

Uncertainty and the inconvenient facts of diagnosis



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ABSTRACT

One common contemporary usage of the term “diagnostic uncertainty” is to refer to cases for which a diagnosis is not, or cannot, be applied to the presenting case. This is a paradoxical usage, as the absence of diagnosis is often as close to a certainty as can be a human judgement. What makes this sociologically interesting is that it represents an “epistemic defence,” or a means of accounting for a failure of medicine’s explanatory system. This system is based on diagnosis, or the classification of individual complaints into recognizable diagnostic categories. Diagnosis is pivotal to medicine’s epistemic setting, for it purports to explain illness via diagnosis, and yet is not always able to do so. This essay reviews this paradoxical use, and juxtaposes it to historical explanations for non-diagnosable illnesses. It demonstrates how representing non-diagnosis as uncertainty protects the epistemic setting by positioning the failure to locate a diagnosis in the individual, rather than in the medical paradigm.

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Introduction

In *On the Heels of Ignorance* (2019), Owen Whooley concludes his analysis of how psychiatry has dealt with its ignorance over time by challenging social scientists to explore and reveal how other groups “manage and mitigate the effects of ignorance” (p. 200). He believes that it is only by shedding light on how groups deal with the failures of their systems, and the limitations of their knowledge, that we can truly understand their paradigms, practices, and politics. He ponders why the broader public has been so ready to accept psychiatry and its myriad reinventions, even when each subsequent reformulation, from the asylum to psychobiology, psychoanalysis, and diagnostic psychiatry is but an admission of its limitations, a kind of apology for the previous paradigm.

As a practicing clinician in a clinical school, I have become aware of an intriguing exemplar of ignorance management in professional publications written by, and directed at—in contrast with Whooley’s practitioners of psychiatric medicine—doctors of physical medicine. The term “diagnostic uncertainty” is becoming increasingly used to describe a failure of medicine’s diagnostic paradigm: that is to say, when a condition cannot be diagnosed within a diagnostic classification system. In the present article, I undertake an exposé in line with Whooley’s call. Rather than, however, focussing on general ignorance *per se* I focus on the individual cases that cannot be explained within medicine’s paradigm. I explore how the notion of uncertainty is implemented

to protect medicine’s diagnostic underpinning when it appears to be inadequate.

When a patient presents a complaint to the medical system, it is usually in pursuit of a diagnosis. This is because diagnosis can be key to explaining the complaint, providing therapy, and describing the future implications (prognosis) for the ailment.

All diagnoses are not equal in their explanatory potential. A patient may receive a “pathological diagnosis,” that is to say, one which explains the mechanism which has made the patient ill. For example, “myocardial infarction” is a pathological diagnosis which explains a symptom of heartburn or chest pain by the narrowing of the coronary arteries resulting in cardiac tissue necrosis. When a recognizable explanation cannot be found for the ailment, a patient may receive instead a “symptomatic” diagnosis. The chief complaint becomes the diagnosis; “back pain” and “chest pain” are classic examples. They do not explain, as the pathological diagnosis does, rather, they describe. Or, the patient may be offered the diagnosis of “medically unexplained symptoms” a kind of wastebasket diagnosis that explains what medicine cannot explain with a new diagnostic label. This label is superficially inoffensive, but it has been demonstrated to be used synonymously with, and increasingly as a substitute for, psychosomatic disorders, placing the explanation for symptoms in the patient’s psyche (Jutel, 2010). I will demonstrate how, in addition to these diagnostic forms, the concept of uncertainty has inserted itself into the discussion of the complaints without pathological explanations, and then discuss the meaning of this discursive construction.

To be clear, diagnosis is not an easy process, and there are situations where, to arrive at a diagnosis, the diagnostician is justifiably uncertain and must weigh different diagnostic options

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to decide which one, among a range, fits the case. There are many reviews of the challenges of differentiating between for example, dissimilar tumours of the parotid gland (Castelijns & Leemans, 2009), idiopathic aplastic anaemia and low-risk myelodysplastic syndrome (Nakao & Gale, 2016), Lyme disease and neuropsychiatric disorders (Koster & Garro, 2018), and many more.

The process of diagnosis requires temporary uncertainty because a decision between options is part of the diagnostic task; by the time a decision is made, the uncertainty is quelled. The focus of this current article, however, is on the cases where the *absence* of diagnosis is represented by the word “uncertainty.” It is important to differentiate between these uses of the term because they are so fundamentally dissimilar. One describes transient confusion that accompanies the decision-making process; the other, however, is an at-least-for-the-moment *certainty* of no available diagnosis. The latter, paradoxical use, is illuminating in relation to the revelation of ignorance management.

I therefore focus on “diagnostic uncertainty” as a salient way in which physical medicine deals justifies its shortcomings. I will demonstrate how today it discursively brushes off complex and debilitating conditions which it cannot explain. I will present this review against a backdrop of an historical analysis that describes different ways of representing these same shortcomings and develops an argument to explain the difference. But, ultimately, I will show how the usage is actually an *epistemic defence*, that is to say, one way of accounting for the limitations of the medical paradigm to explain all problems of health and illness, by transferring the responsibility for non-diagnosis to a cognitive domain.

