Avoiding False Positives: Zones of Rarity, the Threshold Problem, and the DSM Clinical Significance Criterion

Rachel V Cooper, PhD (History and Philosophy of Science)

False positives arise when people without disorders are diagnosed as having disorders. Various approaches for avoiding false positives have been suggested. This review critically assesses the roles of zones of rarity, the threshold problem (the problem of determining the boundary of disorder in cases that shade into normality), and the Diagnostic and Statistical Manual of Mental Disorders (DSM) criterion that requires that a disorder cause clinically significant impairment or distress (the harm criterion). The lack of zones of rarity in much of psychiatry gives rise to the threshold problem. The DSM harm criterion is frequently presented as offering a solution to the threshold problem. However, I argue that the harm criterion cannot offer a general solution to the threshold problem, as harm is not always correlated with the intensity and frequency of symptoms. Still, the harm criterion is essential to ensure that people who are merely different are not diagnosed as having a disorder. The threshold problem can be addressed by selecting symptom-based cut-off points to distinguish between disorder and normality. These cut-off points are frequently arbitrary in the sense that they often reflect no natural division between disorder and normal, but they may be more or less wisely chosen. Where possible, the thresholds should be set so that the advantages of diagnosis can be expected to outweigh the disadvantages.

Éviter les faux positifs : les zones de rareté, le problème du seuil, et le critère de significativité clinique du DSM

Les faux positifs surviennent lorsque les personnes exemptes de troubles se font diagnostiquer des troubles. Diverses approches ont été suggérées pour éviter les faux positifs. Cette revue évalue de façon critique les rôles des zones de rareté, du problème du seuil (le problème de déterminer la limite du trouble dans les cas qui se confondent avec la normalité), et le critère du Manuel diagnostique et statistique des troubles mentaux (DSM) qui demande qu'un trouble cause une incapacité ou une détresse cliniquement significative (le critère des dommages). L'absence de zones de rareté dans une grande partie de la psychiatrie donne lieu au problème du seuil. Le critère des dommages du DSM est fréquemment présenté comme offrant une solution au problème du seuil. Toutefois, je fais valoir que le critère des dommages ne peut offrir une solution générale au problème du seuil, car les dommages ne sont pas toujours corrélés à l'intensité et à la fréquence des symptômes. Tout de même, le critère des dommages est essentiel pour faire en sorte que les personnes qui sont simplement différentes ne se fassent pas diagnostiquer un trouble. Le problème du seuil peut être traité en sélectionnant des points d'inclusion basés sur les symptômes pour distinguer entre trouble et normalité. Ces points d'inclusion sont souvent arbitraires au sens où ils ne reflètent souvent aucune division naturelle entre trouble et normalité, mais qu'ils peuvent être choisis avec plus ou moins de discernement. Autant que possible, les seuils devraient être déterminés de manière à ce que les avantages du diagnostic puissent l'emporter sur les désavantages.
False positives occur when nondisorder is mistaken for disorder. False positives lead to people without disorders being unnecessarily exposed to the costs of psychiatric diagnosis (for example, drug side effects and stigma) and to inflated prevalence rates that distort health care planning. Classic examples show that false positives arise for different reasons. In the case of homosexuality, mere difference was mistaken for disorder. Going further back, in the medicalization of runaway slaves, a normal reaction to oppression was mistaken for individual pathology. Currently, many worry when conditions that shade into everyday difficulties are diagnosed, for example, at the boundary between shyness and social anxiety disorder. A key claim of this paper is that as false positives arise for different reasons no one solution will suffice to rule them out; a battery of approaches to avoiding false positives will be required.

The Role of Zones of Rarity

The idea that zones of rarity should separate valid disorders from other disorders and normality has a long history, championed most notably by Robert Kendell (with Jablensky). If one imagines plotting cases of disorder in multidimensional property space (as in cluster analysis), zones of rarity occur whenever sparsely populated space separates clusters. When classificatory boundaries align with zones of rarity then nature is cut at its joints, and the informational content of categories is maximized. If zones of rarity could be identified between disorders and normality then one may hope this could help avoid false positives. Some marker may be identified that securely located a case to either the disorder or the normality side of the zone. The major concern for people adopting this picture is the seeming scarcity of zones of rarity within psychopathology. This triggers calls for more research in the hope of locating zones, or leads to resignation that psychiatry must resort to a dimensional system.

