Who will develop dyslexia? Cognitive precursors in parents and children

van Bergen, E.

Citation for published version (APA):
Writing – the art of communicating thoughts to the mind, through the eye – is the great invention of the world. Great, very great in enabling us to converse with the dead, the absent, and the unborn, at all distances of time and space; and great, not only in its direct benefits, but greatest help, to all other inventions.

- Abraham Lincoln -
At this very moment, precisely controlled eye movements make you process a few black marks on a white page from the general introduction of this thesis. The message I convey from my brain to your brain is in fact coded as just a collection of straight and curved lines. Yet such a collection of tiny lines can transfer knowledge or bring a story to life. Our ability to read and write is an impressive feat and life today is impossible to imagine without it.

From an evolutionary perspective, reading and writing are relatively recent inventions. Homo sapiens have existed for 200,000 years, but reading and writing dates back roughly 5,000 years (Powell, 2009). Nevertheless, it has been only some 100 years since nearly all people in Western societies have become literate. It probably does not come as a surprise that the start of compulsory education, and hence reading education for all, roughly coincides with the birth of its study, the science of reading.

From a neuroscientific perspective, simply reading a single word is already a very complex task, let alone reading this thesis. Learning to read is not something that comes naturally to us, like learning to walk or talk. Instead, it takes a couple of years of deliberate practice. But once mastered, we recognize words instantly. A 10-year old skilled reader can read a word that is flashed on a screen for just 200 milliseconds (Yap & van der Leij, 1993). On seeing the word a cascade of orchestrated neural firing is triggered, making us recognize the black marks as a meaningful word. The beauty of it is that this all happens very quickly and automatically; you cannot help but read.

1.1 Dyslexia

In view of the complexity of word recognition it is perhaps not surprising that not all children learn to read without difficulty. Although the exact definition and diagnostic criteria of dyslexia are still hotly debated (e.g., Brown Waesche, Schatschneider, Maner, Ahmed, & Wagner, 2011; Fletcher, Coulter, Reschly, & Vaughn, 2004; Kleijnen et al., 2008; Snowling & Hulme, 2012), there is consensus among researchers and practitioners that individuals with dyslexia experience difficulty in mastering word-level reading skills and show persistent problems in the accuracy or fluency of word-reading. These problems in word-level reading often go hand in hand with problems in word-level spelling.

Throughout this thesis I will use ‘reading’ to refer to word-level reading, rather than reading comprehension.
There are large individual differences in reading fluency, that is, how quickly and accurately single words are read. Individuals with dyslexia are at the bottom of the distribution of normal variation in reading fluency. Slow speed of processing and relatively high error rates indicate their lack of automatic decoding (van der Leij & van Daal, 1999). It can readily be seen that the prevalence rate of dyslexia reflects the cut points established as criteria for identification, as well as the exclusion criteria that are used. According to the criteria proposed in the Dutch Dyslexia protocol, it is estimated that 4% of Dutch primary school children have dyslexia (Blomert, 2006).

Children with dyslexia might experience great difficulty in education. After the initial stage of learning acquisition, an educational transition takes place from learning to read to reading to learn. Struggling with the task of reading itself hampers a child with dyslexia to employ reading to learn about for example science or history. Indeed, dyslexia negatively impacts long-term academic outcomes (Willcutt et al., 2007). Moreover, dyslexia is associated with increased risks of anxiety disorders, depression, attention-deficit/hyperactivity disorder (ADHD), conduct disorder (Bosman & Braams, 2005; Carroll, Maughan, Goodman, & Meltzer, 2005; Willcutt et al., 2007), and even criminal behaviour (Kirk & Reid, 2001; Macdonald, 2012).

Given the trouble that dyslexia might cause, it is very important to study in what way individuals with dyslexia differ from their peers without dyslexia. The ultimate goal is to unravel the causal pathway of aetiological risk factors leading to dyslexia. Insight into these pathways will inform the development of effective interventions to ameliorate the impact of dyslexia.

From research comparing individuals with and without dyslexia, we know that those with dyslexia generally show deficits in specific cognitive areas, like the processing of speech sounds in spoken words. Finding a cognitive deficit linked to dyslexia raises the question as to whether the deficit is a consequence of children’s reading problems or whether it was already present before they came to the task of learning to read. If so, the deficit is said to be a precursor of dyslexia. Furthermore, to better understand the developmental pathways we need to find out whether a precursor of reading ability or disability is specifically linked to reading development or whether it is shared with for instance arithmetic development. Beyond studying these cognitive characteristics intrinsic to the child, the current research will explore a new avenue by investigating the cognitive risks that parents pass on to their child.
1.2 Theoretical Framework

The issues pursued in the current thesis were guided by previous empirical work on cognitive deficits associated with dyslexia and by theoretical work, specifically the multiple deficit model and the generalist genes hypothesis. This theoretical framework will be discussed in the following two sections, concluded by a third section about the proposed hybrid model.