Diagnosis and the profession

The absence of diagnosis is a problematic situation for a field that defines itself by its diagnoses. If, as Andrew Abbott, offers in his *The System of Professions* (1988), the “task of professions are human problems amenable to expert systems” (p. 87), then understanding how diagnosis provides access to those systems is pivotal. He explains that key to the jurisdictional claims of a profession is the ability to classify, reason, and take action about a given problem—vital to the establishment and maintenance of a profession and defining its purview (p. 94).

Diagnosis is medicine’s primary classification tool (Jutel, 2011a). It determines treatment and prognosis, allocates resources, differentiates lay from professional, and provides a hierarchy of authority within the professions. Diagnosis enables evidence-based practice, epidemiology, and gives access to the sick role. It is a fundamental component of medicine’s episteme, as it determines what counts as disease, the “truth” of medicine. Yet, today, as in yesteryear, there are illnesses for which a diagnosis cannot be provided.¹ Such non-diagnosable ailments require an epistemic defence (“mitigation of ignorance”) because medicine’s epistemic setting is anchored by diagnosis. An epistemic setting is a field within which there are parameters for what constitutes knowledge, and what counts as acceptable information, practice or protocols (see Whooley, 2013).

Medical practice is based on identifying the general in the individual: how does the particular clinical profile (i.e., the presenting symptoms) fit into a more generic disease framework (diagnosis)? All actions in relation to the presenting symptoms emerge from that framework; the diagnosis provides at once an

explanation, a treatment, and an idea of what the outcomes may be (prognosis). It makes order out of a picture of disarray, which in itself, gives authority to medicine, a powerful profession indeed, to be able to provide such structure (Jutel, 2011a, 2011b).

A contemporary reader might be hard pressed to imagine a medicine without diagnosis, but nineteenth and early twentieth century medical doctors battled to promote the clinical function and social importance of the diagnostic process they espoused. Doctors identified scientific diagnosis and its study as what distinguished them from non-scientifically qualified practitioners over whom they were struggling to gain supremacy. Diagnosis protected their jurisdictional boundaries. More than just the process of naming an ailment, it was a rational process that doctors felt they alone truly mastered. Diagnostic skill was lauded as the mark of a doctor’s success (Cathell, 1885).

While other professional categories may have claimed to diagnose, medical doctors held non-medical (“quack”) diagnosis in contempt. The Berlin special correspondent to the *British Medical Journal* wrote dismissively of “. . . a quack who is in the habit of diagnosing his patients’ diseases from the stockings they have worn” (Berlin Correspondent, 1897, p. 171). And, in his 1926 lecture, entitled *Doctors and the Public*, Dr E. Graham Little emphasised the danger in allowing “unqualified and ill trained persons to undertake the diagnosis of disease” (Little, 1926, p. 9). Little found popular support for his belief in *The Times* where an unnamed author wrote: “. . . [T]he fatal weakness of the unqualified practitioner is not his lack of a medical degree, but his lack of knowledge of disease which, however imperfectly, enables a qualified man to interpret the signs presented by this patient, to distinguish one sign from another, and to relate the signs to prognosis and treatment. Diagnosis in short must precede therapy” (in Little, p. 8).

Yet, even within the rational, scientific epistemic system of medicine, dependent upon diagnosis, there were, and still are, many cases which do not fit within the diagnostic parameters set by the profession, where generalization could not then, and cannot today, take place. These cases are what Whooley would call the “inconvenient facts” of the medical episteme. They are the inconsistencies of an epistemic setting which have to be dealt with by its proponents, either by ignoring them, treating them as problematic or setting them aside (Whooley, 2013, p. 43).

In the paragraphs which follow, I draw on a wide range of publications by and for doctors that discuss non-diagnosis. These publications include textbooks for young doctors, professional discussions in peer reviewed journals, editorials, and original research which communicate about the inconvenient facts of non-diagnosis. I will start by reviewing historical writings about cases that were identified as non-diagnosable, as well as by exploring the use made of the term uncertainty in previous eras. Even though my interest is ultimately in contemporary practices, I take this step backwards, to draw out both contrasts and similarities. It is often difficult to find the value-content in contemporary expressions and practices, as it requires us to remove ourselves from our own context whose values we take for granted. The critical distance of history underscores difference (Martin, 1992). But at the same time, it also provides us with a way of seeing persistent social patterns related to institutions, norms, and cultural values (Zerubavel, 2007). My first intent in providing the historical counter example is to underscore difference: demonstrate other ways of discussing non-diagnosis.

However, as the conclusion will affirm, regardless of the era, medicine has an epistemological defence to mount if it is to remain the social arbiter of health and disease. Having established its claim to professional authority via diagnosis, it must convince a now diagnosis-expectant public that medicine retains its expertise even when a diagnosis is not possible.

¹ Here, I distinguish illness from disease, the former being the patient’s sense of unwellness, and the latter the officially recognised category which accounts for the patient’s complaints.

“The laws of medicine too undecided still”

I chose as the historical period the late nineteenth to twentieth century, a time period which Light (2004) refers to as the “formative period of the American health care system.” It is after the establishment of both British and American Medical Associations, but well-distanced from the contemporary era of evidence-based practice. What makes that particular era interesting is that it was a time during which medicine was committed to promoting its epistemic superiority against the “ruinous competition” of myriad competing alternative healers and dispensaries (Warbasse, 1912, in Light, 2004). It provides examples of “epistemic defence” or accounting for its failings.

