Who will develop dyslexia? Cognitive precursors in parents and children

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Don’t become a mere recorder of facts, but try to penetrate the mystery of their origin.

- Ivan Pavlov -
Chapter 6

General Discussion
Who will develop dyslexia? The series of studies reported in this thesis study cognitive precursors of dyslexia in parents and children. We investigated, firstly, the cognitive profile characteristic of children who go on to develop dyslexia. As a related issue we examined whether children’s skills assessed prior to Grade 1 are specifically predictive of later reading development or are also involved in later arithmetic development. The second goal was to examine the impact of the cognitive profile of parents and the literacy environment parents create on children’s reading outcome. The studies employed a prospective design, in which the progress of children at high and low family risk was followed. Children at high risk had a family history of dyslexia. After some years of reading instruction they were categorized as either dyslexic or non-dyslexic (below or above the 10th percentile cut-off on word-reading fluency, respectively). Subsequently, they were compared concurrently and retrospectively with each other and with typically developing children without such a family background. The studies reported in the current thesis include two independent samples of the Dutch Dyslexia Programme (DDP), labelled the Amsterdam and the national sample (see also the General Introduction, Section 1.5.1). The Amsterdam sample (N = 79) covers children’s development from kindergarten to Grade 5 (11 years old). Findings are described in Chapter 2. Chapters 3, 4, and 5 report findings on the national study (N = 212), including the development from 3½ to 9 years of age (Grade 3).

In what follows I will review the findings regarding precursors and literacy skills in children and precursors in families. Each topic will be concluded by linking the findings to Pennington’s (2006) multiple cognitive deficit model, as outlined in the General Introduction (Section 1.2.1 and Figure 1.1). Family-risk studies provide the opportunity to test implications of the multiple deficit model and to add more details for the model of dyslexia. Do the findings of the current set of studies lend support for this model and can the findings allow us to start on specifying the model for the case of dyslexia? Thereafter I will extend Pennington’s model to the intergenerational multiple deficit model, which includes an extra level to account for parental effects. This chapter will be closed by suggestions for future research and concluding remarks.
6.1 Precursors in Children

Two categories of child precursors of dyslexia were studied: general cognitive ability (IQ) and preliteracy skills. The first were studied at kindergarten entry (age 4) in Chapter 5 (the national sample). Preliteracy skills were assessed during the last year in kindergarten (age 5 or 6) and reported in Chapter 2 for the Amsterdam sample and Chapter 4 for the national sample.

6.1.1 IQ

The relation between verbal and nonverbal IQ around the age of four, and reading outcome at the end of Grade 2 was the focus of Chapter 5. It was found that at-risk children who go on to become dyslexic were impaired relative to controls on both verbal and nonverbal IQ, with the gap being larger for verbal IQ. The at-risk children who do not become dyslexic showed good nonverbal abilities, but their verbal IQ was slightly but significantly lower than that of controls. Furthermore, it appeared that nonverbal IQ was equally strongly related to later reading achievement (e.g., word-reading fluency) as to later arithmetic achievement (e.g., mental-arithmetic fluency), while verbal IQ was specifically predictive of reading.

An unresolved issue is the nature of the link between early IQ and subsequent reading ability. Three possibilities are 1) the inclusion of reading-related tasks in the IQ battery, 2) IQ and reading being independent consequences of the same aetiological factors, and 3) a causal link from IQ to reading. Each of these possibilities will be discussed below for verbal and for nonverbal IQ.

According to the first possibility, verbal IQ and reading are related because verbal IQ tests include subtests that assess component skills that are known to be related to reading ability and known to be deficient in individuals with dyslexia (cf. Siegel, 1999). Note that this first account is compatible with both a shared aetiology account and a causal account. That is, the component skill that is supposedly involved in both verbal IQ and reading could be associated with reading through partly shared aetiology or could causally influence reading ability. A component skill shared by verbal IQ and reading might be verbal short-term memory (deficient in dyslexia, see e.g. de Jong, 1998), which was indeed part of the verbal IQ battery of the study presented in Chapter 5. Nevertheless, group differences on this subtest did not exceed
group differences on the other verbal IQ subtests (see Chapter 5, Figure 2), a finding that is not in agreement with this first account. Concerning nonverbal IQ, this first account cannot explain the relation between early nonverbal IQ and subsequent reading as there is no common component skill known.