1.2.1 The Multiple Deficit Model

Research into dyslexia was dominated for a long time by the quest for the Holy Grail: the single cognitive deficit that is necessary and sufficient to cause all behavioural characteristics of the disorder. In the case of dyslexia the dominant hypothesis about a single causal factor has been the phonological-deficit hypothesis (e.g., Snowling, 1995; Wagner, 1986); other hypotheses include core deficits in rapid auditory processing (Tallal, 1980), amplitude-envelope rise-time discrimination (Goswami et al., 2002), visual attention span (Bosse, Tainturier, & Valdois, 2007), visuo-spatial attention (Vidyasagar & Pammer, 2010), magnocellular processing of fast sensory information (Stein & Walsh, 1997), the ability to perform skills automatically due to cerebellar deficits (Nicolson, Fawcett, & Dean, 2001), and the forming of stimulus-specific anchors (Ahissar, 2007).

However, single cognitive deficit models have a number of shortcomings (see Pennington, 2006, for a comprehensive overview). First, there is no single cognitive deficit found that can explain all behavioural symptoms of all cases with dyslexia. For example, not all individuals with dyslexia show a phonological deficit (e.g., Pennington et al., 2012; Valdois et al., 2011), which is the main candidate for a single cognitive deficit explanation. Conversely, not all individuals with a phonological deficit have dyslexia (e.g., Bekebrede, van der Leij, Schrijf, & Share, 2010; Snowling, 2008). This questions a one-to-one mapping and points to the possibility that various constellations of underlying cognitive deficits can lead to the behavioural symptoms of dyslexia.

Second, these models cannot readily explain the phenomenon of comorbidity. In the case of dyslexia, comorbidity refers to the fact that dyslexia co-occurs more often than expected by chance with other developmental disorders, like dyscalculia, specific language impairment, speech-sound disorder, or ADHD. To illustrate this point, suppose disorder A and B each have a prevalence of 5% in the general population. If disorder A and B were
independent, then the chance that A and B co-occur would only be 0.05 * 0.05 = 0.0025, or 0.25%. However, comorbidity rates for developmental disorders are commonly in the order of 30%, for example between dyslexia and speech-sound disorder or dyslexia and ADHD (Pennington, 2006). The huge discrepancy between these figures (0.25% vs. 30%) implies that developmental disorders are not independent.

The single deficit model requires for each comorbidity (pair of disorders) a distinct account. Pennington (2006) discusses as an example the comorbidity between dyslexia and speech-sound disorder. Speech-sound disorder is defined by difficulties in the development of spoken language, especially problems with the intelligible production of speech sounds. Approximately 30% of children with early language or speech problems go on to develop dyslexia. A parsimonious single deficit model to explain this comorbidity is the severity hypothesis. The severity hypothesis states that speech-sound disorder and dyslexia have the same underlying phonological deficit, with speech-sound disorder being an earlier developmental manifestation of this deficit than dyslexia. Comorbid cases will have the most severe phonological deficit. If the phonological deficit is less severe, speech-sound disorder will not reach clinical boundaries but dyslexia will. To account for cases with early speech-sound disorder but without later dyslexia, the model must pose a subtype of speech-sound disorders that is caused by a phonological deficit distinct from the phonological deficit as seen in cases with dyslexia. Alternatively, the phonological deficit in such cases must be resolved by the time they come to the task of learning to read. However, Snowling, Bishop, and Stothard (2000) followed a group of former language-impaired children into adolescence. Those with early speech-sound disorder (isolated phonological impairments at 4 years of age) had normal reading skills at age 15, but continued to show phonological deficits. Similar results were obtained by Peterson, Pennington, Shriberg, and Boada (2009). In their study many children with early speech-sound disorder went on to learn to read normally despite a lasting phonological deficit. Thus, in both studies the children with early speech-sound disorder had a phonological deficit similar to children with dyslexia. This conclusion is inconsistent with the single cognitive deficit severity hypothesis.

