University of Groningen

Autism's anatomy

Verhoeff, Berend

IMPORTANT NOTE: You are advised to consult the publisher's version (publisher's PDF) if you wish to cite from it. Please check the document version below.

Document Version
Publisher's PDF, also known as Version of record

Publication date:
2015

Link to publication in University of Groningen/UMCG research database

Citation for published version (APA):
Verhoeff, B. (2015). *Autism's anatomy: A dissection of the structure and development of a psychiatric concept*. University of Groningen.

Supplement[24]

Challenging a mechanistic model of mental disorders

A fundamental question in the philosophy of psychiatry is: What kind of things are psychiatric disorders? This issue is being discussed extensively in a philosophically oriented literature, but there is still no consensus as to the best answer. Can psychiatric disorders best be conceived of as; objects that exist in nature independent of psychiatric classifications (natural kinds, see, for example, Haslam, 2003; Cooper, 2004); scientifically constructed tools or instruments that help to achieve important goals (practical kinds, see, for example, Zachar, 2002); or maybe as kinds that are brought into being by societies and cultures through the practice of classifying human behavior as distinct kinds (socially constructed kinds, see, for example, Young, 1995)?

Current assumptions, understandings and practices in the field of autism, I suggest, are compatible with a permissive account of natural kinds, namely the *mechanistic property cluster* (MPC) account of natural kinds recently proposed by Kendler, Zachar and Craver (2011) as *the* model for understanding psychiatric disorders in general. However, despite the attractiveness of a value-free mechanistic model, the MPC model has certain limitations. In this supplement to the previous chapter, I illustrate how these limitations relate to the traditional separation of two types of demarcation problems in (the philosophy of) psychiatry – between distinct mental disorders on the one hand, and between normality and pathology on the other hand. A mechanistic model of psychiatric

disorders only concerns the former demarcation problem while it is indifferent with respect to the latter. Due to this limitation, this model is unable to account for the way in which social and cultural norms, and shifting boundaries of normality and pathology shape and transform autism as a psychiatric entity.

Mechanistic property cluster (MPC) kinds

In a recent essay in *Psychological Medicine*, Kendler, Zachar and Craver (2011) argue for a mechanistic model for understanding psychiatric disorders. Inspired by the philosopher Richard Boyd (1991; 1999) they suggest that psychiatric disorders can best be viewed as *mechanistic property cluster* (MPC) kinds. Boyd developed the concept of homeostatic property clusters (HPC) to challenge a stringent essentialist model of natural kinds in which a necessary and sufficient property or structure (an essence) directly and causally determines all key features of a kind (Kendler and colleagues replaced the term 'homeostatic' with 'mechanistic' to avoid possible confusion due to different meanings of the term 'homeostatic').

According to Boyd, there are scientifically important kinds – biological species for instance – that are characterized by a cluster of often co-occurring characteristics and by the underlying mechanisms that bring about their co-occurrence. These clusters do not have invariable and exclusive essences and the members of a kind do not need to overlap in a fixed set of characteristics. Rather, kind membership is defined by some set of empirically discoverable causal mechanisms that explain, in the case of biological species, 'the imperfectly shared and homeostatically related morphological, physiological and behavioral features which characterize its members' (Boyd, 1991: 142). Similar stable patterns of often complex causal mechanisms that involve interactions between multiple possible levels of explanation – such as physiology, behavior and environment – instantiate the imperfect co-occurring characteristics of the members of a species. They are considered imperfect because 'kind definitions must conform to the (sometimes messy and complex) causal structure of the world' (Boyd, 1991: 143). Members of a species need not share all their characteristics, and differences between species can be vague. However, this doesn't imply that there are no stable explanatory mechanisms to be discovered underlying common characteristics of individual members of a species.

Kendler, Zachar and Craver (2011) suggest that Boyd's HPC model of kinds should be the key model for understanding what kind of things psychiatric disorders are. They ask us to consider a multi-dimensional matrix that reflects human mind/brain states. The properties included in this matrix may include genes, neural systems, psychological states, symptoms themselves and environmental inputs. They argue that there are only a finite number of mind/brain states that 'are cohesive and temporally stable, some proportion of which represents "psychiatric syndromes"' (p. 1147). For them, psychiatric disorders are best conceived of as sets of symptoms that are connected through a system of causal mechanisms. Ultimately, these causal mechanisms are what define and sustain the disorder.

