The hazards of diagnosis

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Received 21 March 2010 and in revised form 29 April 2010

Summary

Background: Making a diagnosis is perceived as a climax of clinical effort. Little consideration is being given to potential down sides after the diagnosis is achieved.

Methods: The authors’ personal experience, extensive discussions with peers and a search of the literature.

Results: Several common and significant pitfalls after making a diagnosis were identified. They include ‘locking’ and disregard of further data pointing to possible diagnostic error; pursuing a diagnosis when further information has little practical significance; a severe impact of the news on the patient; a ‘stigma’ effect; loss of the patient’s individual narrative; stopping the diagnosis short of the full pathogenesis or of its finer characterization; disregarding additional medical issues; losing interest in the patients once the diagnostic problem is solved; and assuming full control of future decisions at the expense of the patient’s autonomy.

Conclusions: Physicians need to be cognizant of the impact and significant potential for harm associated with closure of diagnosis and its delivery to their patient. Humility, double-checking and sensitivity to the patient’s predicament may improve quality and communication and prevent ‘the hazards of diagnosis’.

The quest for a name, a diagnosis, remains one of physicians’ major goals. Naming a condition affords the doctor complete control, an anchor in a sea of endless possibilities, a port out of which patient management can safely depart. Definitely it enables us to proceed and make optimal, evidence-based decisions whereas without it we feel lost and may experience overt uneasiness or anxiety. Thus, the quest for diagnosis is often at the center of attention. The time-honored clinicopathological conference (CPC) is devoted to it as are many ‘clinical problem-solving’ articles, quizzes and case reports. An astute diagnostician enjoys recognition and the admiration of his or her peers. The diagnosis is the uncontested jewel in the crown of physicians’ efforts.

With so much emphasis on the diagnosis, physicians may hardly be aware that there may also be significant down sides to making a diagnosis. The possibility that making a diagnosis may also incur adverse effects for the patient is seldom seriously considered. The trends in modern medicine of immersion in laboratory science at the expense of clinical skills, professional values and teaching capabilities¹,² have made the identification of the ‘Hazards of diagnosis’ particularly timely. We identify and draw attention to several common potential pitfalls after making a diagnosis to increase physicians’ awareness, avoid prevalent errors and prevent patient harm.
The diagnosis is erroneous but stops all diagnostic efforts

Once the disease responsible for the patient’s symptoms and signs is identified, physicians’ concerns quickly shift to its management. An established diagnosis creates in the physician a sense of security. Further tests are often not requested and if they are done, or results of prior test are received, they may not be given full, open-minded consideration. Hence, ‘locking’ in the presumptive diagnosis (‘premature closure’) may persist despite bits of evidence suggesting otherwise which is a common source of diagnostic error. Considering the diversity and frequency of obstacles to proper diagnosis including time constraints, bias in decision making, incidental findings, physician overconfidence and patient-related factors—it is hardly surprising that diagnostic errors often occur and need to be continuously considered.7

Humility arouses more trust than overconfidence and should prompt continuous checking and re-checking of the data to ensure the appropriateness of conclusions.

A presumptive diagnosis is pursued by potentially harmful tests when full verification will have no impact on the patient

Giving a name to the illness is only a tool. The real goal is to find the optimal way to understand the illness and provide proper help and support to the patient. The emerging quality of life construct highlights outcomes and allows for palliative care in selected cases.8 Thus, an accurate diagnosis may not always be necessary and patients may be spared the pain, risks and anxiety of a protracted workup whose practical yields (‘utility’) are questionable.9,10 Cost-benefit balance is an additional important down side of unwarranted diagnostic procedures. Although diagnostic futility is hardly discussed in the literature, there is little doubt that it exists.

The patient’s best interests should direct decisions and ‘academic curiosity’ or tests of doubtful significance need to be restrained.

The diagnosis has a severe impact on the patient

Learning the diagnosis may have an overwhelming effect on the patient.11,12 This applies to many diverse conditions, which are often chronic, incurable and worsen with time. Not only malignancies but many benign disorders may carry a bad prognosis of considerable morbidity and possibly premature mortality, as in congestive heart failure or rheumatoid arthritis. Even disclosures of stable treatable conditions such as diabetes, migraine or irritable bowel syndrome constitute a significant burden and may have a serious effect on the patient’s quality of life.13 Moreover, the ever-present risk of recurrence after recovery from cancer, a vascular event or chronic obstructive pulmonary disease (COPD) exacerbation constantly looms over the patient who is often painfully aware that any minor symptom, any day, may herald recurrence and a dramatic change. Contents such as loss of good health, uncertain survival, prospects of possible pain or restriction, the need of further tests or treatments, expected changes in body image, as well as personal and financial implications may cause confusion, anxiety or depression. Some patients may actually feel guilty of causing their own disease. Physicians’ sensitivity to the patient’s predicament seems particularly important since in the circumstances, patient’s understanding and ability to make informed decisions may become significantly compromised,14 and having to pick among many treatment options may seem an overwhelming burden.15

Effective empathy by the physician is invaluable for the patient and can be cultivated by the simple trick of looking at things from the patient’s point of view. Physicians who present to the patient and family as partners, providing hope while preserving honesty16 will successfully support their patients and enable coping.

