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Issues in diagnosing dyslexia

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Since the first descriptions of children with congenital word blindness or dyslexia, the proper criteria for diagnosis of dyslexia have been debated. Issues in this debate concern, among others, the role of underlying causes of reading and spelling and the use of a discrepancy between reading ability and intelligence. This chapter will consider recent evidence from family risk studies of dyslexia that speaks to these issues. We conclude that current evidence on the etiology of developmental disorders neither supports a specific underlying cognitive profile (e.g., phonological deficits), nor the requirement of a discrepancy with intelligence. Deciding factors in diagnosis should be lack of learning opportunity, other exclusion factors, and naturally the degree of reading and spelling difficulties.

Keywords: dyslexia, diagnosis, family risk, multiple deficit model, RTI

1. Introduction

Dyslexia is generally regarded as a disorder in the acquisition of reading and/or spelling at the word level. The disorder becomes manifest after a few months to a few years after the start of reading and spelling instruction and typically persists into adulthood. Despite the seemingly clear symptoms of dyslexia, i.e., reading and spelling problems, the criteria for a diagnosis have been hotly debated. In this chapter we consider two issues that are important to this debate: the role of underlying causes of reading and spelling in the diagnosis and the use of a discrepancy between reading ability and intelligence as a criterion for dyslexia.

A fundamental issue is the use of underlying deficits to further pinpoint reading problems. A group of 58 American scholars argued that such deficits should be considered in diagnosing dyslexia (Hale et al., 2010). In the Netherlands deficits on the underlying causes of dyslexia are even required to become eligible for government funded treatment (Blomert, 2006). The inclusion of an underlying deficit
criterion in the diagnosis of dyslexia is a way to delineate a special group of poor readers that can be considered dyslexic.

The importance of a reading ability–intelligence discrepancy in the diagnosis of dyslexia is already implicit in the first case descriptions of the disorder. Pringle Morgan (1896) describes the 14 year old Percy F., who is not able to read, despite being a bright and intelligent boy of intelligent parents. According to his school-master, ‘he would be the smartest lad in the school if the instruction were entirely oral’ (p. 1378). Given his intelligence, Pringle Morgan considered the reading disorder of Percy F. as ‘so remarkable’ that he is sure that the disorder ‘is due to some congenital defect’ (p. 1378). The discrepancy between reading ability and overall ability, intelligence, has long been included as a criterion in the diagnosis of dyslexia (Fletcher, Lyon, Fuchs, & Barnes, 2007). According to this criterion, individuals with a reading ability that is considerably lower than to be expected on the basis of their intelligence belong to a special group of poor readers. These poor readers qualify for the diagnosis of dyslexia whereas the other poor readers do not. The latter have often been denoted as garden-variety readers (Stanovich & Siegel, 1994).

Below, we will consider these issues. Evidently, both issues stem from the same question: Are dyslexics a special category within the group of poor readers and spellers? Even if this question is answered affirmatively, there might be alternative criteria. In a final section we will briefly discuss these alternatives.

2. Including deficits in the diagnosis

Dyslexia is often qualified as a specific learning disorder. ‘Specific’ denotes that the problem is restricted to reading. It is also used to designate that the poor reading performance is due to specific neurobiological and cognitive deficits that cause the development of dyslexia (Lyon, Shaywitz, & Shaywitz, 2003). As a next step, it has been argued that these deficits are valid indicators for the assessment of dyslexia and that they should be incorporated in the diagnostic criteria (Hale et al., 2010; Tannock, 2013). There are three issues at stake here: The nature of causes in developmental disorders, task-impurity in the measurement of indicators of deficits, and whether all underlying deficits are known. Below, we will argue that none of these issues can be solved satisfactory and therefore it is not (yet) warranted to include underlying deficits in the diagnosis of dyslexia.
2.1 The nature of causes

