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The disease entity in psychiatry: fact or fiction?

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Background. The current debate concerning the forthcoming revisions of the International Classification of Diseases (ICD) and the Diagnostic and Statistical Manual of Mental Disorders (DSM) lacks sufficient historical perspective on groundwork concepts in psychiatry, such as the nature of the disease entity, categorical typologies, dimensional models and their validity and utility.

Objective. To offer an overview of the evolution and metamorphoses of the conceptual basis of classification in psychiatry, with particular focus on psychotic disorders.

Method. Discursive, proceeding from history of ideas to a critique of present dilemmas.

Results. Much of the present-day discussion of basic issues concerning the classification of mental disorders is a replay of debates that took place in the earlier periods of scientific psychiatry.

Conclusion. The mainstream nosological paradigm adopted in psychiatry since early 20th century is in need to be critically examined and transcended with the help of concepts and methodological tools available today.

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The term 'nosology' refers to the theory about the nature of medical conditions and the principles and rules of their classification. In psychiatry, we are facing today the recurrent question about the nosological status of the brain and mind disorders that constitute the professional turf of the discipline. Are we dealing with discrete, discontinuous entities, or with graded continuous phenomena to which we can apply a set of thresholds in order to separate 'pathology' from 'normal variation' and determine the need for treatment? What is the relationship between the clinical manifestations of a disorder and the underlying pathological processes or predisposing genetic aberrations? Such questions have been asked since the earliest stages of clinical research in psychiatry, yet notwithstanding the recent advances in the neuroscience and genetics of psychiatric disorders, many of the present-day answers to these basic questions are a replay of debates that took place in the earlier periods of scientific psychiatry. This suggests that there may be inherent shortcomings in the mainstream nosological paradigm adopted in clinical psychiatry since the beginning of the 20th century that need to be critically examined and, possibly, transcended with the help of concepts and methodological tools that are available today. The present discussion of these issues will focus primarily on the taxonomy of the major psychoses since its history highlights many of the controversies, conjectures and refutations that continue to cast a long shadow on the forthcoming classification revisions involving both DSM and ICD.

Classification in science

The term classification denotes 'the activity of ordering or arrangement of objects into groups or sets on the basis of their relationships' (Sokal, 1974). Classification in medicine has been defined as the 'procedure for constructing groups or categories and for assigning entities (disorders or persons) to these categories on the basis of their shared attributes or relations' (Millon, 1991). The term 'taxonomy', often used as a synonym for classification, should refer properly to the meta-theory of classification; in medicine the corresponding term 'nosology' denotes the concepts and theories that support the classification of symptoms, signs, syndromes and diseases, whereas the term 'nosography' refers to the activity of describing and naming such entities. The names jointly constitute the nomenclature within a particular field of medicine.

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What is the unit of classification in psychiatry?

Medical classifications are created with the primary purpose of meeting pragmatic needs related to diagnosing and treating people experiencing illnesses. Their secondary purpose is to assist the generation of new knowledge relevant to those needs, though progress in medical research usually precedes, rather than follows, improvements in classification. Simply stating that medical classifications classify diseases (or that psychiatric classifications classify disorders) begs the question since the status of concepts such as 'disease' and 'disorder' remains obscure. As pointed out by Scadding (1996), the concept of 'disease' has evolved with the advance of medical knowledge, and at present, is no more than 'a convenient device by which we can refer succinctly to the conclusion of a diagnostic process which starts from recognition of a pattern of symptoms and signs, and proceeds, by investigation of varied extent and complexity, to an attempt to unravel the chain of causation'. 'Disease', therefore, is an explanatory construct integrating information about: (a) statistical deviance from the population 'norm'; (b) characteristic clinical manifestations; (c) characteristic pathology; (d) underlying causes; and (e) the extent of reduced biological fitness. For a constellation of observations to be referred to as 'a disease', these parameters must be shown to form a 'real-world correlational structure' (Rosch, 1975), which is stable and distinct from other similar structures. The typical progression of knowledge starts with the identification of the clinical manifestations (the syndrome) and the deviance from the 'norm'; understanding of the pathology and aetiology usually comes much later. However, there is no fixed point or agreed threshold beyond which a syndrome can be said to be 'a disease'. Today, Alzheimer's disease, with dementia as its clinical manifestation, specific brain morphology, tentative pathophysiology and at least partially understood causes, is one of the few conditions in psychiatric classifications that are defined by their pathology rather than their syndrome. Schizophrenia, however, is still better described as a syndrome (Kendell & Jablensky, 2003).