My goal in this historical exploration is to establish the general patterns used by doctors writing about diagnosis in this era to describe, justify, explain, and discursively construct the non-diagnosable ailment. Using standard historical methods, I explored specialized virtual and physical historical collections and archives, focussing on publications and papers pertaining to the difficulties of diagnosis. I started with shelf searching at the Wellcome Library, perusing books classified under the shelfmark WB141 (General Diagnosis). These tomes were mainly technical guidebooks explaining diagnostic techniques, introducing how to examine a patient, and derive a diagnosis from that examination. I scanned the shelves from 1880 to 1920 and found, usually in the introductory chapters, discussion in some of these books, about diseases that could not be diagnosed. The terms used included “puzzling,” “unknown,” and “undiagnosable” diseases. I used these terms and their variants, then extended this search to the digital indexes of Wellcome, the Medical Heritage Library, the *New England Journal of Medicine*, and the *Lancet*. Curious about the absence of reference to uncertainty, I also searched for “*diagnostic uncertainty*” as well as *diagnosis AND uncertainty* in these same digital databases, but this search did not reveal information about non-diagnosis. I repeated this search at the Philadelphia College of Physicians Library, and I extended my search to the archives, i.e., among medical notes and physician day books. Specialist archivists and history of medicine librarians supported my searches and suggested further material. While I focussed predominantly on the late nineteenth, and early twentieth century, some earlier references were also available and I included them in my material.

Many doctors of the last century (and the one before that), even while profoundly committed to diagnostic medicine, saw the absence of diagnosis not as a problem of diagnosis *per se*, rather of the still developing state of medical knowledge. Even though scientific medicine had become the dominant way of understanding health and disease, and the need for medicine to justify its ascendancy was on the decline, there were still visible flaws in its explanatory paradigm for which it had to account. Doctors who wrote about non-diagnosis acknowledged the youth of medicine as a practice, and the need to continually garner more information to support its development. In 1844, still early days for scientific medicine, Dr Henry Patterson explained to graduates of the Medical Department of Pennsylvania College, “The laws of medicine are too undecided still to be susceptible of a perfect codification” (Patterson, 1844, p. 11). He chided those who tried too hard to fit everything into diagnostic terms.

The term “uncertainty” surfaced, not in relation to decision making, but rather in reference to medicine’s systems of classification. Uncertainty referred, not to indecision, but to the solidity of the classifying structure. In front of a class of graduating doctors, the president of the American Medical Association, Dr A. H. Stevens, explained that medicine was “. . . less uncertain than law, and settled as much as theology.” He argued that certainty was not within the doctor’s purview because “The Omniscient only, without effort and intuitively, knows all things,” and that the

unsettled nature of medical knowledge was important to its prestige, explaining, “If medicine were a perfect science, it would cease to be progressive; its votaries would become mere artisans; the excitement of hopeful labour would become lost in certainty” (Stevens, 1847, p. 6).

The belief that medicine’s explanatory systems still needed refinement was carried well into the nineteenth century by doctors such as Lyle Motley, of Nashville, Tennessee, who lamented that “that there are conditions undiagnosable by the means at present at our command.” He did not lay guilt upon the individual doctor for his inability to diagnose. “If the physician has exhausted every means of examination,” he wrote, “and has carefully studied all the phenomena presented, and then fails to make a diagnosis, he has done his full duty” (Motley, 1923).

But at the same time, individual doctors were held responsible for helping to elaborate the *diagnostic system* upon which their profession resided. They were urged to become research workers, whose observations of even the individual case could contribute to reinforcing medicine’s system. Paradoxically, the success of medicine was a barrier to its full realisation, as “the earning of his [the general practitioner’s] livelihood occupied almost every moment of his time,” preventing the doctor from engaging in systematic research the likes of which would result in the establishment of the “laws that govern [the production of symptoms] and the physiological processes that underlie them” (Paton, 1924, pp. 1–2). The St. Andrews Institute for Clinical Research was founded in Scotland by the cardiologist Sir James Mackenzie specifically to enable research on diagnosis by “. . . the men in contact with the facts” of disease, in other words, the general practitioners.² Its goal was to “lead eventually, through the discovery of the laws governing its phenomena, to the elevation of medicine from an empiric art to its true place as an applied science” (Mackenzie, 1922, p. 8). In their first biennial report (of only three) they actually reported that only ten percent of consultations produced diagnoses (Macnaughton, 2002, p. 567, n. 94). With such dismal outcomes, Mackenzie (1920, p. 156) exhorted, “there is an urgent need for some method in the investigation of disease, different from that which has been pursued in the past.”

There were other ways of accounting for the absence of diagnosis beyond the structure of medicine itself. Cases took time to reveal themselves fully. Pivotal symptoms might not be visible in early stages of disease, and illness might not present straight away with what Philadelphia neurologist, Silas Weir Mitchell, referred to as the disease’s “definite shape.” It is sometimes undesirable, he wrote “to give explanations until they can be securely correct, or haply the sick man is too ill to receive them. Then we are apt, and wisely, to treat some dominant symptom, and to wait until the disease assumes definite shape” (Mitchell, 1888, p. 31).

