In this fifth chapter we will summarize the findings of the research presented in chapter 2, 3, and 4 in order to evaluate if and how well we could answer the research questions presented in the general introduction of the present thesis (chapter 1). Following this summary, chapter 6 will provide a general discussion and relevant conclusions of the research presented, debating the relevance of the introduced cognitive vulnerability markers of future reading failure in children with a family history of dyslexia.

The research in chapter 2 aimed to test the causality claim of the phonological deficit hypothesis of dyslexia by exploring the directionality of the relationship between kindergarten phonological awareness (PA) and grade 1 reading fluency in children with and without familial risk of dyslexia. The phonological deficit hypothesis proposes that difficulty with the representation, storage and/or retrieval of speech sounds, in other words a PA deficit, is the main deficit causing and preceding reading failure (Ramus, 2003; Snowling, 2000; Adams, 1990; Goswami & Bryant, 1990; Vellutino et al., 2004). There is ample support for this hypothesis across reading researchers and sufficient empirical evidence indicating a reciprocal influence between reading and PA (Morais, Cary, Alegria, & Bertelson, 1979; Perfetti et al., 1987). Nevertheless, clear evidence for the generally assumed PA-to-reading causality claim is surprisingly ever so elusive. Children at familial risk of dyslexia pose the best possible candidate for such an investigation before and after acquiring reading skills (i.e., kindergarten and grade 1 when formal reading instruction starts, respectively) since it is known that approximately 40% up to 65% of children with a least one first-degree dyslexic family member will develop a serious future reading deficit (Badian, McAnulty, Duffy, & Als, 1990; Blomert, 2005; Byrne, Shankweiler, & Hine, 2008; Pennington & Lefly, 2001; Shaywitz, Shaywitz, Fletcher, & Escobar, 1990; present thesis chapter 2). We addressed the following basic claims of the phonological awareness deficit:

1. **Is a phonological awareness (PA) deficit characteristic for a familial dyslexia risk?**

   Difficulties with PA did not characterize our sample of kindergarten children at familial risk of dyslexia. More specific, only a small proportion (i.e., 14%, 7 of 48) of these at-risk children showed a kindergarten PA deficit. Moreover, a kindergarten PA deficit was also not characteristic for at-risk children who turned out dyslexic (i.e., 19%, 4 of 21).
2. **Is a PA deficit causally linked to a reading deficit?**

Our results showed no support for the strong claim of the phonological deficit hypothesis that all children with a PA deficit prior to reading instruction turn out dyslexic. The large majority of the current at-risk sample who developed a reading deficit in grade 1 performed within the normal range on kindergarten phonological awareness (i.e., 81%, 17 of 21). Of all at-risk children, only 8% showed a PA deficit in kindergarten and subsequently developed a reading deficit in grade 1 (i.e., 4 out of 48). In our control group, only 4 children showed reading problems in grade 1 (i.e., 9%, 4 of 44) and none of them showed a preceding kindergarten PA deficit.

3. **Is 'phonological insensitivity' linked to a reading deficit?**

Difficulty with phonological processing skills other than phonological awareness (i.e., working memory and lexical processing) did not precede the development of subsequent reading deficits in the present at-risk and control cohort. More specific, only a small proportion of kindergarten at-risk children showed poor phonological lexical processing (i.e., 6%, 3 of 48) or poor phonological working memory (i.e., 2%, 1 of 48). In fact, the majority of at-risk children who developed a reading deficit revealed normal kindergarten lexical processing skills (i.e., 86%, 18 of 21) and normal kindergarten working memory (i.e., 95%, 20 of 21). Similar to at-risk, the proportion of control children with 'phonological insensitivity' in kindergarten also showing subsequent reading problems in grade 1 was low (lexical processing (i.e., 14%, 1 of 7 for lexical processing and 20%, 2 of 10 for working memory).

