

Differential diagnosis and current polythetic classification

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The introduction of polythetic diagnostic classification (DSM-III and ICD-10) in psychiatry was anticipated to improve the reliability of psychiatric diagnoses, facilitate research, and eventually allow the then future DSM-IV to become anchored in objective, etiological criteria. However, the preparations and release of DSM-IV and DSM-5 highlighted the fact that the etiological promise has not materialized. Psychiatric classifications will continue in the foreseeable future to be based mainly on clinical descriptions.

This has stimulated a broad range of reflections and critiques of psychiatric nosology (e.g., 1,2). Yet, the criticism is typically confined to concrete, technical issues, e.g. discussing a necessity for novel categories, modification of existing criteria, correcting definitions and misunderstandings, etc.. The polythetic-operational *foundation* of current classification remains largely unchallenged (3). Thus, despite a nearly universal consensus about the etiological stalemate, psychiatrists continue to believe that the polythetic system is epistemologically adequate and that it has indeed broadly improved clinical diagnostic reliability.

I wish to question the alleged improvement of reliability and to challenge the epistemological adequacy of the polythetic approach. The issue of differential diagnosis will serve as a concrete clinical embodiment of this critique. Needless to say, a full discussion of the theoretical and clinical ramifications of these topics (e.g., the issue of “comorbidity”) is beyond the scope of this article.

IMPROVED RELIABILITY

Diagnostic reliability is typically reported as interrater agreement for selected disorders in the so-called “field trials”, accompanying the construction of diagnostic criteria, or in research studies. The data behind such reports stem from somewhat artificially constructed measurement contexts that formalize what actually happens in the ordinary, everyday clinical practice. Such reports also tend to embellish or even inflate the presented reliability levels (4).

We have no data on the general quality (reliability and concurrent validity) of contemporary clinical diagnostic practices, or data comparing the general utility of successive diagnostic systems. We lack anthropologically oriented research, emphasizing ecological aspects of reliability, i.e. examining the actual reliability of working psychiatrists as they assign a diagnosis, situated in their daily environment.

Instead, we may use other sources of information that, at least indirectly, may shed some light upon these issues. Several, disparate signs jointly indicate a still existing and serious problem with diagnostic reliability.

First, we witness the epidemic-like explosion of certain diagnoses (e.g., autistic spectrum, attention-deficit/hyperactivity disorder). Such dramatic increases may be due to an actual and *true* rise in the incidence, changes in treatment seeking behavior, or availability of novel and more efficacious treatments. However, common clinical experience suggests that quite often such “popularities” emerge because physicians become unduly impressed by newly circulated checklists targeting specific disorders while failing to perform a comprehensive diagnostic assessment, or because physicians are unaware of or simply ignore the diagnostic rules.

Thus, a study of referrals to a mental health center in Netherlands (5) found that, among 242 first-contact patients reporting at least one unequivocal psychotic symptom, only 44% were diagnosed with psychosis, whereas 56% received a non-psychosis diagnosis or no diagnosis at all. In another study of patients discharged with a diagnosis of schizoaffective disorder from two Danish university clinics (6), only 10% of cases actually fulfilled operational criteria for that disorder, whereas the remainder suffered from schizophrenia or bipolar disorder.

Finally, the forensic-psychiatric odyssey of the Norwegian mass-murderer A. Breivik, independently assessed by two teams of psychiatric experts with the resulting ICD-10 diagnoses of paranoid schizophrenia and personality disorder respectively, does not testify to a dramatic improvement of reliability (7,8).

THE POLYTHETIC-OPERATIONAL DIAGNOSTIC SYSTEM

A polythetic diagnostic category of current DSM/ICD is based on a list of symptoms and signs believed to be characteristic for the diagnosis in question. Typically, a certain number of diagnostically equivalent symptoms or signs from a given list is sufficient to arrive at a diagnosis. These “diagnostic criteria” are, contrary to a widespread belief, not “operational” in any epistemological or scientific sense. They are just briefly described in an ordinary non-technical lay language at “the lowest order of inference” (3).

