G., & Gullberg, B. (2002). Language impairment in Swedish bilingual children: a comparison between bilingual and monolingual children in Malmö. *Acta Paediatrica*, 91(2), 229-234.
- Shriberg, L. D., Tomblin, J. B., & McSweeny, J. L. (1999). Prevalence of speech delay in 6-year-old children and

- comorbidity with language impairment. *Journal of Speech, Language and Hearing Research*, 42(6), 1461.
- Snowling, M., Bishop, D., & Stothard, S. E. (2000). Is preschool language impairment a risk factor for dyslexia in adolescence? *Journal of Child Psychology and Psychiatry*, 41(5), 587-600.
- Snowling, M. J., Bishop, D. V., Stothard, S. E., Chipchase, B., & Kaplan, C. (2006). Psychosocial outcomes at 15 years of children with a preschool history of speech-language impairment. [Research Support, Non-U.S. Gov't]. *J Child Psychol Psychiatry*, 47(8), 759-765. doi: 10.1111/j.1469-7610.2006.01631.x
- Snowling, M. J., Muter, V., & Carroll, J. (2007). Children at family risk of dyslexia: a follow-up in early adolescence. *Journal of Child Psychology and Psychiatry*, 48(6), 609-618.
- Spaulding, T. J., Plante, E., & Farinella, K. A. (2006). Eligibility criteria for language impairment: Is the low end of normal always appropriate? *Lang Speech Hear Serv Sch*, 37(1), 61.
- Stark, R. E., & Tallal, P. (1981). Selection of children with specific language deficits. *Journal of Speech and Hearing Disorders*, 46(2), 114-122.
- Stokes, S. F., Wong, A. M. Y., Fletcher, P., & Leonard, L. B. (2006). Nonword repetition and sentence repetition as clinical markers of specific language impairment: The case of Cantonese. *Journal of Speech, Language, and Hearing Research*, 49(2), 219.
- Stothard, S. E., Snowling, M. J., Bishop, D., Chipchase, B. B., & Kaplan, C. A. (1998). Language-impaired preschoolers: A follow-up into adolescence. *Journal of Speech, Language, and Hearing Research*, 41(2), 407.

- Stromswold, K. (1998). Genetics of spoken language disorders. *Hum Biol*, 70(2), 297.
- Tallal, P., Hirsch, L. S., Realpe-Bonilla, T., Miller, S., Brzustowicz, L. M., Bartlett, C., & Flax, J. F. (2001). Familial aggregation in specific language impairment. [Research Support, U.S. Gov't, P.H.S.]. *J Speech Lang Hear Res*, 44(5), 1172-1182.
- Taylor, L. C., Clayton, J. D., & Rowley, S. J. (2004). Academic Socialization: Understanding Parental Influences on Children's School-Related Development in the Early Years. *Review of General Psychology*, 8(3), 163.
- Thordardottir, E., Kehayia, E., Mazer, B., Lessard, N., Majnemer, A., Sutton, A., . . . Chilingaryan, G. (2011). Sensitivity and specificity of French language and processing measures for the identification of primary language impairment at age 5. *Journal of Speech, Language and Hearing Research*, 54(2), 580.
- Tomblin, J. B. (2008). *Validating diagnostic standards for specific language impairment using adolescent outcomes. In Understanding developmental language disorders. From theory to practice.* Hove, East Sussex ; New York, NY.
- Tomblin, J. B., & Nippold, M. A. (2014). *Understanding individual differences in language development across the school years:* Psychology Press.
- Tomblin, J. B., Records, N. L., Buckwalter, P., Zhang, X., Smith, E., & O'Brien, M. (1997). Prevalence of specific language impairment in kindergarten children. [Research Support, U.S. Gov't, P.H.S.]. *J Speech Lang Hear Res*, 40(6), 1245-1260.

