

## IN THE SERVICE OF GOOD WRITING

# What's in a Name? (Diagnosis)

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Shakespeare's Juliet asked, "What's in a name? That which we call a rose / by any other name would smell as sweet." (*Romeo and Juliet* Act II, Scene II, Line 46-47) But Juliet was a love-struck teenager. The Chinese philosopher Confucius gave advice that is more suitable for medical writers: "If names be not correct, language is not in accordance with the truth of things. If language be not in accordance with the truth of things, affairs cannot be conducted successfully." (*Analects* Section 3, Part 13) A diagnosis is a kind of name—a name that must be used accurately. Real estate professionals tell us that the value of a property is determined by 3 factors: location, location, and location. Likewise, the care that a patient receives will hinge on diagnosis, diagnosis, diagnosis. As medical writers, we need to understand what a diagnosis is, and what the process of diagnosis entails.

When I am sick, I want my clinician (physician, nurse practitioner, or physician assistant) to give me 4 vital pieces of information:

- What is wrong with me? (diagnosis)
- Why did this happen? (etiology)
- What can be done about it? (therapeutics)
- What will happen to me because of all this? (prognosis)

Sometimes, the answer to the first question will practically dictate the answers to the rest of the questions. If I receive a diagnosis of influenza, it means that my clinician thinks that my illness resulted from infection with an influenza virus. This diagnosis will affect how my illness will be treated and the predictions that will be made about what will happen to me next.

### WHAT IS A DIAGNOSIS?

The word *diagnosis* came from ancient Greek. *Dia-* came from a root that meant "through, in different directions, or between," but could also mean "thoroughly," and *gnosis* meant knowledge. So, the word *diagnosis* implied a form of knowledge that was chosen from among a set of possibilities. Likewise, *pro-* meant "before," so a prognosis meant foreknowledge—a prediction. Many things can go wrong with the human body. A diagnosis implies that the clinician

knows something about what has gone wrong in a particular case. A prognosis is the clinician's educated guess of what will happen in the future.

*Diagnosis* is a noun, but it has been transformed into the verb *to diagnose*. Thus, diagnosis can be viewed as a process, not just as the label that is applied to the patient's illness at the completion of the process. *Differential diagnosis* can mean the process of choosing from among 2 or more conditions that produce a similar clinical picture. However, a *differential diagnosis* can also mean the list of possible explanations for a patient's condition. Each item on the list is a diagnostic differential.

The ancient Greek philosopher Plato suggested that knowledge is a justified true belief.<sup>1</sup> A diagnosis is a belief that is (one hopes) justifiable. Ideally, there would be some consensus within the scientific community and the medical profession about how each diagnosis can or should be justified. Some diagnoses can be justified by some sort of pathognomonic sign or symptom (ie, something that is distinctively characteristic of that condition). Others are justified by the result of imaging or a laboratory test. The justification could be a formal case definition—a set of criteria that must be met. Yet even if a diagnosis seems to meet the appropriate criteria, it could still be wrong.

Medical students are taught heuristics for diagnosis. A heuristic is an approach to problem-solving. The word *heuristic* came from the Greek verb *heuriskein*, which means to discover. One approach is to consider the most common conditions first—yet rare diseases do occur, albeit rarely. Another approach is to try to find a single diagnosis that explains every aspect of the patient's condition, because the simplest explanation is most likely to be correct. This is in line with the principle called Occam's Razor: "entities should not be multiplied beyond necessity." Yet Occam's Razor must be balanced against Hickam's Dictum: "a patient can have as many diseases as he damn well pleases."

Clinicians can make errors in diagnosis for countless reasons. However, those errors all fall into 2 basic categories:

- A *misdiagnosis* means that the patient was given a diagnosis of a condition that he or she did not really have. A misdiagnosis is easy to make when several different

diseases produce similar signs and symptoms. A misdiagnosis can also result from a false-positive result from a diagnostic test.

- A *missed diagnosis* (also known as a *failure to diagnose*) means that the clinician failed to provide the correct diagnosis. This problem can be due to a false-negative result from a diagnostic test. It can also result from a failure of imagination called premature closure of the differential: a failure to include the correct diagnosis among the differentials being considered.<sup>2</sup>

These individual errors can add up to a systematic under- or overdiagnosis of a disorder.

- *Underdiagnosis* means that the condition is being missed in a significant proportion of cases. Under-diagnosis is common when the population has poor access to medical care, when there is poor awareness of the condition among the population and/or the clinicians, or when the correct diagnosis is hard to make.
- *Overdiagnosis* means that too many patients are being given the diagnosis. The diagnosis is being given to patients who do not have the condition at all and/or it is being given to patients whose cases are too mild to warrant medical attention. Overdiagnosis can be a result of disease mongering, which is the practice of inappropriately widening the diagnostic criteria for a medical condition and aggressively promoting public awareness, to expand the market for diagnostics and treatments.