The first issue concerns the nature of causes in the development of dyslexia, as well as in many other neurodevelopmental disorders. For decades there has been a search for a single cause of dyslexia (e.g., Vellutino et al., 2004). However, a consensus has not been reached. In a recent review of theories on the causes of dyslexia, Ramus and Ahissar (2012) concluded that the many cognitive deficits that have been associated with dyslexia cannot be encompassed by one single theory. Probably several deficits can lead to the disorder. Moreover, it is increasingly realized that a deterministic view of causes is unable to explain several findings on the development of dyslexia. Not a single cause has been found that is both necessary and sufficient to cause the disorder. For example, phonological processing deficits are considered to be a major cause of reading problems (e.g., Vellutino et al., 2004; Hulme & Snowling, 2009), but nevertheless many persons with such a deficit do not seem to have problems in reading (e.g., Pennington et al., 2012). Deterministic single deficit explanations of dyslexia do not fare very well either in explaining the comorbidity of dyslexia with other disorders such as SLI and ADHD (Pennington, 2006). A substantial number of children with SLI, for example, tend to have phonological deficits but do not develop reading problems (Bishop, McDonald, Bird, & Hayiou-Thomas, 2009). The behavioral findings are in line with findings on the genetic origins of dyslexia. Many genes are assumed to be involved in the development of reading problems. Gene variants associated with dyslexia tend to have general rather than specific effects (Plomin & Kovas, 2005), and these variants are also found in nondyslexic groups (Bishop, 2015).

The above described findings are better captured by multiple deficit models (Pennington, 2006; van Bergen, van der Leij, & de Jong, 2014). Causes in these models are conceived as probabilistic. Some causes, i.e. risk factors, increase the likelihood of a disorder, whereas other causes, i.e. protective factors, decrease the risk that the disorder manifests itself. Importantly, a risk factor does not necessarily lead to the disorder. Instead, a disorder is assumed to be the outcome of the interplay of many risk and protective factors. At the genetic level, Plomin, De Fries, McClearn and MacCuffin (2008) have shown that the involvement of only a few genes results in a continuous distribution of risk. Thus, multiple deficit models can easily explain that reading ability is continuous, that presumed deficits are not found in all individuals with the disorder (and vice versa), and that comorbidity among disorders is common. Comorbidity is explained by common risk factors. A larger number of common risk factors increases the risk that dyslexia becomes comorbid with another disorder. Multiple deficit models can also account for findings of family risk studies on dyslexia. For example, mild problems in reading and spelling are often found in non-dyslexic children from dyslexic parents (e.g.,
Snowling & Melby-Lervåg, 2016; van Bergen, van der Leij, & de Jong, 2014). This is to be expected as the familial risk of these children will have resulted, through genetic or cultural transmission, in a number of risk factors and accompanying deficits, although not enough to develop dyslexia. Thus, multiple deficit models can readily explain a range of findings that are hard to understand from a deterministic single-deficit point of view.

What is the consequence of a multiple deficit perspective on developmental disorders for the inclusion of deficits in the definition of dyslexia? Evidently, the probabilistic nature of causes implies that even in the absence of a particular deficit there is still a chance of developing the disorder. But what should be concluded in case no deficit can be demonstrated? Here it should be kept in mind that relations among causal indicators of dyslexia, as well as the relations of these indicators with reading and spelling, are moderate at best (Pennington et al., 2012). Thus, purely on a statistical basis, a substantial number of individuals with reading and/or spelling problems that do not have any deficit is to be expected. In the study by Pennington et al. (2012) the percentage of children with dyslexia but without any known deficit varied between 21 and 32 percent.