The hypothesis of nosological entity

The basic unit of the present classification of the psychoses is the concept (or hypothesis) of the nosological entity, remaining more or less in the form in which it was first formulated by Kahlbaum (1863). This notion postulates a correspondence between presumed aetiology, brain pathology, symptom patterns and outcome as a criterion for bringing together seemingly disparate clinical states and syndromes as manifestations of a single 'disease'. Based on this approach, Kraepelin (1899) grouped together several clinical syndromes into the nosological entities of manic-depressive insanity and dementia praecox, respectively, and laid down the foundation for the dichotomy of the major psychoses. Among the several criteria defining the nosological entity, the criterion of outcome, defined crudely in terms of 'curability' and 'incurability' reigned supreme. Looking back to Kraepelin's early writings, it becomes clear that the criterion of outcome as the cornerstone of his system was largely a matter of convenience – he realized that knowledge of aetiology and brain pathology at the time could offer little guidance, while outcome was a variable accessible to the clinician and could be described in observational terms. Thus, the nosological system based on the 'disease entity' concept established itself firmly in the mainstream of clinical psychiatry, where it remains to the present day. Bleuler's (1911) revision of the concept of 'dementia praecox' and the re-naming of it as 'schizophrenia' did not change substantially the underlying nosological principle, although it extended the diagnostic boundaries of the entity by suggesting that 'the group of schizophrenias' could include non-psychotic, or pre-psychotic variants, such as 'latent schizophrenia'.

Shortcomings of the 'classical' nosological model

Attempts at developing alternative approaches to the systematics of psychotic disorders were undertaken in the last century, but none of them proved as successful as the Kraepelinian nosological theory. Its building blocks fall into two categories: (i) phenomena accessible to clinical inquiry – symptoms, syndromes, patterns of course, and a range of outcomes; (ii) non-clinical correlates, including such describing individuals (e.g. biological variables or behavioural characteristics) or populations (e.g. epidemiological or culture-related). The essential task in the construction of a nosology of discrete disease entities is to identify internally cohesive clinical groupings based on established inter-correlations between symptoms and syndromes (the cross-section) and patterns of course and outcome (the longitudinal aspect). The ultimate test of the validity of such groupings is the degree to which they are found to be associated with variables of potential aetiological significance – pathogenetic mechanisms, treatment response and the degree of stability they possess *vis-à-vis* demographic and cultural variation. However, the nosological entities, defined in this way, and the 'entity' of schizophrenia in particular, soon met with difficulties. The requirement of a close correspondence between

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the cross-section, i.e. the symptoms and syndromes of the disorder, and the characteristics of its course and outcome, was never fully met. The criterion of 'curability' *v*. 'incurability' proved to be too crude, and attempts to identify in the initial stages of a schizophrenic illness symptoms and signs that would reliably predict its outcome, have on the whole failed. It was furthermore demonstrated that a proportion of the 'typical' schizophrenias recover completely, and that a proportion of 'typical' manic-depressive illnesses may run a chronic and disabling course. These observations could not be easily reconciled with the assumptions of the 'dichotomy' model. The inadequacy of the 'curability' criterion became even more obvious after the introduction of psychopharmacological treatment which created a huge 'grey zone' of outcomes that could not be clearly classified as either 'recovery' or post-psychotic 'defect'. This situation was described by Leonhard & von Trostorff (1964): 'If two psychiatrists have different diagnostic opinions about an endogenous psychosis, the decision can be reached only through a catamnesis, or not at all. If one of them diagnoses a manic-depressive illness, then only the demonstration of a defect can prove him wrong…the other, who diagnoses schizophrenia, cannot be refuted at all. He may stick to his diagnosis, even if the catamnesis shows no evidence of defect'.

