Understanding Psychopathology
Melding Behavior Genetics, Personality, and Quantitative Psychology to Develop an Empirically Based Model

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ABSTRACT—Research on psychopathology is at a historical crossroads. New technologies offer the promise of lasting advances in our understanding of the causes of human psychological suffering. Making the best use of these technologies, however, requires an empirically accurate model of psychopathology. Much current research is framed by the model of psychopathology portrayed in current versions of the Diagnostic and Statistical Manual of Mental Disorders (DSM; American Psychiatric Association, 2000). Although the modern DSMs have been fundamental in advancing psychopathology research, recent research also challenges some assumptions made in the DSM—for example, the assumption that all forms of psychopathology are well conceived of as discrete categories. Psychological science has a critical role to play in working through the implications of this research and the challenges it presents. In particular, behavior-genetic, personality, and quantitative-psychological research perspectives can be melded to inform the development of an empirically based model of psychopathology that would constitute an evolution of the DSM.

KEYWORDS—classification; DSM; statistics; comorbidity; dimensions; categories

Psychopathology research is at a historical crossroads. Powerful technologies, such as molecular genetics and sophisticated statistical models, now exist to aid us in our attempts to understand the origins of psychological suffering. To fully exploit these technologies, however, we need to know how to best conceptualize psychopathology. We need an empirically based model of psychopathology that can guide our inquiries into its origins.

Most psychopathology research is currently framed by the system provided in the fourth edition (text revision) of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV-TR; American Psychiatric Association, 2000). A number of specific assumptions underlie the classification of all disorders described in the DSM-IV-TR. A cardinal assumption is that mental disorders are categorical: The manual lists a large number of categories of mental disorders, and for each category, a series of criteria for category membership are listed. People are assumed to be either members of these categories or nonmembers; graded degrees of category membership are not permitted. Importantly, the DSM-IV-TR itself acknowledges potential limitations of this categorical approach to conceptualizing psychopathology, noting that “a categorical approach to classification works best when all members of a diagnostic class are homogeneous, when there are clear boundaries between classes, and when the different classes are mutually exclusive” (p. xxxi).

Each of these areas has proven problematic for DSM categories. Members of specific diagnostic classes tend to be heterogeneous, boundaries between classes are often unclear, and classes are rarely mutually exclusive. This is the sense in which psychopathology research is at a historical crossroads. DSM-defined categories are the most frequent targets of psychopathological inquiry, yet reliance on DSM-defined categories often results in significant problems in research design and interpretation. To pick a specific example for illustrative purposes, if one wants to understand depression, what should be done about the fact that the boundary between depression and other DSM categories is often unclear (e.g., depression overlaps with dysthymia; Klein & Santiago, 2003) and many people who meet criteria for depression meet criteria for other disorders as well (e.g., anxiety disorders; Kessler, DuPont, Berglund, & Wittchen, 1999)? Is it possible to develop an empirically based approach to psychopathology that could overcome these limitations?

The development of such a system is a tractable goal, and the pursuit of this goal involves integrating a number of areas of inquiry that represent quintessential strengths of psychological science. Some broad outlines of such a system can be seen by tying together recent research findings from these areas: Specifically, research strategies, concepts, and findings from
quantitative psychology, behavior genetics, and personality psychology provide the tools needed to develop an empirically based model of psychopathology.

CONTRIBUTIONS OF QUANTITATIVE PSYCHOLOGY TO UNDERSTANDING PSYCHOPATHOLOGY

One prominent movement in psychology during recent decades has been the use of explicit quantitative models to describe and predict psychological phenomena. Quantitative models are sets of mathematical and statistical equations describing and predicting psychological phenomena. Structural-equation models, item-response models, growth-curve models, and other latent-variable models have allowed tremendous increases in the sophistication of theories that can be tested and in the confidence of our conclusions about those theories. These methods also hold promise for understanding psychopathology, because they allow empirical comparison of different classification paradigms. Such paradigms can be represented by different quantitative models, and can be rigorously compared by comparing the fit of those models to psychological data.

Empirical comparisons between different factor models, for example, have indicated that common forms of psychopathology in adults can be understood in terms of a hierarchical factor model (Krueger & Markon, 2006; see Fig. 1) that bears a strong resemblance to influential factor models in child-psychopathology research (Achenbach & Edelbrock, 1984). At a high level of the hierarchy, psychopathological variation and covariation are organized by two broad, correlated dimensions, Internalizing and Externalizing. Internalizing psychopathology represents a spectrum of conditions characterized by negative emotion and includes phenomena such as depression, anxiety, and phobias. At a lower level of the hierarchy, the Internalizing spectrum splits into a Distress subspectrum and a Fear subspectrum; the former is characterized by ruminative disorders such as depression and generalized anxiety, the latter by paranoid disorders such as phobia and panic disorder. Externalizing psychopathology, in contrast, is characterized by disinhibition; this spectrum includes phenomena such as antisocial behavior and substance-use disorder. This hierarchical model provides a better account of patterns of psychopathology than do many competing factor models, including ones that contain fewer or more factors.

