



# From single to multiple deficit models of developmental disorders

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## Abstract

The emerging etiological model for developmental disorders, like dyslexia, is probabilistic and multifactorial while the prevailing cognitive model has been deterministic and often focused on a single cognitive cause, such as a phonological deficit as the cause of dyslexia. So there is a potential contradiction in our explanatory frameworks for understanding developmental disorders. This paper attempts to resolve this contradiction by presenting a multiple cognitive deficit model of developmental disorders. It describes how this model evolved out of our attempts to understand two comorbidities, those between dyslexia and attention deficit hyperactivity disorder (ADHD) and between dyslexia and speech sound disorder (SSD).

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## 1. Introduction

This paper presents a multiple cognitive deficit model for understanding developmental disorders, and contrasts this new model with an earlier single cognitive deficit model. The need for a multiple deficit model arises partly from advances in understanding the complex genetics of behaviorally defined developmental disorders like dyslexia, autism, and attention deficit hyperactivity disorder (ADHD). It has become increasingly clear that the etiology of such disorders is multifactorial and that these

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multifactors partly overlap, such that various pairs of such disorders share some etiological risk factors but not others. This partial overlap of risk factors produces a greater than expected co-occurrence of these disorders, which is called comorbidity.

At the same time that a probabilistic, multifactorial model of the etiologies of these disorders is widely accepted, our cognitive analyses of them often relies on a deterministic, single deficit model. So, there is a potential contradiction between our etiological and cognitive models for understanding such disorders. The main goal of this paper is to attempt to resolve this contradiction by proposing a probabilistic, multiple cognitive deficit model for understanding such disorders.

In what follows, I will trace the evolution of this model, which grew out of our attempts to understand the comorbidities of dyslexia or reading disability (RD). I will first present the single cognitive deficit model. I will then attempt to use the single deficit model to account for two comorbidities of RD, that with ADHD and that with SSD, and show why it is inadequate for this task. Then I will develop an alternative multiple cognitive deficit model, and discuss ways to test this model. I will end by considering other criticisms of the single cognitive deficit model.

## **2. The single cognitive deficit model**

This paper traces the evolution of my thinking about models of developmental disorders and their relations. My thinking began with a very simple model that rested on the assumption of single causes, both at the cognitive and the etiologic levels of analysis. If a single cause (A) is necessary and sufficient to produce a result (a) at the next level of analysis, then the mapping across these two levels of analysis is 1:1. If another cause (B) at the first level of analysis is also necessary and sufficient to produce a distinct result (b), it also has a 1:1 mapping, meaning that it is independent from the first pair, A–a, at both levels of analysis. If this is the case, then a comparison of the two cause-result pairs, A–a and B–b, will yield a double dissociation. This logic can be applied across different levels of analyses. It could characterize the relation between localized brain regions and resulting cognitive deficits, the relation between cognitive deficits and developmental disorders, or the relation between etiologies and developmental disorders.

Although very simple, this model guided our early work on both the cognitive and genetic causes of developmental disorders. For instance, my colleagues and I tested Hallgren's (1950) hypothesis that a single autosomal dominant gene caused dyslexia (Pennington et al., 1991; Smith, Kimberling, Pennington, & Lubs, 1983). In a book on learning disorders, I presented a single cognitive deficit view of different disorders and advocated testing their relations with the method of double dissociation (Pennington, 1991).

Such a simple model also guided, either explicitly or implicitly, much other early work on developmental disorders. Since a similar model had been the cornerstone of traditional neuropsychology and was assimilated by the new field of cognitive neuropsychology (Shallice, 1988), it was not surprising that the first applications of the theory and methods of neuropsychology to developmental disorders would also use such a model. For instance, both Morton and Frith (1995) and Pennington

(Pennington, 1991; Pennington & Ozonoff, 1991; Pennington & Welsh, 1995) proposed single cognitive deficit models for understanding developmental disorders like dyslexia and autism. Because the long term goal of these models was to provide a complete causal account of the development of such disorders, these models included four levels of analysis: etiology, brain mechanisms, cognition, and behavior. But the main focus of these models was at the cognitive level of analysis, where the goal was to reduce the variety of behavioral symptoms which define such disorders to a single underlying cognitive deficit, such as a phonological deficit in dyslexia or a theory of mind deficit in autism. If such a reduction were possible, then the model would be X-shaped in Morton and Frith's (1995) terminology: diverse biological causes would converge on a single cognitive deficit which would be necessary and sufficient to cause the diverse behavioral symptoms found in a given disorder.

One commonly recognized complication for single cognitive deficit models of disorders is the possibility of cognitive subtypes, which have been discussed for some time in the case of dyslexia. But usually such subtypes do not seriously threaten the premise that a single cognitive deficit is sufficient to explain the symptoms of a disorder because each subtype can be thought of as having its own distinct single cognitive deficit. For instance, some researchers have used a dual process model to postulate phonological and surface subtypes of developmental dyslexia (Frith, 1985; Temple, 1985a, 1985b). In this formulation, dysfunction in the indirect route for word identification, which relies on grapheme to phoneme rules, results in developmental phonological dyslexia. Dysfunction in the direct lexical route for word identifications results in developmental surface dyslexia.

Morton and Frith (1995) also considered other complications for the single cognitive deficit model, such as the case where a single biological cause, like phenylketonuria (PKU), causes multiple cognitive deficits, which geneticists call "pleiotropy". They also considered the role protective factors might play in the development of disorders. In later work, both Morton (2004) and Frith (2003) have postulated that multiple cognitive deficits may be needed to account for all the features of a complex behavioral disorder, such as autism. They recognized that while a theory of mind deficit provides a good explanation of the problems in social interaction and communication that partly define autism, such a deficit does not readily explain the third part of the autism symptom triad: repetitive behaviors and restricted interests. Nor does a theory of mind deficit explain some of the cognitive strengths found in autism, such as those on certain spatial tasks, like embedded figures. So Frith (2003) postulated a second cognitive deficit in autism, in central coherence, to help account for the behavioral characteristics not well explained by a theory of mind deficit.

Pennington and Ozonoff (1991) explored this simple model and considered alternatives to it. Their paper was particularly concerned with the different possible mappings that might exist across levels of analysis. Besides a 1:1 mapping, they discussed other possible mappings: one-to-many (pleiotropy), two types of many-to-one mapping (causal heterogeneity and multifactorial causation), and a many-to-many mapping (equipotentiality). In causal heterogeneity, each single cause would be necessary and sufficient to produce the same result, thus preserving a 1:1 mapping for each subtype. In multifactorial causation, more than one causal factor is required to yield a given outcome. In their evaluation of multifactorial causation, they argued that if

each multifactor were jointly necessary to produce a disorder, then probability theory dictated a mathematical limit to the number of possible multifactors involved in a common disorder. For instance, if five multifactors were involved, each with a population frequency of 30%, then the probability of having the disorder (2.4 per thousand) would be much lower than the roughly 5–10% prevalence of disorders like dyslexia or ADHD. But they did not consider a multiple deficit model in which the cognitive risk factors are neither necessary nor sufficient, which consequently would not face this mathematical problem. That is the multiple deficit model that is developed later in this paper, which can be thought of as a form of the many-to-many mapping model, but with different weights across the paths between levels of analysis.

They also attempted to deal with the complexity posed by the often numerous behavioral symptoms found in developmental disorders by parsing symptoms into those that were primary (direct effects of the single cognitive deficit), secondary (symptoms caused by the primary symptoms), correlated (produced by the effects of the etiology of primary symptoms on other brain systems), and artifactual (symptoms not causally related to the etiology or pathogenesis of the disorder but associated because of referral biases).

They concluded by making a plea for parsimony: that the simpler single cognitive deficit model should be considered first. As we will see, now nearly 15 years later, the simple single cognitive deficit model has been much more thoroughly tested and its shortcomings are now much more evident.