However, I suggest that locating zones of rarity is less important than often thought. Characterizing successful classification as cutting nature at its joints leads one to suppose that cuts should be made at zones of rarity. However, the butchery metaphor is frequently inappropriate. In many domains there are either no joints or too many. The philosopher John Dupré suggests that when thinking about the relation between classification and the world we should be realists, but “promiscuous realists.” Typically, when a domain is plotted in multidimensional space, the pattern of similarities is complex. Out of this complexity, one can extract different classifications that may be useful for different purposes. Rather than thinking in terms of cutting nature at its joints, a better metaphor for thinking about the way in which empirical evidence can guide classification is to think of a mountaineer picking a route. The path chosen is informed by the terrain, but rarely determined by it. Different walkers will prefer different paths; those looking for an easy ascent may pick the shallowly rising ridge, those looking for excitement may opt for the rocky gullies, and those who have forgotten sunscreen may stick to the shade.

Often a domain will either contain no zones of rarity, or at least none that are appropriate for marking the distinction between the normal and the pathological. Kendell and Jablensky claim that disorders defined based on syndromes can only be considered valid if separated from other disorders and normality by zones of rarity. Disorders that shade into normality are merely “arbitrary loci in a multidimensional space.” While such categories may have clinical utility, and enable predictions to be made regarding etiology, prognosis, and so on, as the boundaries of such disorders are chosen as opposed to discovered, they are vulnerable to replacement by different successor classifications. For this reason, Kendell and Jablensky see “utility” as second best to “validity.”

I suggest it is a mistake to think that classifications of domains that lack zones of rarity must be second rate. Many respectable scientific classifications describe continuous domains. Alloys provide a good example; knowledge of the composition of an alloy allows accurate predictions regarding a sample’s behaviour. Similarly, if mental disorders vary continuously, this need not limit the prospects for scientific progress. Kendell and Jablensky are correct that divisions imposed on a continuous domain could have been differently chosen, but they are wrong in thinking that alternative possible classifications undermine the classification that is adopted. Kendell and Jablensky view alternative classifications as competing in a zero-sum game; if one classification is valid, then alternatives must be invalid. From the promiscuous realist account, this is a mistake, and alternative classifications can be equally valid in the sense that they provide equally good mappings of different features of reality.

That said, the absence of zones of rarity does leave us with the question of where the threshold should be drawn between normality and disorder. The approach taken by DSM-IV is

Highlights

- The DSM criterion that requires that a disorder cause clinically significant impairment or distress (the harm criterion) is required to prevent people who are merely different from being diagnosed as having a disorder.
- The absence of zones of rarity between disorders and normality gives rise to the threshold problem (the problem of determining the boundary of disorder in conditions that shade into normality).
- The threshold problem should be addressed by selecting symptom-based cut-off points to distinguish disorder from normal.

Abbreviations

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<tr>
<th>Abbreviation</th>
<th>Definition</th>
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<tr>
<td>DSM</td>
<td>Diagnostic and Statistical Manual of Mental Disorders</td>
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<td>IDD</td>
<td>intellectual disability disorder</td>
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<td>SAD</td>
<td>social anxiety disorder</td>
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to rely on judgments that the syndrome is severe enough to cause clinically significant distress or impairment. The impact of this criterion and whether it can be used to deal with the threshold problem will now be considered.

The DSM Criterion That Disorders Cause Clinically Significant Distress or Impairment

In DSM-IV, both the introductory definition of mental disorder and many of the diagnostic criteria sets contain a clinical significance criterion. The wording varies, but generally symptoms must cause “clinically significant distress or impairment in social, occupational, or other important areas of functioning.” Let us call this the harm criterion. The DSM-IV introduction explains,

This criterion helps establish the threshold for the diagnosis of a disorder in those situations in which the symptomatic presentation by itself (particularly in its milder forms) is not inherently pathological ...

Revisions made in DSM-5 threaten this criterion. The new definition of mental disorder does not require that all disorders cause harm, but rather notes that “Mental disorders are usually associated with significant distress or disability in social, occupational, or other important activities.” Let us call this the harm criterion. The DSM-IV introduction explains,

This criterion helps establish the threshold for the diagnosis of a disorder in those situations in which the symptomatic presentation by itself (particularly in its milder forms) is not inherently pathological ...