- Tomblin, J. B., Smith, E., & Zhang, X. (1997). Epidemiology of specific language impairment: prenatal and perinatal risk factors. *J Commun Disord*, 30(4), 325-343; quiz 343-324.
- Tomblin, J. B., Zhang, X., Buckwalter, P., & O'Brien, M. (2003). The Stability of Primary Language Disorder Four Years After Kindergarten Diagnosis. *Journal of Speech, Language, and Hearing Research*, 46(6), 1283-1296.
- Torgensen, J., Wagner, R., & Rashotte, C. (1999). Test of word reading efficiency (TOWRE). *Austin, TX: Pro-Ed*.
- Ullman, M. T., & Pierpont, E. I. (2005). Specific language impairment is not specific to language: The procedural deficit hypothesis. *Cortex*, 41(3), 399-433.
- Wass, M. (2009). *Children with Cochlear Implants: Cognition and Reading Ability*. PhD thesis, Linköping University, Sweden.
- Wass, M., Ibertsson, T., Lyxell, B., Sahlen, B., Hällgren, M., Larsby, B., & Mäki-Torkko, E. (2008). Cognitive and linguistic skills in Swedish children with cochlear implants—measures of accuracy and latency as indicators of development. *Scand J Psychol*, 49(6), 559-576.
- Wechsler, D. (1991). *WISC-III: Wechsler intelligence scale for children*: Psychological Corporation San Antonio, TX.
- Weindrich, D., Jennen-Steinmetz, C., Laucht, M., Esser, G., & Schmidt, M. (2000). Epidemiology and prognosis of specific disorders of language and scholastic skills. *Eur Child Adolesc Psychiatry*, 9(3), 186-194.
- Weismer, S. E., Tomblin, J. B., Zhang, X., Buckwalter, P., Chynoweth, J. G., & Jones, M. (2000a). Nonword repetition performance in school-age children with and without language impairment. *Journal of Speech, Language and Hearing Research*, 43(4), 865.

- Weismer, S. E., Tomblin, J. B., Zhang, X., Buckwalter, P., Chynoweth, J. G., & Jones, M. (2000b). Nonword repetition performance in school-age children with and without language impairment. *Journal of Speech, Language, and Hearing Research*, 43(4), 865-878.
- Vellutino, F. R., Fletcher, J. M., Snowling, M. J., & Scanlon, D. M. (2004). Specific reading disability (dyslexia): What have we learned in the past four decades? *Journal of Child Psychology and Psychiatry*, 45(1), 2-40.
- Vellutino, F. R., Scanlon, D. M., Sipay, E. R., Small, S. G., Pratt, A., Chen, R., & Denckla, M. B. (1996). Cognitive profiles of difficult-to-remediate and readily remediated poor readers: Early intervention as a vehicle for distinguishing between cognitive and experiential deficits as basic causes of specific reading disability. *J Educ Psychol*, 88(4), 601.
- Vellutino, F. R., Scanlon, D. M., Small, S., & Fanuele, D. P. (2006). Response to Intervention as a Vehicle for Distinguishing Between Children With and Without Reading Disabilities Evidence for the Role of Kindergarten and First-Grade Interventions. *J Learn Disabil*, 39(2), 157-169.
- Vernes, S. C., Newbury, D. F., Abrahams, B. S., Winchester, L., Nicod, J., Groszer, M., . . . Geschwind, D. H. (2008). A functional genetic link between distinct developmental language disorders. *New England Journal of Medicine*, 359(22), 2337-2345.
- Westerlund, M. (1994). Barn med tal-och språkavvikelser: en prospektiv epidemiologisk studie av en årskull uppsalabarn vid 4, 7 och 9 års ålder. *Avhandling för doktorexamen*). Uppsala: Uppsala Universitet.
- Westerlund, M., & Sundelin, C. (2000). Can severe language disability be identified in three-year-olds? Evaluation of a

- routine screening procedure. *Acta Paediatrica*, 89(1), 94-100.
- Whitehouse, A. J. (2010). Is there a sex ratio difference in the familial aggregation of specific language impairment? A meta-analysis. [Meta-Analysis]. *J Speech Lang Hear Res*, 53(4), 1015-1025. doi: 10.1044/1092-4388(2009/09-0078)
- Whitehouse, A. J., Bishop, D. V., Ang, Q., Pennell, C. E., & Fisher, S. E. (2011). CNTNAP2 variants affect early language development in the general population. *Genes, Brain and Behavior*, 10(4), 451-456.
- WHO. (2001). International Classification of Functioning, Disability and Health. Geneva *World Health Organization* (pp. 1-303).
- Viding, E., Spinath, F. M., Price, T. S., Bishop, D. V., Dale, P. S., & Plomin, R. (2004). Genetic and environmental influence on language impairment in 4-year-old same-sex and opposite-sex twins. [Research Support, Non-U.S. Gov't Twin Study]. *J Child Psychol Psychiatry*, 45(2), 315-325.
- Villanueva, P., Newbury, D. F., Jara, L., De Barbieri, Z., Mirza, G., Palomino, H. M., . . . Palomino, H. (2011). Genome-wide analysis of genetic susceptibility to language impairment in an isolated Chilean population. *European Journal of Human Genetics*, 19(6), 687-695.
- Zhang, X., & Tomblin, J. B. (2000). The association of intervention receipt with speech-language profiles and social-demographic variables. *American Journal of Speech-Language Pathology*, 9(4), 345.
- Zhu, X., Need, A. C., Petrovski, S., & Goldstein, D. B. (2014). One gene, many neuropsychiatric disorders: lessons from Mendelian diseases. *Nat Neurosci*, 17(6), 773-781.