As I will explain in the rest of this paper, there are several key reasons for rejecting this very simple single cognitive deficit model: (1) behaviorally defined developmental disorders, like dyslexia or ADHD, do not have single causes, either at the etiologic or cognitive levels of analysis, (2) such disorders, instead of being independent, are frequently comorbid and this comorbidity is due to partly shared genetic and cognitive risk factors, and (3) there are other problems with applying double dissociation logic to developmental disorders, which will be discussed at the end of the paper. Because of the key role that research on comorbidity played in the evolution of my thinking, I will begin with an exploration of how I attempted to use the single cognitive deficit model to account for comorbidity.

### **3. Can the single cognitive deficit model account for comorbidity?**

For more than a decade, we have been studying two comorbidities of RD, that with ADHD and that with speech sound disorder (SSD). What we have found poses significant challenges for the single cognitive deficit model.

Comorbidity is an important topic in both child (Angold, Costello, & Erkanli, 1999; Caron & Rutter, 1991) and adult (Clark, Watson, & Reynolds, 1995) psychiatry, partly because it poses challenges for how we categorize disorders and think about their causes. One of the most comprehensive treatments of different possible reasons for comorbidity was provided by Klein and Riso (1993). Neale and Kendler (1995) used quantitative genetic theory to more precisely specify the Klein and Riso (1993) comorbidity models. These models cover both artifactual (chance, sampling

bias, population stratification, definitional overlap, and rater biases) and non-artifactual (alternate forms, multiformity, three independent disorders, and correlated liabilities) explanations of comorbidity. In a separate paper, we presented methods for testing these models and reviewed results from the application of these methods to several comorbidities, including the two considered here (Pennington, Willcutt, & Rhee, 2005). Since I developed my own models for the comorbidity of RD and SSD before I encountered the work by Klein and Riso (1993) and Neale and Kendler (1995), I will explain how my models relate to theirs.

Angold et al. (1999) distinguish *homotypic* and *heterotypic* comorbidity. Homotypic comorbidity is that between disorders from the same diagnostic grouping, such as the comorbidities among different anxiety disorders or that between dysthymia and major depression. Heterotypic comorbidity is that between disorders from different diagnostic groupings, such as the comorbidity between conduct disorder (an externalizing disorder) and depression (an internalizing disorder). In what follows, we will consider an example of each kind of comorbidity. The comorbidity between RD and SSD is a homotypic comorbidity because both RD and SSD can be thought of as language disorders, and the comorbidity between RD and ADHD is a heterotypic comorbidity because ADHD is an externalizing disorder, not a language disorder.

To be consistent with a single cognitive deficit model, each kind of comorbidity requires a distinct explanation. As we will see, homotypic comorbidity could be explained by each disorder being an alternate form of the same underlying etiologic and cognitive risk factors, perhaps with one form being either a more severe manifestation (as major depression is in relation to dysthymia) or an earlier developmental manifestation (as might be true for the relation between separation anxiety disorder and later social phobia). So our favored hypothesis to explain the homotypic comorbidity between RD and SSD was a *severity* hypothesis in which SSD was both an earlier and more severe form of the same etiology and cognitive deficit underlying RD. But it is a harder task for a single cognitive deficit model to account for a heterotypic comorbidity, such as that between RD and ADHD. It must explain the comorbidity as either an artifact of some kind, as a superficial relation (the phenocopy hypothesis, which corresponds to a multiformity model in Neale and Kendler's account), or as a disorder distinct from either of the "pure" disorders (the three independent disorders hypothesis). These were the hypotheses we initially tested when we first encountered the unexpected comorbidity between RD and ADHD. As we will see, we were forced to abandon our favored hypotheses for each kind of comorbidity, homotypic and heterotypic, thus leading us to question the single cognitive deficit model.

### 3.1. *Dyslexia and ADHD*

Somewhat surprisingly, dyslexia is comorbid with ADHD, even though our single deficit theories of each disorder implicate what seem to be distinct cognitive processes: a phonological deficit in dyslexia and an inhibition deficit in ADHD (Pennington, Groisser, & Welsh, 1993). In what follows, I will first review the symptom

and genetic overlap between dyslexia and ADHD. Then I will review what we know about their cognitive overlap and then discuss how what we have learned about this comorbidity argues for a multiple deficit model over a single cognitive deficit model.

### 3.1.1. *Symptom overlap*

Dyslexia and ADHD co-occur more frequently than would be expected by chance and this comorbidity is found in both clinical and community samples. Across studies, around 25–40% of children with either dyslexia or ADHD also meet criteria for the other disorder (August & Garfinkel, 1990; Dykman & Ackerman, 1991; Semrud-Clikeman et al., 1992; Willcutt & Pennington, 2000).

Two artifactual explanations of this comorbidity can be rejected. Because the comorbidity is found in community samples, it is not due to a selection artifact (e.g., Berkson's bias). Because dyslexia is defined by cognitive tests whereas ADHD is defined by behavior ratings (usually from teachers and parents), the comorbidity is not due to definitional overlap.

### 3.1.2. *Apparent cognitive independence*

We began our studies of the relation between dyslexia and ADHD using a single cognitive deficit model. Since there was evidence for distinct cognitive deficits underlying each disorder, we hypothesized there ought to be a double dissociation between them. Dyslexia would have a phonological deficit but not an executive deficit, whereas ADHD would have an opposite profile: an executive deficit but not a phonological deficit. To explain their surprising comorbidity, we hypothesized that dyslexia as a primary disorder caused *just* the symptoms of ADHD, which is the symptom phenocopy hypothesis (an example of one of Neale & Kendler's, 1995, multiformity models).

We tested both the hypothesized double dissociation between the two pure disorders and the symptom phenocopy hypothesis using a  $2 \times 2$  design in which ADHD (presence vs. absence) was crossed with dyslexia (presence vs. absence), yielding four groups: those with neither disorder (controls), those with dyslexia only or ADHD only, and those with both disorders (comorbid group). We gave these groups multiple measures of phonological processing and executive functions and compared their performance across the two resulting cognitive composites (Pennington et al., 1993). We found the hypothesized double dissociation and a profile in the comorbid group that fit the symptom phenocopy hypothesis: the comorbid group was impaired on the phonological composite but not on the executive composite. Hence, this comorbid group had the symptoms of ADHD, but not the underlying executive deficit, as the phenocopy hypothesis would predict. So, these results fit a single deficit model of each disorder.

But subsequent studies using this same design (Nigg, Hinshaw, Carte, & Treuting, 1998; Reader, Harris, Schuerholz, & Denckla, 1994), including one of our own (Willcutt et al., 2001), did not replicate the Pennington et al. (1993) result for the comorbid group, although they generally replicated the double dissociation found between the two "pure" groups. The comorbid group in these subsequent studies had a generally additive combination of the deficits found in each pure group. That is, they had both

phonological and executive deficits. So these replicated results rejected the phenocopy hypothesis. They could be consistent with a version of the three independent disorders hypothesis in which one etiology leads to a phonological deficit and RD only, a second etiology leads to an executive deficit and ADHD only, and a third etiology pleiotropically produces both cognitive deficits and both disorders. Since genetic methods provide a clearer test of the three independent disorders hypothesis, such methods are considered next.

### 3.1.3. Genetic overlap

We have used genetic methods in several studies to test hypotheses for the comorbidity between RD and ADHD. We have found that this comorbidity cannot be explained by assortative mating (Friedman, Chhabildas, Budhiraja, Willcutt, & Pennington, 2003), which is an example of a population stratification artifact. Instead, there is evidence for a genetic overlap between dyslexia and ADHD. One commonly used method to test for a genetic overlap tests whether the relation between two traits is greater in MZ pairs than in DZ pairs. If it is, we say there is “bivariate heritability” for the two traits, meaning that some of the genetic influences on the first trait are the same as some of the genetic influences on the second trait. Several studies have found significant bivariate heritability between dyslexia and ADHD (Light, Pennington, Gilger, & DeFries, 1995; Stevenson, Pennington, Gilger, DeFries, & Gillis, 1993; Willcutt, Pennington, & DeFries, 2000). In addition, we also found that this bivariate heritability between RD and ADHD is more pronounced for the inattentive symptoms of ADHD than the hyperactive/impulsive ones (Willcutt et al., 2003). Since these studies found genetic overlap between RD and ADHD even when the index case (proband) had *only* RD or ADHD, these findings reject the version of the three independent disorders model described above. Instead, they support the correlated liabilities model, in which some, but not all, of the etiologic risk factors for each disorder are shared by both disorders. But accepting this comorbidity model for a heterotypic comorbidity is problematic for a single cognitive deficit model, which holds that the two “pure” disorders are independent.

