Studies in History and Philosophy of Biological and Biomedical Sciences 60 (2016) 15–24

Contents lists available at ScienceDirect

Studies in History and Philosophy of Biological and Biomedical Sciences

journal homepage: www.elsevier.com/locate/shpsc

Diagnosis and causal explanation in psychiatry

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A R T I C L E I N F O

Article history:
Received 22 January 2016
Received in revised form
8 September 2016
Available online 20 September 2016

Keywords:
Psychiatry
Diagnosis
Causal explanation
Causal heterogeneity
Major depressive disorder

A B S T R A C T

In clinical medicine, a diagnosis can offer an explanation of a patient's symptoms by specifying the pathology that is causing them. Diagnoses in psychiatry are also sometimes presented in clinical texts as if they pick out pathological processes that cause sets of symptoms. However, current evidence suggests the possibility that many diagnostic categories in psychiatry are highly causally heterogeneous. For example, major depressive disorder may not be associated with a single type of underlying pathological process, but with a range of different causal pathways, each involving complex interactions of various biological, psychological, and social factors. This paper explores the implications of causal heterogeneity for whether psychiatric diagnoses can be said to serve causal explanatory roles in clinical practice. I argue that while they may fall short of picking out a specific cause of the patient's symptoms, they can nonetheless supply different sorts of clinically relevant causal information. In particular, I suggest that some psychiatric diagnoses provide negative information that rules out certain causes, some provide approximate or disjunctive information about the range of possible causal processes, and some provide causal information about the relations between the symptoms themselves.

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1. Introduction

When a patient presents to the clinic with a set of symptoms, one of the physician's tasks is to make a diagnosis that explains these symptoms. In somatic medicine, the diagnosis usually fulfils this explanatory role by indicating the cause of the patient's symptom presentation (Cournoyea & Kennedy, 2014; Schwartz & Elstein, 2008). For example, the diagnosis of myocardial infarction (MI) explains a patient's chest pain by indicating that the cause is ischaemic necrosis of the myocardium. This model of causal explanation suggests essentialism regarding disease kinds, whereby a diagnosis is taken to pick out a "disease entity" that can be a treated as a distinctive cause (Hucklenbroich, 2014). Moreover, this cause is taken to be invariant across cases, such as the diagnosis of MI referring to a causative pathology, ischaemic necrosis of the myocardium, which is instantiated by every case of MI.

The essentialistic thinking associated with this model of causal explanation continues to influence modern conceptions of psychiatric diagnoses (Haslam, 2014; Hyman, 2010). For example, the following passage from a psychiatric textbook characterises major depressive disorder (MDD) as a distinctive kind of disease that can cause the symptom of depression:

> Depression is more common in older people than it is in the general population. Various studies have reported prevalence rates ranging from 25 to almost 50 percent, although the percentage of these cases that are caused by major depressive disorder is uncertain. (Sadock & Sadock, 2008, p.215)

A popular health information website does so similarly with generalised anxiety disorder (GAD):

> GAD is a long-term condition that causes you to feel anxious about a wide range of situations and issues, rather than one specific event. (NHS Choices, 2016)

Similarly again, the following passage from a research paper on chronic fatigue syndrome (CFS) suggests that MDD refers to a distinctive disease that can explain fatigue symptoms:

> When a well-recognized underlying condition, such as primary depression, could explain the subject's symptoms, s/he was classified as having "CFS-explained". (Jason et al., 2014, p.43)

These sorts of characterisations are not surprising when we consider psychiatry's status as a medical discipline. As noted by Poland (2014, pp. 31–33), psychiatric practice occurs in a context shaped by medical roles and traditions. Hence, as in other medical

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http://dx.doi.org/10.1016/j.shpsc.2016.09.003
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disciplines, disorders in psychiatry are treated as distinctive disease kinds that can be invoked in causal explanations of patients' symptoms.

However, there are reasons to suspect that such essentialistic thinking may be misplaced in psychiatry. At present, it is unclear whether many of our current diagnostic categories actually do pick out stable and distinctive causes. Studies on the genetics and neurochemistry of psychiatric disorders have tended to indicate high degrees of heterogeneity, and while recent advances in cognitive neuroscience and functional neuroimaging have yielded compelling insights into the mechanisms involved in psychopathology, it is disputed whether they on their own could supply individual disease definitions (Hyman, 2010, p. 171). Therefore, we need to at least consider the possibility that many of the major psychiatric disorders could turn out to be causally heterogeneous at every level of analysis (Kendler, 2012; Murphy, 2006; Poland, von Eckhardt, & Spaulding, 1994).