Clinicians were advised to wait, if they wanted to arrive upon a diagnosis. “. . . [M]asterly inactivity is often the most prudent policy . . . the old hunter pauses until, by noting the sun and the trees, and examining the objects around him, he decides upon his course, upon which he then goes forward with cautious and watchful step” (Stevens, 1847, p. 10). Haste in diagnosis was attributed to the quack: “*The more ignorant or dishonest the man, the more dogmatic and hasty are his diagnoses*, for with breadth and depth of knowledge comes its highest give, a conception of its limitations” (Greene, 1910, p. 2. *Italics in the original*).

A Dr Sworder provided an indication of the fact that doctors found it generally acceptable to leave a case undiagnosed when he wrote about the difficulty in diagnosing some forms of Scarlet

² Further on the St Andrews Institute, see the excellent secondary review by Macnaughton (2002).

Fever. He maintained that most doctors “allow the undiagnosable nature of a certain number of cases and do not dub the disease by a new name.” The name, however, remained important to patients, according to W. Stanley Sykes, Late House Surgeon of St Bartholomew’s Hospital. “It is a psychological fact that if you tell a patient that you do not know what is the matter with them” he wrote, “he will immediately begin worrying. He will probably think that he has got some rare and awful disease, and will certainly think that you don’t know your job.” He advocated “Latinizing” symptoms to give a “diagnosis of sorts.” “To put into dog Latin the symptoms of which patient complains is not scientific, but it satisfies him every time . . .” (Sykes, 1927, p. 59–60).

In his textbook, “The process of diagnosis,” E. Ryerson pointed out that even experienced doctors found cases that were not diagnosable. Delaying judgment was an important step in the process: “watching for additional signs and symptoms as the case develops” (Ryerson, 1922). Dr Levy Simpson, physician to Willesden General Hospital, similarly advised “If the evidence is insufficient, or the syndrome not yet unfolded, it is prudent to give a tentative opinion and to bide one’s time. When a diagnosis has been arrived at, and even when it seems a certainty, nothing is lost and sometimes errors prevented, by asking oneself what are the other possibilities and writing them down to make sure they do not escape consideration” (Simpson, 1937, p. 1).

Uncertainty was not represented as a failure of decision-making in most of the historical material. Outside of some discussion of the doubt that a student, or young doctor without extensive experience to draw on in order to make a confident diagnosis might experience, doubting a diagnosis, or being uncertain, was characterised as a positive attribute. According to the Austrian general surgeon, Robert Gersuny, “It is better that the doctor should doubt his own knowledge and judgment, and think of his own possible fallibility. He must not forget that there are symptoms of disease which deceive even the most experienced” (Gersuny, 1889, p. 37). Being too confident could interfere with good diagnostic processes. But, at the same time, confidence was part of the trappings of success, as W. H. A. Jacobson explained in his lecture to the Medical Association of Manchester. “Most people, nowadays, are so knowing,” he orated “I might say so omniscient, so wary, so proud of the accuracy of their diagnosis as the proof of their success, that it requires an habitual and single-eyed love of truth to overcome such obstacles as a not unnatural pride and preference for recording successes only, instead of from time to time faithfully registering errors and mistakes” (Jacobson, 1898, p. 6).

Clamouring agreement, the French physician, Alfred Martinet (whose work was translated and widely circulated for English readers) explained diagnostic mistakes as a result, of among other things, “pride and vanity” and identified those who doubt nothing as being just as dangerous as those who doubt everything, because they “are ignorant of everything” (Martinet, 1922, I, p. 35). Taking this a step further, slow and deliberate diagnosis was a marker of quality, and of the professional doctor. In 1910, University of Minnesota medicine professor C. L. Greene pointed out that “The quack never hesitates to make a diagnosis, but the physician of parts, knowledge, and honesty must often make none or at best a provisional one, and wait for more light” (p. 2). His almost-contemporary, from the British Medical Association, Dr Claude Wilson also emphasised, “. . . [W]e may generally trust the man who is not afraid to say he does not know; when he thinks he does know he is generally right. But, the pronouncements of the pundit who courts the reputation of being able to diagnose every case are necessarily unreliable . . .” (Wilson, 1928, p. 709).

Sir Henry Brackenbury, late Chairman of the British Medical Association, fires a shot at what he sees as the encroachment of diagnostic technology over clinical acumen (for him, “science”)

when he describes diagnostic uncertainty as the lot of young doctors, who resort to science as a way of “making up their minds,” a kind of crutch to make up for their lack of knowledge (Brackenbury, 1935).

Sloppy and idle: diagnostic uncertainty in the twenty-first century

The place of diagnosis has gained an ever-increasing prominence in the practice of medicine. The advent of evidence-based medicine, an epidemiologically-based model that requires classification in order to impose its statistically based analysis, requires cases to make its analysis and impose its order. A case is determined by diagnosis, or the grouping together of like conditions. Diagnosis is required by health services and private insurers for patients to gain access to therapies. Diagnostic manuals and classification systems are constantly expanding with increasing numbers of diagnostic categories into which diagnosticians can classify their cases. The number of possible codes in the World Health Organization’s International Classification of Diseases has increased significantly in the last few decades, with only approximately 13,000 codes in 1978 (American Medical Association, 2015), and 55,000 today (World Health Organization, 2020). The *Diagnostic and Statistical Manual* of the American Psychiatric Association has evolved from a small booklet to 947 pages in its fifth revision (American Psychiatric Association, 1994, 2013). Diagnosis is firmly anchored in both the profession and public mind as the fulcrum of medicine. However, this does not eradicate the inconvenient fact of diseases that do not “fit” into the categories available for use, or the way in which this is typified as being evidence of uncertainty. Medicine’s position as a profession is now well-established and authoritative; the challenge is how it delivers on its promise to be able to diagnose and treat physical ailments. The medical “professional knowledge system” (Abbott, 1988, p. 95) is not only constructed via, but marketed as being able to deliver, diagnoses.