This MPC model of mental disorders is attractive for several reasons. It corrects an empirically inadequate 'gene X causes disorder Y' (essentialist) model, it is compatible with the multicausality, fuzzy boundaries and heterogeneity of most psychiatric disorders, and it provides (unlike pragmatist models) prescriptive guidance for the investigation of objective causal structures that will inform psychiatric nosology in the attempt to carve nature at its joints (ibid.). These joints are not located at the boundaries of single genes, infective agents or local lesions, but at the boundaries of causal mechanisms (Samuels, 2009). Thus, the MPC model facilitates the prospect of discovery and 'true' delineation of specific disorders. Since MPC kinds are grounded in the natural features of the world and are 'not merely imposed upon the world by psychiatrists through their classificatory practices', psychiatric categories will become scientifically valuable in terms of prediction, explanation and control. Even though there is no single causal mechanism or essential property that explains all the superficial properties of a kind, 'the identity of the disease across time and across cultures is grounded in the similarity of the complex mutually reinforcing network of causal mechanisms in each case' (Kendler et al., 2011: 1147).

Furthermore, as both Kendler, Zachar and Craver (2011) and Samuels (2009) – who defends an MPC model for delusions – underline, MPC kinds allow 'that the same cluster of symptoms might arise from different etiological, underlying or sustaining mechanisms in different cases' (Kendler et al., 2011: 1147). There need not be a one-to-one relation between an underlying neurobiological causal mechanism and the resulting cluster of psychiatric symptoms. However, distinct etiological or pathophysiological mechanisms in

different members of the same kind must share a similar causal mechanism at another biological level. Biological heterogeneity is allowed, as long as more homogeneous mechanisms can be identified at other biological levels. Much of the research in autism is, despite profound genetic heterogeneity, directed at identifying unifying neural mechanisms that underlie all – or a subgroup of – autism cases. According to Samuels (2009), identifying such unifying mechanisms comprises perhaps *the* fundamental explanatory challenge for an MPC approach to psychiatric disorders. Kendler, Zachar and Craver (2011) conclude that we are 'far from being able to define plausible stability-producing mechanisms for most psychiatric disorders' (p. 1148).

However, as Chapter 2 also illustrated, contemporary researchers in the field of autism generally, but usually unknowingly, follow the prescriptive guidance of the MPC model. A common neurodevelopmental abnormality is still assumed to unite all – or a subgroup of – autism patients. Functional genomics, epigenetics, molecular genetics and systems biology are among the new hopes in the search for autism's unity. Current developments in autism research fit strikingly well with the MPC model proposed by Kendler et al. (2011). Autism researchers and clinicians need to deal with multiple causes and (genetic) heterogeneity, but autism research is nonetheless directed at identifying 'objective' causal mechanisms that should inform nosologists in their attempt to carve autism's boundaries at its supposed natural joints. However, a convincing mechanistic approach to autism requires a clear separation of two familiar types of demarcation problems in psychiatry.

Two demarcation problems

The first demarcation problem concerns the question of whether and when a certain constellation of signs and symptoms legitimately reflects a distinct category. Is schizophrenia, for instance, a valid disease category and to what extent is schizophrenia distinct from schizo-affective disorder, delusional disorder or any other ('normal') state or trait? A central term in this debate is *validity*. This is a complex construct with several meanings and subtypes, which I do not discuss here in detail. Rather, I briefly focus on how this term has been used in psychiatric nosology.

Robins and Guze (1970) were the first to propose a formal method to improve the validity of psychiatric categories. In their influential article on establishing diagnostic validity for schizophrenia, they proposed five phases in the evaluation of a putative diagnostic category that they thought were an indication of its validity: clinical description, laboratory studies, delimitation from other disorders, follow-up studies, and family studies. These validators were used to show that 'apparent "schizophrenia" with a good prognosis is not a mild form of schizophrenia, but is a different illness' (p. 987). Their findings provided the basis for the distinction between schizophrenia and schizophreniform disorder in *DSM-III* (APA, 1980). Kendler (1990) expanded the set of validators and distinguished between antecedent validators (familial aggregation, premorbid personality, and precipitating factors), concurrent validators (including psychological tests), and predictive validators (diagnostic consistency over time, rates of relapse and recovery, and response to treatment).

