## THE PENETRATING POINT

# The Trivialization of Diagnosis

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Although it is widely recognized that diagnosis plays a central role in clinical medicine, in recent years the primacy of diagnosis has come under attack from several sources. 1. "Billable terms" are replacing traditional medical diagnoses. The former are based on International Classification of Diseases lists, which include many non-diagnoses such as symptoms and signs. 2. Diagnosis often gets short shrift because of the perceived urgency of discharge. 3. The problem oriented record, in practice, has frequently led to a shift in emphasis from synthesis of findings to fragmentation of problems. 4. Presumptive diagnoses frequently metamorphose into established diagnoses in medical records, even if incorrect. 5. A number of authors have apparently disparaged the importance of diagnosis. Nonetheless, it is clear that diagnosis must continue to play a central role in clinical medicine. We propose several ways by which we can resist these forces and assure that diagnosis retains its appropriate position of primacy. Journal of Hospital Medicine 2010;5:116-119. © 2010 Society of Hospital Medicine.

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Historically, diagnosis has been central to patient care. Making the correct diagnosis serves as a guide to the choice of treatment, permits assessment of prognosis, and indicates what complications to expect. Arriving at the correct diagnosis has been a major goal—the Holy Grail, as it were. Accurate diagnosis continues to be a major focus of medical practice, and accurate diagnoses are routinely made every day. Still, many experienced clinicians have the impression that in recent years the primacy of diagnosis has been coming under attack from several sources.

A decade ago, Thomas Szasz pointed out that disease is a fact of nature, while diagnosis is man made. The noun "diagnosis" is derived from the Greek verb diagignoskeinindicating knowledge attained through analysis. As defined in Merriam-Webster's Collegiate Dictionary, "the diagnosis" essentially means the conclusion arrived at by the art of identifying a disease. It is the product of an intellectual effort of a particular analytic type. The response to the question "What is the diagnosis?" has been the name of the specific disease entity with which the patient is afflicted.

Disease entities represent coherent, organizing concepts.<sup>2</sup> A specific disease is a condition with characteristic manifestations—clinical, histologic, or pathophysiologic. If untreated, it results in dysfunction or, in some cases, death. Differentiation of one disease from another is enhanced when there is some sort of understanding, even if incomplete, of the specific pathophysiology at play. Admittedly, concepts of what constitute specific disease entities are not fixed; they evolve with time. Not all diseases have been identified. The underlying etiology may or may not be known. Nonetheless, diseases are recognized as specific entities, distinct from other diseases. Thus, anemia is not regarded as a disease, while pernicious anemia and iron deficiency anemia are diseases. Fever is not a disease, while typhoid fever is. Arthritis is not a disease, while gonococcal arthritis is.

#### "Billable Terms" Are Replacing Traditional **Medical Diagnoses**

The term diagnosis has been redefined to comply with the need to enter a "diagnosis" for billing purposes. Use of this term for this purpose has confused the issue. Diagnoses entered for such purposes are largely derived from International Classification of Diseases (ICD) lists.3 However, the ICD was not intended to definitively identify underlying diseases, nor to serve as a guide to management and prognostication. The 6th revision of the ICD in 1948, the first revision to be widely employed, was designed for epidemiologic purposes and achieved widespread use to obtain mortality and morbidity statistics.<sup>4</sup> It was subsequently also used as a tool to index hospital medical records.

Significantly, it was also employed for billing purposes, with far-reaching pernicious consequences. Although the ICD purports to be a list of diseases, it actually includes symptoms and signs. Consequently, in the billing context, diagnosis no longer necessarily refers to specific disease states; it now refers to "billable terms"-often the manifestation that was responsible for the patient seeking medical assistance. Far from being the product of an intellectual effort, it is often merely a justification for submitting a bill. Examples of such "diagnoses" are shown in Table 1. Many of them represent symptoms, signs, or laboratory abnormalities. The importance of accurate medical diagnosis has been cheapened by this change. The effect is to devalue diagnosis—to lessen its status as the Holy Grail.

The effect of this on trainees is invidious, and predictable. The traditional meaning of diagnosis is being replaced

## TABLE 1. Some Possible "Diagnoses" That Can Be Entered into the Electronic Medical Record in a Major **Teaching Hospital**

Abdominal pain General symptoms Special symptom Abnormal blood test Immune disorders Splenomegaly Back disorder Joint disorder Throat pain Myoneural disorder Coagulation defects Urinary symptoms Visual disturbance Diseases of esophagus Otalgia Eve disorders Pain in joint Vomiting Fluid/electrolyte disorders Right lower quadrant mass Wheezing

in our minds. Physicians in training are tempted to deceive themselves into believing that they have arrived at an understanding of what they are dealing with when they enter such a "diagnosis." After all, have they not responded to the question: "what is the diagnosis?"