In some of the contemporary publications, non-diagnosis is frequently included in a collection of other kinds of uncertainty faced by clinicians. Examples include the necessary uncertainty of preliminary assessment when the diagnostician weighs up a number of different possibilities, progressively eliminating those which do not apply; or, ambiguous diagnoses which are unclear and hard to tease apart. In a prospective study focusing on how certain general practitioners felt about their initial diagnoses of chest pain, comparing their feelings about the diagnosis at initial presentation of the patient with the final confirmed diagnosis, Buntinx, Truyen, Embrechts, Moreel, and Peeters (1991) conflate the unknown with the uncertain, reporting, “Diagnostic uncertainty is not always dispelled, even after many weeks, and following investigation or referral [to specialist services]. Even at this stage, the diagnosis was uncertain or unknown in 12 % of our cases” (p. 123).

Letting non-diagnosis and uncertainty/confusion occupy the same space is common. Than and Flaws (2009) provides an example in a didactic column, “EBM notebook.” This particular paper counsels readers on communicating the absence of diagnosis with frustrated patients. Indeed, they emphasise that diagnosis is not necessarily the outcome of a patient consultation, and they give a mock patient vignette to describe how a diagnostician might discuss the absence of diagnosis. Preparing the patient, they allow that diagnosis may not occur.

Mr Kastagir, we need to do some tests. But you should know that for every 100 patients that come in with symptoms like yours, only 25 of them turn out to have heart problems, while most, the other 75, turn out to have less serious problems, such as indigestion or muscular pain. It may be hard to determine

exactly what those less serious problems are, but we know they are unlikely to come to harm (Than & Flaws, 2009, p. 66).

The scenario offered in conclusion goes on to affirm there is no diagnosis in the end, and at the same time, they are as certain as possible. “As we discussed earlier, we can’t be sure what it is, but with your negative test, we can be almost sure we have not missed anything serious” (p. 67). But, despite this rather certain position that what ails the fictive patient is unknown and at least for the moment, medically unproblematic, the title of their article, “Communicating diagnostic uncertainties to patients,” and its supporting discussion consider their non-diagnosis as a matter of diagnostic uncertainty.

In a similar context, Serbic and Pincus (2014) explore a common adult problem which remains more often than not, undiagnosed: lower back pain. This is a condition which frequently lacks a diagnosable pathophysiological state, and its remedy is far more likely to be in exercise and in occupational modifications than in medical interventions. Current gold standard recommendations are anchored in limiting diagnostic testing (National Institute for Health & Clinical Excellence, 2018). The study in question sought to understand the psychological state of patients with lower back pain. The authors differentiated between participants on the basis of their answer to a question about diagnosis: “I have been given a clear label/diagnosis for my back pain (yes/no)” or “I have been given a clear explanation about why I have back pain (yes/no).” Those who were not given a clear label/diagnosis or explanation were referred to as experiencing “diagnostic uncertainty.”

However, the participants may have been perfectly certain about the absence of diagnosis, particularly if they had read the patient information promoted by the NHS. This material echoes the professional guidelines above, and encourages sufferers to engage in exercise and physical activity rather than in further diagnostic testing or medicalization (National Institute for Health & Clinical Excellence, 2018). The survey did not explore whether they felt certain or not, instead, focused on anxiety, depression, pain intensity and function. The variable attributed to uncertainty was the participant report about having a medical diagnosis or not.

Even those who argue that uncertainty is an inevitable and positive factor in medicine, fall back on the term of “diagnostic uncertainty” to talk about the absence of diagnosis. For example, Simpkin and Schwartzstein (2016) in their perspective article “Tolerating Uncertainty” argue that uncertainty has much to contribute to medicine, and they editorialize for a change to medical practice towards a tolerance of uncertainty. They maintain that obsession with diagnosis is an antithesis to patient-centred care. In their suggestions for reducing the emphasis on diagnosis, they encourage doctors to discuss “uncertainty” with patients.

In an article on diagnostic failure, Weed and Weed (2014) equate uncertainty with the unknown and differentiate “genuine uncertainty (that which is unknown to medical science)” with “personal uncertainty (that which is unknown to the patients’ physician, but which might be known to someone else)” (p. 16). These authors, like those of the twentieth century whose words I will soon discuss, also underline that by learning to avoid doubt, doctors often fail to realise the limitations of medicine for defining what ails individual patients. They refer to it as a “false confidence” (p. 16).

Confirming, as have many of these contemporary authors, the use of the word “uncertainty” to typify the absence of diagnoses, Bhise et al. (2018) use “no diagnosis” as an indirect measure of uncertainty in primary care, and they define diagnostic uncertainty as “subjective perception of an inability to provide an accurate explanation of the patient’s health problem” (p. 113). What is salient in this definition is how it locates the state in “perception,” “inability” and “subjective[ity].” These are cognitive and emotional

spaces linked to the practitioner, in contradistinction to being linked to medicine’s classification systems themselves, or to the principle of classification of individual cases.