Harm has ceased to be necessary for a diagnosis of at least some disorders.

I will argue that the harm criterion is essential for avoiding false positives and should have been retained for all disorders. However, I suggest that DSM-IV makes a mistake in presenting the harm criterion as a general solution to the threshold problem. False positives occur for different reasons. Some occur when harmless conditions are mistaken for disorders, some when errors are made in setting the threshold between normality and disorder, and some for other reasons. In general, false positives that arise in different ways must be dealt with differently. The harm criterion is needed to ensure that harmless conditions are not diagnosed as disorders, but I will show that it cannot provide a general solution to the threshold problem.

To see why the harm criterion is crucial to prevent harmless difference being diagnosed as disorder, let us remember how it came to be introduced. After a period of controversy, homosexuality was removed from DSM in 1973. Evidence that was influential in the debates included data showing that homosexuality is far more common than previously thought, that homosexuality is not linked with indicators of psychopathology, and that many homosexual people are happy and high functioning. Robert Spitzer played a key role in the debates and found defining disorder useful in defending his stance on homosexuality.

Spitzer’s definition of mental disorder requires that patients suffer distress or disability to be considered to have a disorder. As homosexuality need not be linked with distress or disability, homosexuality, per se, cannot be a disorder under Spitzer’s definition. Eventually a version of Spitzer’s definition came to be included in the introduction of DSM-III, and the DSM-IV definition is a direct descendent. The idea that disorders must be harmful has since been influentially defended by Jerome Wakefield, who claims that disorders are harmful dysfunctions, and is also accepted by many others.

The case of homosexuality illustrates why the harm criterion is essential. Homosexuality is statistically unusual, it may occur as a result of some evolutionary dysfunction, and yet, as it causes no harm, we do not want to consider it a disorder. In keeping with such thinking, the DSM-IV approach allows that people who are psychologically different, but who are not harmed by their difference, should not be given a diagnosis. The new DSM-5 definition of mental disorder is less clear on this issue.

Whether a condition causes harm is only sometimes determined by the frequency and intensity of symptoms (note that by symptoms here I do not mean to imply pathology). There are conditions, most obviously depression, where symptoms are intrinsically unpleasant and, in such cases, the degree of harm will plausibly be correlated with symptom severity. In these conditions, measures of either harm or symptom severity may equally well be used to distinguish normality from disorder. However, the symptoms that characterize some other conditions only sometimes cause harm, even when symptoms are intense and frequent. Consider those associated with Asperger disorder. Whether these cause problems depends on multiple factors. Symptom severity plays a role, but so, too, does the material and social environment; for many people with Asperger disorder, the increased availability of computer-based means of communication has facilitated social interaction.

A person’s own likes, interests, and ambitions are also important; a degree of social awkwardness may be career-destroying for an aspiring social worker but acceptable in an engineer.

As there need be no linear relation between symptom severity and degree of harm, the harm criterion does not offer a general solution to the threshold problem (that is, the problem of determining whether symptoms are severe enough to count as disorder). The harm criterion primarily does something different—it prevents the diagnosis of people who are different but who are not harmed by their difference, and these people may not be those whose symptoms are only mild. Note that while harm is necessary for disorder, it is by no means sufficient—no harmless states should be classified as disorders, but not all harmful states are disorders (some are normal but unpleasant states, some are nonmedical problems).

Although their earlier work was influential in securing the place of the harm criterion in DSM-IV, Spitzer and Wakefield have recently been critical of its continuing use. To defend the harm criterion, I will consider and rebut each of their objections.

First, Spitzer and Wakefield hold that the harm criterion is frequently redundant. I admit that the criterion does more work in some contexts than others. As Spitzer and
Wakefield note, the harm criterion often plays little role in clinical practice. However, plausibly this is because most mental health professionals see only patients who believe they need help, or who are perceived by other lay people (for example, the police) to have a problem. This initial screening helps ensure that clinicians see people who are likely to be experiencing symptoms that cause harm. The situation is quite different when DSM is used outside the clinical context. Prevalence rates gained via screening community populations plausibly include many false positives. These arise for various reasons, but some are people who have so-called symptoms but are not bothered by them. In evaluating the importance of the harm criterion, it should also be remembered that DSM plays a cultural role in shaping lay perceptions of disorder and normality. Within the community, there are many people who are physically or psychologically different and yet have flourishing lives. The inclusion of the harm criterion in DSM-IV did real work in helping to legitimate people’s claims that they are merely different and not disordered. In all contexts, the extent to which the harm criterion can help avoid false positives will vary with diagnosis. The effect on diagnoses such as depression is minimal, but the symptoms of depression are intrinsically unpleasant. However, there are many other conditions, including Asperger disorder, transsexualism, attention-deficit hyperactivity disorder, Tourette syndrome, and hypoactive sexual desire disorder, where symptoms may or may not cause problems.