We have begun to test which genes underlie the bivariate heritability of RD and ADHD using molecular genetic methods. We found that the dyslexia risk locus on chromosome 6p21.3 is also a susceptibility locus for ADHD (Willcutt et al., 2002). There are two recent reports of other risk loci shared by RD and ADHD (Gayan et al., 2005; Loo et al., 2004) including loci on chromosomes 3, 10, 13, 15, 16, and 17. Although these results need to be replicated, there is thus accumulating molecular evidence for a partial genetic overlap between RD and ADHD.

So, these results explain the comorbidity between RD and ADHD in terms of partially shared etiologic risk factors, which is what the multiple deficit model predicts. But the cognitive results discussed earlier, which found the two isolated disorders were cognitively independent, do not fit well with the finding of a genetic overlap. So, the next question is whether there are actually other shared cognitive risk factors underlying the comorbidity between dyslexia and ADHD.

#### 3.1.4. Cognitive overlap

In the Willcutt et al. (2001) study which used a  $2 \times 2$  design, we found that the cognitive phenotype in children with both RD and ADHD was essentially the sum of the phenotype in each disorder considered separately. Once full scale IQ and subclinical elevations of RD or ADHD were controlled statistically, we found main effects of ADHD on measures of inhibition (Stop Task and Continuous Performance Test) and main effects of RD on measures of phoneme awareness (PA) and verbal working memory (WM), but no significant interactions. The one caveat is that there was a trend for an RD main effect on inhibition measures, indicating that the double dissociation between RD and ADHD was not totally clean. So the etiology of isolated ADHD presumably led to an inhibition deficit but did not affect the cognitive markers of RD: PA and verbal WM. In contrast, the etiology of isolated RD did have some marginal effects on inhibition, which is the cognitive marker for ADHD. This asymmetric pattern would be consistent with the pleiotropy hypothesis. That is, the RD risk locus on 6p affects two largely separate cognitive processes, phonological processing and inhibition, and individuals with this risk locus are more likely to have deficits on both, thus explaining the comorbidity between RD and ADHD.

However, this cognitive pleiotropy hypothesis is less parsimonious than one in which shared etiologic risk factors lead to shared cognitive risk factors because the cognitive pleiotropy hypothesis must explain why the shared etiological risk factor leads to both cognitive deficits in some individuals and not others. So perhaps by examining other cognitive deficits, we might find more evidence for a cognitive overlap between RD and ADHD.

In a new sample of subjects also using a  $2 \times 2$  design, and now including measures of processing speed, we found more evidence for cognitive overlap between RD and ADHD (Willcutt, Pennington, Olson, Chhabildas, & Hulslander, 2005). The RD only, ADHD only, and RD + ADHD groups had deficits of similar magnitude on the processing speed factor. So a deficit in processing speed might be a cognitive risk factor that is shared by RD and ADHD.

The processing speed measures in this study included the Wechsler Coding and Symbol Search subtests, and the color and word naming baseline conditions of the Stroop. So they included both linguistic and non-linguistic measures of processing speed. Because rapid serial naming (RSN) tasks can also be thought of as measuring processing speed, we included them in a follow-up study (Shanahan et al., in press). A factor analysis of various processing speed measures found two factors: a verbal factor (on which RSN and the Stroop baseline conditions loaded) and a motor factor (on which Wechsler Symbol Search and Coding loaded). Again using a  $2 \times 2$  design, we found main effects of *both* ADHD and RD status on *both* factors. There was also an ADHD  $\times$  RD interaction, such that the comorbid group was *less* impaired on each factor than the sum of the deficits found in each pure group. Given these results, we reasoned that RD and ADHD were *not* cognitively independent in the domain of processing speed. So in this study we did find evidence for a cognitive risk factor that is shared by RD and ADHD.

In sum, our cognitive studies of the heterotypic comorbidity between RD and ADHD have found both cognitive deficits specific to each disorder (in phonological

processing and behavioral inhibition, respectively) and a shared cognitive deficit (in processing speed). This account of their cognitive relation fits well with their partial genetic overlap, but it contradicts the single cognitive deficit model.

The view that behavioral inhibition, or some other executive function (EF) deficit, is the single cognitive deficit underlying ADHD faces other challenges. A recent meta-analysis of studies of executive function measures in ADHD (Willcutt, Doyle, Nigg, Faraone, & Pennington, 2005) found a medium effect size (0.46–0.69) for various EF deficits in groups with ADHD, and such deficits were not accounted for by group differences in intelligence, academic achievement, or symptoms of other disorders. However, because of the medium effect sizes and the presence of many individuals with ADHD without an EF deficit, these authors argued that a single EF deficit (such as a deficit in behavioral inhibition) does not provide an adequate explanation of most cases of ADHD. They suggested a multiple deficit model is needed. Other researchers studying the neuropsychology of ADHD are also advocating a multiple deficit view of this disorder (Castellanos & Tannock, 2002; Nigg, Willcutt, Doyle, & Sonuga-Barke, 2005; Sonuga-Barke, 2005).

### 3.2. *Dyslexia and speech sound disorder (SSD)*

SSD (Shriberg, Tomblin, & McSweeney, 1999) involves difficulties in the preschool development of *spoken* language, specifically problems with the accurate (and therefore intelligible) production of speech sounds in spoken words (it is distinct from stuttering or mutism). SSD was formerly called *articulation* disorder (which emphasized putative problems in the motor programming of speech) and *phonological* disorder (which emphasized putative problems in the cognitive representations of speech). Since each of these terms made a premature commitment to the underlying processing deficit that causes the speech production problem, the neutral and descriptive term SSD is now preferred. SSD is frequently comorbid with specific language impairment (SLI), which is defined by deficits in grammar and vocabulary, and it is also comorbid with RD, which manifests at school age as difficulty learning *written* language, specifically printed word recognition and spelling (see IDA and NICHD working definition of dyslexia, Dickman, 2003). Next we will review the evidence for the comorbidity between SSD and RD and the accumulating evidence for both an etiological and cognitive overlap between them.

#### 3.2.1. *Symptom overlap*

It is already well documented that children with early speech/language problems are at increased risk for later literacy problems (Aram, Ekelman, & Nation, 1984; Bishop & Adams, 1990; Catts, Fey, Tomblin, & Zhang, 2002; Hall & Tomblin, 1978; Magnusson & Naucler, 1990; Rutter & Mawhood, 1991; Scarborough & Dobrich, 1990; Snowling, Bishop, & Stothard, 2000; Snowling & Stackhouse, 1983; Tomblin, Freese, & Records, 1992). In these studies, roughly 30% of children with early speech/language problems go on to have RD. It is also well documented that individuals with literacy problems retrospectively report increased rates of earlier speech and language problems (Hallgren, 1950; Rutter & Yule, 1975). Moreover, the latter

association is not limited to retrospective reports because young children selected for family risk for RD also have higher rates of preschool speech and language problems than controls (Gallagher, Frith, & Snowling, 2000; Lyytinen et al., 2002; Pennington & Lefty, 2001; Scarborough, 1990). But these previous studies have rarely distinguished SSD from specific language impairment (SLI), which is defined by deficits in semantics and syntax. So, it is less clear which subtypes (or components) of SSD per se presage which kinds of later literacy problems.