Taken together, these investigations of the direct claims of the phonological deficit hypothesis do not support that a PA deficit is a causal deficit preceding reading failure in dyslexia. Surprisingly, the phonological deficit hypothesis never proposed an explicit explanation how a PA deficit would in fact cause subsequent reading failure, although the hypothesis does suggest that a child need to acquire some level of PA before being able to link letters to speech sounds (Castles & Coltheart, 2004). One often assumed, indirect claim is that poor phonological representations lead to ‘unstable’ or poor letter-speech sound associations (LS) and as such in turn cause reading deficits (Snowling, 2000; Vellutino, Fletcher, Snowling, & Scanlon, 2004). The research in chapter 2 further investigated this claim:
4. Is a PA deficit causing later reading deficits by causing unstable or otherwise poor letter-speech sound associations?

We found little to no evidence that a PA deficit underlies reading failure as a result of some interference in the relations between letters and their corresponding speech sounds since a PA deficit and a LS deficit rarely accompanied each other in the same kindergarten at-risk children (i.e., 4%, 2 of 48) and never in control children. This was confirmed by a weak concurrent correlation in kindergarten between de PA and LS measures. Instead, PA deficits emerged only after the beginning of reading instruction in grade 1 and only in those at-risk and control children who developed a reading deficit by then (67%, 14 of 21 at-risk children and both reading-disabled control children). The development of letter-speech sound associations was by then well on its way. This finding indicated that a PA deficit seems to develop as a consequence or at least in parallel with the reading deficit.

These results indicated that a PA deficit in the present kindergarten cohort is not causally linked to a LS deficit. In fact, a PA deficit only emerged in grade 1, when children with a reading deficit already developed some level of letter-speech sound knowledge. Since successfully acquiring alphabetical skills is considered the essence of adequately learning to read (Ehri, 2005), the research in chapter 2 lastly explored whether it is a LS deficit, or difficulties with learning the associations between letters and their corresponding speech sounds, that is characteristic of children at increased familial risk of dyslexia. To investigate a potential LS learning deficit, we used a child-friendly, computerized training game aimed at teaching the links between letters and speech sounds to Dutch kindergartners (for an elaborate description of the original Finnish Graphogame method see Richardson & Lyttinen, 2014, pp.; for an elaborate description of the present, Dutch adaptation of the training see Appendix 1, present thesis). If LS learning poses a problem in children at increased risk, we would expect a difference in training effects between these children and control children.

5. Can children at-risk for dyslexia at pre-reading age be differentiated from control subjects on the basis of their letter-speech sound learning ability on a training specifically designed to accurately teaching these associations?

While all of our control children improved their LS skills due to training, more than one third of at-risk children did not profit from training, showing LS learning problems in kindergarten. Interestingly,
although this LS learning resistant at-risk group did not improve their ability to actively couple letters to their corresponding speech sounds, they did improve their letter knowledge, outperforming their untrained counterparts, similar to the trained control children. Also, both at-risk and control children reached ceiling on letter knowledge at a similar pace within half a year of reading instruction in grade 1. Our results finally demonstrated that this kindergarten LS learning deficit revealed in a core at-risk group was not related to a later reading deficit in grade 1. More specific, (a) only a quarter to a third of at-risk LS training resisters actually developed a reading deficit in grade 1 and (b) the proportion of at-risk LS training resistant children and at-risk LS training respondent children with a grade 1 reading deficit did not differ.

Chapter 3 further investigated if PA and LS in addition to the ability to rapidly name highly familial visual objects (RAN) were predictively correlated to subsequent reading in children at familial risk of dyslexia. These three important reading-related markers are known to be uniquely correlated to reading outcome in normally developing children (e.g., de Jong & van der Leij, 1999; Kirby, Georgiou, Martinussen, & Parrila, 2010; Landerl et al., 2013; Lyytinen et al., 2004a; Lyytinen et al., 2004b; Schatschneider, Fletcher, Francis, Carlson, & Foorman, 2004; Schatschneider & Torgesen, 2004). It is unclear however whether predicting reading success is the same as predicting high risk reading outcome. Identifying early risk markers predicatively correlated to at-risk reading outcome is vital given to possibility to promote more accurate and effective early diagnostic and intervention efforts. Moreover, since we explored the correlation between these cognitive markers and reading outcome both in kindergarten and grade 1, we could explore possible changes in these predictive relationships as a consequence of acquired reading context. Finding a differential predictive pattern in the current sample would confirm the findings of chapter 2 that PA deficits emerged only after the start of reading acquisition.