Messer, Sibilia, and Miazhiom’s *Diagnostic Uncertainty and Clinical Decision-Making Strategies* (Messer, Sibilia, & Miazhiom, 2018) is among the most direct in using the term as an explanation for those cases which potentially undermine medicine’s paradigm. “Recommendations and criteria sets for diagnosis or classification may provide guidance but often fall short of converting the specificities of each individual patient into a confident diagnosis” they write (p. 267). A similar position is taken by Kelly and Panush (2017) who, in their article on diagnostic uncertainty, coin a new diagnosis that they can use when there is not a diagnosis, or when a patient presents symptoms from a variety of different disorders. They propose “overlap syndrome.” They lament that their diagnostic systems are inadequate from several perspectives: “Some patients have manifestations of more than a single nosologic entity while others do not display findings which would satisfy diagnostic criteria for any of the currently recognized rheumatic disorders” (p. 1211). They posit that as many as 25 % of rheumatic disease patients cannot be diagnosed. Refreshingly, they explicitly acknowledge the limits of their episteme: “Diagnostic and epistemologic humility should be a more common default position than is usually taken. We need to be comfortable with the uncomfortable. It better serves our science and art, and our patients. While uncertainty can be distressing, the alternative is indeed more perilous” (p. 1212).

“The absence of a codifiable disease ensures uncertainty when the symptoms remain unexplained” pen Scott-Wittenborn and Schneider (2017), even though they describe this as “ruling out disease” (p. E1), a situation of presumed certainty, since “ruling out” clearly implies being able to assertively reject the presence of disease. This terminology paradoxically still anchors the absence of diagnosis, semantically at least, in cognition.

Bioethicist Diane O’Leary’s essay about the limitations of medicine’s diagnostic model also conflates diagnostic uncertainty with the medically unexplained (O’Leary, 2018). The conflation is present not only in her paper’s title, “Ethical Management of Diagnostic Uncertainty: Response to Open Peer Commentaries on ‘Why Bioethics Should Be Concerned with Medically Unexplained Symptoms,’” but also in its content. She explains that diagnostic uncertainty can be related to psychogenic symptoms, as well as to “benign self-limiting biological symptoms, rare disorders, contested conditions, everyday medical conditions that present themselves in unusual ways, more serious medical conditions that are diagnostically challenging (such as autoimmune diseases), and very serious medical conditions that have not yet made their seriousness apparent.” She also provides a link between the mutually individualising medically unexplained symptoms and diagnostic uncertainty, which I will discuss in the conclusion.

I finish up this section with one last example, to which I will also return in the conclusion because it is so explicit in the way in which it discusses non-diagnosis as a failure of the individual, rather than of the system. Roger Jones, editor of the *British Journal of General Practice*, does not share this sense of medicine’s limitation in his editorial “After Achilles” (Jones, 2016). Reflecting on a historical article about the difficulty of diagnosis, he muses about diagnosis as a preface to an issue of his journal. Jones asserts that general practice has “floundered among unhelpful phrases such as ‘tolerating uncertainty,’ ‘using time as a diagnostic tool’ and letting the diagnosis emerge”, which sadly passed into our lexicon.” He continues, “at worst, this approach to diagnosis is sloppy and idle, and seems to lie at the other end of the spectrum from the need to make early, accurate diagnoses in patients presenting with even the most vague symptom complexes” (p. 115). We will come back to this quotation at the end of this paper, as I provide an

argument about how this focus on uncertainty as representative of undiagnosed ailments provides a defence of medicine, and the limitations of its episteme.

Epistemic defence

To link this representation of non-diagnosable ailments to the epistemic defence, we must remember what shapes the epistemic setting of medicine. Diagnosis is the means by which knowledge is adjudicated in medicine, and it is via its diagnostic framework that medicine has gained the authority “to define the real” (Whooley, 2013, p. 20). When its diagnostic methods are insufficient to explain disease, it must still justify that diagnosis is an achievable aim.

Diagnosis is a “mediating act” (Abbott, 1988, p. 94) which triggers the professional knowledge system, and indicates both inference and treatment, the cornerstones to medicine’s jurisdictional claims. Abbott writes, “[The] classification system is a profession’s own mapping of its jurisdiction, and internal dictionary embodying the professional dimensions of classification” (p. 96). So powerful is diagnosis as a trigger it is the metaphor that Abbott uses to describe professional acts generically (outside of medicine).

To support their jurisdictional system, historical and the contemporary practitioners of medicine have found ways to account for the flaws of their classification framework: the diagnostic paradigm. Far from discarding diagnosis as a way of practicing medicine, both have confirmed its power. In the historical accounts, the failure of diagnosis is most frequently reported as a result of the budding state of medicine, still incomplete, still constructing itself, still identifying the laws of generalization into which individual cases of disease can be categorised.

While contemporary medicine too acknowledges the limits of its knowledge base, and makes similar calls as those of its

predecessors about the need to march ever forward in pursuit of more information about disease (“evidence” as it is called today), this move towards characterising the absence of diagnosis as “uncertainty” performs another function. It places the absence of diagnosis in a cognitive space. To be uncertain is to negotiate between multiple options, to be in a position troubled by one, or many, other possibilities. But most importantly by being a cognitive state, it is located in the mind of the individual clinician. When we talk about diagnostic uncertainty, we position the problem as one of expertise rather than one of the diagnostic structure. Locating the problem of non-diagnosis in the individual clinician, as a cognitive problem, it can then become evidence of, as Jones (2016, p. 115) asserts, “sloppy and lazy practice,” rather than of an imperfect system.