The argument that recovering schizophrenias are not 'true' cases of the disorder, and ought to be retrospectively re-diagnosed if lasting recovery occurs, is clearly contradictory in the light of findings from the two major WHO studies, the International Pilot Study of Schizophrenia (IPSS; World Health Organization, 1979) and the subsequent Ten-Country Study on Determinants of Outcome of Severe Mental Disorders (Jablensky *et al.* 1992). In the IPSS, cases were diagnosed in the strictest possible way by applying three sets of criteria: (i) clinician's diagnosis according to ICD-8; (ii) computer diagnosis using the CATEGO algorithm; and (iii) empirical grouping of cases by cluster analysis, on the basis of maximum shared characteristics. Patients who met simultaneously the three sets of criteria were designated as a 'concordant' group of schizophrenia. This group was expected to be more homogeneous than the rest of the cases, including course and outcome. However, the data did not reveal any significant differences in course and outcome between the 'concordant' cases and the 'non-concordant' cases. This finding casts considerable doubt on the prognostic value of a restrictive diagnosis of schizophrenia. Such findings do not stand alone in the recent literature. A number of follow-up studies in the last 20 years confirm the notion that severe deterioration is not the 'typical' outcome of schizophrenia, even if a very long (10–65 years) follow-up period is considered (Jablensky *et al.* 2011). Moreover, some of these studies have shown that many of the symptoms and syndromes that are often regarded as indicators of a poor prognosis, actually undergo a reversal in a significant proportion of cases. This was strikingly demonstrated by the long-term follow-up of the patients of the WHO IPSS (prevalence cohort, follow-up at 26 years after the episode of inclusion) and the WHO Ten-Country Study of Schizophrenia (incidence cohort, follow-up at 15 years after the first psychotic episode). According to the WHO report (World Health Organization, 2007), 'the most striking overall finding…is that the current global status of over half of these subjects – 56% of the incidence group and 60% of the prevalence group – is rated as 'recovered'. Nearly half have experienced no psychotic episodes in the last 2 years of follow-up… These percentages accord fairly well with ratings of both current symptoms and functioning'.

The 'classical' nosological hypothesis could be sustained if it were demonstrated that within the broad range of possible outcomes, particular patterns were predicted by specific constellations of symptoms and signs. In the WHO studies, attempts to identify predictors of patterns of outcome by multivariate statistical techniques led to two conclusions. First, the low percentage of variance explained by 56 putative predictor variables suggested that either the set of predictors was incomplete, or that unidentified factors intervening after the initial assessment had a significant impact on the outcome. Secondly, among the variables of proven prognostic significance, only a few were mental state variables – most of the predictors were indicators of personality characteristics or of the social situation of the patient. These findings suggest that the prognosis of schizophrenia is an open-ended dynamic process whose direction can, within limits, be modified at any point. The presumed 'characteristic' psychopathological phenomena, such as the Schneiderian first-rank symptoms (Schneider, 1959), did not appear to have prognostic significance.

Another shortcoming of the 'classical' nosological system is its failure to separate consistently the two entities of schizophrenia and affective disorders. This has been known for a long time but the difficulty was thought to reside in the imprecise definition of the diagnostic criteria, rather than in the existence of a large group of conditions which simply defy the dichotomy and exhibit the features of a clinical 'hybrid'. This group attracted a variety of diagnostic labels, including 'schizoaffective disorder' (Kasanin, 1933) or 'unsystematic schizophrenias' (Leonhard, 1999), was classified alternately with the schizophrenias or with the affective disorders, but never found a comfortable place in either category. The existence

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of such 'hybrid' cases poses the problem of defining the borderline between the two nosological entities. This remains today as an open question of how far one could stretch the boundaries of one or the other category in order to accommodate the 'intermediate' or 'atypical' cases without damaging beyond repair the internal cohesiveness of the diagnostic groups. Alternative solutions that have been proposed are: (i) the designation of a third group of psychoses and (ii) treating the 'poor prognosis' schizophrenia and the 'good prognosis' affective disorders as two extremes on a clinical (and presumably genetic) continuum that could accommodate all of the intermediate forms (Owen *et al.* 2007).