### 3.2.2. *Cognitive and etiological overlap*

It is well known that the large majority of children with RD have deficits on measures of phonological processing (Wagner & Torgesen, 1987), including measures of both explicit (i.e., phoneme awareness) and implicit (i.e., phonological memory and rapid serial naming) phonological processing. There is also accumulating evidence that many children with speech and language problems have phonological processing problems, such as deficits on measures of phoneme awareness and phonological memory (Bird & Bishop, 1992; Bird, Bishop, & Freeman, 1995; Bishop, North, & Donlan, 1995; Clarke-Klein & Hodson, 1995; Edwards & Lahey, 1998; Kamhi, Catts, Mauer, Apel, & Gentry, 1988; Leonard, 1982; Lewis & Freebairn, 1992; Montgomery, 1995).

Support for a shared etiology for SSD and RD has been provided by Lewis and colleagues (Lewis, 1990, 1992; Lewis, Ekelman, & Aram, 1989), who found that SSD and RD are co-familial, meaning that both disorders run in the same families. Co-familiality could be explained by either genetic or environmental risk factors shared by family members, so finding co-familiality does not necessarily mean the two disorders share genetic risk factors (i.e., are co-heritable). Using a twin design, we have found that SSD and RD are co-heritable as well (Tunick & Pennington, 2002).

### 3.2.3. *Hypotheses to explain SSD/RD comorbidity*

For a single cognitive deficit theorist, the etiological and cognitive overlap between SSD and RD suggests the parsimonious *severity* hypothesis (cf. Harm & Seidenberg, 1999), which was discussed earlier. The severity hypothesis holds that many cases of SSD and RD lie on a severity continuum in which shared etiological risk factors lead to a shared underlying phonological deficit. In other words, SSD and RD are alternate forms of the *same* disorder. If the phonological deficit is severe enough, it first produces SSD and then later RD. If it is less severe, it does not produce diagnosable SSD (though it may lead to subclinical speech production problems), but it does produce later RD, because reading requires more mature phonological representations than does speech. So this hypothesis posits that RD without earlier SSD is a less severe variant of SSD. To account for the many children with SSD who do not develop later RD, the severity hypothesis must posit that they have a subtype of SSD that is caused by different underlying cognitive deficit. If they had the same underlying phonological deficit as SSD children with later RD, and if this single deficit were sufficient to produce RD as the severity hypothesis posits, then there would be a contradiction. Because phonology is complex, SSD children without later RD could conceivably have a different kind of phonological deficit than SSD

children with later RD, such as a deficit in output phonology as opposed to input phonology, or they could have an altogether different cognitive deficit. But the main point is that the single cognitive deficit severity hypothesis must be rejected if SSD children without later RD have a phonological deficit similar to that found in SSD children with later RD or RD children without earlier SSD.

In sum, the already documented etiological and cognitive overlap between SSD and RD supports this severity hypothesis, but the fact that well over half of children with preschool SSD do not develop later RD poses a serious problem for the severity hypothesis, one that only can be solved by positing that they represent a distinct cognitive subtype of SSD. Moreover, because SSD has not been clearly distinguished from SLI in previous etiological and cognitive studies, there are other possible hypotheses to explain the relation between SSD and RD.

Some years ago, in an NIH grant application, we proposed five competing hypotheses (all but one of which were single cognitive deficit hypotheses) to account for the comorbidity of SSD and RD (Fig. 1). The first of these was the severity hypothesis that we have just discussed. The logic of these hypotheses can be more clearly understood by thinking of them in a  $2 \times 2$  contingency table formed by crossing two distinctions, common vs. specific etiologies and common vs. specific cognitive phenotypes. In the severity hypothesis, there is both a common etiology and a common cognitive phenotype (except for the subtype of SSD without later RD). In the synergy and assortment hypotheses, both etiologies and cognitive phenotypes are specific. The synergy hypothesis (Scarborough & Dobrich, 1990) postulates that SSD only leads to later RD when SSD co-occurs with SLI. It also postulates that RD without earlier SSD is a distinct disorder from RD with earlier SSD. Hence, the synergy hypothesis hinges on an explanation for the comorbidity between SSD and SLI, which in the diagram is explained by a partial etiologic and a partial cognitive overlap between these two disorders. The assortment or non-random mating hypothesis is an example of the population stratification artifact mentioned earlier, which is one of the explanations for comorbidity posited by Klein and Riso (1993). In the assortment model, SSD and RD are distinct disorders in terms of both etiology and cognitive phenotype, but comorbidity arises because a parent with SSD is more likely to marry a spouse with RD (e.g., non-random mating). Because each disorder is

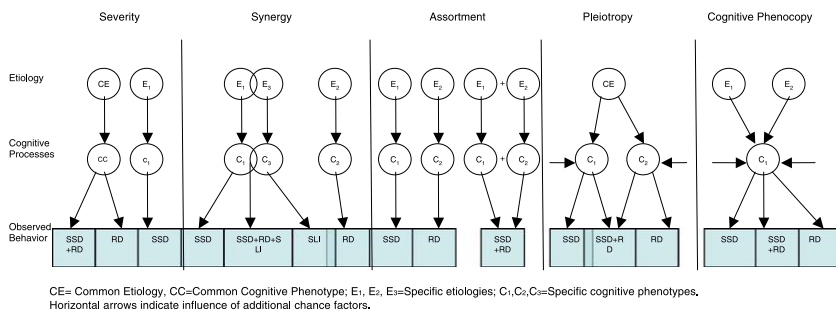


Fig. 1. Schematic representation of five hypotheses.

heritable on its own, offspring of such a mating are at increased risk for *both* disorders, and thus some of them are comorbid.

In the pleiotropy hypothesis there is a common etiology, but specific cognitive phenotypes, whereas in the cognitive phenocopy hypothesis, there are specific etiologies but a common cognitive phenotype. We have discussed pleiotropy earlier as a possible explanation for the comorbidity between RD and ADHD. The pleiotropy hypothesis posits that a shared etiology can variably lead to two distinct cognitive phenotypes, one of which is sufficient to produce SSD, and one of which is sufficient to produce RD. Because of chance factors (sideways arrows), some individuals will have only one of these cognitive phenotypes (pure cases) and some will have both (comorbid cases). So, the disorders will be cognitively independent and thus the phenotype in comorbid cases will be the sum of that in pure cases (which is essentially what we found for EF and PA deficits in the case of RD and ADHD, as discussed earlier). Finally, we come to the cognitive phenocopy hypothesis, which is admittedly the least transparent of the five. The cognitive phenocopy hypothesis posits that there are two distinct etiologies for the same cognitive phenotype (e.g., etiological heterogeneity), which is perfectly plausible. But it next posits that because of chance factors (sideways arrows), this common cognitive phenotype has variable manifestations at the symptom level, sometimes producing RD or SSD alone (pure cases) and sometimes producing both (comorbid cases).

In sum, these five hypotheses were: (1) severity (both etiology and cognitive phenotype are shared, but comorbid children have a more severe phonological deficit and SSD only children have a distinct etiology and a distinct cognitive phenotype); (2) synergy (the etiologies and cognitive phenotypes of SSD and RD are distinct, but comorbidity between SSD and *SLI* produces later RD); (3) cross-assortment or non-random mating (both the etiology and cognitive phenotypes are distinct, but individuals with SSD or RD are more likely to select mates with RD or SSD, thus transmitting risk alleles for both disorders to their children); (4) pleiotropy (a shared etiology leads to two distinct cognitive phenotypes, which co-occur in comorbid children); and (5) cognitive phenocopy or genetic heterogeneity (distinct etiologies lead to a shared cognitive phenotype, thus producing comorbidity). These hypotheses were generated without knowledge of the Klein and Riso (1993) hypotheses. Nonetheless, all but one of them (cognitive phenocopy) correspond to one of their hypotheses. The severity and pleiotropy hypotheses correspond to different versions of the alternate forms hypothesis. Synergy is partly similar to the three independent disorders hypothesis, and assortment is an example of a population stratification artifact as discussed earlier. It can also be seen that of the five hypotheses, only the synergy hypothesis clearly involved the interaction of multiple deficits.

