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The validity of psychiatric diagnoses: The case of 'specific' developmental disorders

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ABSTRACT

We tested whether developmental coordination disorder (DCD) and mixed receptive expressive language disorder (RELD) are valid diagnoses by assessing whether they are separated from each other, from other childhood disorders, and from normality by natural boundaries termed zones of rarity. Standardized measures of intelligence, language, motor skills, social cognition, and executive functioning were administered to children with DCD ($n = 22$), RELD ($n = 30$), autistic disorder ($n = 30$), mental retardation ($n = 24$), attention deficit/hyperactivity disorder ($n = 53$) and to a representative sample of children ($n = 449$). Discriminant function scores were used to test whether there were zones of rarity between the DCD, RELD, and other groups. DCD and RELD were reliably distinguishable only from the mental retardation group. Cluster and latent class analyses both resulted in only two clusters or classes being identified, one consisting mainly of typical children and the other of children with a disorder. Fifty percent of children in the DCD group and 20% in the RELD group were clustered with typical children. There was no evidence of zones of rarity between disorders. Rather, with the exception of mental retardation, the results imply there are no natural boundaries between disorders or between disorders and normality.
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1. Introduction

Planning for DSM-V has been strongly influenced by the fact that the syndromes defined in DSM-III and its successors have not been validated (Kupfer, First, & Regier, 2002). Addressing the validity criteria proposed by Robins and Guze (1970) and Kendler (1980), Kupfer et al. summarised almost three decades of research:

> "despite many proposed candidates, not one laboratory marker has been found to be specific in identifying any of the DSM-defined syndromes. Epidemiologic and clinical studies have shown extremely high rates of comorbidities among the disorders, undermining the hypothesis that the syndromes represent distinct etiologies. Furthermore, epidemiologic studies have shown a high degree of short-term diagnostic instability for many disorders. With regard to treatment, lack of treatment specificity is the rule rather than the exception" (p. xviii).

Kendell and Jablensky (2003) questioned whether the criteria used to establish the validity of psychiatric diagnoses are appropriate. They argued that the Robins and Guze (1970) and Kendler (1980) criteria were based on the implicit and possibly unjustified assumption that psychiatric disorders are discrete, mutually discriminable, disease entities. Kendell and

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Jablensky suggested that "the possibility that disorders might merge into one another with no natural boundary in between . . . was simply not considered" (p. 5). Kendell and Jablensky's critique does not imply that DSM-defined disorders are invalid. Rather, they state that "although there is a growing assumption, at least within the research community, that most currently recognized psychiatric disorders are not disease entities, this belief has never been demonstrated, mainly because studies of the appropriate kind have rarely been mounted" (p. 7).

According to Kendell and Jablensky (2003), the key characteristic of valid disease entities is that they are separated from each other and from normality by natural boundaries. Natural boundaries are defined as *zones of rarity* (cf. Sneath, 1957) such that the characteristics or symptoms of one syndrome are discontinuous with normality and the symptoms of other syndromes. If zones of rarity cannot be identified, the implication is that there are no natural boundaries between syndromes and they "are merely arbitrary loci in a multidimensional space in which variation in both symptoms and etiology is more or less continuous" (Kendell & Jablensky, 2003, p. 7).

Based on available evidence, there is reason to doubt that there are zones of rarity between specific developmental disorders (learning disorders, communication disorders, motor skills disorder) and between specific and pervasive developmental disorders (autistic disorder, asperger's disorder, pervasive developmental disorder not otherwise specified) or other disorders usually first evident in childhood. As Kupfer et al. (2002) noted about all DSM disorders, comorbidities among specific development disorders are high, as they would be between specific and pervasive developmental disorders if they were not precluded by hierarchical classification rules in DSM-IV. Indeed, comorbidity among developmental disorders is so high that a child who is diagnosed with any developmental disorder is more likely than not to also meet diagnostic criteria for one or more other developmental disorders (e.g., Cohen et al., 2000; Frazier et al., 2001; Hofvander et al., 2009; Kadesjo & Gillberg, 2001; Kaplan, Dewey, Crawford, & Wilson, 2001; McGrath et al., 2008; Miniscalco, Nygren, Hagberg, Kadesjo, & Gillberg, 2006; Watemberg, Waiserberg, Zuk, & Lerman-Sagie, 2007).