This enables the situation in question here—the ailment without a name—to exist without maiming the expert system. While the point of this paper was to review how uncertainty is used as a kind of epistemic excuse, its parallels with a similar excuse should not be overlooked. Undiagnosable disease is also often referred to as the “medically unexplained symptom” as I mentioned above. As I have written elsewhere (Jutel, 2010), and as have many others before me (for example, Dumit, 2006; Malterud & Taksdal, 2007; Nettleton, 2006), the term “medically unexplained symptoms” (MUS), which seems to acknowledge the limit of the medical episteme is actually, performing another role. The term is used as a diagnosis of its own to refer to psychosomatising disorders, or the physical expression of psychological distress. By so doing, the unexplained (i.e., the inconvenient facts of diagnosis) become evidence here of another individual failing. When the case cannot be explained, it is explained as a result of the psychiatric makeup of the patient, who is projecting his or her angst onto bodily function. Despite the wide array of seemingly disparate symptom presentations which may be referred to under this banner, they are clumped by the medical literature in a discrete diagnostic category of their own: MUS. This category, rather than

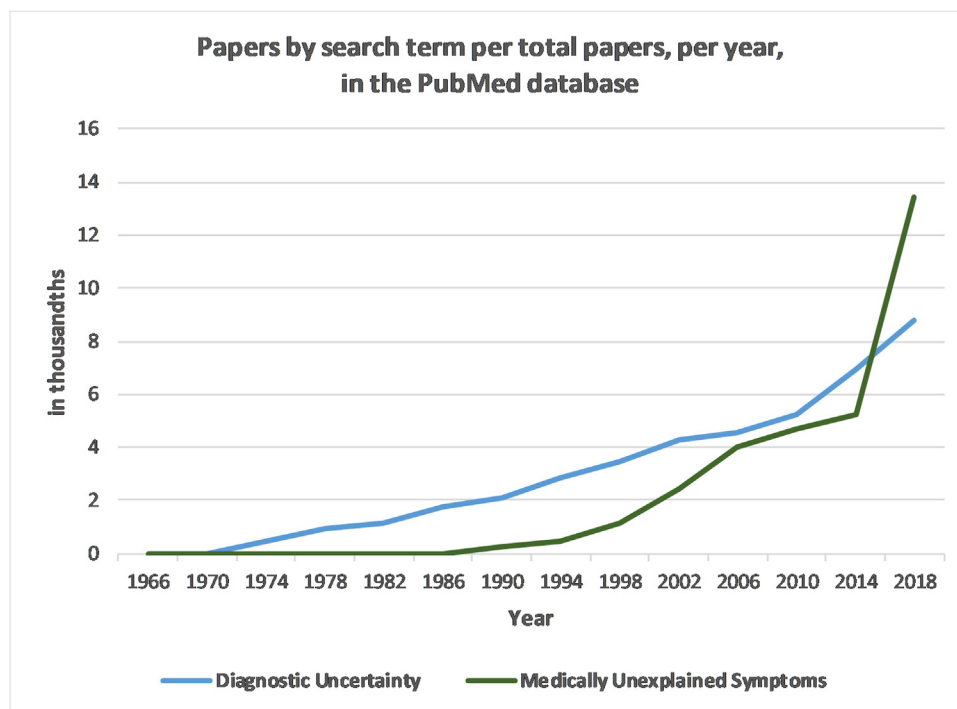


Fig. 1. Searches on the terms *medically unexplained symptoms* and *diagnostic uncertainty* in the PubMed database show parallel and significant increases in relative frequency over the period 1965–2018. Graph created by author, based on yearly distributions of search results obtained from Alexandru Dan Corlan, Medline trend: automated yearly statistics of PubMed results for any query (2004), Web resource at <http://dan.corlan.net/medline-trend.html>, Accessed 18 March 2021.

reflect the limitations of medicine, implies the infallibility of the physician and confirms the power of diagnostic medicine (Jutel, 2010). Fig. 1 demonstrates the concomitant rise of relative frequency of both terms “medically unexplained symptoms” and “diagnostic uncertainty” in the PubMed database.

While a limited number of contemporary medical publications is less dismissive of the unexplained symptom, and seek to justify a non-psychiatric explanation for MUS, these few publications generally do not step back from the diagnostic model. Meador (2005), for example, harkens back to early twentieth century publications when he refers to these symptoms as “not explained yet,” reflecting the epistemic hope for what medicine will one day be able to reveal.

There is, however, a paradox in the use of the term uncertainty to characterise non-diagnosis. Fox (1980), in a retrospective review of her own 1957 seminal work on uncertainty, explained “To be puzzled, ignorant, unable to understand; to lack needed knowledge or relevant skill; to err, falter, or fail, without always being sure whether it is ‘your fault’ or ‘the fault of the field’ (as one medical student put it), is especially painful and serious when the work that you do is medical” (p. 5). She summarised a typology of uncertainty that included imperfect mastery of knowledge, limitations in available knowledge, and the difficulty in distinguishing among the first two.