Second, the literature suggests that early language and later reading ability indeed share aetiological factors, which causes the phenotypic correlation between them. Hayiou-Thomas, Harlaar, Dale, and Plomin (2010), for example, reported that language (or verbal IQ) at age 4 and subsequent reading performance are largely influenced by common genetic and shared environmental influences. Knowing that the genetic and shared environmental correlations are strong does not, however, reveal whether early verbal IQ and later reading are causally linked, with language supporting reading development, or whether they independently draw upon common aetiological influences.

Regarding nonverbal IQ and a shared-aetiology account, we found that nonverbal IQ was related to later reading as well as later arithmetic skills. This finding fits with the generalist genes hypothesis (Kovas & Plomin, 2007; Plomin & Kovas, 2005 - see also the General Introduction, Section 1.2.2). The generalist genes hypothesis states that genes influencing different academic domains and IQ partly overlap (Kovas, Haworth, Dale, & Plomin, 2007). Evidence from molecular genetic studies more clearly points to the possibility that reading, arithmetic, and nonverbal IQ are related because of partly shared aetiological factors. The candidate genes that have been linked to dyslexia all play a role in early brain development: they can be responsible for anomalies in neuronal migration and axon growth (Galaburda, LoTurco, Ramus, Fitch, & Rosen, 2006). The resulting subtle cortical malformations are probably more widespread than just cortical areas involved in reading. Therefore, it is expected that cognitive deficiencies are not solely restricted to the reading process.

Lastly, I will consider a causal account. Verbal IQ was found to be specific for reading development, which is consistent with a partly causal link, with the developing reading system building on the language system. Indeed, models for word decoding generally include aspects of language skills, like the phonological lexicon in the dual-route cascaded model (Coltheart, Rastle, Perry, Langdon, & Ziegler, 2001) and phonological and semantic units in the triangle connectionist model (Seidenberg & McClelland, 1989). Moreover, a causal link for the phonological aspects of language is often proposed (e.g., Hulme,
Bowyer-Crane, Carroll, Duff, & Snowling, in press; Vellutino, Fletcher, Snowling, & Scanlon, 2004), although the relation between phonological skills and reading could also be reciprocal (e.g., Burgess & Lonigan, 1998). Interventions to promote broader language skills in kindergarten children that would result in a subsequently faster reading development, would provide evidence for a causal interpretation. In the absence of such evidence, explaining the relation between verbal IQ and reading in terms of shared underlying factors is just as plausible. In sum, our study showed that early verbal IQ is a specific predictor of word-reading fluency, but the nature of this association remains elusive.

Moving on to nonverbal IQ, one could defend a causal link by arguing that nonverbal IQ indicates an individual’s ability to acquire new insights and skills. However, this is at best an explanation for early literacy development, like grasping the alphabetic principle. It is unclear how an individual’s ability to acquire new insights and skills would translate into developing such a specific skill as word-level reading fluency, and hence, such an unspecified explanation is difficult to defend. Therefore, explaining the relation between early nonverbal IQ and subsequent reading in terms of common aetiological factors seems most plausible.

Although our data do not reveal the nature of the link between IQ and reading, they do show that children who go on to develop dyslexia have a lower IQ than typically developing children. Because at the time their IQ was assessed they could not yet have failed to learn letters or word reading, their lower IQ at this age cannot be a consequence of reading failure and reduced print exposure. In previous prospective studies IQ differences were either neglected or were not possible to investigate (e.g., due to matching on IQ as in de Jong & van der Leij, 2003). For that reason, the relative low IQ of four-year-olds who go on to be dyslexic is in itself a unique finding.

### 6.1.2 Preliteracy Skills

As mentioned above, preliteracy skills were assessed during the last year in kindergarten (age 5 or 6) for both samples. Chapter 2 reports the results for the Amsterdam sample and Chapter 4 for the national sample. The studies included the most important preliteracy skills: phonological awareness, rapid naming, and letter knowledge. Again, the longitudinal character ascertains that these cognitive deficiencies are due to aetiological factors rather than due to poor reading and less print exposure.
In the Amsterdam sample, children were categorised as dyslexic or non-dyslexic in Grade 5. The at-risk dyslexic group read less fluently than the at-risk non-dyslexics and the controls across reading tasks and throughout primary school. Retrospectively, in kindergarten the at-risk group with dyslexia was slower on rapid naming and knew fewer letters than the other two groups. However, group differences on phonological-awareness measures did not reach significance. The absence of a difference could be due to low levels of phonological awareness in Dutch children of this age (see also de Jong & van der Leij, 1999). For many children the tasks were too difficult, which might have obscured existing group differences in the latent ability. Another factor that might play a role is the timing of identifying dyslexia. Reading outcome was not assessed until fifth grade, when word-reading fluency in Dutch is more strongly related to rapid naming than to phonological awareness (Vaessen & Blomert, 2010). Hence, a group categorized as dyslexic at this age is expected to show larger deficiencies in rapid naming than phonological awareness. In line with this, the children with dyslexia were impaired on rapidly naming familiar pictures and colours during kindergarten.