Second, Spitzer and Wakefield suggest that the harm criterion causes false negatives. They offer examples of patients who they think should be considered disordered, but who cannot be diagnosed if the harm criterion are enforced. These range from a high-functioning drug user to a man with erectile dysfunction who is not distressed by his impotence. I deny these are false negatives. High-functioning people who voluntarily take drugs are not disordered (though they may be considered to be foolishly risking their health). Harmless impotence should be understood as a mere difference rather than a disorder. This may rankle, as impotence is plausibly an evolutionary dysfunction, but in so far as it causes no harm, it should not be considered a disorder; to say anything else here raises issues of consistency with the homosexuality case (why would it be okay to be gay, but not to be impotent?).

Third, Spitzer and Wakefield, and other critics, worry that the harm criterion is hard to operationalize. This worry is partly powered by a misunderstanding of the work of the harm criterion. If one assumes that the harm criterion deals only with false positives produced by the threshold problem, then it is easy to think that it could be replaced by more specific guidance regarding required symptoms. For example, instead of requiring that, say, hypoactive sexual desire causes harm, one may precisely specify the symptoms required (for example, having been uninterested in sex for such and such a period). However, as I have argued, false positives occur for various reasons. Not all are related to problems with thresholds. Some people are highly symptomatic but are not harmed by their symptoms. The harm criterion is crucial to ensure that such people are regarded as merely different rather than disordered.

Given that the harm criterion is required, how might it be operationalized? It is easy to get bogged down by the apparent difficulties. Of course, there are deep philosophical problems with determining the nature of harm, or its converse, the nature of the good life. These are real issues, but it is also important not to focus on difficulties to the extent that one overlooks how many judgments can be made easily and with very widespread agreement. Within psychiatry, the key questions do not concern what sorts of lives are the very best, but which are good enough, and often judgments of this sort are easy to make. There is very broad agreement about the components of a good enough life, which will include the following: freedom from persistent unpleasant experiences (pain, panic, and so on); being able to engage in those activities that are essential for self-maintenance, such as washing and cooking; having some friends; and being able to engage in some sorts of meaningful activity (which may be a job or a hobby). Such notions can be operationalized via one or more of the many existing scales that seek to measure quality of life or impairment.

An alternative approach is suggested when one remembers that false positives are plausibly more frequent in community prevalence studies than in the clinic. People who are not harmed by their symptoms neither refer themselves for treatment nor are they sent by concerned others. This suggests the second possibility for operationalizing the harm criterion—one simply asks respondents whether they have sought or would like treatment. Of course, some people lack insight and believe themselves to have acceptable lives when they do not. In an attempt to deal with such cases, one might either automatically consider people with certain symptoms (for example, maybe delusions) to be in need of care, or also ask the respondents’ friends or relatives whether they think the person needs help. Something like this approach was taken in the National Institute of Mental Health Epidemiologic Catchment Area Program and the National Comorbidity Study, which asked respondents about health care usage (and also the degree to which symptoms interfere with their lives).

I am hopeful that an acceptable method, or methods, for operationalizing the harm criterion will be found. Still, even if it turns out that the harm criterion cannot be neatly operationalized, it is still required. This conclusion may seem more palatable if it is remembered that psychiatric diagnosis already and essentially relies on numerous notions that cannot be cleanly operationalized, for example, whether a response is culturally expectable or is due to social conflict, or whether a delusion is bizarre.

Returning to the Threshold Problem

I have argued that the harm criterion is essential but that it cannot deal with the threshold problem; as in many conditions, whether symptoms cause harm will not be
threshold problem be dealt with? The only solution is to pick some objectively specifiable but ultimately arbitrary cut-off point. In the same sort of way that the cut-off point for high blood pressure is chosen to be a particular figure, the cut-off point between having drinking and alcoholism may be taken to depend on a certain number of units of alcohol being consumed during a certain number of days. For a disorder to be diagnosed, it would both be necessary that it cause harm and also that the symptoms meet the cut-off point for symptom severity.