If it does turn out to be the case that psychiatric diagnoses cannot be understood essentialistically, then this would cast doubt on whether such diagnoses genuinely are of any causal explanatory value in clinical practice. This is not only an epistemological problem for philosophers, but is relevant to psychiatric researchers and practitioners. First, it calls into question the validity of our current diagnostic classification in psychiatry. That is, if it turns out that our current diagnostic categories do not represent distinctive disease kinds, then it is questionable whether they can be used to support inductive inferences and formulate laws (Cooper, 2005). Second, as argued by Haslam, the essentialisation of psychiatric disorders can encourage harmful stigma "because it represents sufferers as categorically abnormal, immutably afflicted, and essentially different" (Haslam, 2014, p. 25). Hence, if it turns out that diagnostic categories in psychiatry do not correspond to distinctive causal essences, then there is a possibility that such essentialistic conceptions of psychiatric disorders not only mislead patients, but also harm them.

In light of these concerns, it is worth asking whether there are other ways to think about the explanatory roles of diagnoses that do not encourage problematic reification. This will be the focus of this paper. I shall argue that even in the case that psychiatric diagnoses turn out to be causally heterogeneous at every level of analysis, they can still provide information that is explanatorily valuable in the clinical setting. Moreover, while these other forms of explanation do not fit the standard model of causal explanation whereby a diagnosis specifies a distinctive cause, I shall show that they are nonetheless causal in satisfying ways.

The paper proceeds as follows. I begin in Section 2 by distinguishing two types of explanatory question, which are the explanation of a syndrome in general and the explanation of the clinical presentation of a particular patient with appeal to a diagnosis. Using MDD as a case study, I explore the potential challenges that psychiatric disorders pose for these explanatory questions. While philosophers of psychiatry have offered promising approaches to the first kind of explanation that handle the challenges of heterogeneity and multilevel complexity, these problems continue to affect the second kind of explanation. Nonetheless, I argue in Section 3 that even though psychiatric diagnoses may turn out not to pick out homogeneous causal essences, there are other ways in which they might offer causal explanatory information. I suggest that some can provide negative information that excludes certain causes, some can provide partial explanations involving possible causal processes, and some can provide information about the causal relations between the symptoms themselves.

It should be made clear from the outset that my intention is not to argue that MDD definitely is a heterogeneous phenomenon. Rather, it is to explore the philosophical implications for the explanatory role of the diagnosis if it were to turn out to be causally heterogeneous. I use MDD as a case study, because it typifies a scenario where, given our current incomplete understanding of the causal processes involved, there remains a real possibility that there is no single causal essence that defines the disorder. However, even if it were to turn out that MDD is associated with a stable causal structure, there are other psychiatric diagnoses that are likely to be causally heterogeneous, and so my analysis of causal explanation would still be applicable. For example, some researchers suggest schizophrenia (Wheeler & Voineskos, 2014) and bipolar disorder (Maletic & Raison, 2014) might turn out to be causally heterogeneous.

2. Challenges for explanation in psychiatry

2.1. Two explanatory questions

Throughout this section, I use the example of MDD to highlight some of the challenges facing explanation in psychiatry. Before I turn to the case study, it is important to distinguish two kinds of explanatory question regarding diagnoses in medicine (Thagard, 1999, p. 20). The first kind, which I henceforth call "disease explanation", belongs to medical research. This is the explanation of a clinical syndrome in general. The goal here is to develop a general model that brings together the relevant causal factors and mechanisms responsible for the syndrome. For example, the disorder characterised by swollen limbs and bleeding gums known as scurvy is explained by defective collagen synthesis due to ascorbic acid deficiency (Thagard, 1999, pp. 120–122). The second kind of explanation, which I henceforth call "diagnostic explanation", occurs in the context of clinical practice. This is where a patient presents with such and such symptoms, and the physician makes a diagnosis that explains these symptoms. Take the example mentioned in Section 1 of patient's chest pain being explained by the diagnosis of MI. Here, the *explanandum* is not the clinical syndrome in general, but the clinical presentation of the particular patient.

These two explanatory questions are connected. In diagnostic explanation, where a diagnosis is invoked to explain a patient's symptoms, the understanding of the disorder picked out by the diagnosis comes from the general model that is constructed through disease explanation. For example, disease explanation informs us that MI in general involves rupture of an atherosclerotic plaque and thrombus formation leading to occlusion of a coronary artery and ischaemic necrosis of the myocardium, and it is in virtue of this knowledge that the diagnosis of MI functions as a causal explanation of the occurrence of chest pain in a particular patient. Hence, what the general model of a disorder looks like has implications for the explanatory function of the diagnosis in the particular case.

Much of the philosophical literature on explanation in psychiatry has focused on disease explanation, rather than diagnostic explanation. Theorists have expressed concerns that high degrees of heterogeneity and complexity could present significant challenges for developing comprehensive models of many major psychiatric disorders (Hyman, 2010; Murphy, 2006; Poland, 2014). However, as we shall see, this also has implications for diagnostic explanation. Note that it is not so much the heterogeneity of symptoms that is the problem, as many medical disorders that have been successfully modelled can present in several different ways. For example, syphilis has protean manifestations, which can include ulceration, gastric dysmotility, cardiac disease, and paresis, but these many different manifestations are unified by a singular cause, namely *Treponema pallidum* infection. Rather, the concerns

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