Moving on to the national sample, Chapter 4 presents the findings regarding preliteracy skills in kindergarten. In Chapter 4 reading status was assessed in third grade and preliteracy skills at the end of kindergarten. The at-risk dyslexic group was impaired on all preliteracy skills. The at-risk children without later dyslexia showed normal letter knowledge and rapid naming, but performed below controls on phonological awareness. Note that the two independent samples converge on the conclusion that at-risk dyslexic children are impaired, compared to the other two groups, on rapid naming and letter knowledge in kindergarten. The findings across the samples differ, however, for phonological awareness: no group differences in the Amsterdam sample and a step-wise pattern (at-risk dyslexia < at-risk no-dyslexia < controls) in the national sample. Although this may seem contradictory, it should be mentioned that in the national sample the phonological-awareness tasks were more appropriate for the children's ability level. For this reason, we attach more importance to the group differences in the national sample.

With regard to the multiple deficit model (Pennington, 2006), the current set of studies shows that the cognitive processes associated with dyslexia are phonological awareness, rapid naming, and verbal and nonverbal IQ. Note that these associations are not necessarily causal (see the discussion...
on IQ in Section 6.1.1). Letter knowledge might be added to the list of cognitive processes as an underlying skill of reading. However, it could also be regarded as belonging to the symptom level, being a forerunner or autoregressor of reading.

The multiple deficit model predicts that normal reading children with a family risk do slightly poorer on reading-related skills than normal reading children without such risk. This is because there is evidence that the underlying cognitive processes of reading are also complex traits, influenced by multiple genetic and environmental factors (Naples, Chang, Katz, & Grigorenko, 2009; Petrill, Deater-Deckard, Thompson, De Thorne, & Schatschneider, 2006), so the at-risk no-dyslexia children are expected to receive some of the involved risk factors. A difference between the two normal reading groups was indeed found for verbal IQ and phonological awareness, but not for rapid naming. How their surprisingly good rapid naming can be explained aetiologically remains puzzling. It seems that verbal IQ and phonological awareness are related to both reading status and family-risk status, whereas rapid naming only differentiates between children differing in reading status but not between children differing in risk status. The at-risk children without dyslexia are good at rapid naming and, in anticipation of Section 6.3.2, their affected parents are relatively good at rapid naming. This suggests that good rapid naming somehow decreases the probability of becoming dyslexic, but how remains unclear.

The findings regarding preliteracy skills of the at-risk children who went on to become dyslexic confirmed that phonological awareness, rapid naming, and letter knowledge are cognitive risk factors for dyslexia. Chapter 4 additionally investigated comorbidity with dyscalculia and whether the investigated preliteracy skills are also cognitive risk factors for dyscalculia. Dyslexia and dyscalculia (defined as scoring in the bottom 10% on word-reading fluency or arithmetic fluency, respectively) were indeed comorbid, as evidenced by the 42% rate of dyscalculia in the at-risk dyslexic group. In comparison, these figures were 20% and 8% in the at-risk and not at-risk normal readers, respectively. The trio of preliteracy skills was shown to be predictive of later arithmetic achievement as well. Rapid naming was equally strongly related to reading and arithmetic, but phonological awareness and letter knowledge were more specific precursors of reading. After controlling for rapid naming and letter knowledge in a regression analysis, phonological awareness dropped out as a predictor of arithmetic. Therefore, our data did not
provide unequivocal support for the hypothesis of De Smedt and colleagues (Boets & De Smedt, 2010; De Smedt & Boets, 2010; De Smedt, Taylor, Archibald, & Ansari, 2010) that individuals with dyslexia have difficulties with retrieving arithmetic facts from long-term memory because these facts are phonological in nature. Our data suggest that the speed of processing or speed of retrieving phonological codes might be more important in predicting later achievement in arithmetic fluency.