While research at the cognitive level of explanation was still searching for a single deficit, studies at the genetic level converged on the conclusion that the aetiology of dyslexia and other developmental disorders is genetically
complex (Pennington, 2006). So instead of a single gene determining dyslexia, many genes act probabilistically, each having only a small contributory effect to the aetiology of dyslexia (Bishop, 2009). Moreover, behavioural genetic studies showed for certain developmental disorders that the relation between two traits (like reading ability and inattention) is larger in monozygotic twin pairs than in dizygotic twin pairs (Willcutt, Pennington, & DeFries, 2000). Such a bivariate heritability supports genetic overlap between the conditions, in this example between dyslexia and ADHD. The partly shared aetiology of dyslexia and ADHD does not yet rule out the possibility of a distinct single cognitive deficit for each disorder. However, studies have demonstrated that a processing speed impairment is not only a characteristic of dyslexia, but also of ADHD (e.g., Willcutt, Pennington, Olson, Chhabildas, & Hulslander, 2005), suggesting that processing speed is a shared cognitive risk factor (McGrath et al., 2011). The accumulating evidence for aetiological and cognitive overlap between dyslexia and ADHD speaks against a single deficit model for explaining their frequent co-occurrence. Also for other dyslexia comorbidities, shared cognitive deficits are found, for example a phonological deficit in specific language impairment (e.g., Bishop, McDonald, Bird, & Hayiou-Thomas, 2009) and a processing-speed deficit in dyscalculia (e.g., van der Sluis, de Jong, & Leij, 2004).

It seems that single deficit models are untenable and must give way to multiple cognitive deficit models for understanding developmental disorders. The multiple cognitive deficit model proposed by Pennington (2006) is depicted schematically in Figure 1.1. In his model, multiple genetic and environmental risk factors operate probabilistically by increasing the liability to a disorder; conversely, protective factors decrease the liability. These aetiological factors produce the behavioural symptoms of developmental disorders by influencing the development of relevant neural systems and cognitive processes. Importantly, there is no single aetiological or cognitive factor that is sufficient to cause a disorder. Instead, multiple cognitive deficits (each due to multiple aetiological factors) need to be present to produce a disorder at the behavioural level. Some of the aetiological and cognitive risk factors are shared with other disorders. Consequently, comorbidity among developmental disorders is to be expected, rather than something that requires additional explanations. Finally, from Pennington’s multiple deficit model it follows that the liability distribution for a given disorder is continuous and quantitative, rather than being discrete and categorical. Therefore, thresholds to define disorders are rather arbitrary.
Figure 1.1. Pennington's multiple deficit model. Double headed arrows indicate interactions. Causal connections between levels of analysis are omitted.

G = genetic risk or protective factor
E = environmental risk or protective factor
N = neural system
C = cognitive process
D = disorder
Pennington (2006) concludes his paper by remarking that it remains challenging to test the multiple cognitive deficit model. The model is much more complex than single deficit models, which are attractively parsimonious, but this complexity is needed to account for the observations at the different levels of analysis. The model is universally applicable to developmental disorders, but therefore remains abstract. It is not specified which aetiological factors, neural systems, and cognitive processes interact to produce a given disorder.

We would like to argue that a line of inquiry that can contribute to testing and specifying the multiple deficit model are family-risk studies. In family-risk studies, children are followed who are at risk of dyslexia by virtue of having an immediate dyslexic family member (usually a parent). Such studies have shown that 34 to 66% of them develop dyslexia (Elbro, Borstrøm, & Petersen, 1998; Pennington & Lefly, 2001; Scarborough, 1990; Snowling, Gallagher, & Frith, 2003; Torppa, Lyytinen, Erskine, Eklund, & Lyytinen, 2010), depending on the stringency of the dyslexia criteria. The much higher prevalence of dyslexia among offspring of parents with dyslexia is consistent with twin studies showing moderate to strong heritability of dyslexia (e.g., Olson, Byrne, & Samuelsson, 2009).

From the multiple deficit model it follows that children at family risk experience at least some of the aetiological risk factors: they inherit genetic risk factors and might experience a less rich literacy environment. Hence, it is hypothesized that at-risk children have a higher genetic and environmental liability than children without a family history of dyslexia (labelled control children). Furthermore, the at-risk children who go on to develop dyslexia are expected to show cognitive deficits in several processes. Some of these cognitive processes are expected to be affected even before the onset of reading instruction, as a consequence of aetiological risk factors and deficient neural systems.

A key prediction of the multiple deficit model for family-risk studies concerns the at-risk children who do not develop dyslexia. If liability to dyslexia were discrete (as would happen if only one factor, say a gene, were involved), at-risk non-dyslexic children would not differ from controls. However, according to the multiple deficit model, liability is continuously distributed. This also follows from the fact that reading ability is influenced by many genes of small effect, producing normal distributions of phenotypes (Plomin, DeFries, McClearn,
McGuffin, 2008, p. 33). Consequently, the multiple deficit model predicts that at-risk children without dyslexia also inherit at least some disadvantageous gene variants from their dyslexic parents, giving them a higher liability than control children, although still lower than at-risk dyslexic children. At the behavioural and cognitive level this should translate into mild deficits in literacy skills and some of its cognitive underpinnings. When plotting mean performances of the three groups, a step-wise pattern (i.e., at-risk dyslexic < at-risk non-dyslexic < controls) would support a continuum of liability, one of the characteristics of the multiple deficit model.