A third problem for which the 'classical' nosological theory has failed to find an acceptable solution is the classification of the sub-threshold, practically non-pathological forms of cognitive and affective deviations and the unusual personalities which are encountered among biological relatives of schizophrenia patients. Their importance and frequency were recognized by Bleuler (1920), who coined the term 'latent schizophrenia'. A bewildering variety of diagnostic labels has been proposed for such conditions: 'abortive schizophrenia' (Bleuler, 1920); 'ambulatory schizophrenia' (Zilboorg, 1941); 'pseudoneurotic schizophrenia' (Hoch & Polatin, 1949); 'borderline schizophrenia' (Kety, 1988), but none has been universally accepted, or the diagnostic criteria have been unequivocally defined. Subsequent evidence has provided support for a genetic link to schizophrenia, which strengthened the concept of a schizophrenia 'spectrum' (Kendler *et al.*, 1994). Both the place of these conditions in the psychiatric classification and their definition are still a matter of contention. The DSM-III solution, which removed the non-psychotic syndromes from the category of schizophrenia and assigned them, as 'schizotypal personality disorder' to the personality disorders, runs counter to the principle that conditions of a presumably common origin should be classified together. The 'borderline' or 'spectrum' forms related to the affective psychoses have so far received less attention than the non-psychotic satellites of schizophrenia, but the recognition of a syndrome of 'masked depression' (Kielholz, 1979), and the notion of an affective or cyclothymic personality disorder suggest that similar problems also exist on the affective side of the 'classical' diagnostic dichotomy of the major disorders. At present, the 'borderline' forms are of limited therapeutic interest, since most cases do not require pharmacological treatment, and there is little evidence that, if provided, treatment is effective. Their theoretical and research importance, however, is considerable, especially from the point of view of the genetics of the major psychoses. If, along with the manifest major psychoses, the existence of clinically subliminal disorders produced by a shared genotype could be demonstrated, the search for endophenotypic traits and mechanisms would be facilitated. In this sense, the study of the 'borderline' disorders may even hold the key to a better understanding of the pathogenetic aspects of the major psychiatric syndromes.

Post-Kraepelinian remedial approaches

The difficulties referred to above resulted in attempts to make adjustments to the classification while still retaining the basic notion of the nosological entity as the mainstay of the whole system. One approach involved the shifting of the boundary of the dichotomy in such a way as to absorb the majority of 'atypical' cases within a modified version of the one or the other group of disorders. This approach has produced a variety of 'narrow' and 'broad' re-definitions of the scope of schizophrenic and affective disorders. Examples are Langfeldt's (1937) notion of 'nuclear' schizophrenia and 'schizophreniform' disorders; and the antithetic suggestion that most 'atypical' schizophrenic illnesses are, in fact, variants of the affective psychoses (Pope & Lipinski, 1978). The Feighner criteria (Feighner *et al.* 1972), and subsequently DSM-III, introduced a restrictive definition of schizophrenia, retaining implicitly the idea that 'true' schizophrenia is a condition whose hallmark is a deteriorating course, and shifting a large proportion of the less deteriorating cases into an expanding intermediate category of schizoaffective disorders. This strategy would be justified if it were possible to demonstrate that re-defining the categories on symptomatological grounds would yield more homogeneous patient populations in terms of course and outcome. Several studies, including the WHO IPSS (World Health Organization, 1979), have compared the prognostic implications of alternative diagnostic definitions of schizophrenia based on varying scope of the category. The results have, by and large, demonstrated only marginal advantages for some of the proposed sets of diagnostic criteria, insufficient for the practical purposes of a prognostic diagnosis.

Another approach involved attempts at a wholesale rejection of the dichotomy and, instead, fragmenting the large group of psychoses into multiple 'micronosological' entities for which symptomatological, prognostic, genetic and treatment response validating criteria were sought. This strategy is best represented by the Wernicke–Kleist–Leonhard tradition that defined a 'third' group of 'cycloid' psychotic disorders. Within the group of affective disorders, this approach

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introduced a split between 'bipolar' and 'unipolar' disorders (Leonhard, 1999). A similar development in Scandinavian psychiatry has been the development of the concept of 'psychogenic' or 'reactive' psychoses (Strömgren, 1987). The common feature of these undertakings is that they retain both the categorical approach to the subdivision of the field and the 'classical' criteria of the nosological entity, while rejecting the original dichotomy. The degree to which this approach captures real biological discontinuities among the multiple entities remains uncertain.

