Developing Tolerance for Ambiguity

FROM THE INCEPTION OF MEDICAL EDUCATION, STUDENTS ARE TRAINED to recognize and categorize disease. Through the formulation of differential diagnosis we learn which constellations of symptoms are characteristic of certain pathologies, and we structure our investigations, treatment plans, and prognostic predictions accordingly. These activities ground and direct the clinical encounter. Yet, as the articles in this issue of MSJAMA illustrate, the concept of disease is inherently elusive and ambiguous, and the implications of classifying and labeling disease extend far beyond the typical patient-physician interaction.

Often the recognition and treatment of disease is straightforward; the previously well individual with a fever and a cough productive of purulent sputum or acute abdominal pain with rebound at McBurney’s point occur within a well-defined paradigm, and do not challenge that paradigm’s assumptions or limits. Yet how does one make sense within the clinical encounter for the more troubling boundary cases that resist easy classification? How should one think about a patient with newly diagnosed cancer who feels well yet harbors occult pathology, or a patient with debilitating symptoms that have no evident organic basis? As Winston Chiong discusses, the naming and diagnosis of disease involve evaluative judgments as well as scientific fact and are as much a social phenomenon as a scientific practice.

Assigning the label of disease to a patient is an intrinsically subjective exercise. Disease is constructed by each patient’s personal histories and beliefs, by professional and lay individuals who surround and interact with the patient, and by society at large. Each construct has the power to influence the others, and each relationship has the ability to shape the way that disease is understood. In this context, patients and physicians each have the ability to affect the construction of meaning. A case in point is the individual with a genetic predisposition that increases his or her risk of developing a certain disease.

As Kelly Smith writes, there is a common perception that certain genes destined one to develop particular diseases, yet this statistical probability cannot be taken as a certainty. The implications of this are vividly illustrated by "Anonymous" who carries the BRCA mutation; she writes of inhabiting an indeterminate zone in an uncertain "world beyond the white coat." As her article poignantly demonstrates, these issues matter because attribution of labels such as "diseased," "healthy," "susceptible," or "pre-disposed" engender a wide range of consequences from the private and interpersonality to the larger spectrum of society. Access to health care, employment and insurance options, and lifestyle choices are all mediated by the labeling of disease and the legitimation of those labels. As Katerina Christopoulus discusses, those who occupy the sick role, whether voluntarily or not, inhabit a particular and distinctive position within society.

This issue of MSJAMA explores the ambiguity that surrounds the understanding and diagnosing of illness, and the social consequences entailed by the attribution of disease labels. It is our hope that by becoming aware of and attentive to these ambiguities, medical students as both future clinicians and future patients will have a greater awareness and sensitivity to the unknown and uncertain in medicine.
Diagnosing and Defining Disease

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The patient and his wife are seated when the doctor enters the room. They are holding hands; the heel of his foot taps erratically against the leg of his chair. The doctor closes the door behind him while glancing at his chart.

“Mr Richardson, Mrs Richardson, how do you do? Sorry to keep you waiting. It’s been a busy morning.”

“No, no, it’s no problem, Doctor.” The patient laughs nervously. “I’ve been waiting two years to find out what’s wrong with me. A couple of minutes . . . ” he trails off and shrugs.

“I know what you mean.” Pages rustle as the doctor flips through the chart. “Well, you know, when you first came in to see me, I was as confused about your symptoms as you. Maybe more.” He looks up and smiles ironically—patient and wife nod politely but do not smile back. “But with these test results and the changes you’ve been noticing over the last few months, I think we have a better idea of what’s going on. Mr Richardson, I think that you have a condition that we call . . . ”

The patient and his wife lean forward slightly, eyes wide: an image of pregnant anticipation.

What happens when a patient is given a diagnosis? A diagnosis is, first of all, a description of the patient as a sufferer of a particular disease process. A diagnosis can also be an explanation for patients who have had symptoms but do not know their cause. Giving a name to the problem may seem to resolve the mystery, such that even patients with intractable, chronic diseases may feel relief when diagnosed. And of course, a diagnosis often offers a prediction about the future course of illness, or about the genetic or infectious risk to others.

All of the above belong to what might be considered the “science” of medicine—but diagnosing diseases has more subtle implications as well. Identifying a disease can confer social legitimacy on a patient’s symptoms, relieving patients of the suspicion that they are malingering or exaggerating their woes; conversely, it may be seen to imply that the patient is somehow abnormal or requires intervention. Having a disease might mark the patient as a member of a community of sufferers, as demonstrated by the many patient and family associations that exist for various illnesses. Finally, diagnosing a disease can have important legal and economic consequences, such as conferring eligibility for disability benefits or even (in the case of the insanity defense) exempting patients from responsibility for their actions.