The conclusion that some of the cognitive processes of importance to reading are also important for arithmetic, whereas others are distinct to reading, is in line with the multiple deficit model and the generalist genes hypothesis (Kovas & Plomin, 2007; Plomin & Kovas, 2005 - see also the hybrid model in the General Introduction, Section 1.2.3). In sum, nonverbal IQ and rapid naming are shared and therefore contribute to the correlation between arithmetic and reading. Likewise, at the lower end of the distribution, they contribute to the comorbidity between dyscalculia and dyslexia. Verbal IQ, phonological awareness, and letter knowledge were found to be skill-specific cognitive processes, contributing to the dissociation between arithmetic and reading. The inclusion of arithmetic and its associated disorder dyscalculia in the study of the specificity of dyslexia precursors underscore the value of studying comorbidity. Other comorbidities are also of interest, but were not dealt with in the current studies.

6.2 Literacy Skills in Children

Chapter 3 compares for the national sample the three groups of children at the end of Grade 2 on literacy skills and their main cognitive underpinnings: phonological awareness and rapid naming. On all five reading tasks, which measure accuracy and fluency of word and pseudoword reading, the at-risk children with dyslexia were severely impaired compared to the control children, as indicated by large effect sizes (1.74 to 2.83). In addition, they made many errors in spelling words and in phoneme deletion, and were slow in serial naming of familiar items.

Although the at-risk group without dyslexia had literacy skills within the normal range for their age, they read significantly less accurately and fluently than controls on all of these reading measures (with effect sizes in the
range of 0.43 to 0.74). The same step-wise pattern was found for spelling skills and phonological awareness. A salient finding was the good performance of the at-risk children without dyslexia on rapid naming. Despite the fact that they fared significantly worse than controls on literacy and phonological awareness, they were equally good on naming digits and colours rapidly.

The group differences for phonological awareness and rapid naming in second grade mirror those for kindergarten, so the picture did not change. Apparently, phonological awareness is associated with both reading and risk status, while rapid naming is only related to reading status. The at-risk children who go on to develop normal reading skills might do well despite their family risk because the efficiency of the processes that rapid naming tap might protect them against dysfluent reading. On the other hand, one could argue that they still have mild literacy problems due to their mild phonological awareness deficit.

The multiple deficit model states that developmental disorders, like dyslexia, are influenced by many genetic and environmental risk factors that each increase the liability for the disorder without being necessary or sufficient for causing it. From this it follows that the multiple deficit model predicts a liability or risk function that is continuously distributed in the normal population. For our three groups this would mean a high liability for the at-risk dyslexia group, moderate liability for the at-risk no-dyslexia group, and low liability for the control group. At the behavioural level these differences in liability should be observable as differences in literacy skills, with the at-risk no-dyslexia group taking up an intermediate position. This hypothesis was confirmed: on all literacy tasks we found that the at-risk children who did not meet the dyslexia criterion still performed significantly weaker than controls. Nonetheless, as described in the previous section, this pattern was not systematically found at the cognitive level.

6.3 Precursors in Families

In addition to predictors of dyslexia residing in children we examined possible predictors in their families. More specifically, we studied effects of home literacy environment and parental literacy skills on children’s reading outcome.
6.3.1 Home Literacy Environment

Children’s socio-economic status, as indexed by parental level of education, can be seen as a distal family background characteristic. The literacy environment that parents provide at home might act as a mediating proximal process between socio-economic status and children’s reading development (cf. Leseman, Scheele, Mayo, & Messer, 2007). Both these distal and proximal family characteristics were investigated.

In the Amsterdam as well as in the national sample the families with a parent with dyslexia (i.e., at-risk families) had lower levels of education compared to the families with average to good reading parents (i.e., control families). Within the at-risk sample, the parents of the affected children had slightly lower educational levels than those of the unaffected children.

Parental level of education could assert an influence on children’s reading development via the proximal family environment. Hence, it was investigated whether there is a relation between the literacy environment that parents provide at home and children’s family history of dyslexia and reading outcome. In the Amsterdam sample (Chapter 2) home literacy environment was assessed during the second (and final) year of kindergarten. The three groups were compared on library memberships, frequency of shared reading, and on the number of books in the home, but they did not differ significantly on any of these measures. In the national sample (Chapter 4) the groups did not differ either on cognitive stimulation by parents, but there was a tendency for parents of control children to own more magazines, newspapers and books. The two at-risk groups did not differ in any of the measures of home literacy environment.