But even though the other four hypotheses are based on a single deficit model, it can readily be seen that several of them require additional factors to explain the diversity in outcomes at the symptom level. For the *severity* hypothesis, children with SSD + RD must have more of the shared etiologic risk factors than those with RD alone and these must be a distinct pathway leading to SSD without later RD. For the *pleiotropy* hypothesis, the additional factor, chance, does all the “heavy lifting” because it determines which cognitive phenotypes are affected. The *cognitive*

*phenocopy* hypothesis also requires that one postulate chance factors which determine whether a common cognitive deficit is expressed as SSD, RD or both at the symptom level. So in trying to extend the single deficit model to the problem of comorbidity, we were forced to postulate additional risk factors. In sum, SSD illustrates well the potential contradiction posed by the intersection of a single (phonological) deficit model of RD and the phenomenon of comorbidity. Although SSD is comorbid with RD and appears to share a phonological deficit with it, the contradiction arises because the comorbidity is not complete: not all children with SSD develop later RD and not all children with RD had earlier SSD.

### 3.2.4. *Tests of the five key hypotheses*

To distinguish these five hypotheses, three questions need to be addressed. (1) Do SSD and RD share a common genetic etiology? (2) Do they share an underlying cognitive phenotype? (3) Is there assortative mating between individuals with SSD and those with RD? In what follows, I present what we and others have learned about the answers to these questions.

First, there is now stronger evidence for a shared genetic etiology between RD and SSD, which rejects the phenocopy, synergy, and assortment hypotheses, all of which posit distinct etiologies for RD and SSD. Several replicated risk loci or QTLs for RD have been identified, on chromosomes 1p, 2p, 3p-q, 6p, 15q, and 18p (Fisher & DeFries, 2002). Two groups have now tested whether some of the risk loci already identified for RD are also risk loci for SSD. Stein and colleagues found that SSD is linked to the RD locus on chromosome 3 (Stein et al., 2004). They tested several related phenotypes, including SSD itself, phonological memory, phonological awareness, and reading. All of these phenotypes were linked to the RD risk locus on chromosome 3, indicating that this locus affects phonological development and contributes to the comorbidity between SSD and RD. We have also found that SSD is linked to RD risk loci on chromosomes 6 and 15, and perhaps 1 (Smith, Pennington, Boada, & Shriberg, 2005). We also tested multiple phenotypes, including SSD itself, phonological memory, and phonological awareness, all of which provided evidence for linkage.

In sum, while there appear to be QTLs shared by SSD and RD, we do not expect that all the QTLs affecting either disorder will be shared (a genetic correlation of 1.0), nor that all the environmental risk factors affecting either disorder will be shared. Instead, it is more likely that some of the genetic and environmental risk factors will be shared and some will be distinct. So, while we can reject the hypotheses in Fig. 1 that posit completely distinct etiologies for SSD and RD, that does not mean that we can confirm a complete etiological overlap.

The second test of these five hypotheses is whether RD and SSD share an underlying cognitive deficit. To perform this test, we examined preliteracy skills, including phoneme awareness, in a large sample of preschool children with SSD (Raitano, Pennington, Tunick, & Boada, 2004). Since we were also interested in whether the cognitive deficit in SSD varied by subtype, we divided the sample along two dimensions, presence vs. absence of SLI, and persistent vs. normalized speech problems. We found that a phoneme awareness (PA) deficit was pervasive across the four resulting

subtypes of SSD, although its severity varied in an additive fashion as a function of each subtype dimension. Those with SLI had a worse PA deficit than those without SLI; those with persistent speech problems had a worse PA deficit than those whose speech had normalized. A similar pattern of results was found for alphabet knowledge. Intriguingly, the SSD group as a whole had a less pronounced deficit in rapid serial naming than in PA or alphabet knowledge. So the results of this study, along with other evidence reviewed earlier, indicate that there appears to be a shared underlying phonological deficit in SSD and RD, and that this shared deficit is found in all four subtypes of SSD. However, it is always possible that the subtypes performed poorly on the PA measures for different reasons. Otherwise the results of this second test reject the synergy, assortment, and pleiotropy hypotheses and only partially support the severity hypothesis, which requires a fairly common subtype of SSD (those without later RD) with a different cognitive deficit. The fact that the PA deficit is not restricted to the group with both SSD and SLI is also inconsistent with the predictions of the synergy hypothesis, which is also contradicted by the genetic results just discussed.

To address the third question regarding assortative mating, we examined the parents in our large sample of children with SSD (Tunick, Boada, Raitano, & Pennington, submitted). Relative to control parents, parents of SSD probands reported higher rates of both speech and reading problems, indicating that SSD was familial in this sample and that SSD and RD were co-familial. We also found similar results in the siblings of probands; they had higher rates of speech problems and worse scores on preliteracy measures than controls. These results indicate an etiological overlap between SSD and RD, consistent with the studies discussed earlier. In contrast, we found low rates of cross-assortment in these parents. Moreover, SSD probands with comorbid preliteracy problems rarely came from cross-assorted parents. So we did not find support for the assortment hypothesis.

In sum the results of these three tests reject all but the severity hypothesis. But despite the fact that the severity hypothesis garners some support from these data and that of previous studies reviewed earlier, there are still significant challenges to how it accounts for the nature of the comorbidity between SSD and RD. The severity hypothesis proposes that SSD and RD are comorbid because they share etiological risk factors (some of which are genetic) and these lead to a shared phonological deficit, which is more *severe* in children with comorbid SSD and RD than children with RD only. To account for SSD children who do not become RD, the severity hypothesis must postulate a subtype of SSD with a distinct etiology and a different underlying cognitive deficit. If SSD children without later RD nonetheless have the same underlying phonological deficit, the severity hypothesis must be seriously questioned. But the results of Raitano et al. (2004) just discussed suggest there is not a common subtype of SSD without a PA deficit. Clearer evidence on this point is provided by long term follow-up study (Snowling et al., 2000) of SSD children initially identified by Bishop at preschool age. These researchers found there were former SSD children with a persistent deficit in PA in adolescence who are nonetheless normal readers. Both these results are inconsistent with the severity hypothesis.

New data from a dissertation project in our lab by Tunick (2004), also questions the severity hypothesis. Her project involved two comparisons of SSD and RD, one between probands at age 5 and one between siblings around age 8. The goal of the proband comparison was to test which deficits are shared and specific to each disorder before the onset of literacy instruction. The sibling comparison tested whether a similar pattern of shared and specific deficits is also found in siblings, at a later age.

Since both SSD and RD vary in the severity of the symptoms that define them diagnostically, it is important to compare SSD and RD groups that are similar in severity. Consequently, we matched the SSD and RD proband groups on severity, as well as on age and gender. The 23 SSD probands were selected from the entire sample of SSD probands in our current study so as to match the 23 RD probands from our earlier longitudinal study of children at high family risk for RD (Pennington & Lefty, 2001). The RD probands were all the children in the high family risk group who were later diagnosed as RD at follow up. For the sibling comparison, we recruited a separate sample of RD siblings and matched them to a subset of our current sample of SSD siblings on (1) proband sibling's diagnostic severity, (2) their own diagnostic severity, and (3) age and gender.

The comparison of the profiles of phonological processing deficits in probands and siblings tests the severity hypothesis, which predicts similar profiles in each disorder, with greater impairment in the SSD group. The profiles consisted of three phonological processing constructs: phonological awareness, phonological memory, and rapid serial naming. In the proband comparison, somewhat different measures of the same constructs had been used with each group, so their *z*-scores relative to matched controls were used to compare the SSD and RD proband groups. In the sibling comparison, the same measures were used in each group. Tunick (2004) found that SSD and RD probands share a deficit of similar magnitude (relative to their controls) on the phonological awareness composite, but had significantly different profiles across the other two phonological constructs, rapid serial naming (RSN) and phonological memory (PM), producing a significant group  $\times$  domain interaction. This interaction arose because the SSD proband group performed significantly better than the RD proband group on the RSN composite, and non-significantly worse on the PM composite. This interaction replicated in the sibling comparison, in which the *same* measures of these constructs were used in each group. The relative strength on the RSN composite in both SSD groups is a somewhat surprising finding, given that one would expect a slower articulatory rate in SSD. So, it will be important to replicate this result in another SSD sample. But this finding could help explain why not all SSD children develop later RD, despite having a PA deficit. In sum, Tunick's (2004) results do not support the predictions of the single deficit, severity model because the PA deficit is not more severe in the SSD group.