We do not mean to imply that physicians are doing anything wrong by entering ICD terms for billing purposes. What must be done for billing purposes must be done. It is important to be aware, however, and to continually remind ourselves, that what has been entered for this purpose is often not a true medical diagnosis.

Further, when the diagnosis is not yet known, it is not possible to enter a true diagnosis. There is no way to say "I don't know." It would be preferable to simply admit that the diagnosis is not yet established, as a medical resident has recently emphasized.<sup>5</sup>

### Diagnosis Often Gets Short Shrift Because of the Perceived **Urgency of Discharge**

The emphasis on diagnosis several generations ago may have resulted, at least in part, from the relative paucity of effective therapeutic interventions before the 1930s. Things have changed; therapeutic capabilities are much more powerful now. Making the correct diagnosis seems to have lost its urgency. Instead of the major question being "what is the diagnosis?" it now is often "what do we do now?" The diagnosis is often an afterthought. Indeed, it is sometimes not even mentioned in discharge summaries, where, not uncommonly, one sees nondiagnoses such as "blood in stool" or "polyarthritis."

In addition, we are under pressure to shorten the inpatient stay of hospitalized patients. At least a portion of the public is aware of this; thus, it has been noted in the New York Times that: "The pressure to get patients out of the hospital rapidly can focus medical attention on treatment rather than diagnosis."6 We commonly seek to ameliorate the patients' status to permit discharge before (or often without) learning what we are dealing with. Sometimes one senses that the primary question has become "how soon can we discharge this patient?"

A price is paid for this. In the absence of a valid diagnosis, patients may be subjected to a broad array of nonessential investigations and therapeutic interventions, each with its own possible complications. Patients are often discharged without a diagnosis having been made, presenting a serious challenge to outpatient physicians who are left to manage them without a clear idea of what they are dealing with. It often falls to the outpatient physicians to make the diagnosis. This is somewhat problematic, since they themselves are under harsh time pressure. Patients often require rehospitalization for the same as-yet-undiagnosed condition.

#### The Problem-Oriented Record Poses Problems

The widespread use of the problem-oriented record, originated by Lawrence Weed,<sup>7</sup> has led to problems of its own.<sup>8</sup> It has evolved, away from its original intent. In practice, its major emphasis often seems to be on identification of problems and tracking their progress, rather than on synthesis. This often leads to muddy rather than clear diagnostic thinking. Assessments and progress notes frequently consist of lists of symptoms, organs, abnormal laboratory findings, or even medical specialties. The net effect is often fragmented thinking—as Weed<sup>7</sup> put it, "failure to integrate findings into a single entity." Synthesizing diverse findings into a single entity, when possible, is necessary to define a diagnosis. Failure to do so may have serious consequences. In a recent study of diagnostic errors in internal medicine, cognitive errors were frequently found to contribute to such errors.9 The most common cognitive problem was faulty synthesis. How much worse than faulty synthesis is failure to synthesize at all!

#### Presumptive Diagnoses, Even if Incorrect, Metamorphose into Established Diagnoses

We must often treat empirically. When there is no firm diagnosis, presumptive diagnoses must be made and acted upon. Unfortunately, there are not always mechanisms for the physician to make it clear that his or her diagnosis is only presumptive. (A common example is "acute viral syndrome," generally an educated guess.) All too often, presumptive diagnoses are entered, without qualification, as definitive diagnoses, and then achieve immortality. Thus, if a patient is incorrectly diagnosed as having rheumatoid arthritis, all subsequent presentations will start: "A so-and-so year-old woman with rheumatoid arthritis for many years .... " Presumptive diagnoses are frequently not questioned. It is easier to assume that they were arrived at after due consideration. Once entered in the medical record, they may be difficult to remove.

It is true that the need to arrive at a precise diagnosis is less pressing for some medical specialties than for others. Emergency physicians, critical care physicians, and frequently, surgeons, must commonly act on the basis of presumptive diagnoses. In contrast, internists, family physicians, psychiatrists, and indeed all physicians who care for patients with chronic illnesses can, with time, be expected to sort out accurate from inaccurate presumptive diagnoses.

