The DSM-ICD diagnostic approach as an essential bridge between the patient and the "big data"

G.B. Mitropoulos

5th Department, Psychiatric Hospital of Attika, Athens, Greece

The use of diagnostic manuals in psychiatry is generally necessitated by the lack of tests that would corroborate psychiatric diagnosis. Criticism towards the today prevailing DSM-ICD diagnosis traditionally regards among others such problems as hyponarrativity, biologism, "death of phenomenology", and a questionably valid over-fragmentation of diagnosis. Lately, and especially after the appearance of the 5th edition of DSM (2013), criticism focuses at such issues as lack of validity, having failed to adopt a dimensional model, not adequately relying on genetics and neurobiology, and impeding, rather than facilitating, research into the etiology of mental disorders, the DSM becoming an "epistemic prison". The former problems seem to derive from the fact that the operationalist criteria are often uncritically adopted as the ultimate authority in diagnosis, instead of being merely guides, as intended originally and explicitly; the latter problems have been made more evident since the emergence of the American RDoC research initiative, which not only points to an alternative, more valid classification of mental disorders, but also aspires to signal a move of psychiatry towards precision medicine, having as its main dogma that mental disorders are disorders of brain circuits, which are expressed as complex syndromes. In this paper, the historical and epistemological context of the emergence of DSM is examined; its achievement in terms of diagnostic reliability as well as clinical utility is not negligible, especially taken into consideration the climate of virtual diagnostic arbitrariness which characterized the (American) psychiatry before 1980, with obvious consequences for the authority of the specialty. Then, the potential of the new era of genetics, neurobiology and analysis of the "big data" for generating a novel approach to psychiatric diagnosis and classification is put into consideration, while it remains unknown in what way the findings of RDoC could lead and be translated into a new classification system. Moreover, the particularity of the psychiatric object, the clinical significance of the categorical approach to diagnosis, as well as the need for a "irreducible psychological level of explanation" are discussed. In our view, today, the DSM–ICD diagnosis lies between two different and potentially opposing demands and tendencies: on the one hand, the demand for the individual, subjective and phenomenological particularity of the mentally ill to be taken into consideration (a demand that sometimes underestimates the need for clinical communication); on the other hand, the (largely future) vision for more and more analysis of biological data in the name of a yet to be clarified personalized therapy (the very notion of diagnosis becoming potentially redundant). Finally, considering the particularity of the psychiatric object, we conclude that

the DSM-ICD approach, with its categorical diagnoses and its descriptive operational criteria, despite its inherent imperfections and inadequacies, continues to have a place in psychiatry as an essential bridge/interface between clinic and research data, as a common clinical language, and as an epistemic hub; and that prerequisites for diagnostic validity should be sought both in the cells of RDoC and in those theoretical approaches which examine human subjectivity as such, included phenomenology and psychoanalysis.

Key words: Diagnosis, DSM, categorical, big data, phenomenology, subjectivity.

Psychiatry's reliance on diagnostic manuals places it in a unique position among medical specialties and stems from the absence of useful diagnostic tests.[1] DSM and chapter V of the ICD are the prevailing diagnostic classifications today and, despite individual differences, share a common philosophy and character.[2] Here, the focus will be on the former classification, because its latest edition (DSM-5) is the most recent, and is the one that has provoked the greatest controversy. However, the discussion presented here may also of interest in view of the oncoming 11th revision of the ICD.