There is also reason to doubt that there are zones of rarity between these disorders and normality. Specific developmental disorders are diagnosed when a child's measured performance in academic skills, language, or motor skills is substantially lower than the child's verbal and/or non-verbal intelligence. This means that a child's specific ability score, the child's intelligence score, and the discrepancy between these two ability scores all represent points on normal distributions of scores and the scores of children with a disorder are continuous with those of children with no disorder. This continuity makes it unlikely that there is a zone of rarity between disordered and normal children. Rather, just as mental retardation is partly defined by very low scores that represent the bottom tail of the normal distribution of intelligence test scores, children with specific developmental disorders have scores that place them at the bottom tail of the normal distributions of specific ability scores and/or at the bottom tail of the normal distribution of discrepancy scores.

According to Kendell (1989), discriminant function analyses are the most appropriate test to assess for zones of rarity separating syndromes from each other and from normality. The aim is to assess whether members of groups with and without disorders can be accurately classified, as would be expected of discrete entities, and also to see if the distribution of discriminant function scores is bimodal. It is the trough between the two peaks in the distribution that represents the zone of rarity, the intermediate cases between the two syndromes or between a syndrome and normality.

A second approach to validating hypothetically discrete syndromes is cluster analysis. It was initially thought by Robins and Guze (1970) that cluster analysis would be useful in the first stage of the validation process, that is, in the initial description of syndromes. The procedure was expected to group individuals based on shared characteristics, the sets of correlated symptoms that would define different disorders, and these groups would then be shown to have other characteristics in common (like course and response to treatment) that would validate the grouping. A weakness of the approach was that effective clustering depends on prior knowledge of which symptoms need to be included in the analysis, something that cannot be known until the validity of a syndrome has been demonstrated. Cluster analysis is more useful as a way of confirming that individuals who share a diagnosis are grouped with each other on the basis of the defining symptoms, and not with healthy people or people with another diagnosis.

A third approach is latent class analysis, a method for finding subtypes of related cases (referred to as latent classes) from multivariate ordinal or categorical data (Lazarsfeld & Henry, 1968). In most respects, latent class analysis is similar to cluster analysis, except that the assumption of conditional independence leads to more natural and meaningful groups (if any). Conditional independence simply means that within a latent class that corresponds to a distinct syndrome, the presence or absence of one symptom is viewed as unrelated to the presence or absence of all other symptoms.

In this study, five disorders were sampled for their value in assessing the validity of two specific developmental disorders: developmental coordination disorder (DCD) and mixed receptive expressive language disorder (RELD). DCD and RELD are defined by delays in acquiring either motor skills or receptive and expressive language skills relative to either general intelligence (DCD) or nonverbal intelligence (RELD). If these disorders represent discrete disease entities, there should be a zone of rarity separating children with these disorders from each other, from typically developing children, and from children with one of three other disorders. Attention deficit/hyperactivity disorder (ADHD) is partly defined by delays in the ability to sustain attention and/or to inhibit behavior, and the delays are inferred from behavior rather than from ability testing. Autistic disorder (AD) is partly defined by qualitative impairments in social interaction and communication and by restricted repetitive and stereotyped behavior, interests, and activities. However, DSM-IV precludes the diagnosis of DCD and RELD when a pervasive developmental disorder is present, which implies that clinically significant problems with motor skills and language are common in autism. To the extent that motor or language problems are present, this implies that both DCD and RELD may be continuous with autism. Finally, mental retardation (MR) is partly defined by general intellectual

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