Although behavioural genetic studies point to substantial heritability of reading, they also estimate that roughly 30% of individual differences is due to environmental factors (Byrne et al., 2009; Petrill et al., 2006; Taylor, Roehrig, Hensler, Connor, & Schatschneider, 2010). The moderate environmental influences do not leave much room to find effects of home literacy environment, especially because our volunteer sample probably does not represent the full environmental range. Indeed, our data about children’s home literacy environment did not reveal significant group differences. It should be admitted however, that home literacy environment was only measured superficially and subjective. When the home literacy environment is assessed more thoroughly, preferably with objective observations, the question whether children’s literacy
environment at home influences their reading development can be answered more reliably. Also other environmental factors warrant further investigation, such as pre- and perinatal factors and school and classroom characteristics. Despite the mentioned limitations, our findings are in agreement with findings from other family-risk studies, which also failed to show effects of home literacy environment on children’s reading outcome (Elbro, Borstrøm, & Petersen, 1998; Snowling, Muter, & Carroll, 2007; Torppa et al., 2007). Thus, no environmental risk factors of substantial effect have been identified that would have been easy targets for intervention.

To summarize, we did find an effect of parent’s educational level, but this did not translate into detectable effects of literacy practices at home. This conclusion might seem paradoxical. However, the lower educational level of the parents of the at-risk sample might be partly a consequence of their reading difficulties. This could be the main reason for a relation between parental education and offspring’s reading outcome, rather than via environmental differences in literacy stimulation. Moreover, environmental effects of parental education are smaller than what might be expected intuitively. Genetic factors account for about two-thirds of individual differences in parental education (Rowe, Vesterdal, & Rodgers, 1998) as well as in reading attainment (Byrne et al., 2009; Petrill et al., 2006; Taylor et al., 2010). Therefore, the intergenerational relation between parental education and their offspring’s reading ability might well be largely genetically mediated. In line with the given explanations, Walker, Petrill, and Plomin (2005) found that school achievement shows substantial genetic influence (i.e., 69%). Shared environmental influences accounted for only 12% of the variance. While controlling for genetics, most of these shared environmental influences were due to effects of social-economic status (parental education and occupation) rather than school characteristics.

It should be borne in mind, though, that the high heritability of reading performance does not imply at all that educational improvements are pointless. Instead, they positively impact on almost all children’s reading achievement and raise the average of standardized scores of a class receiving effective reading intervention. Nonetheless, it is likely that individual differences among children remain largely genetically driven (Olson, Byrne, & Samuelsson, 2009). This suggests that children with a genetic constraint on their reading development need increased reading instruction, a notion that I will come back to in Section 6.5.
6.3.2 Parental Skills

The key innovating factor of the present family-risk studies is probably the inclusion of cognitive abilities of the parents. We went beyond using parental literacy for the sole purpose of dichotomizing children into high and low family-risk samples by examining the relation between reading and reading-related skills of the parents and reading skills of their children. We had objective measures of the parents with dyslexia. Although all children in the at-risk sample have a parent with dyslexia, they might still vary in their degree of family risk for dyslexia. We tested this in both the Amsterdam and the national sample by comparing the groups of at-risk children with and without dyslexia on the reading skills of their affected parent. Since parents pass on their genes to their offspring and shape their environment, parental reading skills might be taken as an indicator of the offspring's liability to dyslexia.

In the Amsterdam sample (Chapter 2) the dyslexia of the parents of the affected children was more severe than the dyslexia of the parents of the unaffected children. This was indicated by group differences on reading fluency of words and pseudowords, with effect sizes of 0.78 and 1.71, respectively. This is an intriguing finding, because the affected parents read on average at the fifth percentile compared to national norms. Yet in this restricted range group large differences were observable.

Employing data of the national sample (Chapter 3), we had the opportunity to replicate the above findings, and in addition, to extend them by investigating parental spelling, nonword repetition, and rapid naming. As we lacked a direct measure of phonological awareness, nonword repetition was used as a proxy. The difference between the at-risk children with and without dyslexia was replicated for the affected parent's word-reading fluency, although with a smaller effect size (Cohen's $d$ was 0.48). The two at-risk groups did not differ in parental pseudoword reading, spelling, and nonword repetition, though both groups were impaired compared to controls. Interestingly, however, the parents of the at-risk dyslexia children were slower on alpha-numeric rapid naming than those of the at-risk no-dyslexia children. This underscores the special role of rapid naming, at least in transparent orthographies. In regression analyses parental word-reading fluency and rapid naming also predicted at-risk children's word-reading fluency, even after controlling for children's concurrent phonological awareness and rapid naming.