These difficulties with the severity hypothesis led us to develop an alternative *multiple* cognitive deficit model of RD and SSD, which is presented later. In this multiple deficit model, comorbidity between these two disorders arises from a shared cognitive deficit (in phonological representations), which interacts with other non-shared cognitive deficits to produce the symptoms that distinguish the two disorders.

The severity and the multiple deficit hypotheses make competing predictions about the literacy outcome of children with SSD. The severity hypothesis predicts (1) that SSD children who do not develop later RD (SSD-only children) have a distinct form of SSD without the kind of underlying phonological deficit found in either SSD + RD or RD alone, and (2) that SSD children who do develop later RD (comorbid children) have a more severe phonological deficit than both RD children without earlier SSD (RD-only children) and RD children in general (since only about 30% of RD children had earlier SSD). In contrast, the multiple deficit hypothesis predicts (1) that SSD-only children may have a similar phonological deficit but differ on other cognitive risk and protective factors, and (2) that comorbid children will not necessarily have a more severe phonological deficit than RD-only children or RD children in general, but they must have an additional cognitive risk factor to explain why they have RD. We will test these competing hypotheses in the longitudinal follow-up of the SSD probands in [Raitano et al. \(2004\)](#).

In sum, both comorbidities considered here appear to be explained by partly shared and partly distinct genetic and cognitive risk and protective factors. So a developmental disorder like RD represents the interaction of multiple cognitive risk factors, some of which are shared with other comorbid disorders. This multiple cognitive deficit model of developmental disorders is quite different from the single cognitive deficit that guided our earlier research. Next we turn to the task of specifying this multiple cognitive model more explicitly. We begin by first considering other earlier multiple deficit views of dyslexia.

#### **4. Other multiple deficit views of dyslexia**

The possibility of multiple deficits in dyslexia is itself not new and has appeared repeatedly in different forms in the literature. This possibility probably first appeared in early attempts to identify subtypes of dyslexia characterized by different neuropsychological profiles on clinical test batteries (e.g., [Doehring, Trites, Patel, & Fiedorowicz, 1981](#); [Mattis, French, & Rapin, 1975](#); [Satz, Morris, & Fletcher, 1985](#)). Some of the subtypes that were identified had multiple deficits. Research on these early subtype proposals uncovered methodological and statistical problems that will need to be addressed by the new multiple deficit model proposed here. One was the problem of distinguishing associated cognitive deficits from causal ones. For instance, because there are individual differences in spatial cognition, there will almost inevitably be some poor readers who are also poor at spatial cognition. But this association could be irrelevant for understanding their reading problem. The second was the assumption that subtypes would correspond to discrete bumps or clumps in the univariate or multivariate distribution. Much of this subtyping work relied on statistical techniques, like cluster analysis, to identify subtypes which occupied particular regions of the multivariate continuum. But it soon became evident that these techniques would also find clusters in random data. So, while the subtypes identified might be statistically reliable, there was no guarantee that they were valid. Without a strong cognitive theory of reading development, which specifies how multiple cognitive processes

interact to produce skilled reading, it is difficult to interpret subtypes identified by such statistical methods.

Somewhat later, Vellutino, Scanlon, and Tanzman (1991) hypothesized that subtypes of dyslexia might be characterized by a weighted profile of more than one deficit. In this formulation, a deficit in one area might be tolerated unless it co-occurs with other deficits. Watson and Willows (1993) argued for a multiple deficit model in the context of considering possible visual deficits in dyslexia, which could interact with a phonological deficit to produce the disorder. Badian (1997) proposed a triple-deficit model of dyslexia, in which the combination of phonological, orthographic, and rapid naming deficits produced a more severe form of dyslexia than would be found with only double or single deficits. Morris et al. (1998) used cluster analytic methods to identify seven multiple deficit subtypes of RD. In terms of the single deficit model, it is striking that a phoneme awareness (PA) only subtype did *not* emerge. Instead, although PA deficits were found in all but one of the subtypes, they occurred with other deficits. But we are still left with uncertainty about which of the co-occurring deficits play a causal role in which kinds of reading deficits. For instance, Ramus (2003) reviews evidence for co-occurring sensory and motor deficits in dyslexia, but argues they play a limited role in causing the disorder.

The next section presents previous work involving the idea of multiple deficits in dyslexia that is particularly relevant to the new model proposed here. This previous work has contributed several ideas which are part of this new model, including the idea of a multivariate continuum of reading-related cognitive skills, interactive processing and consequent tradeoffs in development, and the importance of unitization and automaticity in reading development. There is also relevant evidence for multiple deficits in dyslexia in languages other than English.

#### 4.1. *Multivariate continuum*

Various researchers (e.g., Olson, Kliegl, Davidson, & Folz, 1985; Stanovich, 1988) have proposed that a multivariate continuum characterizes cognitive skills related to reading, meaning that the boundaries between putative subtypes would be arbitrary. Based on this finding, Stanovich (1988) proposed a “phonological-core variable-difference model”. In this model, individuals with relatively pure developmental phonological dyslexia occupy a region of multivariate space. As one moves away from that region, the specificity of the underlying phonological deficit decreases and one encounters poor readers with other associated cognitive deficits. According to this model, pure phonological dyslexics are more likely to be found among those poor readers who meet an IQ discrepancy definition, whereas those that do not are more likely to be “garden-variety” poor readers. Although Stanovich (1988) acknowledged that some of the associated deficits found among less pure poor readers were likely to be consequences rather than causes of poor reading, the article did not propose a model of how non-phonological deficits could cause reading problems or how phonological and non-phonological deficits might interact. The double-deficit model (Wolf & Bowers, 1999) which is considered later, addressed these issues.

#### 4.2. Interactive processing

Another multiple deficit view of dyslexia comes from connectionist models of single word reading. These models have certainly produced surprises for single deficit theories of dyslexia syndromes. When Hinton and Shallice (1991) modeled deep dyslexia, the a priori prediction was that semantic errors would be produced by damage to a particular component of the network. Instead, it turned out that damage practically anywhere in the network produced such errors because of the interactive nature of the processing involved in going from print to semantics.

A similar conclusion was reached for errors in reading pseudowords and exception words, respectively, in the application of connectionist models to dyslexia subtypes (Manis, Seidenberg, Doi, McBride-Chang, & Petersen, 1996). Each error type is the hallmark of a different dual-process subtype of developmental dyslexia (i.e., phonological vs. surface) and is thought to reflect damage to distinct mechanisms (e.g., Temple, 1985a, 1985b). What Manis et al. (1996) demonstrated was that although the network could be altered in different ways to produce each subtype, these alterations did not produce clean dissociations. An alteration that mainly affected exception word reading also impacted the accuracy of reading pseudowords and *vice versa*. Once again the interactive nature of network dynamics is responsible.

Drawing on this connectionist work, Snowling and colleagues (Snowling, 2002; Snowling et al., 2000) have proposed a multiple deficit model that has many similarities to the multiple deficit model proposed here. Based on the connectionist models of normal and abnormal reading developed by Seidenberg and colleagues (Harm & Seidenberg, 1999; Plaut, Seidenberg, McClelland, & Patterson, 1996; Seidenberg & McClelland, 1989), these researchers emphasized the interactive nature of reading development. In their view, a strength in semantic codes may offset a phonological deficit, so it is the interaction of multiple cognitive factors that determines whether one has dyslexia. They also used this model to understand poor comprehenders, who are normal at decoding single printed words but impaired in reading comprehension, and to criticize the severity hypothesis discussed earlier because it only focuses on one component of the several that interact in reading.