Comparing the three groups of children on behavioural measures sheds light on cognitive deficiencies and behavioural symptoms, the bottom two levels in Figure 1.1. These three groups have also been compared on neural processing of visual and auditory stimuli (e.g., Leppänen et al., 2010; Plakas, van Zuijen, van Leeuwen, Thomson, & van der Leij, 2012; Regtvoort, van Leeuwen, Stoel, & van der Leij, 2006), the second level of the multiple deficit model. Some family-risk studies have also examined aspects of the home environment, which belong to the aetiological level. However, specific genetic risk factors remain hidden in family-risk studies. As genetic screening of children for their dyslexia susceptibility is still far away, we propose an indicator of their genetic risk. Since reading ability is moderately to highly heritable and children receive their genetic material from their parents, we argue that cognitive abilities of parents can partly reveal their offspring’s liability. The parents of at-risk children will have weaker reading skills than those of control children, reflecting selection criteria in family-risk studies, but the key issue is whether parental reading skills differentiate between at-risk children with and without dyslexia. Based on the multiple deficit model it is expected that at-risk children who develop dyslexia have inherited more genetic risk variants than at-risk children without dyslexia and that this difference can be revealed by lower reading performance of parents of the at-risk dyslexic children. In Section 1.4.2 I will elaborate upon parental effects.

1.2.2 The Generalist Genes Hypothesis
In our family-risk study we seek to identify cognitive processes playing a role in the developmental pathways that lead to dyslexia. The multiple deficit model states that some cognitive deficits are shared among disorders. This raises the question of which cognitive precursors of dyslexia are distinct and which are shared with other disorders. With regard to learning abilities, like reading
ability, there is a hypothesis that addresses this specificity issue: the generalist genes hypothesis (Kovas & Plomin, 2007; Plomin & Kovas, 2005).

The generalist genes hypothesis stems from behavioural genetic studies employing the twin design. The twin design is the major method to quantify genetic and environmental influences on a trait. It compares the resemblance of monozygotic twins, who are genetically identical, with dizygotic twins, who are on average 50% similar genetically. If for a certain trait monozygotic twins are more similar than dizygotic twins, genetic factors must play a role. If there is no difference in resemblance heritability is negligible. For dyslexia, the concordance rate (that is, the likelihood that one twin will be affected if the other twin is affected) is 75% for monozygotic twins and 43% for dizygotic twins (Stromswold, 2001), suggesting substantial heritability. Estimates for the heritability of reading ability are in the range of .47 to .84 (Taylor, Roehrig, Hensler, Connor, & Schatschneider, 2010, and Byrne et al., 2009, respectively).

Recently, the field of behavioural genetics has moved beyond quantifying genetic and environmental influences on a trait to studying genetic and environmental overlap between traits. For the three learning abilities reading, arithmetic, and language, empirical data have shown that the genes important for one learning ability largely overlap with the genes important for the other learning abilities. The genetic correlation is the measure that quantifies this: it indexes the extent to which genetic influences on one trait correlate with the genetic influences on another trait (independently of the heritability of the traits). The genetic correlation between learning abilities is about .70 (Kovas & Plomin, 2007; Plomin & Kovas, 2005). This suggests that half (0.70^2) of the genes associated with reading ability are generalists: they also influence other learning abilities. Hence Plomin and Kovas (2005) named their hypothesis the ‘generalist genes hypothesis’. As genetic correlations are not 1.0, there are also specialist genes: genes that contribute to dissociations among learning abilities. However, most genes associated with reading are generalists, also in another respects: twin studies have demonstrated that genetic influences on the lower end of the reading distribution (containing children with dyslexia) are largely the same as the genetic influences on the entire distribution (Kovas, Haworth, Dale, & Plomin, 2007; Plomin & Kovas, 2005).

Observed differences in learning abilities among individuals are also partly due to differences between the environments in which individuals were born, were brought up and live. Behavioural genetics subdivides
environmental influences into those that make family members similar (called shared environmental effects) and those that do not contribute to resemblance among family members (called non-shared environmental effects). Shared environmental effects include shared family experiences or sharing the same school or community. Examples of non-shared environmental effects are accidents, illnesses, hobbies, and influences of individuals such as peers and teachers. Also for these environmental components statistics exist analogous to genetic correlation. Shared environmental correlations among learning abilities are as high as genetic correlations, so shared environmental effects are also largely general effects (Kovas & Plomin, 2007). In contrast, non-shared environmental correlations are low. This indicates that these effects primarily act as specialists, contributing to performance differences in learning abilities within a child (Kovas & Plomin, 2007).