A third approach (Ey *et al.* 1923) represented a more radical departure from the Kraepelinian nosology. French psychiatry has never been bound by the notion of 'curability' *v.* 'incurability', or has ever fully accepted the dichotomy of psychotic disorders. Instead, psychoses were classified along two axes: (i) the depth and locus of psychotic disorganization ('disorders of consciousness' and 'disorders of personality') and (ii) the distinction between acute and chronic course of the conditions.

Was the nosological entity, after all, a fiction?

None of the above attempts to re-shape the nosology of the major psychiatric disorders has been entirely successful. Yet there can be no doubt that the 'classical' nosological hypothesis was a major step forward that introduced order and parsimony in a field that had previously been chaotic or arbitrarily subdivided. As Jaspers (1963) pointed out, 'the idea of the disease entity is in truth an idea in Kant's sense of the word: the concept of an objective which one cannot reach since it is unending; but all the same it indicates the path for fruitful research and supplies a *valid* point of orientation for particular empirical investigations'. The least that could be said is that the nosological hypothesis helped to formulate clearly, and bring into focus, issues that critics could oppose or endorse, thus contributing to a healthy diversity of viewpoints that was necessary for a developing discipline such as psychiatry.

A more fundamental re-thinking of the nosological theory underlying the classification of psychotic disorders would require, on the one hand, resolving the kind of contradictions outlined above and, on the other hand, developing concepts that would allow a better interdigitation of clinical, neurobiological, genetic and behavioural data. Some basic premises of such an approach had been actually outlined by Kraepelin in one of his last articles, 'Die Erscheinungsformen des Irreseins' (Kraepelin, 1920). Kraepelin's position can be summarized in three key points: (i) it is necessary 'to turn away from arranging illnesses in orderly well-defined groups, and to set ourselves the undoubtedly higher and more satisfying goal of understanding their essential structure'; (ii) the affective and schizophrenic forms of mental illness do not represent the expression of particular pathological processes, but rather indicate the areas of our personality in which these processes unfold; (iii) we must accustom ourselves to the idea that the phenomena of illness which we have hitherto used are not sufficient to enable us to distinguish reliably between manic-depressive illness and schizophrenia in all cases. It must remain an open question whether this is due to general human psychological mechanisms operating in combination with pathological changes, or whether hereditary factors make certain areas more susceptible and accessible to pathological stimuli'.

The present state of the nosological issues in psychiatry: DSM-IV and ICD-10

Classifying involves forming categories or taxa for ordering natural objects or entities and assigning names to these. Ideally, the categories of a classification should be jointly exhaustive to account for all possible entities, and mutually exclusive. In biology, despite continuing arguments between proponents of evolutionary systematics, numerical taxonomy and cladistics, there is agreement that classifications reflect fundamental properties of biological systems and constitute 'natural' classifications. This is not so with psychiatric classifications. First, the objects being classified in psychiatry are not 'natural' entities but explanatory constructs. Secondly, the taxonomic units of 'disorders' in DSM-IV and ICD-10 do not form hierarchies and the current classifications contain no supraordinate, higher-level organizing concepts. Therefore, DSM-IV and ICD-10 are certainly not systematic classifications in the usual sense in which that term is applied in biology. A closer analogue to current psychiatric classifications can be found in the so-called indigenous or 'folk' classifications of living things (e.g. animals or plants) or other material objects (Cantor & Genaro, 1986). 'Folk' classifications do not consist of mutually exclusive categories and have no single rule of hierarchy (but may contain many rules applicable *ad hoc*). They are useful because they are pragmatic and adapted to the needs of everyday life. Essentially, they are augmented nomenclatures, i.e. lists of names for conditions and behaviours, supplied with implicit or explicit rules of how these names should be assigned and used. As such, they are useful tools of communication and can play an important role in psychiatric research, clinical management and teaching.