For the individual patient, an error in diagnosis can lead to bad treatment decisions and bad outcomes. On a societal level, these individual errors may result in poor public health and large-scale misallocation of resources.

## DIAGNOSES ARE LABELS

A diagnosis is a label that is applied to a patient's illness. This labeling allows clinicians and scientists to sort cases of illness into categories and to sort patients into groups, which makes it possible to do clinical studies. Some diagnostic labels imply the kind of knowledge that the clinician believes that he or she has about the patient's condition. Others refer to the process through which the clinician's belief was justified (see Types of Diagnosis).

## Types of Diagnoses

**admitting diagnosis**—the diagnosis reported by the clinician who decided that the patient needs to be hospitalized. The admitting diagnosis may be tentative because the patient has yet to undergo examinations and testing. The admitting diagnosis is recorded for administrative purposes.

**biologic diagnosis**—a diagnosis based on a test performed on a laboratory animal (eg, inoculation of a cell culture, egg, or laboratory animal with a patient's specimen to isolate a virus).

**clinical diagnosis**—a diagnosis based on the patient's symptoms and clinical signs at examination, as well as the patient's history. Symptoms are phenomena (eg, pain) that are subjective (ie, only the patient can observe them). Clinical signs are phenomena (eg, swelling, heat, or redness) that are objective (ie, the examining clinician can observe them). The term *clinical diagnosis* can also mean a diagnosis that was based on all the information (including laboratory and imaging results) available during the patient's lifetime, as compared with the autopsy findings.

**cytologic diagnosis**—diagnosis based on examination of exfoliated (shed) cells.

**definitive diagnosis**—a final diagnosis based on the results of confirmatory testing.

**diagnosis of exclusion**—a diagnosis made by eliminating all of the other known differentials.

**diagnosis ex juvantibus**—a diagnosis based on the result of treatment, such as antibiotic therapy.

**direct diagnosis**—a diagnosis made by observing structural lesions or pathognomonic signs or symptoms. Pathognomonic means distinctly characteristic of a particular disease.

**discharge diagnosis**—the diagnosis recorded when the patient leaves the hospital.

**etiologic diagnosis**—a diagnosis that implies the underlying cause of an illness (eg, a diagnosis of measles means that the clinician believes that the patient has a measles virus infection), or the process of identifying the underlying cause of the illness (eg, isolation of a particular strain of bacteria in the case of an infection).

**faux diagnosis**—a false diagnosis that is made for some administrative purpose.

**laboratory diagnosis**—a diagnosis based on the results of tests or examinations performed in a laboratory on specimens (eg, of tissue or bodily fluids) taken from the patient.

**niveau diagnosis**—localization of the exact level (*niveau* in French) of a lesion, such as in the spinal cord.

**pathologic diagnosis**—a diagnosis based on the observed presence of structural lesions. The clinicopathologic method meant comparing the records of the patient's condition during lifetime with the autopsy findings.

**physical diagnosis**—a diagnosis based on looking at (inspection), feeling (palpation), tapping on (percussion), and listening to (auscultation) the patient's body.

**presumptive diagnosis**—a diagnosis based on the known probability of a known condition, such as the antemortem diagnosis of a disorder that can be confirmed only at autopsy. (Compare with **definitive diagnosis**.)

**provocative diagnosis**—a diagnosis based on a provocative test, such as exposure to an allergen to confirm an allergy.

**radiologic diagnosis**—a diagnosis based on the results of imaging, such as x-ray imaging, computed tomography, or magnetic resonance imaging.

**serum diagnosis**—a diagnosis based on testing of serum (the fluid left after a blood sample coagulates).

**syndromic diagnosis**—the recognition of a group of signs and symptoms that characterize a clinical condition that might not always result from the same cause. The word syndrome came from the Greek for "occurring together." If research shows that a particular syndrome always results from a particular cause, the syndrome becomes a disease.

**wastebasket diagnosis**—a vague and possibly meaningless diagnosis given when a patient's condition cannot be easily classified.

**working diagnosis**—the differential (or set of differentials) that is considered most likely to be the true diagnosis.