2.2 Task impurity

A second, and related, issue is that the cognitive measures used to assess deficits cannot be regarded as pure indicators. Performance on these measures also depends on abilities that are irrelevant with respect to the deficit as well as to reading or spelling (Tannock, 2013). This is nicely illustrated by a recent study of van Viersen, de Bree, Kroesbergen, Slot, and de Jong (2015) on the underlying cognitive deficits of dyslexia in gifted children. Van Viersen et al. made two rival predictions. Following the phonological-core variable-difference model (e.g., Stanovich & Siegel, 1994) they expected that gifted children with dyslexia, compared to averagely intelligent children with dyslexia, would have similar deficits in phonological processing skills but perform higher on tests that are more related to intelligence. The phonological-core variable-difference model has been mainly tested by comparing dyslexic children with or without a discrepancy with intelligence. It is unknown how the model fares with gifted children, evidently having a huge discrepancy between reading ability and intelligence. As an alternative, van Viersen et al. argued that gifted dyslexics might have some abilities, for example a larger vocabulary, that have a beneficial effect on their word reading. Accordingly, their reading abilities are overestimated and these children need to have relatively larger phonological deficits to obtain a reading level that is commensurate with dyslexia.
At first sight the results of the study were only partly in agreement with the core deficit model. In accordance with this model, the dyslexia groups (matched on reading ability but differing on IQ) performed similarly on the rapid naming of alphanumeric and non-alphanumeric symbols. However, on phonological awareness the gifted dyslexics performed better than the averagely intelligent children with dyslexia. This result may suggest that gifted children have a less severe deficit. However, this interpretation would run counter all evidence on the importance of phonological awareness for learning to read (e.g., Vellutino et al., 2004). A more likely explanation is that gifted children perform better on the task specific aspects of the task that are unrelated to reading. These task specific aspects might require abilities that are related to intelligence. If average and gifted dyslexic groups are properly matched on reading, as was done by van Viersen et al., then a higher performance on the task is to be expected. The result can be regarded as another demonstration of a phenomenon that has been lively discussed with respect to the measurement of working memory functioning, the ‘task impurity problem’ (e.g., van der Sluis, de Jong, & van der Leij, 2007). With hindsight, it should be noted that this prediction also follows from the phonological-core variable difference model, but here extended to the task level. Tasks are not pure. Performance on the phonological core aspects of the tasks that are related to reading might not be affected by intelligence. Other aspects of the task might be related to intelligence or other individual differences and also influence performance, and as far as intelligence is concerned, increase performance.

2.3 Unknown causes

A third issue with respect to the inclusion of underlying deficits in the diagnosis concerns the question whether all deficits are known. There is probably no study in which the currently known underlying cognitive correlates of dyslexia, like rapid naming and phonological awareness, are shown to account for all variation in reading ability. Obviously, there are more causes of poor reading. However, one might argue that these causes are likely to be irrelevant to the diagnosis of dyslexia. For example, poor reading due to school refusal, motivation, or lack of opportunity in general does not qualify for a diagnosis of dyslexia. In diagnosing dyslexia, environmental explanations of poor reading and spelling should be discarded and only causes that relate to the genetic origins of dyslexia are of relevance.

It seems hard to prove that all relevant causes of dyslexia, or of any other behavioral disorder, are known. However, there is a way out. Many studies have shown that reading is a highly heritable ability (see Olson, Keenan, Byrne, & Samuelson, 2014). As said, dyslexia is assumed to be a disorder of neurobiological origin. If
all relevant underlying deficits are known, it follows that all genetic variance in reading should be captured by the currently known causes of dyslexia. Results from behavior genetic and family risk studies, however, show that this is not the case and thereby suggest that hereto unknown relevant causes exist.

Byrne et al. (2009; see also Byrne, Olson et al., 2006) followed twins from kindergarten through second grade. Well-known underlying abilities of reading (print knowledge, phonological awareness, and rapid naming) were assessed in kindergarten. Modern behavioral-genetic techniques enable the estimation of genetic variance that abilities have in common (e.g., Plomin & Kovas, 2005). Byrne et al. used this technique to estimate the common genetic variance of the kindergarten abilities and second grade abilities in reading and spelling. As expected, genetic variance of the abilities in kindergarten overlapped with genetic variance in later reading and spelling. More importantly, however, there was also a substantial amount of genetic variance in second grade reading and spelling that could not be accounted for by predictors from kindergarten. This suggests that some novel genes get involved in reading and spelling development after the start of instruction. Thus reading (and spelling) and its known underlying abilities are genetically partly different.