Recent latent-variable-modeling studies have suggested that, indeed, these common forms of psychopathology are best thought of as continuous, rather than categorical, in nature. Generally speaking, continuous models classify people by locating them along graded dimensions, whereas categorical models classify people into distinct groups. Explicit comparisons of continuous and categorical models of the occurrence and co-occurrence of externalizing disorders indicate that this broad domain of psychopathology is continuous in nature, reflecting a liability or underlying level of risk for disorder that is graded in severity rather than discrete and categorical (Krueger, Markon, Patrick, & Iacono, 2005; Markon & Krueger, 2005). Although, to our knowledge, continuous and categorical models of the overarching internalizing domain have not been compared directly, relevant research does exist for specific internalizing syndromes. For example, continuous models of depression have increased validity over categorical models (Aggen, Neale, & Kendler, 2005), and depressive symptoms appear to be continuously distributed (Hankin, Fraley, Lahey, & Waldman, 2005).

Finding that common forms of psychopathology are best conceptualized as continuous in nature calls into question the

![Fig. 1. A model of common forms of psychopathology. The numerical values are path coefficients, representing the strength of associations between constructs; stronger relationships are associated with larger values. The data on which the figure is based come from a meta-analysis presented in Krueger and Markon (2006). Reprinted from "Reinterpreting comorbidity: A model-based approach to understanding and classifying psychopathology," by Robert F. Krueger and Kristian E. Markon, 2006, Annual Review of Clinical Psychology, 2, page 126. Copyright 2006 by Annual Reviews (www.annualreviews.org). Reprinted with permission.]
current assumption of the DSM that psychopathology is always categorical.

Continuous models of psychopathology help delineate informativeness (how much a disorder or symptom is indicative of a dimension underlying multiple symptoms or disorders, akin to a factor loading in traditional factor analysis) and severity (where a disorder or symptom is located along a dimension underlying multiple symptoms or disorders). For example, such models delineate these characteristics of different disorders within the Internalizing and Externalizing spectra, as well as the informativeness and severity of specific symptoms with regard to individual disorders. Aggen et al. (2005), for example, evaluated the informativeness and severity of different symptoms of depression. They demonstrated that the most informative symptoms were depressed mood, lack of interest, and duration greater than 2 weeks. The most severe symptoms, however, were suicidal ideation, inability to concentrate, and feelings of worthlessness. This picture of the differential informativeness and severity of depression symptoms is in contrast to the classification approach taken in the current DSM, in which different symptoms are mostly equal in their usefulness as indicators of disorder categories.

CONTRIBUTIONS OF BEHAVIOR GENETICS TO UNDERSTANDING PSYCHOPATHOLOGY

Models that have proven useful in understanding psychopathology can be extended to include information on genetic and environmental influences. By including data on the relatedness of different individuals in families, similarities in patterns of psychopathology across individuals can be modeled as a function of how related the individuals are. For example, to the extent that distinct patterns of psychopathology are manifested more frequently among genetically related individuals than among unrelated individuals, taking into account potential environmental reasons for resemblance, those patterns reflect genetic influences. Such research thereby helps inform the understanding of psychopathology by incorporating information on the origins, or etiology, of disorders.

Evidence suggests that patterns of etiologic influence on common forms of psychopathology generally mimic observed, or phenotypic, patterns. Kendler, Prescott, Myers, and Neale (2003) modeled patterns of psychopathology among twins and concluded that genetic influences have the same hierarchical internalizing–externalizing factor structure seen phenotypically. Their results indicate, for example, that if one identical twin has one internalizing disorder, his or her twin is more likely to have another internalizing disorder than to have an externalizing disorder, and vice versa. These findings are important because they suggest that etiologic influences on common forms of psychopathology share the same organization as psychopathology itself—psychopathology appears to derive its observed structure from the structure of its underlying etiology. That is, the internalizing and externalizing spectra are observable not only in the phenotypic patterning of mental disorders, but also in the patterning of underlying genetic risk factors for these disorders.

As our understanding of molecular neurobiology and genetics improves, it will also become possible to delineate the physical nature of the biological structures underlying psychopathology and its etiology. A greater understanding of the molecular-genetic substrates of psychopathology will help refine psychopathology models by providing details about the structures underlying the phenotypic organization of psychopathology. In this regard, molecular genetics not only helps explain why psychopathology occurs but also what psychopathology is—how it is best thought about and best organized conceptually. Along these lines, recent research indicates that genes are organized in functional systems of variation—that is, genes are inherited together in sets that parallel the functions of the proteins they encode (Petkov et al., 2005). In the future, it may be possible to link gene-expression variation in these systems to dimensions of psychopathology.