In the two above mentioned studies data were reported of the
parent with dyslexia. Chapter 4 (about the national sample) completes this by examining the influence of the parent without dyslexia for the first time. It was unfortunate that we did not have objective measures of the literacy skills of the parent without dyslexia; we only had their report on whether they experienced literacy difficulties. Nevertheless, this measure appeared to be a very good indicator of reading fluency: in a subsample we found a strong correlation (.85) between assessed and self-reported literacy skills. In addition, this measure yielded interesting findings. As hypothesised, also for the non-dyslexic parents there was a difference between the two at-risk groups: the parents of the affected children reported more literacy difficulties compared to those of the unaffected children. Moreover, the literacy difficulties of the non-dyslexic parent explained additional variance in children's reading fluency after controlling for literacy difficulties of the dyslexic parents.

Thus, the findings in the Amsterdam sample regarding the affected parent were only partly replicated in the national sample. However, the results concerning the unaffected parent further support the conclusion that children at family risk for dyslexia differ in their liability, as indicated by differences in parental reading skills between at-risk children with and without dyslexia. Moreover, differences between the two family-risk groups in the severity of the dyslexia of the affected parent have now been replicated in Finnish, in the Jyväskylä Longitudinal Study of Dyslexia. Torppa, Eklund, van Bergen, and Lyytinen (2011) showed differences in parental reading fluency, accuracy, and spelling. Regression analyses showed that these measures also predicted children's literacy skills in Grade 3; parental literacy skills even predicted children's reading accuracy over and above children's preliteracy skills.

Do the findings regarding precursors in families lend support for the multiple deficit model? According to this model, the aetiology of dyslexia (and other developmental disorders) is multifactorial and probabilistic. Multiple genetic risk variants interact with each other and with multiple environmental risks to ultimately produce the disorder at the behavioural level. Regarding environmental risks, we investigated aspects of one environmental factor: home literacy environment. However, we did not find effects on children's reading outcome. Children's social-economic status (as indexed by parental level of education) did differ among groups, arguably asserting influence on children's reading development via environmental as well as genetic pathways (see Section 6.3.1).
Genetic risk factors were not measured directly. Although there is now a huge body of evidence indicating that genes contribute importantly to individual differences in reading ability (e.g., Byrne et al., 2009; Hayiou-Thomas et al., 2010), the specific gene variants found thus far only explain a tiny part of these differences (see Bishop, 2009, for the example of the KIAA0319 gene), despite substantive work in the field of molecular genetics. This phenomenon also applies to other common traits and is called the mystery of the missing heritability (see e.g., Manolio et al., 2009). Genetic screening is therefore not (yet) informative about a child’s genetic liability to dyslexia (Bishop, 2009). Nevertheless, we proposed (see General Introduction, Section 1.4.2) that something can be said about children's liability. Since parents pass on their genetic material to their offspring and shape their environment, cognitive abilities of parents could be used as an indicator of the genetic and environmental risk and protective factors in the multiple deficit model.

The current studies provide two kinds of support for parental skills being an indicator for children's liability. First, as in other family-risk studies, two samples of children were recruited based on having or not having a parent with dyslexia. The current and previous family-risk studies found a large effect of having a family history on children's risk of becoming dyslexic. For example, in Chapter 3 it was found that the rate of dyslexia was 30% in the high-risk group and only 3% in the low-risk groups, whereas by definition 10% of an unselected sample would meet the employed dyslexia criterion. Thus, having a parent with dyslexia increases the risk (and having a second parent with literacy difficulties increases the risk even more, see Chapter 4). Conversely, having average to good reading parents decreases the risk.

Secondly, within the at-risk sample it was found that affected and unaffected parents of the affected children had more literacy problems than those of the unaffected children. Moreover, when considering at-risk children's reading fluency on a continuous scale (rather than having or not having dyslexia), parental reading skills were significant predictors of children's reading skills. The current investigation of the impact of parental skills on children's skills are more fine-grained than in previous family-risk studies: Previous family-risk studies only assessed reading skills of the parents (or even relied on self-report of dyslexia) for the purpose of dichotomising their sample into children with and without family risk (Boets et al., 2010; de Bree, Wijnen, & Gerrits, 2010; Pennington & Lefly, 2001; Scarborough, 1990; Snowling, Gallagher, & Frith,
2003; Torppa, Lyytinen, Erskine, Eklund, & Lyytinen, 2010 - but see Snowling, Muter, & Carroll, 2007, for an exception). In contrast, we studied parent-child resemblance for both parents for reading and reading-related skills.