However, the term “diagnostic uncertainty” as revealed in this essay, is used, not in relation to anxiety about whether an outcome chosen is correct. To be certain is not necessarily to know everything. For example, to say “I don’t know” can be a statement of certainty. It does not characterize a speaker torn between knowing and not knowing (troubled by various possibilities), rather, one who acknowledges the limitations of their capability, or of their episteme.

What is also paradoxical is that, as sociologist Light (1979) expounded in his seminal work on uncertainty, “socialization for uncertainty takes on particular significance in professional training because the professions depend on the public believing that they know what they are doing” (p. 310). By using the term uncertainty, precisely the opposite is happening. It is possibly better to suggest that there is a problem of cognition than it is to admit that the field cannot account for particular forms of illness, that it is “the fault of the field.” Uncertainty is a way of deflecting threats away from the field as a whole, looking carefully, instead, at the individual practitioner. Light has commented recently that he does not find the term uncertainty very helpful in any case. “What’s at issue,” he muses “are the various forms of ambiguity, ambivalence, risk, and perhaps bewilderment and mystery . . . at the borders of certainty.” He sees these as places of potential action, rather than of the limitation that “un” certainty implies.³

The field remains, to a certain extent and always, under threat from a range of external forces. Warbasse’s 1912 observation of the “ruinous competition” of the early twentieth century (in Light, 2004) today comes in the form of artificial intelligence, patient self-diagnosis, and other pathways to diagnosis. So, what we see here is not a jurisdictional battle between this profession and that, rather a battle to maintain public belief in the diagnostic framework “sold” to the public in medicine’s youth, now firmly engrained in social systems. Diagnosis remains pivotal to medicine’s work, and at the same time, is widely (over-?) used by potential patients to explain what ails them, blurring the boundaries in the doctor-patient relationship, changing the premises of the consultation and challenging authority (Jutel, 2017).

Protecting medicine becomes an important priority for medicine. We see this clearly in the words of Roger Jones who opined that tolerating uncertainty was evidence of sloppy practice; it is a way of shifting the difficulties of medicine to the shoulders of its practitioners. While the words of Jones are direct and unequivocal, the word usage I have described above is, for the most part, unwitting. It is an example of how words we use are often instilled with meaning beyond their simple usage. It is also in line with concurrent thinking about the responsibilities of the neoliberal subject (Trnka & Trundle, 2017).

A better stance for the preservation of medical epistemic authority in the face of its inherent shortcomings might be to distance the field from diagnosis, and to be transparent about the limits of medicine. Propst (1939, p. 83) opined, “It is sometimes impossible to adequately summarize in a name the whole state of a patient’s disequilibrium.” This view is echoed by Jerome Kassirer in an era closer to our own (albeit in cognitive terms): “absolute certainty in diagnosis is unattainable, no matter how much information we gather, how many observations we make, or how many tests we perform . . . more tests do not necessarily produce more certainty” (Kassirer, 1989, p. 1489).

This article adds a new leaf to the sociology of diagnosis. The point of this subdiscipline has been to acknowledge the degree to which diagnosis shapes understandings of health, illness, and disease (Brown, 1995; Jutel, 2011b). It recognises both the categories of disease which are available for assignment, the process by which diagnoses are assigned, and the consequences of diagnostic labelling (Blaxter, 1978).

The sociology of diagnosis has for its objective not only an understanding of how diagnostic categories are socially shaped, but also how diagnosis works as a social process—the diagnosis-as-category and -as process referred to by Blaxter (1978). But diagnosis has consequences as well. My work (Jutel, 2011b), and those of many others in this field,⁴ explores how diagnosis as a system legitimizes, stigmatizes, exploits, and positions the diagnosee and the diagnoser; as well as explaining myriad social understandings of health, illness and disease. Diagnosis assigns social roles in the health care encounter, with the ability to diagnose pivotal in the power relation. But germane to the present essay, diagnosis grounds the profession. That Abbott uses it as a metaphor for how *all* professions implement information from their knowledge systems underscores the need for this epistemic defence.

Here, with the exploration of the discursive use of diagnostic uncertainty, we have something new to consider. We have a comment on how diagnosis constructs knowledge, but at the same time, how its absence can be palliated in this same knowledge construction.

It is an important moment to open this discussion on diagnostic uncertainty. While on the one hand we are experiencing a proliferation of new diagnoses, at the same time, diagnosis is also under threat from a range of perspectives: some concrete, some speculative. As mentioned above, artificial intelligence and apps offer simultaneously real and imagined (or misrepresented) benefits to the practice of medicine, both of which trouble, however, the diagnostic power previously vested primarily with doctor. The Star Trek Tricorder (a device, which placed on the patient’s forehead revealed what was wrong with them) remains a thing of science fiction (Jutel & Jutel, 2017).

Diagnosis is a very useful medical tool, because as it generalises, it also provides a pathway to treatment, explanation and prognosis.

⁴ See for example Ebeling (2011), Hayes, McCabe, Ford, Parker, and Russell (2021), Horwitz (2011), McGann and Hutson (2011), Nettleton, Kitzinger, and Kitzinger (2014), Weinberg (2020); and Whooley (2010).

³ D. W. Light (personal communication, 19 December 2020).

But it also obfuscates, as it seeks to represent the individual in a generic category which clearly, cannot always suit. The old adage “you must treat the patient and not the disease” characterises medicine’s amazing potential while at the same time, recognising the limitations of the diagnosis to explain all that ails us.

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