6. Are kindergarten PA, RAN and LS knowledge important predictors of subsequent grade 1 and 2 reading ability in at-risk children learning to read in a relatively transparent orthography?

LS knowledge was the only kindergarten reading-related marker correlated to grade 1 reading fluency in our at-risk sample. Kindergarten RAN was correlated to at-risk reading outcome in grade 2. PA in kindergarten was not correlated to subsequent at-risk reading outcome in first or second grade.
7. Does the emerging correlation pattern in kindergarten change as a result of acquiring basic literacy skills in grade 1 (i.e., grade 1 prediction of grade 2 reading fluency)?

Grade 1 results indicated that LS knowledge remained the only marker correlated to reading fluency of children at increased familial risk for dyslexia a year later. The correlation between kindergarten and grade 1 LS knowledge and reading outcome of at-risk children in our sample, combined with the finding that kindergarten RAN was predicatively related to grade 2 at-risk reading, was interpreted as a possible early cross-modal orthographic-phonological association deficit that was already noticeable in at-risk children prior to the start of reading instruction.

8. Is the correlation pattern found over kindergarten and grade 1 in children at-risk for a deficient reading development comparable to the pattern found in their normally developing peers without such a family history of dyslexia?

Before the start of reading instruction, the at-risk and control children both shared LS knowledge as the sole kindergarten marker correlated to subsequent reading outcome in grade 1. After the start of reading instruction in grade 1, we saw that LS knowledge remained predictive of grade 2 reading in our at-risk sample. At that point, PA emerged as the sole predictor in our control sample. RAN showed no predictive relationship after reading instruction started in both groups. Together, these results revealed that the relevance of kindergarten predictive markers changed as a result of acquiring reading skills, specifically in those children without risk.

Our findings of chapter 3 indicated that once reading instruction started a differential correlation pattern between potential predictive risk markers and reading outcome emerged for children with and children without a familial risk for dyslexia. This is possibly due to a different developmental pace of reading acquisition in the former at-risk group, in which early identification of subsequent reading failure is most evident and necessary. Our collective results of chapter 2 and 3 indicate that PA, LS and RAN are all relevant markers of at-risk and normal reading outcome. However, the contribution of these markers seems to differ on the developmental stage of the reading acquisition process that is reached, which in turn seems to vary with the presence or absence of familial risk for dyslexia (for similar results see, Cardoso-Martins & Pennington, 2004; Pennington & Lefly, 2001). Chapter 4 therefore further investigated to which extent these reading-related vulnerability markers differentiate within a large, heterogeneous sample of already established reading disabled children with or without such a familial
predisposition to the experienced reading failure. To this aim, chapter 4 explored whether we could identify distinct patterns of cognitive deficits underlying reading problems based on these three important reading-related markers, next to working memory and general cognitive abilities (non-verbal IQ and receptive vocabulary). A data-driven clustering attempt was conducted in a large heterogeneous sample that includes slightly older poor reading children attending second to fourth grade. We investigated whether emerging cognitive subtypes of poor reading are specifically characterized by an increased familial risk of dyslexia or instead by more environmental risk factors (i.e., socio-economic status, reading performance level of attended schools, or co-morbidity factors like arithmetic deficits and ADHD). In doing so, the present study was an application of the first bottom-up classification taking into account recent major genetic advances in the reading field that indicate that familial risk undermines the development of reading in dyslexic families (e.g., Grigorenko, 2001; Castles, Datta, Gayan, & Olson, 1999). The questions addressed in chapter 4 were:

9. Can a data-driven classification based on the cognitive reading-related vulnerability markers PA, RAN, LS processing and VWM differentiate distinct and coherent cognitive deficit profiles within a large sample of grade 2 to 4 poor readers?