This conclusion was supported by a recent study of van Bergen, Bishop, van Zuijen and de Jong (2015) about the relationship of reading ability between parents and children. Family-risk studies have shown that having a dyslexic parent increases the likelihood that a child becomes dyslexic (e.g., Snowling, Gallagher, & Frith, 2003; van Bergen, de Jong, Plakas, Maassen, & van der Leij, 2012; see for a review Snowling & Melby-Lervåg, 2016). Unlike family-risk studies in which selected samples of children with and without a family risk of dyslexia are involved, van Bergen et al. (2015) included an unselected sample covering the whole range of parental reading ability. More importantly, they assessed the reading ability of both parents as, evidently, both parents transmit genes to their children and also jointly provide a ‘reading’ environment. In children, measures of reading and of its putative causes were included. The latter concerned commonly accepted causes, such as phonological awareness and rapid naming, as well as visual attention span (e.g., van den Boer, van Bergen, & de Jong, 2014). The main question was whether the influence of parents, reading ability on their offspring was fully mediated by the underlying causes of reading.

As to be expected, parents’ reading abilities were related to those of their offspring. A novel finding was that the influence of fathers and mothers was similar. In addition, the parental effects were only partly mediated by the underlying abilities of the children. Indirect effects of parents on their children accounted for just over half of the total effect of parental reading ability on children’s reading ability. However, it should be noted that these familial effects could be the result of genetic or cultural transmission or both. For two reasons, however, the parent-child
resemblance for reading is probably largely genetic. Firstly, van Bergen et al. computed the parent-offspring correlation that is to be expected if transmission is purely genetic. Assuming that about two third of the individual variation in reading is genetic (Olson et al., 2014) this correlation is .33. This corresponded very well with the observed correlation of approximately .34. Secondly, Wadsworth et al. (2002) showed that in adoptive families, in which parents and children have no genes in common, parent-offspring relations were absent. More recently, Swagerman et al. (in press) modeled the relationships of reading among twins, siblings and parents. They did not find any effect of cultural transmission either. Together, these studies strongly suggest that the transmission of reading skills from parents to children is mainly genetic. Returning to the topic of unknown causes, it seems that parental reading can be seen as an indicator of children’s genetic risk. In the van Bergen et al. (2015) study the effect of parent reading on child reading (i.e., genetic risk) was only partly mediated by known cognitive causes. This suggests that the other half of the genetic risk should be mediated by cognitive correlates that are to date unknown.

3. Intelligence achievement discrepancy

Case descriptions like the one by Pringle Morgan (1896) and anecdotal evidence on intelligent children who, unexpectedly, appear unable to learn to read have had a major influence on the definition of dyslexia. For many years a discrepancy between overall intelligence and reading or spelling ability was argued for and used by practitioners as a requirement to qualify for a diagnosis of dyslexia (see for example DSM-4). However, for several reasons the use of a discrepancy criterion in diagnosing dyslexia has been largely discarded (e.g., Fletcher et al., 2007; Tannock, 2013), although it is still vehemently advocated by some (e.g., Reynolds & Shaywitz, 2009).

In short, reasons against the use of the discrepancy criterion are as follows. Firstly, reading and spelling problems in children with and without a discrepancy with their intelligence have similar etiology (Fletcher et al., 2007). Moreover, both groups appear about equally responsive to treatment (Fletcher et al., 2007). More generally, except for baseline reading ability, other baseline cognitive characteristics do not predict how well children respond to intervention (Stuebing et al., 2015). Furthermore, there are problems with assessing a discrepancy reliably. It is based on two measures, reading ability and intelligence, and as a result measurement error is doubled. Finally, there is evidence that reading problems might negatively affect the development of skills that are relevant to measures of intelligence, like vocabulary (Cunningham & Stanovich, 1997). As a result, the paradoxical situation might arise that the reading problem remains over time whereas the discrepancy disappears.
Recent evidence from a family risk study provides additional reasons to discard the IQ-reading discrepancy criterion. Van Bergen, de Jong et al. (2014) tested whether the reading problems of dyslexic children were unexpected by considering IQ before the onset of reading. The ‘unexpectedness’ of reading problems refers to the assumption that dyslexic children’s cognitive development does not differ from their normal reading peers up to the onset of reading instruction. Longitudinal family risk studies are especially suited to test this assumption as in these studies dyslexic and normal readers are usually not matched on IQ. Accordingly, in these studies IQ can be regarded as an outcome. Family risk studies also provide information about a group of children with a family risk who do not develop dyslexia. Hence, it is possible to consider whether differences in IQ at an early age are related to dyslexia, risk only, or both.