Our findings support the view that skills of parents indicate their offspring’s liability, which in itself is the combination of all genetic and environmental factors that affect reading development. Thus, parental skills might shed light on the aetiological level in the multiple deficit model. It is not possible to disentangle the genetic and environmental contribution to the intergenerational transmission of skills. However, according to our data the transmission of risk seems to be mainly via genes. It is important to note that genes are inherited, not phenotypic traits. Thus, although our data reinforce the view that parental skills are indicative of their offspring’s liability, parental skills will never completely specify it.

### 6.4 The Intergenerational Multiple Deficit Model

In the discussion of Chapter 4 an extension of the multiple deficit model was proposed: the intergenerational multiple deficit model. Below I will elaborate on this model. The model is depicted in Figure 6.1. In the figure it can be seen that a top layer is added to Pennington’s multiple deficit model (see Chapter 1, Section 1.2.1), which represents characteristics of parents. Note that again influences between layers are omitted from the figure.

Cognitive abilities of parents, for instance reading ability, form part of their phenotype (PT in Figure 6.1). Their phenotype is the result of their genotype (GT) in interaction with their environment. Genes do not code for cognitive and behavioural traits but for the structure of proteins and the regulation of gene expression, which in highly complex ways and in interaction with the environment guides the building and maintenance of the brain (Fisher & Francks, 2006; Fisher, 2006). Despite this gap between genes and cognition, for traits that show genetic influences in behavioural genetic studies there must be a relationship between genotypic and phenotypic variation. In other words, for heritable traits parental phenotype is a proxy for their genotype.

As both parents pass on half of their genes to their offspring, the genotype of both of them determines the genotype of their offspring. It follows that the phenotype of parents must be related to some extent to the genotype
of children, which includes children's genetic risk and protective factors for a particular developmental disorder.

In addition to transmission of parental skills via genetic pathways, parental skills could be passed on via environmental pathways. Parents largely shape their children's childhood environment, which creates a relation between parents' characteristics and children's environment. For example, good reading parents are more likely to spend a lot of time reading (Chapter 4), thereby providing a role model to their children. Moreover, they appear to be better educated (Chapter 2 and 3), and as a result, might live in better neighbourhoods and might send their children to better achieving schools. Hence, the cognitive phenotype of parents could be one of the factors that determines children's environmental risk and protective factors. However, other aspects of the phenotype of parents might also influence children's reading development. For instance, the behaviour of parents and the interaction between them determines how structured or chaotic the household is, something that has been shown to be related to children's school performance (Hanscombe, Haworth, Davis, Jaffee, & Plomin, 2011). In conclusion, the phenotype of parents must also be related to a certain degree to children's environmental risk and protective factors.

Given the above two lines of reasoning, the phenotype of parents is informative about children's genetic and environmental factors. Focussing on developmental disorders, this suggests that certain aspects of the phenotype of parents can inform us about a child's liability to a particular developmental disorder. Regarding dyslexia, the aspects of the phenotype of parents that are expected to shed light on children's liability to dyslexia are skills in accurate and fluent reading, spelling, and their cognitive underpinnings like phonological awareness and rapid naming. Related skills (such as language and arithmetic) and their underlying cognitive abilities might also play a role. The ability of parents on each of the relevant continua can be conceptualised as a position in multivariate space. The position of father and mother in multivariate space is proposed to be indicative of a child's predisposition towards dyslexia. The intergenerational transfer of skills from parents to children goes through genes, environment, and their interaction. Family studies can point to the skills that show intergenerational correlations; genetically sensitive studies can ultimately disentangle the contributions of genetic and environmental effects to such an intergenerational correlation.
**Level of analysis**

Figure 6.1. The intergenerational multiple deficit model (an extension of Pennington's multiple deficit model). Double headed arrows indicate interactions. Causal connections between levels of analyses are omitted.

- GT\textsubscript{p1} = genotype of parent 1
- PT\textsubscript{p1} = phenotype of parent 1
- G = genetic risk or protective factor
- E = environmental risk or protective factor
- N = neural system
- C = cognitive process
- D = disorder

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**Aetiological risk and protective factors**
- G\textsubscript{1}
- E\textsubscript{1}
- G\textsubscript{2}