Thus, while diseases are commonly thought to be scientifically defined, the naming and diagnosis of diseases is also a social practice with implications that extend beyond the clinic doors. We might wonder, given these pressures, if “disease” is truly a unified concept. Some diseases, such as tuberculosis or embolic stroke, identify a highly specific etiologic agent or process; others, like Alzheimer disease or scleroderma, indicate pathologic changes of unclear cause; while syndromes and functional disorders simply describe collections of symptoms and signs that frequently occur together. What do these labels all have in common?

This question is complicated because there is a tension in medical concepts of health and disease. On one hand, they are meant to represent objective, scientific facts—this patient is healthy, that patient has disease. But on the other hand, they also involve evaluative judgments about good and bad—a healthy state is better than a diseased state, a diseased state is in some way abnormal or dysfunctional—that are more than matters of simple scientific fact. To illustrate: when a patient is diagnosed with coronary artery disease, part of what is implied is that subendothelial accumulations of lipids and macrophages are present in his or her coronary arteries, increasing the likelihood of coronary ischemia, and these are more or less objective descriptions. But another part of what is implied is that the patient has a disease, an abnormality that results in impairment of his or her quality of life, and this is an evaluative judgment.

“Quality of life” is a telling phrase, because it reveals how concepts like health and disease, function and dysfunction, are interwoven with conceptions of the good life. As Caplan et al note, “what will count as minimal standards of function or as special levels of excellence will depend on value judgments concerning what is important to be able to do as a human being.” Indeed, ideas about what counts as health and disease have changed to fit changes in value judgments and therapeutic options. For instance, contraception is now thought of as part of medical care because the medical community considers it important for people to plan and control reproduction, while physicians of an earlier time would have considered reproduction to be the necessary physiologic function of sex. Impotence was once considered by most people to be simply an unfortunate occurrence, but with less problematic therapies it is now considered a treatable medical condition. Both of these changes reflect changed cultural attitudes about the role of sex and sexuality in human life.

Thus, the concept of health involves a descriptive component (what someone is able to do) as well as an evaluative component (what it is important to be able to do, in order to be able to live a good life). Similarly, the concept of disease involves a duality between a description (a physiological or functional difference between the patient and the “healthy” norm) and an evaluation (the judgment that this difference is abnormal or dysfunctional, and not just different). However, this duality is obscured in everyday practice, in part due to the scientific aspirations of medicine and scientific assumptions built into the medical model.
For instance, consider the standard “SOAP” progress note, in which the patient’s report of symptoms is termed “subjective,” while physical findings and laboratory results are termed “objective.” This subjective/objective distinction is based on a standard understanding of scientific observation, in which the subjective corresponds to how things seem from a particular perspective (in this instance, the patient’s experience) while the objective corresponds to concepts that are not tied to any particular point of view (such as temperature or the concentration of ions in the blood). In Western science, the subjective and objective are often reconciled by a “reduction,” in which the subjective appearance is explained by reference to the objective account of how things “really are.”

The classic example is the Copernican heliocentric model of the cosmos, in which the subjective appearance that the sun travels around the earth is explained by an objective description of the earth orbiting the sun.

Medical practice employs a related model in the notion of a “disease entity,” inherited in large part from the scientific aspirations of earlier physicians such as Koch and Virchow. In this model, diseases are conceptualized as distinct, objective entities that are common to afflicted patients. While the patient often cannot directly perceive the presence of the disease entity itself (such as a microorganism or histologic change), the patient does perceive the subjective symptoms that are caused by its presence. Thus, the subjective symptoms are explained by reference to objective changes in the body, in much the same way that subjective appearances in the physical world are explained by reference to objective concepts such as matter, energy, and force.

The disease entity model has had great success in explaining the symptoms associated with infectious diseases, certain cancers, and poisons, which are understood as distinct entities and which often produce a “classic” set of symptoms and signs in the afflicted patient. These are widely taken as ideal examples of disease processes, and they influence a paradigm in which physicians are seen (or see themselves) as disease-hunting scientists or detectives collecting data to identify the etiologic agent. (For instance, it is not surprising that most of the vivid short pieces in Berton Rouèche’s classic The Medical Detectives involve bacteria, parasites, and poisons.) This paradigm then structures the expectations of patients and physicians. Both may see the physician as someone who can or should provide “answers” to the patient’s problems, and both can be frustrated when this expectation is not met.