1. DSM-III and the need for communication

Common to all DSM editions is a categorical character of diagnosis: the various disorders are more or less distinct clinical syndromes which can be empirically described. However, it was the third edition (1980) of the manual that revolutionized psychiatric diagnosis both in America and internationally. DSM-III (as well as its successors) differed from its predecessors mainly in two ways: first, it was a-theoritical as regards the etiology of the disorders (especially avoiding the psychoanalytical etiological hypotheses which informed much of DSM-I and II) and, second, diagnoses did not rely on general, albeit representative, descriptions of clinical syndromes, but on clear diagnostic criteria.[3]

If DSM-I was a response to post-war statistical needs, DSM-III was born of the need for diagnostic reliability, that is the need for a common diagnostic language, which would be understood and used by everyone, in clinical practice, in medical education, in research and in epidemiology; in short, it was born of the need for communication. At the same time, DSM-III had the explicit goal to conform to the principles of evidence-based medicine, which at the

time was still a movement in its first steps. It should be reminded that, according to its explicit goals, the evidence-based medical model "de-emphasizes intuition, unsystematic clinical experience, and pathophysiologic rationale as sufficient grounds for clinical decision making and stresses the examination of evidence from clinical research".[4]

During the 1970s American psychiatry was undergoing a serious "crisis of legitimization", which is reflected in the rise of the anti-psychiatric movement.[5] Studies conducted at that time showed that American psychiatrists not only tended to diagnose schizophrenia more easily than their British colleagues,[6] but that they could also not distinguish diagnostically pseudo-patients from schizophrenic patients.[7] The unreliability of American psychiatry is generally attributed to the complete domination of post-war psychiatry by (the American version of) psychoanalysis, as well as Adolf Mayer's biopsychosocial model, which resulted to a limited interest in accurate diagnosis and nosology among psychiatrists.[8,9] It should be noted that nothing of the kind ever took place in European psychiatry.

The new diagnostic manual would eventually appear in 1980, and its emphasis would be on description, the givens of observation and reliability. At the same time, and under the influence of the neopositivist philosophy of science, it would render psychiatric diagnosis operationalist.[10,11] DSM-III succeeded completely in restoring the authority of psychiatry as a medical specialty. Similarly, it achieved the goal of reliability, of a common language and of communication between clinicians. During the last 37 years, DSM has been an invaluably useful tool: its diagnostic categories facilitate diagnosis, clinical decisions, research on the treatment of mental disorders, medical training, epidemiology, as well as the evaluation of such mat-

ters as treatment efficacy, clinical course, remission, relapse and prognosis of mental disorders.[12] At the same time, DSM has received extensive criticism, the most salient points of which I will try to summarize.

2. Death of phenomenology, hyponarrativity and clinical prototypes

The intention of the architects of DSM-III (and the following editions) was to create a set of diagnostic criteria which would be merely "guides" to clinical diagnosis, which in any case is based on "clinical judgement" and requires "clinical training and experience".[3,13,14] Nevertheless, what actually happened, in the words of Nancy Andreasen, a distinguished member of the DSM-III task force, is that, after 1980, DSM "was universally and uncritically accepted as the ultimate authority in psychopathology and diagnosis"[8] and its descriptions ended up being used, not as abstractions, but as descriptions complete and sufficient in themselves. This resulted, among other things, in what Andreasen diagnosed as "the death of phenomenology" in America: research in psychopathology "is a dying (or dead) enterprise", medical students are taught a poor version of the clinical picture, and psychiatric history taking is limited to a dry checking of symptoms and signs.[8]

Besides that, many commentators accuse DSM of what they call "hyponarrativity". The term was coined in 2006 by psychiatrist John Z. Sadler, and means that, for the manual, signs and symptoms of a mental disorder can be assessed empirically and independently of the subjective experience of the patient, of the circumstances of his life, and of the personal meaning the patient ascribes to them as well as to his symptoms.[15] With the publication of DSM-5 in 2013, the discussion about hyponarrativity was exacerbated, as the manual's new edition was accused (exaggeratedly, in my opinion) that, by removing the bereavement exclusion from the diagnosis of major depression, it virtually abolishes the fundamental distinction between mourning and depression.[16–18]