A common assumption underlying discussions about validity and proposals to increase the validity of psychiatric categories is that a 'truly' valid psychiatric disorder reflects genuine underlying (pathophysiological) differences in relation to other disorders and normal brain functioning. Kendell and Jablenski (2003) argue that while the diagnosis of psychiatric disorders is still based on clinical observation, a distinct syndrome will be valid if we reasonably expect that it can be defined by physiological, anatomical, chromosomal, histological or molecular abnormalities. Besides increasing reliability, since *DSM-III* the aim of psychiatric classification systems has been to create psychiatric categories that facilitate the identification of genes, neurotransmitter mechanisms and other neurobiological markers related to psychiatric disorders. In line with this aim, the ultimate goal of psychiatric taxonomy, as the research agenda for *DSM-5* Kupfer et al. (2002) concluded on this issue, has become 'to translate basic and clinical neuroscience research relating brain structure, brain function, and behavior into a classification of psychiatric disorders based on etiology and pathophysiology' (p. 70).

The MPC model of psychiatric kinds is in line with this effort. By informing nosologists, the MPC model attempts to increase the validity of psychiatric categories, where validity depends on whether a certain psychiatric category captures genuine underlying differences. The MPC model is supposed to bring us closer to the ultimate goal of current psychiatric nosology, which is a system

based on etiology and pathophysiology with neuroscience providing the foundation for classification and possibly individual diagnosis. However, the MPC model and conventional discussions on validity are largely indifferent towards another central demarcation problem in psychiatry. This second demarcation problem which will be discussed below, concerns a more general question: How can the distinction between normal and pathological mental functioning be made?

In a comprehensive monograph on this contested topic, Bolton (2008) discusses several possible ways to make this distinction. One way, for instance, is to conceive pathological mental functioning as 'a matter of breakdown of meaningful connections in mental life' (p. 16). Examples of a breaking down of meaningful connections include emotions that are excessive or have no appropriate object, behavior that is not under the control of the person's will, and beliefs that have no basis in experience. Another possibility, inspired by the work of Jerome Wakefield (1992), is to conceive of pathological mental functioning as 'not functioning as it has been naturally designed to do in the evolutionary process' (Bolton, 2008: 17). Despite the value of some of the theories he discusses, Bolton concludes that there is not one single theory that adequately distinguishes all forms of mental pathology from normality. Furthermore, in line with a widespread consensus among philosophers of psychiatry, Bolton concludes that how the line between what is normal and what is pathological in mental functioning is drawn depends on social, cultural and individual values and circumstances. Even Jerome Wakefield (1992), who is considered to be on the naturalist side concerning mental disorders, acknowledges that a biological dysfunction needs to be *harmful* in order to become pathological, and harm cannot be understood independent of sociocultural circumstances.

In defending the value-free MPC approach for delusions, Samuels (2009) is well aware of the two potentially conflicting demarcation problems. However, he argues that the normativity of pathology is not necessarily but only contingently connected with delusions. Without some reason to suppose that this connection is a necessary one, this normativity does not pose a threat to the MPC model regarding delusions (ibid.). Kendler, Zachar and Craver (2011) are equally aware of the evaluative nature of mental pathology as they acknowledge that 'values are intimately involved in determining which psychiatric kinds deserve clinical attention' (p. 1147). However, values are not

only involved in determining whether the condition we have come to call autism deserves clinical attention, they are also involved in defining and delineating this psychiatric kind in the first place. Taking the distinction between normality and pathology into account is crucial for understanding the way in which autism emerged, transformed and is currently defined as a diagnostic entity.