1.2.3 The Hybrid Model
The generalist genes hypothesis and the multiple cognitive deficit model complement each other well. The multiple deficit model is more general because it holds for all common developmental disorders, while the generalist genes hypothesis specifically pertains to learning abilities and disabilities. Furthermore, the multiple deficit model includes four levels of explanation, whereas the generalist genes hypothesis only concerns the aetiological level. Nevertheless, the generalist genes hypothesis details for learning abilities the degree of overlapping and unique influences in each of the three aetiological components (genetical, shared environmental, and non-shared environmental influences). I have visualised the generalist genes hypothesis and incorporated it into the multiple deficit model, yielding the hybrid model depicted in Figure 1.2. In this model only the first and the fourth layer are further specified because the generalist genes hypothesis only deals with these two levels. The aetiological factors of the first level influence the behavioural manifestations at the fourth level by acting through the second and third level.

The hybrid model quantifies the overlap in aetiological factors between learning abilities: genetic and shared environmental effects are largely shared by the three learning domains, whereas the non-shared environmental effects are largely distinct. These differential overlaps are visualised in the hybrid
Figure 1.2. Hybrid model for learning (dis)abilities, consisting of the generalist genes hypothesis (Kovas & Plomin, 2007) and the multiple deficit model (Pennington, 2006). Double headed arrows indicate interactions. Causal connections between levels of analyses are omitted.

N = neural system
C = cognitive process
RD = reading disorder; AD = arithmetic disorder; LD = language disorder
model as the degree of overlap between the circles. Despite this quantification of aetiological overlap, the hybrid model does not specify which aetiological factors are relevant. Regarding genetic factors, molecular genetic studies will ultimately inform us which genes are implicated in dyslexia. Knowledge of specific genes contributing to dyslexia susceptibility promises to help bridge the gap from genes to neural systems, cognitive processes, and behavioural outcomes (Fisher & Francks, 2006). Until now, only a handful of candidate genes are identified, each of small effect. As an example of the small effects, the most common form of the candidate gene KIAA0319 is found equally frequent in people with and without dyslexia. The high-risk version is found in 35% of those with dyslexia and 27% of those without, whereas the figures for the low-risk version are 24% versus 36% (Bishop, 2009).

Insight into which specific neural systems, cognitive skills, and behavioural symptoms are implicated in dyslexia can be gained from family-risk studies. The hybrid model points to the opportunity to study reading in combination with arithmetic or language to increase insight into shared and distinct factors. We chose to focus on reading and arithmetic, both basic school skills central during early primary school. As the model suggests, its disorders, dyslexia and dyscalculia, indeed often co-occur (Landerl & Moll, 2010). Moreover, this pair of comorbidity is under-researched compared to the comorbidity of dyslexia with ADHD or language disorders. We aimed to study the comorbidity issue at the cognitive level of explanation. Which cognitive factors are specific and which are shared between the development of reading and arithmetic?

1.3 Precursors in Children

This thesis will deal with two types of cognitive skills: general cognitive skills and preliteracy skills. The key questions regarding cognitive precursors in children are: Do the children who go on to develop dyslexia show cognitive deficits before they come to the task of learning to read? And do the at-risk children who do not develop dyslexia perform at the same level as the control children?

Reading performance is the outcome of the combined action of multiple factors, as depicted in Figure 1.2. Knowledge about these factors can
be obtained by studying predictors of reading outcome. In addition, knowledge about predictors of dyslexia is of clinical importance because it enables early identification and therefore remedial teaching of children at heightened risk of dyslexia.

1.3.1 IQ
Reading ability and IQ are correlated. Therefore, when selecting a group of children with dyslexia based on low reading achievement, it can be expected that their group mean on IQ falls below that of a group of children who do not show reading problems. However, scores on IQ tests might be adversely affected by reading problems and, as a result, less print exposure or print exposure to less complex texts (Cunningham & Stanovich, 1997). Consider for instance conceivably poorer performance on the Verbal Comprehension Index (including vocabulary, similarities, comprehension, information, and word-reasoning subtests) of the WISC (Wechsler, 2004), a world-wide used IQ battery. Thus, the correlation between reading ability and IQ might partly be a consequence of reading ability. Therefore, longitudinal data from before and after reading onset are required. The only two previous family-risk studies that report IQ before Grade 1 (Snowling et al., 2003; Torppa et al., 2010) report that the at-risk group with later dyslexia performed lower on verbal IQ, but the studies are inconsistent regarding nonverbal IQ. Moreover, IQ scores are given but the papers are silent about their interpretation.

A partly causal relation between early verbal IQ and later reading development is plausible, since the cognitive system for written language builds on the cognitive system for oral language (Hulme, Snowling, Caravolas, & Carroll, 2005). Regarding nonverbal IQ, if it is associated with later reading, a causal interpretation is less self-evident. It might be that nonverbal IQ indicates a child’s ability to acquire cognitive skills in general. It cannot be readily seen how this general ability translates into a specific problem in learning to decode words. Alternatively, a possible relation between early nonverbal IQ and later reading might be explained by a third common underlying variable. It might be that both abilities share aetiological factors, as do different domains of learning abilities according to the generalist genes hypothesis (see Figure 1.2). Chapter 5 and Chapter 6 (General Discussion) will further discuss the interpretation of longitudinal relations between IQ and reading.