Finally, a further criticism is that the DSM polythetic diagnostic criteria differ greatly from the way a clinician actually thinks when making a diagnosis: in reality, diagnosis as a mental process consists not in checking symptoms in a list, but in comparing between the particular case which is being examined and clinical prototypes, that is representative exemplars in the sense of Gestalt, which are invariably formed and acquired mentally and enriched continually with growing experience.[19,20] In this sense, and –I would say– fortunately, the way we diagnose in practice is never so "hyponarrative" as the DSM operationalism would have us believe. Nevertheless, the "loss of subjectivity and interpersonal context" of the patient (with which Castiglioni & Laudisa charge DSM) is not only a, so to speak, side-effect of operationalist diagnosis, but bears upon the philosophy evidence-based itself, to the extent that it considers the patient's subjectivity as "a disturbance factor to be eliminated in order to purify scientific analysis of mental disorders".[18]

3. Reliability and validity. Categorical and dimensional diagnosis

It has been said that the major weakness of DSM is its lack of validity.[21] To begin with, we ought to acknowledge that many DSM categories do display some construct validity.[2] It is nevertheless a fact that, as Allen Frances, architect of the DSM-IV, has remarked, "the DSM is necessarily more about forging a common language than finding a truth" (quoted in Haslam 2013).[22]

No-one denies that diagnostic reliability is of paramount importance, at least to clinical practice. Reliability does not guarantee validity; nevertheless, reliability is a pre-condition for validity.[22] Moreover, as Jeffrey Bedrick points out, "any diagnostic system has to abstract away the particular experiences of patients and form idealizations if its goal is to develop a shared framework for understanding and treating the individuals encountering the same condition".[23] Any diagnostic system thus aims to form a common conception of psychopathology, to function as a common means of communication and, be accepted by clinicians and researchers of varying theoretical orientations.[3] These goals are probably more likely to be achieved with diagnostic criteria which are clear and operationalist.[22] Finally, categorical diagnoses may be uncertain in terms of validity, but still be clinically useful.[12]

Despite all that, it is still a fact that the clearly descriptive and atheoretical approach of DSM had a number of side-effects: splitting of diagnosis, questionable grouping of disorders, proliferation of diagnostic categories, emergence of diagnoses of doubtful validity,[24,25] inflation of the vexing problem of comorbidity.[2] Even more importantly, it was proved that the consensual neo-Kraepelinian descriptions of syndromes not only failed to serve as an adequate basis for etiological research, but to a great extent they also became an unintended "epistemic prison" for clinical and translational researchers.[2]

Relevant to this subject is the trade-off between categorical and dimensional diagnosis. Although today most psychiatrists would generally agree on the usefulness of a dimensional model, there is much less agreement on exactly which dimensions should be used in diagnosis.[26] DSM-5 (much like DSM-IV), while acknowledging the (theoretical) necessity of a dimensional diagnosis, eventually retained its categorical character, since the proposal of alternative dimensional definitions was eventually considered to be "immature scientifically",[14] even in the domain of personality disorders.[26]

4. The RDoC as an alternative research paradigm

As a consequence of the above developments, the American NIMH prioritizes research which is not based on DSM diagnostic criteria. This rationale that justifies this policy is that "diagnostic categories based on clinical consensus fail to align with findings emerging from clinical neuroscience and genetics".[27] Holding as a fundamental tenet that "mental illnesses are brain disorders expressed as complex cognitive, emotional, and social behavioral syndromes",[28] the NIMH has since 2010 adopted the Research Domain Criteria (RDoC) initiative as its research framework. At present, "RDoC is not a diagnostic system, it's merely a framework for organizing research";[29] its explicit goal is, however, "to ultimately provide a framework for classification based on empirical data from genetics and neuroscience".[27] According to its initiators, the primary focus for RDoC is on neural circuitry, with levels of analysis progressing in one of two directions: upwards from measures of circuitry function to clinically relevant variation, or downwards to the genetic and molecular/cellular factors that ultimately influence such function.[27]