The ’stigma’ effect

The mere pronunciation of a diagnosis may not only ‘brand’ the patient in his or her own eyes but also carries a ‘stigma’ for the family and caregivers that can have far-reaching consequences.17 Illnesses that have unequivocal external manifestations apparent to all (such as obesity, depression, Parkinson’s disease or psoriasis) demonstrate the potential high impact of the ‘stigma’ effect. Even medical professionals are not immune and tend to show lesser regard or low respect for patients with certain conditions.18,19 Patients whose behavior had a dominant part in the pathogenesis of illness (alcohol abuse, human immunodeficiency virus (HIV) infection, etc.) may be especially vulnerable. Guilt, shame and depression were prevalent after the diagnosis of lung cancer. Even nonsmokers felt stigmatized.20,21 Once ‘the name’ is given to the patient, it is there to stay. It may signify the
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Start of daily dependence on medications and loss forever of the freedom associated with health as well as serious future consequences. Conditions such as hearing loss or emphysema might be suspected by the patient but once a test is done and the name is pronounced, the effect is irreversible. A patient who is informed that he has emphysema often experiences heightened awareness causing him to feel breathless during activities that were just recently asymptomatic and to avoid others that he is perfectly capable of performing. Thus, the power of naming has an effect of its own. The medical label itself is frequently perceived as being more severe than its lay synonym22 and even clinical judgments may be affected by it.23

Encounters with patients who might be stigmatized need special attention and care on the part of physicians to avoid their judgment and behavior being tainted by stigma.

The diagnosis stops short of the full pathogenesis or goes too far

A patient who presented with protracted headache and low-grade fever was diagnosed with chronic meningitis due to Cryptococcus neoformans. Cryptococcosis is unlikely in a patient with an intact immune system. HIV was negative but marked CD4 depletion due to ‘occult’ Sjogren’s syndrome was found and this had important practical implications.24 Stopping too early may hinder deciphering the full pathogenesis and examples are prevalent. Sometimes, the opposite occurs and physicians get carried away in their investigations. A 75-year-old Prime Minister presented with a minor stroke. Transesophageal echocardiography (TEE) revealed a small patent foramen ovale. Although he had multiple vascular risk factors, heparin and anti-platelet agents were given, until severe intracranial hemorrhage due to cerebral amyloid angiopathy developed. With the more likely diagnosis of in situ thrombosis, the patient would have had no further investigations and sent home safely on aspirin.25

Maintaining the exact balance in each case between under-investigation and over-investigation requires careful consideration and is one of the arts of clinical medicine.

Looking for the coin under the lamp post

Focusing on the patient’s main diagnosis may well leave other, less central but no less important medical issues unattended. For example, a patient diagnosed with stroke may have associated depression or abnormal liver enzymes that remain neglected.26 Similarly, while concentrating on the primary diagnosis, important preventive measures are likely to be disregarded.27

Taking care of the patient’s health does not stop at the main diagnosis and mandates a ‘wide angle’ approach.

The doctor loses interest

Some physicians enjoy the ‘hunt’ and become immersed in the riddle of getting at the diagnosis but may lose enthusiasm, dedication and interest once diagnosis is achieved. Such aftermath is not that unexpected considering that diagnostic efforts are sharp and focused (sometimes instantaneous)28 while treatment of many conditions is prolonged, slow and often disappointing.

It is after the diagnosis that the patient needs us the most and maintaining good communication and strong patient–physician relationship29 remains utterly important.

The diagnosis fails to convey its finer biological characteristics

The farther we move from gross pathological diagnosis, the more we understand disease heterogeneity. The modern concept of ‘patient-tailored’ medicine epitomizes an evolving response to unique individual features of disease in an attempt to improve patient outcomes.30 Thus, a patient with lymphoma associated with Helicobacter pylori infection is entirely different from a patient with lymphoma and chronic hepatitis C. Breast cancer patients with BRCA mutation or HER2 receptor positivity need different treatment strategies than negative patients.31

Naming a patient’s condition should not replace recognition of finer individual biological differences that affect patient’s prognoses and response to treatment.

The patient’s individual narrative becomes lost

As long as the patient has a cluster of poorly understood symptoms and signs, they are likely to be thoroughly explored. The patient’s unique detailed ‘narrative’ gets full attention.32 Once the patient is diagnosed however, she may well become ‘that
breast cancer in room 8’. Most components of the patient’s story and suffering may become blurred, if not entirely effaced and replaced by a single lacunar term, the diagnosis. This name is identical for all patients with the disease; however, the patients are very different.

This limitation of assigning a name to a patient’s illness needs to be acknowledged and an effort made to recognize and retain the unique personal elements in each patient’s illness.

The doctor takes the wheel

With the diagnosis secure, important decisions regarding different treatment options lie ahead. The danger that the doctor would decide routes for the patient or ‘frame’ the alternatives for the patient to ‘decide’ in a way that is governed by the doctor’s values rather than the patient’s, remains very real.33 This approach significantly undermines not only patient satisfaction but also patients’ rights and health outcomes.34

Physicians need to be constantly aware that it is the patient’s legal and ethical right to be well informed and decision making must be shared.

In conclusion, the period ‘after the diagnosis’ may constitute a ‘locus minoris resistentiae’ in patient care and needs to be recognized as such. Physicians have to continue assessment of new data to verify that no diagnostic error has been made and ensure that ‘digging’ for the full pathogenesis has not stopped too early or is being carried too far. Heightened sensitivity to the impact of the diagnosis on the patient as well as continued interest in the patient as an individual with unique circumstances and narrative, not a diagnosis, cannot be overemphasized.35 Additional health issues other than the main diagnosis need addressing. Finally, decision making after the diagnosis is established is often more complex than during the diagnostic process itself.36 It remains our role to inform, counsel and guide our patient’s decisions but never to assume that we can decide for them.

Conflict of interest: None declared.

References


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