Diagnosis: one useful method among many

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In their Comment, Boyle and Johnstone[1] argue that the "paradigm" of psychiatric diagnosis "has comprehensively failed" and is potentially countertherapeutic and untenable. Their claim is based on the argument that biomarkers for specific diagnoses do not exist, substantial stigma is associated with diagnosis, and diagnosis results in a necessarily medical approach to so-called functional mental distress, interpreted by clinicians as one of various clearly delineated biological diseases needing treatment with potentially disabling drugs. If this characterisation was true, we would wholeheartedly agree, but clearly it is not, and much of their effort is directed at a straw man (misrepresentation of the position of diagnosis then refutal of the misrepresentation).

Diagnosis is not, in any sense, a "paradigm", but a series of classifications. Although many difficulties exist with the present set of diagnoses,[2] these difficulties do not constitute an argument against diagnosis itself because each individual classification should be judged on its own merits. The same absence of argument applies equally to other forms of classification, including the conceptual categories that the authors mention in their own critique.[1] In the place of diagnosis, they argue for an individualised approach that encompasses the richness of individuals' lives and the personal intelligibility of their problems.

Although not explicitly named, Boyle and Johnstone are advocating a formulation-based approach. We agree that this approach is a useful and important method, which is undoubtedly why it is recommended for both psychiatrists[3] and clinical psychologists[4] by their respective professional bodies. However, we do not see formulation as either mutually exclusive or necessarily a successor to diagnoses, as both can be complementary. In addition to diagnoses being used within formulations, we note that many evidence-based psychotherapies use psychological formulations drawn from diagnoses, such as obsessive compulsive disorder[5] and post-traumatic stress disorder.[6] Formulations in themselves do not necessarily avoid difficulties with diagnosis. We agree with Johnstone and Dallos'[7] earlier warning that "there is no guarantee that formulations will not be used in a stigmatising, objectifying, un-collaborative way", and concerns about reliability and validity apply equally.[8] In the same way that diagnoses can be used badly, so can formulation.[4] Both diagnoses and formulations are useful but imperfect methods. We do not argue for either to be jettisoned wholesale, but neither should be used uncritically or be exempt from continued research and critique.

Boyle and Johnstone argue that to make a psychiatric diagnosis is synonymous with labelling of a single presumed disease entity, validated by identification of a specific biomarker. However, most clinicians and professional bodies, following the existing body of research, now agree that clinical diagnoses in psychiatry are most likely to represent syndromes that have multifactorial and overlapping causes that include personal, social, and biological factors.[9]

Diagnoses are sometimes stigmatising, but not always. Different attitudes to specific diagnoses between and within people have been recorded widely in the literature,[10] and we note that Horn and colleagues (including Johnstone)[11] showed exactly this difference in research on individuals diagnosed with borderline personality disorder. The authors correctly state that psychiatric drugs generally fail to target underlying disease processes or correct chemical imbalances. However, in truth, very few drugs meet these criteria. We do not argue against pain relief, chemotherapy, or blood pressure drugs on these grounds; however, like these drugs, psychiatric ones can improve quality of life when used appropriately.[12]

Boyle and Johnstone suggest that to see people as having "intelligible reasons for thoughts, feelings and behaviours" is somehow in opposition to diagnosis, largely based on their "every diagnosis a disease" fallacy. However, we would argue for a less narrow definition of the term "intelligible" in this context. Many causes of emotional distress and mental illness exist, but not all will be fully understandable at the level of personal meaning. For example, cerebrovascular changes, seizure activity, autoimmune disorders, and prescription corticosteroid use are some of the many confirmed causes of psychosis,[13] but an individual's experience of these effects might not be a reliable guide to their significance for recovery. Conversely, in instances in which a clear-cut medical diagnosis exists, psychosocial factors have an important role and similarly need to be understood and formulated. For example, HIV acts at

the level of DNA, but social and psychological factors are equally important in acquirement of the infection and management of the disorder. "Intelligible" can include both medical explanation and lived experience, which seems to be a richer and more plausible view of the individual.

In essence, the distinction between purely functional and purely organic does not exist, but is instead a false dichotomy that pervades the authors' argument. All of our thoughts, feelings, and behaviours should be understood in terms of biological, psychological, and social causation. We therefore argue that medical approaches and appropriate diagnoses are a necessary, but not sufficient, component of a partnership for better mental health.

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New treatment targets for autism spectrum disorders: EU-AIMS

Autism spectrum disorders are one of the most common and severe neurodevelopmental disorders, but no effective treatments for core symptoms are available.[1] The main reasons for the absence of effective treatments are the high clinical and genetic heterogeneity between affected individuals, restricted knowledge of the underlying pathophysiological mechanisms, and the lack of reliable diagnostic biomarkers. Hence clinical trials, which have largely been unsuccessful so far, rely on biologically diverse groups of patients, operationally defined according to the Diagnostic and Statistical Manual of Mental Disorders and the International Statistical Classification of Diseases and Related Health Problems, 10th revision. The identification of more homogenous biological subgroups is therefore essential for the development of novel treatments based on the molecular mechanisms underpinning autism spectrum disorders.

Recent advances in genomics and new methods to model pathophysiological mechanisms in vitro and in vivo might now make identification of new treatment targets and the stratification of patients according to biological biomarkers possible. Hence, in 2012, the Innovative Medicines Agency set up a large-scale public–private partnership—EU-AIMS—to harness these advances in an integrated translational research programme aiming to identify new biomarkers and treatment targets for autism spectrum disorders.

EU-AIMS brings together 14 academic partners, seven members of the European Federation of Pharmaceutical Industries and Associations, three small-to-medium enterprises, and patient organisations.[1] The consortium has five interlinked themes (figure). Our programmes on cellular assays and animal models capitalise on the discovery of rare monogenic forms of autism spectrum disorders to identify pathophysiological mechanisms. Because many of these risk genes seem to converge on common molecular pathways,[2] this approach promises to identify treatment targets for larger groups of patients. We mainly (but not exclusively) focus on genes involved in synaptic development and function, and their effect on excitatory–inhibitory imbalance and brain connectivity.[3] A translational research programme links this work to human beings by using electrophysiological, neuroimaging, and molecular imaging methods.

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