## Cause or Effect?

Diagnoses are labels that allow us to sort cases. However, diagnostic labels themselves can be sorted into categories, such as etiologic (ie, pertaining to cause) vs syndromic (ie, pertaining to the pattern of observable effects). Some diagnoses (eg, the classic exanthems) start off as a syndromic diagnosis, only to become an etiologic diagnosis as its cause is revealed. An exanthem is an acute, sudden illness that involves a widespread rash along with other symptoms (eg, fever and headache). By the early 10th century, the Persian physician Abū Bakr Muhammad ibn Zakariyyā al-Rāzī (known in the West as Rhazes) knew that smallpox and measles were separate diseases. Yet the causes of those 2 diseases remained unknown for another millennium. In the early 20th century, pediatricians assigned numbers to the most common childhood exanthems (Box: The Classic Exanthems).<sup>3</sup> These disorders began as clinical syndromes. But in the 20th century, researchers found that each of these classic exanthems (except for fourth disease, which might not exist as a separate entity) was due to a different infectious agent. Thus, 5 out of the 6 classic exanthems became diseases, and the diagnoses changed from syndromic to etiologic diagnoses.

### The Classic Exanthems

In the early 20th century, pediatricians numbered the most common childhood exanthems (diseases that caused a rash and other symptoms, such as fever)<sup>3</sup>:

- **First disease** was rubeola (measles)
- **Second disease** was scarlet fever
- **Third disease** was rubella (German measles)
- **Fourth disease** was Duke's disease (unknown today)
- **Fifth disease** (erythema infectiosum or slapped cheek syndrome) is still called fifth disease
- **Sixth disease** was roseola infantum.

Measles, rubella, fifth disease, and roseola are caused by viral infections. Scarlet fever results from a bacterial infection (group A *Streptococcus*).

## Names and Numbers

Medical conditions often had 2 names: the popular name and the scientific name (eg, measles and rubeola). A medical condition was often named after the person who first described it in the medical literature (eg, Cushing's syndrome). Today, AMA style requires us to drop the 's' after the person's name (Cushing syndrome). Sometimes, the name of a condition changes. For example, *dementia praecox* became *schizophrenia*, and *manic-depression* became *bipolar disorder*. Sometimes, the name is changed because the cause is discovered (eg, "De Vivo syndrome"

became "glucose transporter type 1 deficiency syndrome"). In May 2015, the World Health Organization recommended that new diseases should not be named after persons or refer to geographical locations, animals, foods, cultural or occupational groups, or populations or industries and should not include terms that incite undue fear (eg, *fatal*).<sup>4</sup> If you are uncertain of the current name of a condition, you might look it up under the Medical Subject Headings thesaurus of the National Library of Medicine (<https://www.ncbi.nlm.nih.gov/mesh>). Genetic disorders are catalogued in Online Medical Inheritance in Man (<https://www.omim.org/>), a joint effort between the National Library of Medicine and Johns Hopkins University.

To make it easier to record medical diagnoses and compile statistics on public health, the World Health Organization introduced the International Classification of Diseases and Related Health Problems. The 11th edition, which is currently being implemented, provides roughly 55,000 unique alphanumeric codes for diseases, injuries, and causes of death. For example, the code for a distal fracture of right radius with dorsal tilt and joint involvement after falling on the sidewalk would be NC32.50 & XK9K & XJ5GS / PA60 & XE53A.<sup>5</sup> Similarly, the American Psychiatric Association's *Diagnostic and Statistical Manual*, currently in its fifth revision (DSM-5), is used in the United States for coding mental disorders.

## Binaries, Scales, and Spectrum Diagnoses

Many common diagnoses involve some sort of binary. A binary is some variable that can have one of 2 values: on or off, yes or no, alive or dead, male or female, pregnant or not pregnant. Yet in practice, even a yes-or-no question has 3 possible answers: yes, no, and no answer. Although a binary involves 2 categories that are separate conceptually, the boundary between the 2 categories may be fuzzy in practice. Thus, it can be hard to classify some individuals. For example, a child's biological sex is a diagnosis that used to be made at birth but is now often made much earlier, during an ultrasound examination. Nearly every human being can easily be classified as either male or female on the basis of their phenotype (their anatomy) or their karyotype (presence or absence of a Y chromosome). Nevertheless, there are a few cases (about 1 in 5,500 live births)<sup>6</sup> in which a newborn's external anatomy does not match the chromosomal sex or cannot be easily classified. These rare cases do not prove that sex is nonbinary. Rather, they just illustrate that the boundary between the 2 categories is slightly fuzzy.

Even if the boundary between categories is a single clear criterion, some cases can be misclassified if the criterion is

hard to document. For example, a woman is either pregnant or not pregnant. By definition, a pregnancy begins when an embryo implants itself on the inner lining of the uterus. Yet the pregnancy cannot be detected by blood or urine tests until a few days after implantation.