Our results revealed four distinguishable clusters of reading disability, specifically three of which showed a distinct underlying cognitive deficits pattern in addition to below average spelling scores, while a fourth subtype showed no cognitive deficits despite their reading failure. This latter reading-only impaired reader type, although performing below average on reading fluency, showed significantly less impaired reading ability than the other three poor reader types and was the only type with average range spelling skills. The other three subtypes shared a PA deficit but showed a variable and discriminative expression across the other included cognitive vulnerability markers: (a) general poor readers with deficits across all reading-related and general cognitive markers, (b) PA-LS specific poor readers with an additional LS deficit next to the common PA deficit and (c) PA-RAN specific poor readers with an additional RAN deficit besides PA problems.
10. Are possible emerging cognitive profiles of deficits underlying different types of poor reading distinguishable based on genetic influences, environmental influences or a combination?

Although only genetic testing (e.g., twin study designs) can detangle how much variation in a trait is related to genes or environmental factors, and familial risk of dyslexia is not a pure genetic trait since both genes and environments are shared by families (Pennington & Olson, 2008; Plomin, Reiss, Hetherington, & Howe, 1994; Rutter & Silberg, 2002), it is one of the strongest predictors of the development of reading disorders (Scarborough, 1989, 1990; Snowling, Gallagher, & Frith, 2003; Thompson et al., 2015). The results in the current poor reader sample revealed that the general poor readers and the PA-LS specific poor readers were characterized most by poor socio-economic status, while the PA-RAN poor readers showed a very strong familial risk of dyslexia. Interestingly, this familial risk PA-RAN subtype showed high IQ scores discrepant to their reading skills, while the other two more environmentally characterized types showed relatively poorer intellectual skills (i.e., approximately 1 standard deviation below the PA-RAN subtype). The fourth reading-only impaired showing a less severe reading deficit compared to the other three subtypes and being the only poor reader type without spelling difficulties were significantly younger although they did not attend a significantly lower school grade. This indicates that their exposure to certain levels of grade specific reading instruction was comparable to that in the other subtypes and therefore could not have been a factor explaining their reading difficulties. The children in this subtype did however attend schools with a significantly poorer overall reading level. Taken this into account, together with their less severe and non-specific (i.e., no underlying cognitive deficit pattern) reading problems and average spellings skills, indicate that these children are influenced by external factors instead of a specific altered cognitive development. Reading difficulties as a result of less adequate reading instruction at school seems more easily counterattacked by more adequate or intensive instruction.

Together, the research described in chapter 4 identified four homogeneous and distinguishable clusters within the current poor reading sample, which strengthened the notion that different cognitive subtypes of reading disability exist (for previous clustering attempts see for example Morris et al., 1998; King et al., 2006). Our results moreover show that data-driven classification solely based on cognitive vulnerability markers commonly used for the diagnostics of dyslexia can differentiate between poor reading as a result of increased familial risk for dyslexia and reading failure as a result of poor environmental factors. This confirms previous neuroimaging results indicating a primarily genetic poor
readers type and an environmentally influenced impaired type (Shaywitz & Shaywitz, 2005). Moreover, although diagnostics of dyslexia solely based on an IQ-reading discrepancy cut-off criteria has been discarded over the past two decades (Stanovich & Siegel, 1994; Stuebing et al., 2002), our results did indicate that the way IQ forms distinct patterns with cognitive and familial risk markers seems informative. More specific, our findings supported the idea that IQ can provide more information on the cause of the reading deficit experienced by the child (Friend, DeFries, & Olson, 2008; Wadsworth, Olson, & DeFries, 2010; Wadsworth, Olson, Pennington, & DeFries, 2000) as well as have a valuable attribution for exclusion purposes in diagnostic processes (Coltheart & Jackson, 1998; Rack & Olson, 1993). Being able to identifying homogeneous subtypes within the heterogeneous reading disabled group is a promising way forward for future neuroimaging and genetic studies as well as future diagnostic and remediation attempts. Focusing on meaningful subtypes creates the possibility to design research and intervention efforts more specifically and avoids potential downfalls of generalizing based on a cognitively mixed group.
References