A specific example of the problem of presumptive diagnosis is of interest. It is not uncommon, following a first encounter, for a diagnosis to be entered based on the patient's history alone. While such diagnoses are frequently correct, they are not invariably correct. The patient may have arrived at the conclusion herself; she may have misunderstood what she was told by her physician, or her physician may have been in error. Such inaccurate diagnoses also often achieve immortality in the medical record.

#### **Apparent Disparaging of the Importance** of Diagnosis

Further trivialization has come from a number of publications expressing concerns about the importance of diagnosis. Thus we read that there are negative consequences of emphasis on diagnosis. When we know what is wrong, we focus less on the individual and more on the disease. 10 In his recent book Our Present Complaint. American Medicine, Then and Now, the scholar C.E. Rosenberg<sup>11</sup> includes a chapter with the provocative title "The Tyranny of Diagnosis." He points out that even a century ago the fear was expressed that burgeoning scientific medicine would lead to denigration of physicians' holistic and intuitive skills.<sup>11</sup> Other authors maintain that firm diagnoses may be misleading, since many diseases are a matter of degree in a continuum—a spectrum—that are best defined employing a statistical model of risk prediction.<sup>12</sup> The suggestion is made that the usefulness of diagnostic tests should not be related to the presence or absence of a disease, but rather to whether they influence outcome.<sup>13</sup>

"Scientific medicine" is focused on diagnosis. Denigration of diagnosis has often come, as a philosophical posture, from opponents of reductionist thinking. As Rosenberg<sup>11</sup> points out: "It has become fashionable among humanistic and social science-oriented commentators to dwell on the distinction between illness and disease, between the patient's felt experience and the constructions placed on that experience by the world of medicine." Their opposition, he feels, reflects the value-laden mutual incompatibility (real or apparent) of art and science, of holism and reductionism.<sup>2</sup>

It is true that medicine is more than just biology. There is a great deal to be said for the view that scientific medicine tends to deemphasize the humanistic, holistic aspects of medical practice. However, despite all these concerns, most physicians—and, to be fair, most critics—agree that making an accurate diagnosis is important. Thus, though the title of his relevant chapter is The Tyranny of Diagnosis, Rosenberg<sup>11</sup> states: "I might just as well have used the term indispensability." Indeed, the opening words of that chapter are: "Diagnosis has always played a pivotal role in medicine." <sup>11</sup> Other authors cited above issue this disclaimer: "We are not against diagnosis. Diagnosis does and always will play a central role in clinical medicine."12

The importance of diagnosis is underscored by the vigorous debate about how to assess diagnostic tests;14 apparently, diagnosis does indeed matter. While it is true that diagnoses are not always precise, objective, and quantifiable, 10 abun-

dant evidence points to the unavoidable conclusion that identifying the patient's disease is heuristically useful; that is, it works.2 The track record of modern scientific medicine in improving mortality and morbidity speaks for itself. It hardly seems necessary to defend it. In addition to representing a valuable intellectual challenge in its own right, diagnosis is pivotal to the scientific mission of medicine.

#### What Can Be Done?

The net effect of all these forces: the use of "billable terms" as diagnoses, the pressures of managed care, fragmented problem lists, persistence of incorrect presumptive diagnoses in medical records, and antireductionist criticisms is to encourage sloppy diagnostic thinking in some physicians. What can be done to emphasize the proper use of differential diagnosis in arriving at a definitive diagnosis? What can be done to underscore the importance of differentiating between presumptive and definitive diagnoses? Most importantly, how can we instill the respect for the intellectual honesty necessary to acquire and retain these skills?

Above all, we should relentlessly impress on our students and trainees the importance of arriving at an accurate definitive diagnosis. They should be aware that the job is only half done if the diagnosis has not been made. We should do this repeatedly, both by word and by example. We ourselves must display intellectual honesty.

In addition, we ought to be able to enter "diagnosis uncertain," so coded, or to append the phrase "-cause unknown" after the manifestation of concern, when we don't really know what is going on. We should routinely indicate when a diagnosis is merely presumptive. Perhaps we need a way to indicate: "This diagnosis is definitive" or to indicate the specific evidence that led to the diagnosis (eg, biopsy, laboratory result, radiographic finding). Finally, we need to correct the current confusion between diseases and billable terms, to differentiate the disease from the symptom, perhaps by labeling ICD-9-CM codes simply as "billing codes," with a separate entry for actual medical diagnoses.

Although powerful historical forces have brought us to this state, we believe that arriving at the correct diagnosis is at least as important now as it has been in the past, and that its primacy should be recognized, celebrated, and fought for. We owe our patients no less.

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