Limitations of a mechanistic model

As Cooper (2010) convincingly argues, culture-bound syndromes that emerge in highly specific social and historical contexts can still be distinct 'natural' disorders. For instance, similar to different kinds of igneous rocks that are created under specific environmental conditions, a mental disorder can be influenced by cultural and environmental factors such as diet, lifestyle or environmental pollution, and still be a distinct natural (MPC) kind grounded in a network of causal mechanisms. Social and cultural factors can be considered as causal agents that become part of the entire network of causal mechanisms associated with the particular kind. Biology and culture may interact, Cooper argues, 'so as to produce cases of a disorder that are recognizably and reliably similar to each other and such disorders can usefully be recognized by psychiatric classification systems' (ibid.: 331).

Following Cooper's argument, putative culturally and historically specific *causal* factors (for example, child-rearing practices or environmental toxins) and, as a hypothetical consequence, varying prevalences or manifestations of autism all over the world would not necessarily threaten a mechanistic (MPC) model of autism. However, the fundamental requirement of the model, that the *identity* and *boundaries* of a particular disorder are *set* by causal mechanisms, is particularly problematic for autism. As Kendler, Zachar and Craver (2011) argued, 'the identity of the disease … is grounded in the similarity of the complex mutually reinforcing network of causal mechanisms in each case' (p. 1147). 'An MPC kind' is their best answer to the ontological question: What kind of thing is a psychiatric disorder? However, the historically and culturally variable boundaries of 'impairment of social interaction' or 'a lack of ability to understand and use the rules governing social behaviour' – now considered essential features of autism – are clearly not set by causal mechanisms. This

issue of setting the boundaries of autism is not just a matter of demarcating a coherent cluster of signs and symptoms, it is also a matter of demarcating normality from pathology.

Social and cultural values and norms not only influence whether a certain cluster of symptoms is considered as a disorder, but they play, in autism at least, a necessary role in what becomes a recognizable cluster of symptoms in the first place. Defining autism as a nosological entity incorporates the (shifting) needs and discontents of a society regarding how an individual interacts with others, empathizes, makes friends, seeks to share enjoyment, initiates small-talk, and figures out implicit social norms. This blurs the boundaries between the two discussed demarcation problems as demarcating autism (and identifying neurobiological dysfunctions related to autism) *necessarily* involves demarcating undesirable conditions. An MPC model of autism that attempts to ground the identity and boundaries of autism in causal mechanisms has to ignore these normative dimensions.

Mental disorder in *DSM-5*

Both in the definition of mental disorder in *DSM-IV* (APA, 1994) and in the proposal by Stein et al. (2010) for a modified definition of mental disorder for *DSM-5*, the two discussed demarcation problems are reflected in separate criteria (see also Broome and Bortolotti, 2010; Verhoeff and Glas, 2010). In particular, criterion A, that a mental disorder is 'a behavioral or psychological syndrome or pattern that occurs in an individual', implicitly concerns the first demarcation problem of whether a certain cluster of features legitimately reflects a distinct disease (Stein et al., 2010: 1761). Criterion B – 'the consequences of which are clinically significant distress (for example, a painful symptom) or disability (i.e. impairment in one or more important areas of functioning)' – refers to the second general problem of demarcating normality from pathological mental functioning.

The separation of the two demarcation problems in different criteria is compatible with an MPC model of psychiatric kinds, in which a behavioral or psychological syndrome or pattern (cluster) reflects underlying (psychobiological) mechanisms. Whether it 'deserves clinical attention' (criterion B) can be approached as a separate issue. However, for autism, as we

have seen, these two problems are inextricably linked to each other. The phrase 'in an individual' in criterion A is particularly problematic for autism. As Broome and Bortolotti (2010) indicated, the phrase 'in an individual' is complex, controversial and carries conceptual baggage. It may seem evident that certain psychological states and behavioral patterns belong to or reside in an individual. However, as the case of autism illustrates, the recognition and description of an autistic behavioral pattern or particular autism signs and symptoms is profoundly embedded in a social and cultural context. Defining autism depends on historically and culturally variable ideas about deficiency, abnormality and dysfunction, and on the need to demarcate and treat particular discontents and impairments that have appeared. The case of autism, generally considered to be one of the most 'biological' of all mental disorders, illustrates Broome and Bortolotti's (2010) suggestion: 'that at the very least the claim that a disorder occurs "in an individual" warrants further examination' (p. 1784).