Earlier family risk studies had shown that at risk children with and without dyslexia have a lower verbal intelligence before the onset of instruction (e.g., Snowling et al., 2003; Torppa, Lyytinen, Erskine, Erklund, & Lyytinen, 2010). However, these studies lacked power to determine whether nonverbal intelligence was impaired as well. Van Bergen et al. used the data of the Dutch Dyslexia Program (van der Leij et al., 2013), which is a large longitudinal study on children at risk for dyslexia. Intelligence was assessed with a battery of tests at the age of 4. Dyslexia was established by the end of second grade, when the children were about 8 years of age. At this age also the arithmetic skills were measured. Reading and arithmetic skills are related and have common genetic variance (Plomin & Kovas, 2005). As a result, dyslexia often coincides with arithmetic problems (Landerl & Moll, 2010). Inclusion of a measure of arithmetic enabled van Bergen, de Jong et al. (2014) to examine whether a conceivably lower IQ in dyslexic children at the age of 4 was specific to reading or was due to the common aspects of reading and arithmetic problems.

The results of van Bergen, de Jong et al. (2014) were clear cut. At age 4 a stepwise group pattern appeared for verbal IQ. Children who became dyslexic had a lower verbal intelligence than their normal reading peers. Family risk children without dyslexia had mild IQ impairments, but the difference with the control children disappeared when parental education was taken into account. Importantly, controlling for arithmetic performance did not change the results. For nonverbal IQ, results were different. Dyslexic children performed more poorly than control children on the nonverbal intelligence tests, whereas the children with only family risk did not. However the poor performance of the dyslexic children disappeared when statistically controlling for group differences in arithmetic skills. Thus before the onset of reading, children at family risk for dyslexia who become dyslexic have specific impairments in verbal IQ. They also have mild impairments in nonverbal intelligence, but these are related to their lower arithmetic abilities.
The findings of van Bergen, de Jong et al. (2014) clearly show that, unlike suggestions in early case descriptions, development in other domains than reading and spelling was not entirely normal before the start of instruction. But how should these findings be interpreted? It has often been argued that reading builds on the language system, especially as far as the phonologically-dependent abilities are concerned (Hulme & Snowling, 2009; Vellutino et al., 2004). Phonological-dependent abilities are also included in verbal intelligence, such as verbal short-term memory and possibly also vocabulary. Therefore, dyslexics will have impairments in verbal intelligence and these impairments are already manifest before reading (e.g., Scarborough, 1990). Consequently, children with dyslexia have a lower chance to develop an IQ–reading discrepancy because the very same disorder that underlies reading also negatively affects verbal intelligence. These findings speak against the use of verbal IQ in dyslexia criteria.

But what about the use of nonverbal intelligence to establish an IQ–reading discrepancy? This is not straightforward either. Note that there is no well-accepted theory that specifies a causal mechanism that can account for the (weak) relationship of nonverbal intelligence with reading. The relationship might be due to abilities that underlie the acquisition of academic skills in general. This fits well with the finding that the dyslexics’ lower performance on nonverbal intelligence was also related to their lower arithmetic ability, and thereby to what reading and arithmetic have in common. Indeed, Plomin and Kovas (2005; Kovas, Haworth, Dale, & Plomin, 2007) have shown large overlap in the genetic variance of reading and arithmetic. They argued that the majority of genes involved in the development of one learning ability also affect the development of other learning abilities. That is, the common set of genes has pleiotropic effects. They termed such genes ‘generalist genes’. For example, risk versions of genes found to be related to dyslexia are involved in migration of neurons in early brain development (Galaburda, LoTurco, Ramus, Fitch, & Rosen, 2006). This affects areas that are important for reading, but there is also the chance that areas are affected related to the development of arithmetic. The ‘third factor’ (i.e., common genetic variants) leads to a correlation between different domains of learning, even in the absence of a causal link between these domains. More generally, it is likely that genes involved in dyslexia increase the likelihood that other cognitive abilities are impaired. However, these abilities might not causally influence reading development. As far as generalist genes are involved, it is partly a matter of chance which ability, if any at all, in a particular child is affected in addition to reading. Overall, it is hard to justify a discrepancy with IQ or other learning abilities in the definition of dyslexia, as these abilities or disabilities might stem in part from the same underlying causes as the reading and spelling problems.
4. Concluding remarks