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The 'reification' problem

Today, many clinicians are aware that diagnostic categories are simply concepts, justified only by whether or not they provide a useful framework for organizing the complexities of clinical experience to make predictions about outcome and to guide treatment decisions. However, there is the risk that once a diagnostic concept such as schizophrenia has come into general use, it tends to become 'reified' (Hyman, 2010) as an entity of some kind, which can be invoked to explain the patient's symptoms and whose validity need not be questioned. Even though the authors of DSM-IV were careful to point out that 'there is no assumption that each category of mental disorder is a complete discrete entity', the mere fact that a diagnostic concept is listed in an official classification and provided with an operational definition tends to encourage this insidious reification.

The ambiguous status of the classificatory unit of 'disorder'

The term 'disorder', first introduced as a generic name for the unit of classification in DSM-I (1952), has no clear correspondence with either the concept of disease or the concept of syndrome in medical classifications. It circumvents the problem that the material from which most of the diagnostic rubrics in psychiatry are constructed consists primarily of reported subjective experiences and patterns of behaviour. Some of those rubrics correspond to syndromes in the medical sense, but many appear to be sub-syndromal, reflecting isolated symptoms, habitual behaviours, or personality traits. The ambiguous status of the 'disorder' creates conceptual confusion and hinders the advancement of knowledge. Apart from the 'reification fallacy', the fragmentation of psychopathology into a large number of 'disorders', of which many are merely symptoms, leads to a proliferation of comorbid diagnoses, which clinicians are forced to use in order to describe their patients. This blurs the important distinction between true comorbidity (co-occurrence of aetiologically independent disorders) and spurious comorbidity masking complex but essentially unitary syndromes. It is, therefore, not surprising that disorders, as defined in the current versions of DSM and ICD, have a strong tendency to co-occur, which suggests that 'fundamental assumptions of the dominant diagnostic schemata may be incorrect' (Sullivan & Kendler, 1998).

Reinstating the concept of syndrome

Although the range of possible aetiological factors – genetic, toxic, metabolic, or experiential – that may give rise to psychiatric disorders is practically unlimited, the range of psychopathological syndromes, reflecting the brain's responses to a variety of noxae, is limited. Since a variety of aetiological factors may produce the same syndrome (and conversely, an aetiological factor may give rise to a spectrum of syndromes), the relationship between aetiology and clinical syndrome is an indirect one. In contrast, the relationship between the syndrome and the underlying pathophysiology, or specific brain dysfunction, is likely to be much closer. This was recognized long ago in the case of psychiatric illness associated with somatic and brain disorders where clinical variation is restricted to a limited number of 'organic' brain syndromes, or 'exogenous reaction types' (Bonhoeffer, 1912). In the complex psychiatric disorders, where aetiology is multifactorial, future research into specific pathophysiological mechanisms could be considerably facilitated by a sharper delineation of the syndromal status of many current diagnostic categories. This provides a strong rationale for reinstating the syndrome as the basic unit of future versions of psychiatric classifications.

Validity and 'validators'

Although the reliability of psychiatrists' diagnoses may be substantially improved by the use of explicit diagnostic criteria, their validity remains uncertain. What is meant by validity of a diagnostic concept in psychiatry is rarely discussed and few studies have addressed this question directly. There is no simple measure of validity of a diagnostic concept that is comparable to the well-established procedures for the assessment of reliability. The types of validity often mentioned in the context of psychiatric diagnosis – construct, content, concurrent and predictive – are borrowed off the shelf of psychometric theory in psychology. However, few diagnostic concepts in psychiatry meet these criteria at the level of stringency normally required of psychological tests. Despite such ambiguities, a number of procedures have been proposed to enhance the validity of psychiatric diagnoses in the absence of a simple measure. Thus, Robins & Guze (1970) outlined a programme with five components: (i) clinical description; (ii) laboratory studies; (iii) delimitation from other disorders; (iv) follow-up studies; and (v) family studies. This schema was later elaborated by Kendler (1980), who distinguished between antecedent validators (familial aggregation, premorbid personality and precipitating factors); concurrent validators (e.g. psychological tests); and predictive validators (diagnostic consistency over time, rates of relapse/recovery, response to treatment). Andreasen (1995) has proposed 'additional' validators, such as

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molecular genetics, neurochemistry, neuroanatomy, neurophysiology and cognitive neuroscience, and suggested that 'the validation of psychiatric diagnoses establishes them as 'real entities''.