This paradigm is less well suited to multifactorial conditions, such as type 2 diabetes mellitus and coronary artery disease, which are thought to result from the overlap of various contributing factors rather than a single etiologic agent. Still, in these diseases there are objective findings that can confirm and explain the patient’s subjective feelings of illness, so the subjective/objective distinction is preserved. More problematic are functional disorders, syndromes of unclear etiology (such as chronic fatigue syndrome and fibromyalgia), and complaints that do not fit any recognized symptom complex. In these cases, there may be no objective findings to confirm and explain the patient’s subjective feelings of illness—the subjective feeling of illness is all there is. Physicians may then feel pressurized by the seriousness of the patient’s complaints to give a diagnosis that they privately regard as scientifically unsubstantiated; and some have worried that giving names that pretend to explain poorly understood complaints may have the unintended effect of creating new illnesses, as with the “transient mental illnesses” studied by philosopher Ian Hacking. For the patients’ part, they may feel that without the legitimacy conferred by a recognized medical diagnosis, the severity of their symptoms and even their own sincerity will be doubted.

What these tensions suggest is that rather than debating whether or not these syndromes and functional disorders are “real” or “legitimate” medical conditions, scrutiny should instead focus on conventional models of disease and the standard assumptions that patients and physicians bring to the medical encounter. The presumption that disabilities and functional limitations are less real in the absence of an independently observable disease entity reflects an assumption that all real medical conditions must follow the same paradigm. Yet the possibility remains that the modes of explanation appropriate to illnesses like infections and poisons may not be applicable to more complex complaints, such as those involving interactions between mind, body and culture. The tensions felt by many physicians and patients suggest that new modes of characterizing medical problems are needed.

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Genetic Disease, Genetic Testing, and the Clinician

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Modern medicine emphasizes treatment of the sick. It is often said that the widespread genetic testing soon to follow the completion of the Human Genome Project will usher in a new era of preventive medicine. Such changes require new ways of thinking, however. For example, there may be nothing clinically wrong with a healthy patient who requests genetic testing, even if the tests reveal disease genes. Since all individuals have genetic skeletons in their closets, it is important to be careful not to confuse having disease genes with having the diseases that they cause. Unfortunately, many in the public have adopted a kind of genetic determinism that sees genes as destiny; for example, having the gene associated with colon cancer means they will develop colon cancer. Physicians tend to be more careful, yet even they are not immune to subtle versions of genetic determinism.

One example of this is the uncritical categorization of certain diseases as “genetic.” In fact, an adequate concept of genetic disease is extremely difficult to come by. The simplest notion would require a 1:1 correspondence between a disease and its genes, but this is the exception rather than the rule. For example, cystic fibrosis (CF) is often put forward as a good example of a genetic disease, since it seems to result from mutations in a single gene, CFTR. Even in this case, however, the exact relationship between CFTR mutations and disease is not clear, as virtually every possible combination of sweat chloride test results, genetic test results, and symptoms has been observed.1 If a patient presents with the classic symptoms of CF and is found to have a mutation in the CFTR gene, the physician might understandably infer that the mutation caused the disease. But if an asymptomatic patient is discovered to have a CFTR mutation, it is unclear what this means. The physician might tell the patient the gene is abnormal and that he or she is likely to develop specific symptoms, but it is not really known whether even this qualified prognosis is true. This is because current knowledge of CF is based largely on studies of people who have the disease. We do not yet know how likely it is that someone would test positive for a CFTR mutation and remain healthy all his or her life. The claim that these situations are rare is thus based on an implicit genetic determinism and is not supported by data. Such assumptions may be harmful if, for example, they cause patients to make inappropriate treatment decisions. Nevertheless, because they remain tacit and thus escape critical scrutiny, assumptions like this are quite common.2

Moreover, the causal role of a specific gene is typically less direct than in CF. Suppose a woman from the general population, who is not otherwise at risk for breast cancer, tests positive for the BRCA1 gene. She is likely to be told that she has an 85% lifetime risk of developing breast or ovarian cancer. This is serious news, especially since it is offered in the seemingly unassailable, quantitative language of science. However, the 85% figure is actually based on cancer-gene covariance in families that have an unusually high incidence of early-onset breast cancer.3 It is an open question to what extent such families accurately represent the general population. In these circumstances, should the patient even be given a numerical estimate of risk, knowing that quantitative results relayed by a physician are often taken as absolute? Physicians may have an intuition that information is always harmless and desirable, but this is defensible only if the information can be used to make appropriate decisions. Surely, if the results of a genetic test cause a patient to undergo what later turns out to be an unnecessary radical mastectomy, the test information can not be considered harmless.