Two issues deserve attention here. First, the symptoms/signs *shared* by two or several disorders tend to be omitted from the diagnostic lists in order to strengthen the clinical distinctiveness of the categories (e.g., depressed mood and anxiety are exclusively listed in the context of mood and anxiety disorders). Second, the simplification of the psychopathological descriptions to brief, lay language statements converts the symptoms and signs into phenomenological primitives or homogeneous elements. There is only one kind of delusion (i.e., it is assumed that all delusions share the same phenomenological structure), one kind of anxiety, one kind of auditory verbal hallucination, etc.. Consequently the syndromes, solely constituted by aggregates of such elements, lose their characteristic salience, and their boundaries become blurred. A recent study using a network model of DSM-IV symptoms demonstrated that half of the symptoms are connected with short paths. The individual disorders are therefore also mutually proximate, accounting for the high levels of empirically observed comorbidity (9).

The narrative, conceptual and phenomenological descriptions of pre-DSM-III psychopathology were eliminated from the contemporary diagnostic manuals. Those descriptions contained a discussion of the characteristic prototypes of mental disorders, their phenomenological structures and the interdependency of their constituent features (e.g., in the manic syndrome, the potential relations between the global “volatility” of the manic gestalt, increased mood, vitality, psychomotor speed, and grandiosity). They also contained a consideration of the phenomenological structure of the individual symptoms and signs, their relations of implication or entailment, and their context dependence. Such information no longer exists in the diagnostic manuals and is largely gone into oblivion.

For example, a reader of DSM-IV or DSM-5 is told that schizophrenia is a mixture of positive and negative symptoms that happens to satisfy certain inclusion and exclusion criteria. This definition says more about what schizophrenia *is not* (e.g., non-organic, non-affective) than *what it is* (10), i.e. what kind of validity is behind this category (11), what is its characteristic gestalt that constitutes its difference from other potentially similar mixtures of positive and negative symptoms (12), what justifies schizophrenia’s dominating diagnostic rank in the taxonomic hierarchy, or why it is risky to expose a patient with schizophrenia to an orthodox psychoanalysis.

MAKING DIAGNOSIS AND DEFINING CONCEPTS: PROTOTYPES AND GESTALTS

The process of differential diagnosis in the pre-DSM-III era was framed by *prototypical* considerations. Although such considerations still take place or, more exactly, *cannot avoid* to take place in any diagnostic situation (including somatic medicine), in psychiatry they only operate on an implicit, un-reflected level, because they are un-anchored

and incompatible with the philosophy underlying the polythetic-operational classification.

A prototype is a central example of a given category (a sparrow is more typical of the category “bird” than is a penguin or an ostrich), with a graded dilution of typicality towards its borders, where it eventually overlaps with neighboring prototypes. Thus, the prototypical categories exhibit an intrinsic dimensionality (13). However, a prototype is *not just an example* (exemplar), but contains condensed information on its internal configuration of properties and its relations to neighboring prototypes (14). The concept of prototype/gestalt is fit for description of single symptoms and signs as well as larger entities such as diagnostic categories. One can use the concept of prototype-gestalt in a narrow or a wide sense, neither one limited to perception but also involving complex cognitive-affective operations.

In a narrow sense, a gestalt is a unity or organization of phenomenal aspects, that emerges from the interactions among its component features (part-whole relations). The whole is irreducible to a mere aggregate, because it is more than a sum of its parts. In a diagnostic process there are reciprocal dependencies between the whole and its single features. The clinical whole confers on its constitutive features their characteristic diagnostic significance. Conversely, the single clinical features, by instantiating the gestalt, imbue it with clinical concreteness and rootedness (12).

In a wider sense, the notion of gestalt entails an interplay of factors that extend beyond the subject to include not only a mental state, but also the patient’s engagements with the environment and others. For instance, detecting a delusion involves taking into account not only the patient’s verbal contents but also his experiences, way of arguing, relational style and relevant historical information. To use the concept of delusion competently, a psychiatrist must master plenty of other prototypes and concepts (e.g., psychosis, rationality, reality, hallucination, etc.) (8).

The argument for a prototype-based diagnosis is fundamentally linked to the fact that perception is always apperceptively (conceptually) informed: perceiving something is to perceive it *as a something*, as a token of a certain type. A perceptual or cognitive object is always given as a certain gestalt. The unfamiliar is perceived in terms of the familiar, i.e. in terms of the general type or gestalt that is “activated in the particular perception” (15). This process is called *typification* and is intrinsic to all human cognition and hence to the diagnostic process as well.