#### 4.3. Unitization and automaticity

The double deficit model (Bowers, 1993; Wolf & Bowers, 1999) proposes a non-phonological cause of RD. Similar to other subtyping models, this model proposes two single cognitive deficit subtypes of RD, the familiar phonological deficit subtype and a new, naming-speed deficit subtype. Dyslexics with both deficits have a “double deficit” and are predicted to have more severe reading problems than those found in either single deficit subtype, although the model is not explicit about whether the joint effects of the two deficits are additive or interactive. It is well documented that groups with RD perform poorly on measures of rapid serial naming and that such

measures predict later reading skill, especially reading fluency, in longitudinal studies (e.g., Pennington & Lefly, 2001). What is novel about the double deficit model is the claim that rapid serial naming (RSN) deficits are non-phonological in nature and the focus on the combined effects of a double deficit.

The double deficit model proposed that the non-phonological aspect of RSN tasks is in precise timing mechanisms. Unlike the phonological hypothesis which focuses on a spoken-language *representational* deficit that precedes reading acquisition, this aspect of the double deficit hypothesis focuses on a *processing* component necessary for achieving automaticity in reading itself. Bishop (1997) also emphasized the importance of processing accounts of developmental disorders. So, the double deficit model has both a competence deficit and a processing deficit. Automaticity in reading requires unitized orthographic codes which are well integrated with phonological and semantic codes. Automaticity also requires the ability to rapidly redeploy visual attention across individual words in a line of text. So a child could have normal phonological and semantic codes, have the visual cognitive skills necessary for learning orthographic codes, but still have trouble forming *connections* between orthographic codes and the already established phonological and semantic codes. Since RSN tasks require automatic connections between visual representations (of colors, objects, letters, or numbers) and their phonological name codes, and since such tasks require rapid redeployment of attention across a line of items, RSN tasks arguably tap both of these processes that are necessary for automaticity in reading.

While the double deficit model predicts that a single phonological deficit is sufficient to cause dyslexia, the results presented earlier on SSD suggest otherwise. That is because there are SSD children with a persisting phonological deficit but not a reading disability (Snowling et al., 2000). The results from Raitano et al. (2004) and Tunick (2004) indicate that RSN is relatively spared in SSD and that such sparing may be a protective factor that offsets a phonological deficit. So these findings pose a puzzle for both the double deficit model and the single phonological deficit model of dyslexia.

#### 4.4. *Dyslexia in other languages*

A multiple deficit approach to dyslexia can also be found in research on poor readers in other languages besides English. For example, a recent study of dyslexia in Chinese children (Ho, Chan, Tsang, & Lee, 2002) found that over half the sample (about 57%) had triple or quadruple cognitive deficits, whereas only 20% had single deficits. Moreover, phonological deficits were found in only 15% of the sample, whereas deficits in the other three domains were more common (rapid naming: 50%, orthographic: 39%, and visual; 37%). Interestingly, rapid naming deficits are also more common than phoneme awareness deficits among German children with dyslexia (Wimmer, 1993). These results indicate that a phonological deficit is not necessary to produce dyslexia in all languages and suggest that in some languages (i.e., Chinese) multiple deficits are usually required.

## 5. Multiple deficit model

Similar to the complex disease model in medicine (Sing & Reilly, 1993) and the quantitative genetic model in behavioral genetics (e.g., Plomin, DeFries, McClearn, & Rutter, 1997), the current model proposes that (1) the etiology of complex behavioral disorders is multifactorial and involves the interaction of multiple risk and protective factors, which can be either genetic or environmental; (2) these risk and protective factors alter the development of cognitive functions necessary for normal development, thus producing the behavioral symptoms that define these disorders; (3) no single etiological factor is sufficient for a disorder, and few may be necessary; (4) consequently, comorbidity among complex behavioral disorders is to be expected because of shared etiological and cognitive risk factors; and (5) the liability distribution for a given disease is often continuous and quantitative, rather than being discrete and categorical, so that the threshold for having the disorder is somewhat arbitrary. Applying the model to the comorbidities considered here (RD+ADHD and RD+SSD), each individual disorder would each have its own profile of risk factors (both etiological and cognitive), with some of these risk factors being shared by another disorder, resulting in comorbidity.

Fig. 2 illustrates the complex disease model as applied to complex behavioral disorders. There are four levels of analysis in this diagram: etiologic, neural, cognitive, and symptom, where clusters of symptoms define complex behavioral disorders. For

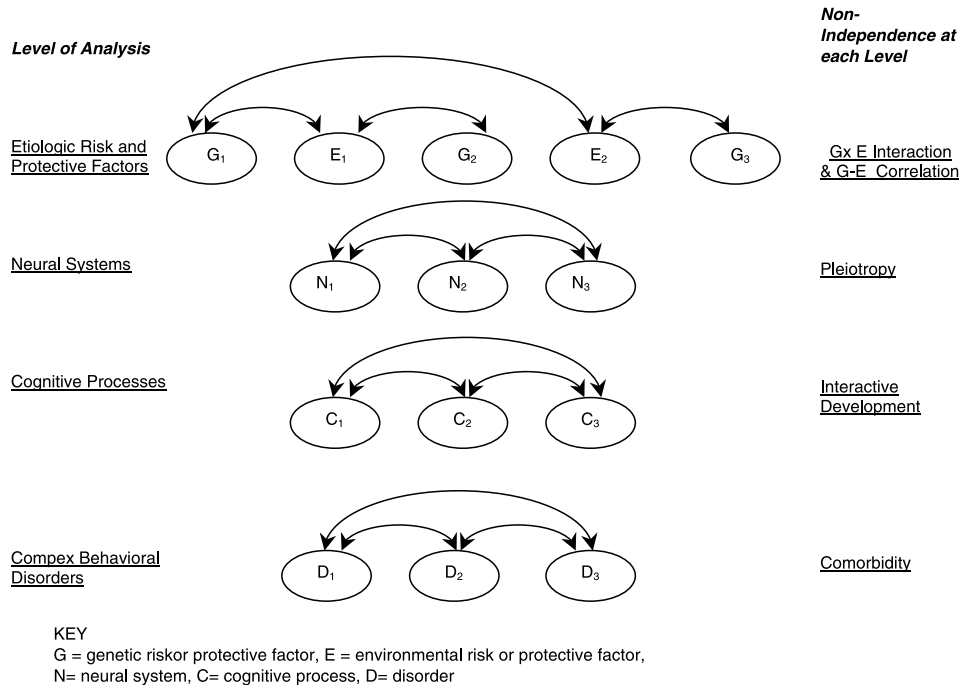


Fig. 2. Multiple deficit model.

any such complex behavioral disorder, it is expected there will be many more risk and protective factors than the five shown here. Bidirectional connections at each level indicate that constructs are not independent. For instance, at the etiologic level, there are likely to be gene-environment interactions and correlations. At the neural level, a single genetic or environmental risk factor will often affect more than one neural system (pleiotropy). Even if the risk factor initially only affects one neural system, this alteration will likely have downstream effects on the development of other neural systems. At the cognitive level, constructs are correlated because their developmental pathways overlap and because cognition is interactive. Overlap at the cognitive level leads to comorbidity at the symptom level. So, while the single deficit model conceptualizes the relation between disorders in terms of double dissociations, the multiple deficit model conceptualizes this relation in terms of partial overlap. At the symptom level, there is comorbidity (i.e., greater than chance co-occurrence) of complex behavioral disorders. Omitted from the diagram are the causal connections *between* levels of analyses, some of which would include feedback loops from behavior to brain or even to etiology. The existence and strength of these various causal connections must be determined empirically. The weights on the connections between levels of analysis will tell us to what extent different etiologic and cognitive factors contribute to comorbidity at the symptom level.

This model makes it clear that achieving a complete understanding of the development of disorders like SSD, dyslexia or autism will be very difficult because of the multiple pathways involved. But this kind of model is needed because it is becoming increasingly clear that there are shared processes at the etiologic, neural, and cognitive levels across such disorders.

Testing the multiple deficit model will benefit from multivariate methods like confirmatory factor analysis and path analysis. Multivariate behavioral genetic analyses that build on these methods will also be highly relevant because they allow one to model the genetic and environmental covariance among both diagnostic dimensions and underlying cognitive processes. Such methods are already being very usefully applied to the problem of understanding the relation between elementary cognitive processes and the complex trait of general intelligence – psychometric *g* (e.g., [Plomin & Spinath, 2002](#)).