To reserve the diagnosis of dyslexia for a special group of readers with specific cognitive deficits or a discrepancy with IQ is not supported by current evidence on the etiology of developmental disorders. The probabilistic nature of causes and the problem of task impurity imply that reading and spelling problems with or without concomitant cognitive deficits might stem from the same underlying disorder. Moreover, not all underlying cognitive deficits are known, rendering the use of a deficit criterion even more problematic. Also an IQ-discrepancy criterion is not warranted, because IQ can be affected by the same underlying etiological factors (certainly at the genetic level) as deficits in reading and spelling themselves.

Does this imply that all poor readers and spellers should be regarded as dyslexic? That is not what we would like to argue. Definitions of dyslexia often exclude individuals with reading and spelling problems that result from other disorders or that can be accounted for by general external factors (see e.g., DSM-V, American Psychiatric Association, 2013). Obvious exclusion criteria concern intellectual disabilities, visual or auditory problems, language problems in the language of instruction, and environmental disadvantages which, for example, result in school absenteeism. All these exclusion criteria concern a lack of opportunity to acquire reading and spelling abilities. It is important to distinguish reading difficulties due to lack of opportunity from those due to constitutional factors.

A specific case of a lack of opportunity is inadequate educational instruction (DSM V; Lyon et al., 2003). Of course ‘inadequate instruction’ is difficult to assess. In the response to intervention approach (RTI; Fletcher et al., 2007; Fuchs, Fuchs, & Compton, 2012) dyslexics are considered as persistent nonresponders. A stepwise approach is taken to select this group. First, in Tier 1, response to regular instruction in the classroom is considered. In a next step, Tier 2, the nonresponders of Tier 1 are enrolled in a standardized and preferably evidence-based intervention. Individuals who do not respond to Tier 2 instruction either are regarded as dyslexic.

RTI is an elegant approach to assess whether reading and spelling development can be accelerated when opportunities are optimal. However, a number of practical problems with its application should be mentioned. Firstly, the approach seems especially suited for children in primary school, and most research on RTI has indeed concerned only this age-group (e.g., Fuchs et al., 2012). Secondly, it is difficult to establish criteria for a lack of response. Moreover, to establish response to intervention there is a need for curriculum-based achievement measures, which are often not available. Thirdly, there is a lack of evidence-based interventions.

In our view, diagnosing dyslexia is an interactive process. As a first step, reading and spelling problems should be assessed. In the second step, exclusion criteria have
to be examined: both the mentioned obvious exclusion criteria, as well as those pertaining to a lack of opportunity, including assessment of adequate instruction. Indeed, the RTI approach is incorporated in the recent DSM-V criteria as reading and spelling problems have to be persistent (for at least 6 months) ‘despite the provision of interventions that target those difficulties’ (p. 66). Our proposed step 1 and 2 are identical to DSM-V. However, in an additional final step, underlying cognitive deficits should be measured. Deficits should not be taken as another criterion to establish a diagnosis, but, as argued by Pennington et al. (2012), can be used to validate the diagnosis. In case no deficits are found, this is a reason to consider once more whether exclusion criteria are not met. If repetition of step 2 leads again to the same conclusion, then the diagnosis of dyslexia is warranted.

References


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