Conclusion

The mechanistic property cluster (MPC) model, which attempts to define and delineate autism in terms of causal mechanisms, is attractive for several reasons: it corrects an empirically flawed essentialist model; it is compatible with the multicausality, heterogeneity and fuzzy boundaries of many mental disorders; it provides prescriptive guidance for the investigation of objective causal structures; and it 'satisfies the intuitions of reductionist psychiatrists' (Kendler et al., 2011: 1148). Current autism research fits the MPC model strikingly well, as autism research – despite the acknowledged heterogeneity of the condition – is guided and regulated by the depiction of autism as a scientific and natural object that can be discovered and identified with systematic neuroscientific investigation. However, the MPC model of natural kinds (needs to) neglect(s) the way in which autism relates to ideas about what kind of behavior is inappropriate and in need of correction or support. As Chapter 2 argued, normative issues concerning disability and impaired social interaction have been and still are inextricably linked to how we recognize and understand autism.

References

APA (1980) *Diagnostic and Statistical Manual of Mental Disorders, 3th Edition (DSM-III)*. Washington, DC: American Psychiatric Association.

APA (1994) *Diagnostic and Statistical Manual of Mental Disorders, 4th Edition (DSM-IV)*. Washington, DC: American Psychiatric Association.

Bolton D (2008) *What is Mental Disorder?* Oxford, UK: Oxford University Press.

Boyd R (1991) Realism, anti-foundationalism and the enthusiasm for natural kinds. *Philosophical Studies* 61: 127-148.

Boyd R (1999) Homeostasis, species, and higher taxa. In Wilson RA (ed) *Species: New Interdisciplinary Essays*. Cambridge, MA: MIT Press, 141-185.

Broome M and Bortolotti L (2010) What's wrong with 'mental' disorders? A commentary on 'What is a mental/psychiatric disorder? From DSM-IV to DSM-V' by Stein et al. (2010). *Psychological Medicine* 40: 1783-1785.

Cooper R (2004) What is Wrong with the DSM? *History of Psychiatry* 15: 5-25.

Cooper R (2010) Are culture-bound syndromes as real as universally-occurring disorders? *Studies in History and Philosophy of Biological and Biomedical Sciences* 41: 325-332.

Haslam N (2003) Kinds of kinds: A conceptual taxonomy of psychiatric categories. *Philosophy, Psychiatry and Psychology* 9: 203-217.

Kendell R and Jablensky A (2003) Distinguishing between the validity and utility of psychiatric diagnoses. *American Journal of Psychiatry* 160: 4-12.

Kendler KS (1990) Toward a scientific psychiatric nosology. Strengths and limitations. *Archives of General Psychiatry* 47: 969-973.

Kendler KS, Zachar P and Craver C (2011) What kinds of things are psychiatric disorders? *Psychological Medicine* 41: 1143-1150.

Kupfer DJ, First MB and Regier DA (2002) *A Research Agenda for DSM-V*. Washington, DC: American Psychiatric Association.

Robins E and Guze SB (1970) Establishment of diagnostic validity in psychiatric illness: its application to schizophrenia. *American Journal of Psychiatry* 126: 983-987.

Samuels R (2009) Delusion as a Natural Kind. In Broome MR and Bortolott L (eds) *Psychiatry as Cognitive Neuroscience: Philosophical Perspectives*. Oxford: Oxford University Press, 49-79.

Stein D, Phillips K, Bolton D, Fulford K, Sadler J and Kendler KS (2010) What is a mental/psychiatric disorder? From DSM-IV to DSM-V. *Psychological Medicine* 41: 1143-1150.

Verhoeff B and Glas G (2010) The search for dysfunctions. A commentary on 'What is a mental/psychiatric disorder? From DSM-IV to DSM-V' by Stein et al. (2010). *Psychological Medicine* 40: 1787-1788.

Wakefield JC (1992) Disorder as harmful dysfunction: a conceptual critique of DSM-III-R's definition of mental disorder. *Psychological Review* 99: 232-247.

Young A (1995) *The Harmony of Illusions: Inventing Post-Traumatic Stress Disorder.* Princeton, NJ: Princeton University Press.

Zachar P (2002) The Practical Kinds Model as a Pragmatist Theory of Classification. *Philosophy, Psychiatry and Psychology* 9: 219-227.