One thing physicians can do for patients is direct them to trained genetic counselors who will discuss the desirability and interpretation of genetic tests in detail. Unfortunately, the dearth of trained counselors and the pressures of managed care, this is not always possible. Primary care physicians forced to deal with these new tests thus have a 2-fold responsibility. First, they must keep themselves up to date concerning the complexities of gene-disease relationships, taking careful note of what is still unknown as well as what is known. Second, they must communicate this information openly and honestly with the patient. In particular, physicians must be prepared to debunk uncritical notions about the causal power of genes and even to argue against the advisability of genetic testing, especially in situations where there is no clear differential treatment based on what the tests reveal. It is better to admit to patients that medical science cannot yet answer their questions than to offer a false sense of certainty through tacit endorsement of information of uncertain quality.

References


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In the Twilight: Life in the Margins Between Sick and Well

Anonymous

My mother learned she had breast cancer in her mid-30s. When she was 39, she was diagnosed with a second primary cancer, this time of ovarian origin. Her dying was protracted and particularly horrific. She was admitted to the hospital late one spring because of “metastatic carcinoma, known recurrent cerebral metastasis, progressive weakness, and inability to void.” As the attending physician wrote in her discharge summary, “In view of the untreatability of the situation, she was approached for her comfort only, given pain medication as required for relief of discomfort, and died quietly.”

I knew long before my first semester course in medical genetics that my family history and my Jewish background did not bode well for my own risk of acquiring the same diseases that killed my mother. In a vacation break during my first year of medical school, I sat in an office on the 6th floor of a major city hospital and decided that I needed to consider knowing for sure. After several sessions with a genetic counselor, I wrote a check, watched as a nurse drew 2 tubes of blood, and went home to wait. Four weeks later, in the same office, I held my husband’s hand as the geneticist sat down with us and said succinctly: “I’m sorry, but in the same office, I held my husband’s hand as the genetic counselor, I wrote a check, watched as a nurse drew 2 tubes of blood, and went home to wait. Four weeks later, in the same office, I held my husband’s hand as the geneticist sat down with us and said succinctly: “I’m sorry, but your test showed a mutation in the BRCA1 gene.”

Language describes possibility; medical language often implies distinct outcomes. Whole lives are altered by a few words coming from the person with the stethoscope and the white coat. Cancer. Heart disease. AIDS. These are words and more than words. They are also metaphors that suggest distinct futures. Some of these futures are determined on the basis of evidence: women with my specific mutation—and with a family history as strong as my own (my mother was not the only member of her family to be diagnosed with these cancers)—are estimated to have a lifetime risk of breast cancer that approaches 85%; the risk for ovarian cancer nears 44% over the lifetime. Other notions of futures are determined by one’s subjective experience. Breast and ovarian cancer have a particular meaning for me because of how the diseases affected my mother, and because of the tremendous suffering I associate with them. I fear a specific type of dying.

What does one do when one is a healthy medical student, enjoying classes, in a great relationship, with beloved friends and a profession that brings great happiness—and suddenly one learns that one is carrying a genetic mutation that is associated with a greatly increased cancer risk? For a long time after I learned my status, I grieved for the life I once thought I had. I kept trying to remind myself I was not actually sick, although the line between health and illness certainly had become blurred. It is true that I am perfectly healthy, yet 2 weeks after learning my status I had my own oncologist and breast surgeon, underwent my first mammogram and breast magnetic resonance scan, and had my first biannual transvaginal ultrasound. I was weighing the options of having children (My doctors: “If you want them, you need to have them soon”), of prophylactic surgery (“Your best option at this point is prophylactic bilateral mastectomies and oophorectomies by age 34”), and worrying about the uncertainty ahead (“Surgery will only decrease your inherited risk by 90% for breast cancer, and 50% for ovarian cancer, because women with your mutation are also prone to primary peritoneal carcinoma after oophorectomy”). It was time to reorder my priorities. Quickly approaching the end of my 20s, I made the choice to postpone my medical training to start a family. I was suddenly part of the world beyond the white coat—the world of most people, who can no longer look at disease as a professional challenge, but as a potential path for their own future. Between the sick and the well is the uncertain, and that is where I now found myself.