Research on psychopathology framed by dimensions such as those described in this article can help delineate the links between phenotypes and functional genetic systems. For example, Stallings et al. (2005) reported that a composite externalizing index provided stronger evidence for linkage to specific areas of the genome, when compared with separate antisocial and substance-dependence indices used alone. This composite index provided the strongest evidence of genetic linkage in a sample of adolescents and young adults, suggesting that a locus on chromosome 9 increases risk for externalizing psychopathology in general, as opposed to risk for only specific externalizing syndromes.

CONTRIBUTIONS OF PERSONALITY PSYCHOLOGY TO UNDERSTANDING PSYCHOPATHOLOGY

Constructs such as the Internalizing and Externalizing spectrums bear a notable resemblance to personality constructs. Like personality constructs, psychopathology-spectrum constructs organize broad domains of human individual differences and provide theoretical coherence for those domains. In addition to these conceptual parallels, data also link personality constructs per se to the model in Figure 1 in a way that is psychologically meaningful. Internalizing-spectrum disorders are associated with the broad personality domain of negative emotionality or neuroticism, whereas externalizing-spectrum disorders are associated both with constructs in that domain and with constructs in the broad domain of disinhibition—a domain that intersects unconscientiousness and disagreeableness (for a meta-analytic perspective on the structure of these personality constructs, see Markon, Krueger, & Watson, 2005; for a recent review of these personality–psychopathology connections, see Krueger, 2005). Psychologically speaking, negative emotionality confers risk for disorders in the internalizing spectrum, whereas a combination of negative emotionality and disinhibition confers risk for
disorders in the externalizing spectrum. Moreover, these connections extend beyond phenotypic associations. Behavior-genetic research supports a genetic basis for these connections, indicating that personality and psychopathology are linked at an etiological level (Krueger, 2005).

CONCLUSIONS

The modern DSMs have been fundamentally helpful in psychopathology research. They have provided explicit definitions of categories of psychopathology. The research we reviewed would not have been possible without the foundation provided by these definitions. Nevertheless, the research reviewed here also underlines the value of some evolutionary steps in the field’s conceptualization of psychopathology to further psychological research on the subject.

One evolutionary focus is the DSM itself. Psychological scientists have important roles to play in pushing for changes to the DSM. The processes that will eventuate in the publication of the next edition of the DSM (DSM-V) are just getting underway (see http://www.dsm5.org/), and there are reasons for optimism regarding the scientific bases for DSM-V. For example, a number of conferences have been organized to discuss research agendas to help place DSM-V on solid scientific footing (see, e.g., Widiger, Simonsen, Krueger, Livesley, & Verheul, 2005, for discussion of a research agenda for personality disorders articulated at one of these conferences).

Yet the DSM is a complex document, shaped in understandable and legitimate ways by considerations that extend beyond psychopathology research per se. To pick a single illustrative example, categories of psychopathology provide labels that are used routinely in facilitating third-party payment for professional services. This record-keeping function of the DSM is conceptually separate from the utility of the DSM as a framework for psychopathology research, but it is no less legitimate. As a result of this understandable multiplicity of influences and purposes, the DSM represents a compromise among diverse considerations.

Such compromises may not optimally serve the needs of the psychopathology research community. As a result, an empirically based model of psychopathology may develop separately from the DSM, to help frame and propel novel research. Some specific steps in developing this kind of model can be gleaned from the current review, and constitute expansions of the conceptualization of psychopathology to further psychological research on the subject.

In developing such databases, it is also necessary to greatly expand the scope of the model in Figure 1. The model developed out of attempts to understand the comorbidity (co-occurrence) of the limited subset of DSM disorders that have been the primary focus of epidemiological inquiry; many psychopathology constructs were not included simply because the relevant data do not exist. Expanding the scope of the model requires coverage of a greater diversity of psychopathological symptoms and personality constructs, most likely using samples in which the prevalence of diverse forms of maladaptive behavior is higher than in the community-dwelling population (e.g., treatment-seeking samples).

Such an expanded and more detailed model would logically lead to novel questions in both treatment-oriented and etiologically oriented psychopathology research. With regard to studies of treatment, one could ask if interventions are impacting specific symptoms, specific syndromes, or broad spectra. Parallel questions would emerge in attempting to understand the etiology of psychopathology. For example, do specific genetic polymorphisms (distinct forms of genes) influence details of symptom presentation or overall risk for a broad spectrum of psychopathologies?

The development of this kind of empirically based model of psychopathology—separate from the DSM—might be viewed as unfortunate, in the sense that it might further separate science and practice. Yet it may also be a necessary step in realizing the promise of psychological science as a foundation for developing effective means to alleviate human suffering.

Recommended Reading

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REFERENCES