As discussed earlier, the relation between cognitive risk factors and comorbid disorders can be expressed as weights on the paths from the cognitive to the behavioral level. Multivariate methods allow one to estimate these weights. Tests of a multiple deficit model should address both phenotypic and etiologic relations between the cognitive and behavioral level and test whether relations are similar for extreme scores (e.g., diagnoses) and individual differences across the whole distribution. A Cholesky decomposition ([Neale & Cardon, 1992](#)) tests which phenotypic and etiologic relations are shared and which are specific across correlated measures, and so is highly relevant for the multiple deficit model. Recently, a multivariate approach has been used in linkage analyses of RD, which has multiple correlated cognitive phenotypes ([Marlow et al., 2003](#)). Previous univariate linkage analyses of these multiple phenotypes has sometimes produced conflicting results or indicated genetic dissociations that did not replicate. In contrast, the multivariate results were much more consistent.

Although these multivariate approaches can seem atheoretical, they allow one to test explicit a priori hypotheses. Just from what has been presented earlier, one can imagine several competing multiple deficit hypotheses for the relation between SSD and RD or RD and ADHD. For instance, one could propose that a PA deficit and a RSN deficit are jointly necessary to produce RD (contrary to existing data (Pennington, Cardoso-Martins, Green, & Lefly, 2001)), and that a PA deficit and a PM deficit are jointly necessary to produce SSD. Or one could propose that (1) they are probabilistic risk factors and that both main effects and interactions of each pair determine the risk for RD or SSD, (2) that they are only additive probabilistic risk factors, or (3) that one must add other risk factors to the model of each disorder. Each of these hypotheses could be tested in a sample selected to include individuals with one, both or neither disorder and who had been given the relevant cognitive measures. What this paper argues is that one gains important constraints in testing hypotheses by having to account for at least two disorders and their relation. But, one would still like to know how the multiple cognitive risk factors are derived from a cognitive theory of the relevant domain for that disorder (e.g., speech production in SSD; single word recognition in RD, or behavior regulation in ADHD), and how the cognitive theory can be broadened to deal with more than one disorder at a time.

Even with powerful multivariate methods, we will still face the difficult problem of distinguishing causal risk factors from secondary or associated ones. So we will still need longitudinal and training studies to test which cognitive risk factors play a causal role. The same cognitive risk factor, say a deficit in processing speed, could be found in two comorbid disorders, and yet have a different causal relation to each disorder. It could be primary in one and secondary or associated in another.

These are some of the complexities we face in moving from single to multiple cognitive deficit models of developmental disorders, and facing them makes it clear why single cognitive deficit models have been so appealing. But, hopefully, the arguments and data presented here provide a convincing case for the need to test multiple deficit models of developmental disorders. In the final section of this paper, we review other problems with the single cognitive deficit model.

## **6. Other problems with the single cognitive deficit model**

Other researchers have pointed out a number of problems with the straightforward application of the theory and methods of cognitive neuropsychology to the case of developmental disorders. One problem with using a standard cognitive neuropsychology approach to understanding developmental disorders is the strong emphasis on dissociations between “pure” cases, such as the double dissociation between the phonological and surface subtypes of dyslexia described earlier. Bishop (1997) argued that this dissociation logic was not well suited to developmental disorders for several reasons: (1) an individual with an apparently pure subtype of such a disorder, like developmental phonological dyslexia, may manifest an opposite subtype at a later age (e.g., Nation & Snowling, 1997; see also Karmiloff-Smith, Brown, Grice, & Patterson, 2003; Thomas & Karmiloff-Smith, 2002 for discussions of why dissociations between

developmental disorders can change with development) (2) interactions between levels of cognitive processing across development can easily convert an initial single cognitive deficit into a complex pattern of impairments, making it difficult to determine which deficit is primary, (3) such interactions also produce compensatory tradeoffs in development, thereby masking initial deficits, and (4) general processing limitations could lead to what appears to be a domain-specific deficit.

Oliver, Johnson, Karmiloff-Smith, and Pennington (2000) criticized applying the “static neuropsychological deficit approach” to developmental disorders, because it assumes that (1) brain–behavior relations are constant across development and hence similar in adults and children; (2) that a mapping exists between damage to or dysfunction in a localized brain structure and a particular cognitive deficit; and (3) that cause runs only from the neural to the cognitive level. As a consequence of these assumptions, such disorders have often been studied at later ages, close to their developmental end state. These authors argue for a different approach to understanding brain–behavior relations in both developmental disorders and the mature brain. In this alternative neuroconstructionist or connectionist approach, the specializations found in the mature brain are *products* of development rather than innate, prewired modules. Atypical development results from subtle, often widespread, differences in the initial state which lead to “alternative developmental trajectories in the emergence of representations within neural networks” (Oliver et al., 2000, p.1).

Van Orden, Pennington, and Stone (2001) criticized the double dissociation logic used in the cognitive neuropsychology approach. Although the vast majority of cases of both acquired and developmental brain dysfunction present with a complex pattern of associated deficits (i.e., they are mixed cases), the cognitive neuropsychology approach focuses on pure cases, each with an apparently single cognitive deficit. A contrasting pattern of single cognitive deficits across two pure cases (i.e., a double dissociation) is taken as strong evidence for the relative independence and hence modularity of each underlying cognitive process.

Van Orden et al. (2001) identified serious problems with this double dissociation logic. Essentially, these problems are circularities, having to do with the notion of pure cases and with the assumption that double dissociations can only arise in modular cognitive architectures. For double dissociations to be theoretically interesting, the cases or groups must be “pure”, such as the contrast between a pure case of phonological dyslexia and a pure case of surface dyslexia. But the notion of a pure case involves a double circularity. The first circularity is that a particular cognitive theory is required to define which cases are pure. Since there is no theory-independent definition of pure cases, contrasting pure cases inevitably confirm the theory which chose them. The second circularity is that the very possibility of a pure case with an isolated modular deficit already assumes that modularity theory is true. These problems might not be insuperable if double dissociations could only arise in a modular architecture. But it has since become apparent that double dissociations, such as the double dissociation between phonological and surface dyslexia, can arise in non-modular architectures (Farah, 1994; Harm & Seidenberg, 1999; Plaut, 1995).

Because processing is interactive in these non-modular, connectionist architectures, double dissociations are rarely absolute. An impairment in process A slightly

degrades process B and vice versa. But the same is true in patients with acquired disorders (see discussion of cases of acquired dyslexias in Van Orden et al., 2001) and is typical of developmental disorders. Connectionist models of dissociations in both typical (e.g., Elman et al., 1996) and atypical development (e.g., Harm & Seidenberg, 1999), as well as in brain damage syndromes (e.g., Farah, 1994) make it clear that a dissociation at the level of behavior does not always require a separate stage or process at the cognitive level. So single and double dissociations are hardly an infallible tool for dissecting the underlying cognitive architecture.

However, the issue of modularity is partly separable from the issue of single vs. multiple deficits. Even if there are innate modules, because the etiologies of developmental disorders act very early in brain development, they would likely affect more than one module, which is the concept of pleiotropy. And even if the etiologies of different developmental disorders were independent (which we have shown is not true for behaviorally defined disorders like dyslexia and ADHD), as long as these etiologies acted pleiotropically there would likely be shared cognitive risk factors across disorders. So two of the main ideas of the multiple cognitive deficit model – multiple cognitive deficits in any given disorder and partly shared cognitive risk factors across disorders – could be derived from a strong innate modularity theory, given pleiotropy. But the multiple deficit idea does contradict the idea of a complete 1:1 mapping between etiologic and cognitive risk factors, which would lead to each disorder having its own single independent cognitive deficit.

In sum, this paper has traced how my thinking has evolved from single to multiple cognitive deficit models of developmental disorders. Multiple cognitive deficit models are more consistent with the multifactorial and probabilistic etiology of such disorders. But challenges remain in how to specify and test multiple cognitive deficit models.

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