**Neural systems**
- N\textsubscript{1}
- N\textsubscript{2}
- N\textsubscript{3}

**Cognitive processes**
- C\textsubscript{1}
- C\textsubscript{2}
- C\textsubscript{3}

**Behavioural disorders**
- D\textsubscript{1}
- D\textsubscript{2}
- D\textsubscript{3}
The intergenerational multiple deficit model is inspired by the current studies on dyslexia, but is generally applicable to other multifactorial developmental disorders with a genetic component. Examples of such disorders include attention-deficit/hyperactivity disorder, developmental coordination disorder, dyscalculia, specific language disorder, and autism spectrum disorder. With respect to autism spectrum disorder, a number of studies (e.g., Bölte & Poustka, 2006; Happé, Briskman, & Frith, 2001; Losh et al., 2009) have studied the cognitive phenotype of parents of probands (as opposed to children of probands, as in family-risk studies of dyslexia) and found in parents similar but milder impairments as in their children, indicating parent-child resemblance. A second example concerns specific language impairment. In a recent study (Bishop, Hardiman, & Barry, 2012), language skills of probands and their parents were found to correlate. In another recent study (Bishop et al., 2012) it was found that a parent’s non-word repetition ability was a predictor of whether the child would develop specific language impairment. These examples provide evidence of intergenerational transfer of cognitive skills other than reading.

6.5 Future Directions

The present collection of studies has yielded some answers, but meanwhile has raised new questions. To start with, our and previous family-risk studies did not show significant effects of the home literacy environment on children’s reading outcome, but behavioural genetic studies do find shared and non-shared environmental influences. Is the home literacy environment important but did we fail to find this because of other reasons (see Section 6.3.1)? Or are other environmental factors more important, like pre- and perinatal factors, peer influences, or characteristics of the school and classroom? Genetic sensitive studies can estimate how ‘environmental’ these factors actually are.

A second interesting area for future studies regards comorbidity. From the (intergenerational) multiple deficit model it follows that comorbidities between developmental disorders are to be expected. This points to the importance of studying developmental disorders that commonly co-occur. By doing so, one can uncover shared and distinct risk factors at each of the levels of explanation. This not only helps to understand the origin of the comorbidity, but also the developmental paths leading to each of the disorders.
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In examining the specificity of precursors for dyslexia we included arithmetic and dyscalculia, but comorbidities with other developmental disorders were not considered. Including more than just a single (dis)ability in future work will enhance our understanding. The same applies for studying intergenerational transfer: parents might confer risks in different domains, like reading, language, and attention. Therefore, future studies are needed to test whether a more complete picture of parents’ cognitive and behavioural profile gives a more reliable indication of their offspring’s liability to a particular disorder.

The third future direction concerns the relation between parental and children’s skills in an unselected sample. The findings reported in this thesis on parent-child resemblance might not be generalizable to the normal population since we had selected samples of high and low family risk. Building on the reported studies, we are currently conducting a study that aims to investigate relations between parents’ and children’s skills across the entire range of reading abilities. For that purpose we test whole families in NEMO – the science museum in Amsterdam – on reading, arithmetic, rapid naming and phonological awareness, using the same tests for all family members. In addition, environmental factors are assessed via questionnaires and genetic factors via saliva samples. Over 600 participants have been tested so far. Their data allows us to examine intergenerational transfer of skills and to study the cognitive, environmental, and genetic factors that influence reading ability and disability.

Finally, our studies have practical implications which warrant further investigation. The current and previous family-risk studies suggest that children who have a combination of weak-reading parents and weak preliteracy skills in kindergarten run the highest risk of developing dyslexia. This group of children is likely to have a high genetic endowment for dyslexia. This suggests that for this group classroom instruction in reading is not sufficient. We are currently investigating whether these children can be assisted by offering a reading intervention, consisting of regular extra practice from mid-kindergarten till mid-Grade 2. Preliminary results (Zijlstra, van Bergen, Koomen, & van der Leij, 2012) confirm that these children are most susceptible for dyslexia, but that early intervention can indeed ameliorate the impact of their high liability, provided that the intervention is intensive and individually tailored.
6.6 Concluding Remarks

The present thesis has characterized the cognitive profile of children with a family background of dyslexia that do and do not become dyslexic readers. This has revealed precursors of dyslexia, and by including arithmetic outcome, the specificity of these precursors. In addition, it has shed light on the cognitive profile of their parents, which provided a start in understanding the intergenerational transfer of reading ability and disability. Together, the findings on child and parental level have advanced our understanding of the factors influencing the liability and the development of dyslexia. However, more research is needed to move the science of reading towards elucidating the causal chain that leads to the behavioural manifestation of dyslexia.
6.7 References


De Smedt, B., Taylor, J., Archibald, L., & Ansari, D. (2010). How is phonological processing related to individual diffe-