The natural unfolding of a comprehensive semi-structured prototype-based diagnostic assessment involves *reflective and critical questioning* of typifications, which become supported, weakened or discarded by explicitly elicited diagnostic information on symptoms, their evolution, social history etc., progressively limiting the number of diagnostic options (16). Typification as such can never be eliminated because it is an automatic aspect of cognition. A recent review of mechanisms involved in concept formation, use and understanding suggests that *concepts* (e.g., psychiatric categories)

are not constituted by a list of criteria, but are organized around prototypes/gestalts (17): “Theory of concepts must be primarily prototype-based... , within a broader knowledge representation scheme in which the concept is positioned both within a hierarchy and within a theoretical framework(s) appropriate to that domain” (14, p. 488). It follows that the more knowledgeable and experienced is the psychiatrist, the more refined is the diagnostic repertoire.

Finally, it needs to be emphasized that prototypically defined and described nosological categories may be enriched and supplemented by lists of criteria. This was, in fact, the original but, unfortunately, unrealized intention behind the DSM-III (3).

CLINICAL REALITY OF DIFFERENTIAL DIAGNOSIS IN A POLYTHETIC SYSTEM

All diagnosis is an instance of differential diagnosis: the task is to pick up, from a larger catalogue of potential options, the one that most adequately fits the patient.

Let us then imagine a young clinician in an open outpatient facility, trained with the DSM/ICD manuals as her exclusive source of psychopathological knowledge. She encounters a self-referred male in his early 20-ies, sitting on the floor of the waiting room in a lotus position, mumbling, and occasionally laughing to himself in a silly manner. How should she proceed after the initial greetings?

Since her diagnostic-cognitive field lacks a prototypical-conceptual grid, she is exposed to what in cognitive science is known as a “frame problem”, i.e. the issue of how to decide what is relevant, indeed what is even the relevant overall context within which to approach a given problem (16). Theoretically, she would therefore need to explore the inclusion and exclusion criteria for nearly all disorders (the number will vary with the degree of diagnostic hierarchy). That is, obviously, not feasible in practice. Instead, she may imitate a digital computer and use a structured interview. Such interview is essentially constructed as a binary decision tree with mandatory probing questions and suggested cut-off points. The epistemological problems and the quite meager pragmatic utility of structured interviews have been amply addressed (16,18). Here, it is important to note that the very nature of structured questioning confers a limited diagnostic utility on the interview, because of the low sensitivity and specificity of the responses. Responding with a “no” or “yes” to the question of “feeling down” neither excludes nor strongly supports any specific diagnosis.

Most likely, our clinician will conduct a so-called “clinical interview”, a conversation starting with the patient’s complaints and reasons for seeking help, and assisted by various symptom checklists locally in use. In this process, the patient may be diagnosed with major depression if he answers affirmatively to five or six criteria of this diagnosis. In other words, for a psychiatrist untrained to impose a conceptual-psychopathological grid on the diagnostic information, the

patient’s initial behavior (suggestive of schizophrenia) may easily fail to display a relevant clinical salience and hence fail to enter into the diagnostic considerations.

Thus, a young psychiatrist, unfamiliar with the prototypical structure of psychopathology, finds herself exposed to a myriad of chaotic, unconnected data, where each individual feature is equally worthy of attention and may therefore become a pivot of a potential diagnostic class. With growing experience, this clinician will invariably acquire her own *private* prototypes, shaped by the local ideologies and habits and by personal inclinations, i.e., in an implicit way that is *not exposed to an academic, rigorous, and peer-shared reflection*. “Private” prototypes become easily activated by single, popping up clinical features that happen to evoke a single aspect of a contingent diagnostic category. Here, a decisive role is often played by the very first verbalizations of the complaint. If a patient mentions a habit of cutting herself, a “borderline” diagnosis will be likely considered.

CONCLUSIONS

The distinctions and concepts in the realm of experience and behavior play now, and will continue to play, a decisive role in psychiatric classifications. These distinctions do not function with the simplicity of causal referents, as it is often the case with signs and symptoms of somatic medicine (e.g. jaundice → bilirubine cycle). Rather, they exhibit a phenomenological-empirical and theoretical complexity, which cannot be adequately represented through the simplifying, reductive approach of the operational-polythetic system.

The differential diagnostic process is not (only) a matter of a digitalized decision tree, but involves context dependencies and complex pattern recognitions. These empirical, phenomenological and theoretical issues constitute the domain of the *science of psychopathology*. In recent decades, research, study and training in psychopathology have been seen as largely redundant, because the polythetic manuals seemed to offer all that was needed for research and practice. These assumptions have proven to be false.

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