It is more than obvious that RDoC, over which there is considerable enthusiasm, focus on biology and observable behavior to a degree unthinkable for DSM, which has been traditionally accused of biologism, behaviorism or even a "decisive denial of the psychic reality" of the patient.[25] The vision of the architects of RDoC is clearly that, in the near future, psychiatry will not treat clinical syndromes, but characteristics or traits of individual patients identified through data mining across genomic, physiologic, imaging, and clinical levels;[29] a vision which most probably relegates to limbo the, very real in the view of many psychiatrists, need for an "irreducible psychological level of explanation".[30] Of course, no-one knows today in which way the RDoC findings will be able to lead and translate into a new classification system. Despite and beyond this uncertainty, RDoC seems to mark or to promise a shift of psychiatry towards the socalled "precision medicine".[29]

5. Precision psychiatry and the potential bypass of diagnosis

There is a partial overlap between the terms "precision medicine", "personalized medicine" and "systems medicine", all of which fall under the umbrella of what is called "big data approach". All these approaches generally try to identify statistical genotype-phenotype associations using large datasets and drawing on omics-based technologies, such as proteomics and metabolomics. They focus on the use of information technologies in medicine without depending on a detailed understanding of biological mechanisms.[31,32] This approach has been considered to be analogous to Amazon's recommendation engine, which utilizes a huge database of past purchasing behavior to predict which items individuals might want to purchase in the future.[31]

It seems that this approach, being a top-down modelling, considers the human body more or less as a "black box", the knowledge of the internal workings of which is rendered abundant vis-à-vis the information derived by big data analysis.[31] Such a neglect of the mechanical science of Galileo

and Harvey is a development of considerable importance not only for medicine but also for the philosophy of science in general. We may be heading towards a totally different conception or even a complete bypass of what we traditionally call "diagnosis".[33,34] Of course, the vision of precision medicine largely remains a future one. In oncology, however, clinical decisions already rely, at least partly, on computer algorithms.[35]

6. DSM as an essential bridge/hub between opposing approaches

As I see it, the landscape concerning psychiatric diagnosis today is roughly the following:

On the one hand, there is the requirement for a diagnosis which takes into consideration as much as possible the individual, subjective and phenomenological particularity of the mentally ill; sometimes, this requirement is expressed in a way which seems to underestimate the need for a common diagnostic language and communication between clinicians. On the other hand, there is the requirement for an approach of mental disorders which would increasingly rely on computational analysis of huge amounts of biological (genetic, neuroscientific) data; no doubt a future vision, the relation of which to diagnosis, clinical practice and clinical meaning[36] remains to be determined. Between these different and possibly contradictory exigencies and tendencies, DSM, with its categorical diagnoses and descriptive operationalistic criteria, despite its innate imperfections and inadequacies, seems to be a necessary bridge and, at the same time, the only means to define "clearly and distinctly" (to recall an old philosophical requirement) the ground and the object of psychiatry as a clinical medical specialty (figure 1).

Psychiatry as a medical specialty has its own, very particular, phenomenologically defined object[37] and, as a result, occupies an intermediate, albeit dominant, position between a psychoanalysis sensitive to the slightest subjective vibrations and a soulless analy-

sis of neuroscientific data. Psychiatry draws valuable information both from theory and research; in order, however, for psychiatry to accomplish its very special clinical mission, psychiatric diagnosis must necessarily be made also in the way that is the most natural to human perception and cognition: namely, the categorical. The mistake of the past is that DSM was used to a great extent as the sole diagnostic truth. Maybe now that its authority, as well as categorical diagnosis as such, are greatly disputed, it is the right moment to redefine its role. DSM (in its present and future editions, and similarly ICD), far from being a law, a doctrine, or a "bible" of diagnosis, can and ought to be:

– A bridge or interface between clinic and research,[36] as well as psychiatric and psychoanalytical clinical practice

– A common diagnostic language, which will not preclude the special use of other languages/classifications[38]