These procedural criteria implicitly assume that psychiatric disorders are distinct entities; the possibility that disorders might merge into one another with no clear boundary in between is rarely considered. There is increasing evidence of overlapping genetic predisposition to schizophrenia and bipolar disorder, as well as to seemingly unrelated disorders, such as autistic spectrum, intellectual disability and, possibly, epilepsy. It is equally likely that the same environmental factors may contribute to several different syndromes. Should such findings be systematically replicated, their repercussion on future psychiatric classifications will be considerable. Variations in psychiatric symptomatology might indeed be better represented by 'an ordered matrix of symptom-cluster dimensions' (Widiger & Clark, 2000) than by a set of discrete categories. However, it would be premature at this time to discard the current categorical entities. Although there is a mounting assumption that many of the currently recognized psychiatric disorders are not disease entities, this has never been demonstrated, mainly because few studies of the appropriate kind have ever been designed and conducted.

Validity and utility

It is crucial to maintain a clear distinction between validity and utility (Kendell & Jablensky, 2003); at present these two terms are often used as if they were synonyms. A diagnostic rubric may be said to possess utility if it provides non-trivial information about prognosis and likely treatment outcomes, and/or testable propositions about biological and social correlates (Meehl, 1959). Many, though not all, of the diagnostic concepts listed in contemporary classifications such as DSM-IV and ICD-10 are extremely useful to clinicians, whether or not the category in question is valid, as they provide information about the likelihood of recovery, relapse, deterioration and social handicap; they guide treatment decisions; they describe symptom profiles; and they guide research into the aetiology of the syndrome. However, there is a critical difference between validity and utility. Validity is an invariate characteristic of a diagnostic category – there may be uncertainty about its validity because of lack of relevant empirical information – but in principle, a category cannot be 'partly' valid (Kendell & Jablensky, 2003). *Utility*, on the other hand, is a graded characteristic that is partly context specific. Schizophrenia may be an invaluable concept to practicing psychiatrists, but of little use to researchers exploring the genetic basis of psychosis. For example, the DSM-IV definition of schizophrenia is useful for predicting outcome, because some degree of chronicity is in-built. But a broader definition, covering a heterogeneous 'schizophrenia spectrum', is more useful for defining a syndrome with high heritability for genetic research.

Categories *v.* dimensions

There are many different ways in which classifications can be constructed. The fundamental choice is between a categorical and a dimensional structure, and it is worth recalling the observation by the philosopher Carl Hempel (Hempel, 1961) that, although most sciences start with a categorical classification of their subject matter, they often replace this with dimensions as more accurate measurements become possible. The requirement that the categories of a typology should be mutually exclusive and jointly exhaustive has never been fully met by any psychiatric classification (or, for that matter, by any medical classification). Psychiatric classifications are eclectic in the sense that they are organized according to several different classes of criteria, e.g. presenting symptoms or traits, age at onset, course, pathology and causes, without a clear hierarchical arrangement. One or the other criterion may gain prominence as knowledge progresses or contextual conditions (social, legal, or service-related) change. However, despite their apparent logical inconsistency, medical and psychiatric classifications survive and evolve because of their essentially pragmatic nature. Their utility is tested almost daily in clinical decision making which ensures a natural selection of useful concepts by weeding out obsolete ideas (Jablensky & Kendell, 2002).

Categorical typologies are the traditional, firmly entrenched form of representation for medical diagnoses. They are familiar to clinicians, and most of the present knowledge of the causes, presentation, treatment and prognosis of mental disorders is stored in a categorical format. They are easy to use under conditions of incomplete clinical information and have the capacity to 'restore the unity of the patient's pathology…into a single, coordinated configuration' (Millon, 1991). The main disadvantage of the categorical model is that it tends to encourage a 'discrete entity' view of the nature of psychiatric disorders. If it is understood, though, that diagnostic categories do not necessarily represent discrete entities, but are simply a convenient way of organizing and presenting information, there should be no fundamental objection to their continued use.