Chapter 5 aims to investigate the longitudinal relations between IQ
and reading (dis)ability by examining IQ performance at age 4 and reading outcome at age 8 years. Another goal of this chapter is to investigate the specificity of the potential predictive relation between early IQ and later reading. Are early verbal and nonverbal IQ uniquely related to later fluency in word reading or also to later fluency in mental arithmetic? As mentioned previously, word reading and mental arithmetic are arguably the most important basic school skills acquired during the early school years. Moreover, impairments in acquiring these skills (i.e., dyslexia and dyscalculia, respectively) often co-occur (Landerl & Moll, 2010), making them a good pair of skills to study shared and distinct links with early IQ. As can be seen in the hybrid model (Figure 1.2), the aetiological and cognitive factors influencing reading and arithmetic are assumed to be partly shared and partly unique. Investigating the relationship of reading and arithmetic with early IQ offers insight into whether verbal and nonverbal ability are cognitive factors related to these school skills and if so, whether they are shared between the school skills.

1.3.2 Preliteracy Skills

Besides studying general cognitive abilities we investigated specific cognitive abilities before the start of reading education. The cognitive skills that are thought of as causally linked to subsequent development of decoding skills and later development of fluent reading are phonological skills and knowledge of grapheme-phoneme –or letter-sound– correspondence (e.g., Hulme, Bowyer-Crane, Carroll, Duff, & Snowling, in press). A third ability found to be a powerful longitudinal (and concurrent) predictor of reading outcome is the ability to rapidly name a matrix of items such as colours, objects, or once mastered, digits or letters (e.g., de Jong & van der Leij, 1999; de Jong & van der Leij, 2003; Lervåg, Bråten, & Hulme, 2009).

The main indicator of phonological skills is phonological awareness, which refers to the ability to detect and manipulate sounds in spoken words, like phonemes or rimes. The first task to measure phonological awareness, phoneme deletion, was developed in 1964 (Bruce) and is still widely used (see e.g., Chapter 3). In phoneme deletion the participant is required to identify how a spoken word would sound if one sound were omitted. For example, “What is smile without /s/?” Other phonological-awareness tasks include phoneme blending (e.g., “What does /c/ /a/ /t/ say?”) and segmentation (e.g., “What sounds do you hear in the word /s/ /u/ /n/?”), both of which are used in
Chapter 2 and 4. Rapid naming is sometimes subsumed under the umbrella of phonological skills (Boets et al., 2010; Wagner & Torgesen, 1987), since fast retrieval of phonological codes is one of its task components, alongside the intermodal coupling of visual input to phonological output (Warmington & Hulme, 2012). Others see rapid naming as a separate cognitive underpinning of reading ability (Wolf & Bowers, 1999).

The mentioned trio of preliteracy skills—phonological awareness, rapid naming, and letter knowledge—will be investigated in this thesis. These skills were measured in kindergarten in both the Amsterdam sample (Chapter 2) and the national sample (Chapter 4; see for the sample descriptions Section 1.5.1). Furthermore, Chapter 3 (national sample) will present findings on the concurrent relations of phonological awareness and rapid naming with reading skills, all assessed at the end of second grade. As literacy is acquired, letter knowledge loses its correlations with reading ability (halfway through Grade 1 all letters are taught, producing ceiling effects on letter-knowledge measures), but phonological awareness and rapid naming continue to show concurrent correlations with reading ability (e.g., de Jong, 2011; Vaessen & Blomert, 2010), even into adulthood (e.g., Bekebrede, van der Leij, Plakas, Share, & Morfidi, 2010). The three groups of children in our family-risk studies, at-risk dyslexia, at-risk non-dyslexia, and controls, will be compared on these preliteracy or underlying cognitive skills of reading.

A couple of previous family-risk studies compared the groups of children at kindergarten age. In general, the at-risk children who later become dyslexic are found to have difficulty with phonological-awareness tasks, are slower on rapid-naming tasks, and are less familiar with the letters of the alphabet compared to their typically developing peers (Elbro et al., 1998; Pennington & Lefly, 2001; Scarborough, 1990; Snowling et al., 2003; Torppa et al., 2010). Moreover, after kindergarten at-risk children with dyslexia perform poorly on tests of phonological awareness and rapid naming, in addition to the known difficulties in reading and spelling (Boets et al., 2010; de Bree, Wijnen, & Gerrits, 2010; Pennington & Lefly, 2001; Snowling, Muter, & Carroll, 2007).