– An epistemic hub,[24] which will mediate between different theoretical approaches of mental disorders (see also Adan-Manes & Ramos-Gorostiza 2014).[39]

No-one seriously doubts the need for clinical communication, and many believe that "a dichotomy between science and practice is false"[40] and that the notion of "utility" is not distinct from that of "predictive validity", but overlaps with it.[41] The scientific requirement for diagnostic validity is nevertheless a strong one. Mario Maj puts it elegantly:

Neurobiological mechanisms are likely to be involved in most or all mental disorders, but the level at which the psychopathological identity of these disorders emerges may be higher than that of the brain machinery, and the elucidation of the higher-order (e.g., psychological, cultural) processes which intervene may be crucial.[41]

Given the particularity of the psychiatric object, the presuppositions for diagnostic validity should no doubt be sought for in the cells of RDoC, but also in those theoretical approaches that examine human subjectivity as such, including phenomenology and psychoanalysis.

More narrativity!
(phenomenology, psychoanalysis) ⟹ **DSM-ICD diagnostic categories** ⟸ **More information!**
(big data, genetics, neuro-science)

Figure 1. DSM-ICD as a bridge between opposing exigencies.

Η κατά DSM-ICD διάγνωση ως αναγκαία γέφυρα μεταξύ ασθενούς και "big data"