Sometimes I am filled with a deep longing for the time before I knew my status, when the possibilities ahead of me were not circumscribed by the realities of my situation. I do not have cancer. I am a healthy young woman. Yet I can no longer make the same choices I might have made when I lived under the illusion that I was young enough, or healthy enough, to be untouchable. In truth, all of us live in the twilight; none are promised a life of infinite length. In spite of the order scientific medicine promises us, there are no real promises to be made—there is only life as it is now, with no certainties about how it will be tomorrow. This is what it is like to live with a genetic predisposition, and I suspect that this is also what life is like from the vantage point of one with a serious illness; but most importantly, this is actually what life is like for us all. From the moment we are born we are on a path toward the eventual end of our physical presence here on earth. And none of us—not the woman with cancer, nor her daughter with the increased risk, nor the medical students in the prime of health who have no looking glass into what the future holds—can escape that. We can only be emboldened by our part in creating what might become.

*Author’s actual name withheld at her request.—Eo.
DISEASE, ILLNESS, AND DEATH HAVE CAPTIVATED THE LITERARY IMAGINATION SINCE ANTIQUITY. A STRIKING EXAMPLE OF ILLNESS AS A FUNDAMENTALLY SOCIAL STATE, IN WHICH BEING VIEWED AS SICK SEPARATES ONE FROM THOSE WHO ARE HEALTHY, CAN BE FOUND IN SOPHOCLES' *PHILOCTETES*, THE DRAMA OF AN ARCHER WHOSE PUTRID WOUND CAUSES HIS COMRADES TO ABANDON HIM ON THE WAY TO TROY. WHILE THE SICK ARE NO LONGER OSTRACIZED IN SUCH AN EXTREME MANNER, ILLNESS STILL CONFRONS A UNIQUE ROLE THAT MERITS CLOSER SCRUTINY: THE SICK ROLE.

Physical or mental suffering garners the label of sickness when others—most powerfully, physicians—acknowledge that it impairs social functioning. In the field of medical sociology, "disease" describes biomedical changes in health that it impairs social functioning, while "sickness" encompasses the social dimensions of illness—how being ill affects one's role in society. ¹

People in the sick role may be excused from other obligations, such as work, school, or parenting. Society is willing to legitimize sickness as long as one follows culturally determined conventions of being ill, such as trying to regain health as quickly as possible, or accepting medical help in overcoming the illness. ² Those who abuse the sick role by feigning illness for a secondary gain strain this social contract. In contrast, people who have a disease with no immediately apparent signs may deny the sick role; in addition, these patients may refuse to comply with therapy because they do not appear sick.

As cultural paradigms and norms of illness shift over time, the socially defined parameters of what it means to be sick change. The fiction, poetry, and drama of a given era provide a useful measure of historical perceptions of sickness, and being alive to these representations can deepen an understanding of how the societal underpinnings of the sick role have evolved. Stories of illness become heavily imbued with social judgments with the advent of Christianity in the Middle Ages, when illness revealed as well as punished human iniquity: Boccacio's account of the Black Death underscores how necessary a World of Pains and troubles is to school an Intelligence and make it a soul?³ before dying of tuberculosis at the age of 25 years.

The discovery of the tubercle bacillus and other microorganisms in the 19th century revolutionized the popular understanding of illness. A new space between patient and disease, in which aggressive cellular "invaders" could breach immune "defenses," led to the use of military metaphors in descriptions of disease. ⁴ This not only resulted in more rigid definitions of disease but also focused therapeutic options on quantitatively measured outcomes. The success of the Pulitzer Prize winning play *Wit* underscores how necessary it is to remember that treating a disease includes caring for a person. The protagonist of the play, a professor of English with terminal ovarian cancer, realizes with heart-breaking clarity that the rational and remote approach of the successful academic can fail to acknowledge that to be sick is to suffer. ⁵

One of the dangers of the detached clinical gaze of modern medicine is that the patient's body becomes only a locus for surveillance, investing the medical profession with the authority to decide who is sick—and how sick—based on signs and symptoms. However, sickness can occur without a known etiology. Syndromes such as chronic fatigue and fibromyalgia⁶ defy easy categorizations of illness and remain difficult to treat; those afflicted by them can be disbelieved and marginalized by physicians and lay people alike.

In approaching sickness, physicians should remember that what ultimately cures Philoctetes of his malady is not medicine, but a young Greek's "recognition of his common humanity with the sick man."⁷ The archer goes on to become a hero in the battle of Troy—the sick man wins his war, both literally and figuratively, because of the human touch.

**REFERENCES**