The at-risk children who do not meet dyslexia criteria nevertheless typically perform less well than control children on reading and spelling tasks (Boets et al., 2010; Pennington & Lefly, 2001; Snowling et al., 2003; but see Torppa et al., 2010 for an exception). When group means are visualized in a graph this yields the mentioned stepwise pattern on literacy tasks: the at-risk dyslexics
perform weakest, the at-risk non-dyslexics take up an intermediate position, and the controls perform best. This pattern of results fits with the continuity of family risk (Pennington & Lefly, 2001; Snowling et al., 2003) and with a multifactorial aetiology of dyslexia (Pennington, 2006, Figure 1.1). Previous research has shown mixed results, however, regarding the performance of the at-risk non-dyslexic group on the cognitive underpinnings of reading. During the early school years this group either performs somewhat lower or equally well as the controls (Boets et al., 2010; Pennington & Lefly, 2001; Snowling et al., 2003), but no significant differences have been reported on phonological awareness and rapid naming during kindergarten (Boets et al., 2010; Elbro et al., 1998; Pennington & Lefly, 2001; Snowling et al., 2003; Torppa et al., 2010). Our national sample is larger, and hence, analyses have more statistical power than those in previous studies. Therefore we have the opportunity to detect also subtle impairments.

Returning to the multiple deficit model (Figure 1.1), this model predicts a normally distributed liability continuum. Relating this liability continuum to our three groups, we hypothesize the at-risk dyslexics to be in the high tail of the distribution, the at-risk non-dyslexics to be somewhat above average (inheriting some disadvantageous gene variants from their dyslexic parents), and the controls to have a somewhat below average liability profile (as their parents are average to good readers). This outlined model is in line with the reported phenotypic findings of a step-wise pattern for literacy skills. Combining this model with the fact that literacy, phonological-awareness, and rapid-naming skills are normally distributed and interrelated, it follows that a step-wise pattern for phonological awareness and rapid naming is also most likely.

As for the relation between general cognitive ability and later reading, we also investigated the specificity question for the relation between the preliteracy skills and later reading. Chapter 4 examines whether the predictive value of the trio of preliteracy skills –phonological awareness, rapid naming, and letter knowledge– is specific for later word-reading fluency or whether these skills are also predictive for later mental-arithmetic fluency. De Smedt and colleagues (Boets & De Smedt, 2010; De Smedt, Taylor, Archibald, & Ansari, 2010) have proposed an hypothesis to explain the correlation between word reading and mental arithmetic, which states that performance on these tasks are related because both depend upon the quality of phonological
representations in long-term memory. Mental arithmetic involves fast retrieval of phonological-based arithmetic facts. Mobilizing those facts inefficiently limits memory resources available for selecting and carrying out suitable arithmetic procedures (Hecht, Torgesen, Wagner, & Rashotte, 2001). Therefore, we expected that both phonological awareness and rapid naming in kindergarten would be significant predictors of third-grade arithmetic, although not as strong as for reading, since these skills are the core precursors of literacy, while the core precursor of arithmetic is early number competence (e.g., Jordan, Kaplan, Ramineni, & Locuniak, 2009).

1.4 Precursors in Families

The previous section focused on an individual child (with or without family risk) and his or her cognitive characteristics or risk factors for dyslexia. In addition, the current thesis encompasses a broader domain of plausible risk factors present before formal reading instruction starts. We will investigate whether characteristics of children’s home literacy environment are related to children’s reading outcome. Last but not least, we will study parent-child resemblance, which signifies intergenerational transfer of cognitive skills of parents to their offspring.

1.4.1 Home Literacy Environment

Does the literacy environment that parents provide at home before school entry influence children’s subsequent reading development? This question will be addressed for both the Amsterdam (Chapter 2) and the national sample (Chapter 4). We tested whether individual differences in home literacy environment before first grade were related to individual differences in reading performance some years later. Home literacy environment is usually quantified as the availability of books and other reading material in the home and how often parents read to their children. Parents are often told that they should read to their child to promote reading development. However, the effect of shared-reading interventions on eventual reading performance is surprisingly under-researched and those studies addressing this issue failed to demonstrate that parent reading enhances reading ability (Sénéchal & Young, 2008). Correspondingly, the family-risk studies that looked into literacy characteristics
of the family environment did not show effects on later reading achievement (Elbro et al., 1998; Snowling et al., 2007; Torppa et al., 2007). These findings are in agreement with findings from behavioural genetic studies showing strong genetic and small shared-environmental influences on reading ability (Byrne et al., 2009; Haworth et al., 2009).

1.4.2 Parental Skills
As mentioned earlier, in family-risk studies two samples are followed, differing in family risk by virtue of having or not having a parent with dyslexia. Accordingly, these studies are implicitly based on a dichotomy of family risk. However, it might well be that within the at-risk sample, the groups of children who do and do not go on to develop dyslexia differ in their degree of family risk for dyslexia, despite the fact that they all have an affected parent.