Γ.Β. Μητρόπουλος

5ο ΨΤΕ, Ψυχιατρικό Νοσοκομείο Αττικής, Αθήνα

Η χρήση διαγνωστικών εγχειριδίων στην ψυχιατρική επιβάλλεται από την έλλειψη εξετάσεων που να επιβεβαιώνουν την ψυχιατρική διάγνωση. Η διάγνωση κατά DSM-ICD, που είναι σήμερα η επικρατέστερη, έχει δεχτεί σημαντική κριτική, η οποία παραδοσιακά αφορά σε ζητήματα όπως, μεταξύ άλλων, η υποαφηγηματικότητα, ο βιολογισμός, ο «θάνατος της φαινομενολογίας» και η αμφίβολης εγκυρότητας υπερκατάτμηση της διάγνωσης. Τελευταία, και ιδίως μετά την 5η έκδοση του DSM (2013), η κριτική αυτή εστιάζει κυρίως σε ζητήματα όπως η έλλειψη εγκυρότητας, η μη-υιοθέτηση ενός διαστασιακού μοντέλου, στο γεγονός ότι το DSM δεν βασίζεται επαρκώς στη γενετική και τη νευροβιολογία, και στο ότι παρεμποδίζει μάλλον, παρά προάγει, την έρευνα πάνω στην αιτιολογία των ψυχικών παθήσεων, αποτελώντας μία «επιστημική φυλακή». Από τα παραπάνω ζητήματα, τα μεν μοιάζουν να απορρέουν από το γεγονός ότι τα οπερασιοναλιστικά (operationalist) διαγνωστικά κριτήρια συχνά υιοθετούνται κατά τρόπον άκριτο ως απόλυτη αυθεντία στη διάγνωση, αντί να αποτελούν απλώς οδηγούς, σύμφωνα με τη ρητή πρόθεση των δημιουργών τους· τα δε έχουν αναδειχθεί ιδιαιτέρως μετά την εμφάνιση του αμερικανικού ερευνητικού προγράμματος RDoC, το οποίο όχι μόνο δείχνει προς την κατεύθυνση μιας εναλλακτικής, περισσότερο έγκυρης, ταξινόμησης των ψυχικών διαταραχών, αλλά επίσης φιλοδοξεί να σημάνει τη μετατόπιση της ψυχιατρικής προς την λεγόμενη ιατρική ακριβείας, έχοντας ως βασικό δόγμα ότι οι ψυχικές διαταραχές είναι διαταραχές εγκεφαλικών κυκλωμάτων, οι οποίες εκφράζονται ως σύνθετα σύνδρομα. Σε αυτό το άρθρο, εξετάζεται καταρχάς το ιστορικό και επιστημολογικό πλαίσιο της εμφάνισης του DSM· τα επιτεύγματά του, όσον αφορά στη διαγνωστική αξιοπιστία (reliability) και την κλινική χρησιμότητα (utility), δεν υπήρξαν αμελητέα, ιδίως εάν λάβουμε υπόψη το κλίμα της οιονεί διαγνωστικής αυθαιρεσίας που χαρακτήριζε την προ του 1980 (αμερικανική) ψυχιατρική, με τις όποιες ευνόητες συνέπειες για το κύρος της ειδικότητας. Στη συνέχεια τίθεται υπό συζήτηση η δυνατότητα της νέας εποχής της γενετικής, της νευροβιολογίας και της ανάλυσης των «μεγάλων δεδομένων» (big data) να οδηγήσει σε μια νέα προσέγγιση της ψυχιατρικής διάγνωσης και ταξινόμησης, ενώ παραμένει προς το παρόν άγνωστο με ποιον τρόπο τα ευρήματά του RDoC θα μπορέσουν να οδηγήσουν και να μεταφραστούν σε ένα νέο ταξινομητικό σύστημα. Επιπλέον, γίνεται αναφορά στην ιδιαιτερότητα του ψυχιατρικού αντικειμένου, στην κλινική σημασία του κατηγορικού χαρακτήρα της διάγνωσης, καθώς και στην ανάγκη για ένα «μη αναγώγιμο ψυχολογικό επίπεδο εξήγησης». Κατά την άποψή μας, η διάγνωση κατά DSM-ICD βρίσκεται σήμερα μεταξύ δύο διαφορετικών και ενδεχομένως αντίθετων απαιτήσεων και τάσεων: αφενός της απαίτησης να λαμβάνεται υπ' όψη η ατομική, υποκειμενική και φαινομενολογική ιδιαιτερότητα του ψυχικά ασθενή (απαίτηση που ενίοτε υποτιμά την ανάγκη για κλινική επικοινωνία)· αφετέρου, του (εν πολλοίς μελλοντικού) οράματος για περισσότερη ανάλυση μεγαλύτερων βάσεων βιολογικών δεδομένων στο όνομα μιας αδιευκρίνιστης ακόμα εξατομίκευσης της θεραπείας (με την ίδια την έννοια της διάγνωσης να καθίσταται ενδεχομένως πλεονάζουσα). Τέλος, δεδομένης της ιδιαιτερότητας του ψυχιατρικού αντικειμένου, καταλήγουμε στο συμπέρασμα ότι η προσέγγιση των DSM–ICD, με τις κατηγορικές διαγνώσεις και τα περιγραφικά οπερασιοναλιστικά κριτήρια, παρά τις εγγενείς ατέλειες και ανεπάρκειες, εξακολουθεί να έχει θέση στην ψυχιατρική ως αναγκαία γέφυρα/διεπιφάνεια (interface) μεταξύ κλινικής και ερευνητικών δεδομένων, ως κοινή κλινική γλώσ-

σα, και ως επιστημικός κόμβος· και ότι οι προϋποθέσεις διαγνωστικής εγκυρότητας θα πρέπει να αναζητηθούν τόσο στα κελιά του RDoC, όσο και στις θεωρητικές προσεγγίσεις που εξετάζουν την ανθρώπινη υποκειμενικότητα ως τέτοια, συμπεριλαμβανομένης της φαινομενολογίας και της ψυχανάλυσης.

Λέξεις ευρετηρίου: Διάγνωση, DSM, κατηγορικός, big data, υποκειμενικότητα, φαινομενολογία.

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Corresponding author: G.B. Mitropoulos, 6 Adonidos street, GR-145 64 Athens, Greece, Tel: (+30) 6944 181 286
e-mail: geobmitro@yahoo.gr