Dimensional models, on the other hand, have the advantage of explicitly introducing quantitative variation and graded transitions between forms of disorder,

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as well as between 'normality' and pathology. Whether psychotic disorders can be better described dimensionally or categorically remains an open question. The difficulties with dimensional models of psychopathology stem from several sources. First and foremost is the absence of an established, empirically grounded metric. Most existing scales of symptom severity are of a psychometrically low level of measurement: they are either nominal or ordinal, where one is simply able to state that $a > b > c \cdots > n$ on some property, and to assign numbers that indicate rank order and nothing more. It seems unlikely that equal-interval scaling, or ratio scales will ever be developed for reported subjective symptoms. Although intelligence, aptitude and personality test scores are also, strictly speaking, ordinal, some of them do approximate interval equality (Kerlinger, 1973). Secondly, there is no sight of an agreement on the number and nature of dimensions required to account adequately for clinical variation. Lastly, dimensional models are too complex and cumbersome for everyday clinical practice. These considerations seem to preclude, for the time being, a radical restructuring of psychiatric classification from a predominantly categorical to a predominantly dimensional model. However, categorical and dimensional models need not be mutually exclusive, as demonstrated by the so-called mixed or class-quantitative models that combine qualitative categories with quantitative trait measurements. For example, there is increasing empirical evidence that a retained (and refined) categorical clinical description of the syndrome of schizophrenia can be supplemented with selected quantitative traits, such as measurements of sustained attention, memory dysfunction, amplitudes and latencies of event-related brain potentials and volumetric deviance of cerebral structures (Hallmayer *et al.* 2005). There is a growing body of clinical and neuroscience data suggesting a possibility to develop nosological models that might combine the advantages of the categorical clinical approach with the refinement provided by research on quantitative traits or endophenotype-based dimensions.

DSM-5, ICD-11 and beyond

In the present process of revision of the DSM and ICD, both the APA and WHO are confronted with a dilemma. The revision process has generated requests to alter the criteria defining many individual disorders, to eliminate some and to add new 'disorders', and so on. In many cases, the reasons for such changes, viewed in isolation, seem cogent. However, all definitional changes have serious disadvantages: they are confusing to clinicians; they create a situation in which the relevance of all previous clinical and epidemiological research into disorders as currently defined is uncertain; and they involve tedious and sometimes costly changes in the content and wording of diagnostic interviews, as well as in the algorithms to generate diagnoses from clinical ratings. It is important to maintain awareness of the fact that most of psychiatry's disease concepts are merely working hypotheses and their diagnostic criteria are provisional. Both extremes – a totally unstructured approach to diagnosis and a rigid operationalization – should be avoided. Defining a middle range of operational specificity, which would be optimal for stimulating critical thinking in clinical research, but also rigorous enough to enable comparisons between the results of different studies in different countries, is probably a better solution. Mere re-shuffling and re-defining the ICD or DSM classification rubrics without sufficient and compelling evidence may be arbitrary and premature. This should not mean that we must wait until robust biological markers and indicators become available. On the contrary, experimenting with research-oriented classifications and diagnostic tools linked to emerging 'research domain' neurobiological and genetic criteria (Cuthbert & Insel, 2010) is a promising novel approach. Although the majority of psychiatric disorders appear to be far too complex from a genetic point of view than it had been assumed until recently, molecular genetics and neuroscience will play an increasing role in understanding their aetiology and pathogenesis. However, the extent of their impact on the diagnostic process and on the classification of psychiatric disorders is difficult to predict. The eventual outcome is likely to depend not only on the knowledge base of psychiatry *per se*, but also on the social, cultural and economic forces that shape the public perception of mental illness and influence the clinical practice of psychiatry.

It will be pertinent to conclude this essay with a quotation from Jaspers (1963), which pointedly summarizes the main message of this paper: 'When we design a diagnostic scheme…we abandon the idea of disease entity and once more have to bear in mind continually the various points of view (as to causes, psychological structure, anatomical findings, course of illness and outcome), and in the face of the facts we have to draw a line where none exists…a classification therefore has only a provisional value. It is a fiction which will discharge its function if it proves to be the most apt for the time'.

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