An example of a diagnosis where the approach I am suggesting has long been accepted is IDD (known as mental retardation in DSM-IV). Diagnosis requires both an IQ score of less than about 70 and for there to be problems in everyday living. Here the IQ score deals with the threshold problem, and the requirement that there be problems in everyday living is a version of the harm criterion. Both are required. Some people with IQs of much less than 70 have flourishing lives. The harm criterion ensures that such people do not receive a diagnosis. Some people with IQs far above 70 face problems in living caused by their lack of intelligence, but the cut-off point ensures that such people are considered to be normal but unintelligent rather than disordered. The example of IDD also makes it clear why a universal cut-off point is required for the threshold, rather than it being acceptable to adopt an individualized approach and to claim that people should be considered disordered whenever their symptoms are severe enough to cause them harm. People who are diagnosed are typically eligible for various services and benefits. For these to be distributed fairly, a universal cut-off point is required.

To take another example, consider the difference between shyness and SAD. Both shy people and those with SAD find social interaction problematic and seek to avoid it. For a diagnosis of SAD, both the harm criterion and criteria relating to threshold must be satisfied. Some people may be very shy, but experience no harm (maybe, for example, they are members of a contemplative religious community). The harm criterion acts to ensure that such people are merely different but not intelligent rather than disordered. The example of IDD also makes it clear why a universal cut-off point is required for the threshold, rather than it being acceptable to adopt an individualized approach and to claim that people should be considered disordered whenever their symptoms are severe enough to cause them harm. People who are diagnosed are typically eligible for various services and benefits. For these to be distributed fairly, a universal cut-off point is required.

Deciding the exact cut-off point for the threshold is often an arbitrary decision, in the sense that frequently there are no natural divisions to be mapped. Still, a choice may be more or less wise. Where possible, the threshold should be set such that the benefits of diagnosis (which may include benefits that accrue from treatment that aims to reduce the risk of future harm) will usually outweigh the disadvantages. Many of the controversies that emerged during the period of proposed changes to DSM-5 can be seen as being rooted in worries that cut-off points for some diagnoses may have been selected unwise, such that people who could more profitably be considered normal would be considered disordered. Examples include psychosis risk syndrome,22 mixed anxiety depression,23 and pedophilic disorder, hebephilic type (arguably adults who have sex with teenagers are more appropriately considered normal criminals than disordered).24 The best way to deal with such controversies is via detailed cost-benefit analyses of the expected consequences of altering the previously accepted thresholds.22,23

Conclusions
I have argued that the harm criterion is essential to prevent people who are merely different being diagnosed as having a disorder. The harm criterion cannot deal with the threshold problem because harm is not always linearly related to symptom severity or other measures of disorderedness. Rather the threshold problem must be dealt with by selecting a symptom-based cut-off point. The cut-off point is arbitrary, in the sense that it will map no natural division between the disordered and normal, but it may be more or less wisely chosen. Where possible, the threshold should be set so that the advantages of diagnosis generally outweigh the disadvantages. Finally, it should be borne in mind that requiring harm, and requiring that symptom thresholds are met, will not be enough, on its own, to prevent false positives. False positives also arise for reasons that are not dealt with here (for example, see Dr Wakefield’s work on the importance of requiring dysfunction25 and Horwitz and Wakefield26). For someone to be considered as having a disorder, all the various criteria that act to rule out false positives should be met concurrently.

If the approach championed here had been adopted then DSM-5 would have included a general requirement that disorder can only be diagnosed when a condition results in harm. Some researchers would prefer to conduct research without considering harm. For such purposes, something like the proposed distinction between paraphilias and paraphilic disorders (where a paraphilia is simply the unusual pattern of sexual arousal, and a paraphilic disorder is diagnosed only on the basis of the unusual pattern of sexual arousal plus harm) might have been introduced across the whole classification.

Acknowledgements
I thank Dr Wakefield and Dr First who commented on a draft version of this paper. This paper was also presented at the 2012 Philosophy of Psychiatry Work in Progress Workshop held at Lancaster University and I am grateful for the comments of those present.

The Canadian Psychiatric Association proudly supports the In Review series by providing an honorarium to the authors.

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