As briefly touched upon before, we propose that the assumption that the at-risk children with and without dyslexia have equal liability to dyslexia is testable by considering the cognitive skills of their parents. Multiple genetic and environmental factors act probabilistically, leading to a continuum (rather than a dichotomy) of liability to dyslexia. Since parents provide both genes and environments for their child, variability among children must be associated at least partly with variability among their parents. Therefore, we studied the relation between parents and offspring in their reading and reading-related skills.

To return to the two groups of at-risk children, we investigated whether the reading outcome of children is related to the severity of affected parent’s dyslexia. This would indicate that within the at-risk sample the affected children have a higher liability than the unaffected children, as the multiple deficit model predicts: affected children inherit and experience more of the numerous aetiological risks. This is tested for the Amsterdam sample in Chapter 2 and for the national sample in Chapter 3 and 4. Replication of findings across two independent samples is important as we are the first to investigate this issue. Furthermore, Chapter 4 also investigates the importance of the literacy levels of the parent without dyslexia. Chapter 6 concludes by proposing an extended version of the multiple deficit model of Pennington (2006; Figure 1.1): the intergenerational multiple deficit model, which includes cognitive risks that parents pass on to their offspring.
1.5 The Current Studies

The current set of studies are based on data of the Dutch Dyslexia Programme (DDP). In what follows I will introduce the DDP and I will conclude this chapter with a brief overview of the issues addressed in the chapters to come.

1.5.1 The Dutch Dyslexia Programme

The Dutch Dyslexia Programme (DDP) is a multidisciplinary and multicentre research programme, funded by The Netherlands Organisation for Scientific Research (NWO), and initiated and executed by the University of Amsterdam, the University of Groningen, and the Radboud University Nijmegen. It brings researchers together from a variety of disciplines, including educational sciences, psychology, linguistics, neurology, and genetics. The DDP consists of three components, namely a prospective longitudinal study, intervention studies, and genetic studies.

The present thesis presents findings of two family-risk samples, which I will refer to as the Amsterdam and the national sample. Chapter 2 presents the Amsterdam family-risk study. This sample of children originates from the two intervention studies of the DDP which were conducted in Amsterdam (Regtvoort & van der Leij, 2007; van Otterloo & van der Leij, 2009). Children in this sample were followed from 5 (kindergarten) till 11 years of age (Grade 5). The other three studies of my thesis employ data of the ongoing national longitudinal study of the three DDP universities. In this larger-scale family-risk study, couples with and without a family history of dyslexia who expected a baby were recruited. In short, children were considered at family risk if (at least) one of their parents and one other family member had dyslexia. The children have been followed from infancy. Chapter 3, 4, and 5 belong to this prospective longitudinal study, covering the development of 3½ to 9 years of age (Grade 3).

1.5.2 Outline of the Thesis

My thesis aims to shed light on the constellation of risks that ultimately leads to dyslexia. In Section 1.2 and 1.3 I have introduced the topics that will be covered. As there is no one-to-one relation between the four topics and the four following chapters, I will provide an overview of the topics per chapter.

Chapter 2 (van Bergen et al., 2011) concerns the Amsterdam sample, which is followed from kindergarten to Grade 5. It examines home literacy environment,
preliteracy skills, reading development, and parental skills. Chapter 3 to 5 are part of the ongoing national study. The focus of Chapters 3, 4, and 5 will be children’s cognitive skills at age 8 (Grade 2), 6 (kindergarten), and 4 years, respectively. **Chapter 3** (van Bergen, de Jong, Plakas, Maassen, & van der Leij, 2012) discusses group differences on literacy and its cognitive underpinnings at the end of Grade 2. Moreover, the effect of the literacy skills of the dyslexic parent on children’s reading outcome are examined. **Chapter 4** (van Bergen, de Jong, Maassen, & van der Leij, submitted) the intergenerational transfer from both parents to their offspring is investigated. Moreover, reading and arithmetic skills in Grade 3 will be predicted from home literacy environment and preliteracy skills measured before learning to read. In **Chapter 5** (van Bergen et al., in revision) focuses on the relation between general cognitive ability at 4 years of age and reading and arithmetic four years later, at the end of Grade 2. Finally, in **Chapter 6** the key findings and theoretical implications of the present thesis will be discussed.
1.6 References


de Bree, E., Wijnen, F., & Gerrits, E. (2010). Non-word repetition and literacy in dutch children at-risk of dyslexia and children...
with SLI: Results of the follow-up study. *Dyslexia*, 16(1), 36-44.


the Dutch foundation of dyslexia]. Bilthoven: SDN.