Many diagnoses depend on some sort of measurement (body weight, blood pressure measurements, IQ scores) that falls somewhere on a scale. This kind of diagnosis raises important questions: is the measurement accurate? Is the scale meaningful? Where should the lines between categories be drawn? Consider the diagnosis of obesity. Obesity means that the body contains too much fat. Yet body fat content (adiposity) is rarely measured directly. Instead, clinicians measure body weight (in kilograms) and divide it by the square of the height (in meters) to calculate the body mass index. Body mass index provides a useful rule of thumb. However, bodybuilders can have a body mass index in the obese range despite having very little body fat. Also, the body mass index tends to underestimate body fat in short people and overestimate it in tall people.<sup>7</sup> Yet even if you developed a better metric than body mass index, you would still have to decide where to draw the lines between the categories of underweight, normal weight, overweight, and obese. Even if you draw those lines in reasonable places, someone whose weight falls close to the boundary between categories could switch from one to the other and back within the course of a single day.

Sometimes, differences in magnitude can seem to produce differences in kind. If a patient receives a different diagnosis than another patient received, the difference in labeling seems to imply that the 2 patients have different *kinds* of problem. Yet the 2 patients might have the same underlying kind of problem but at different levels of *severity*. A spectrum disorder is a set of conditions that look different but are believed to represent the same underlying problem at different levels of severity,<sup>8</sup> just as light at different wavelengths looks like different kinds of light.

### Psychiatric Diagnoses and the Biopsychosocial Model

Psychiatric diagnoses pose a particularly thorny set of problems. In general, a mental illness can be any problem that involves one or more of the following: cognition (perceptions and thoughts), emotion (feelings), or action (behavior). To be considered a mental disorder, a mental illness must cause distress or disability, must not be an expectable or culturally sanctioned response to a particular event, and must not be primarily a result of social deviance or conflict with society. Unfortunately, these concepts are broad and lack precise boundaries.

### Key Points

When writing about any diagnosis, think carefully about what that diagnostic label means.

- Is the diagnostician asserting something about the cause of the patient's condition? Or does the diagnosis simply mean that the signs and symptoms follow a familiar pattern?
- How is that diagnosis justified? Is it supported by physical examination findings, radiologic findings, or the results of laboratory testing?
- How meaningful are the criteria for making the diagnosis, and how reliably are they being applied?
- How severe must a case of a condition be to qualify for the diagnosis? How were the boundaries between normal and abnormal values determined?
- Is the diagnostic label saying something true about the patient? Or is it sending a misleading message that undermines the patient's interests?

Some mental disorders may have a purely biological cause. Others seem to result from a combination of biological, psychological, and social causes. The biopsychosocial model accepts that all 3 kinds of causes can contribute and may be interrelated. Psychiatry is part of medicine because many mental disorders are believed to have at least some basis in biology. Yet once a particular condition is shown to have a clear biological basis (eg, psychosis due to anti-*N*-methyl-*D*-aspartate receptor encephalitis), that condition tends to get reclassified as a neurologic disease as opposed to a mental disorder. As a result, psychiatry is left with a set of conditions whose etiology is unknown or is so complicated that it might never be untangled. For this reason, psychiatric diagnoses are generally syndromic (ie, based on a pattern of signs and symptoms) as opposed to etiologic (ie, a statement of cause). Thus, psychiatrists use diagnostic testing to rule out medical diagnoses, as opposed to confirming psychiatric diagnoses.

When trying to figure out the cause of a mental disorder, some psychiatrists prefer to err on the side of biology whereas others prefer to err on the side of psychosocial causes. Medical writers should be alert for either kind of bias. In particular, medical writers should be skeptical whenever a diagnostic label suggests that a patient's physical symptoms are psychological in origin.<sup>9</sup> Conversely, medical writers should also be skeptical of the use of medical-sounding diagnoses for social or educational problems. In 1851, Samuel A. Cartwright argued that slaves who tried to escape from slavery were exhibiting a mental illness that he called *drapetomania*.<sup>10</sup> Today, children who have trouble with the "3 R's" (reading, writing, and arithmetic) are routinely given the diagnoses of dyslexia, dysgraphia, and

dyscalculia. This use of these medical terms implies that the cause of the problem is in the child's brain, when the cause of the child's poor academic performance could very well be in the child's schooling (eg, bad methods for teaching reading and math and a refusal to teach penmanship). If we are using a medical label for a problem whose cause is pedagogical, not medical, then (to paraphrase Confucius) our language is not in accordance with the truth of things, and our affairs cannot be conducted successfully.

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