Diagnosis – Part Two

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Abstract: The problem with diagnosis centers on the distinction between illness, which features subjective elements (symptoms), and "disease," which is defined purely objectively. Diagnosis, the identification of disease, works very well to explain illness, by situating it within the accumulated body of pathological knowledge. It works much less well as a predictor of illness in the future. Yet medical practice increasingly focuses on identifying and potential disease elements before the patient is aware of them. These data are inherently ambiguous and misleading, both by identifying abnormalities which never materialize as illness ("false positives"), and by failing to detect perceptible signs of actual illness ("false negatives"). Above all, they are dangerous, by inculcating fear, reducing risk to a statistical calculation, and promoting drastic, irrelevant, and injurious treatments to "correct" them.
Keywords: Illness (subjective); disease (objective); diagnosis, identification of disease; "preventive" medicine;

5. A Disease for Every Pill

It is widely understood that the relentless pressure and at times fanatical zeal for preventive screening and the detection of subclinical abnormalities has been assiduously cultivated by the pharmaceutical industry, proceeding in parallel with the development of drugs intended to correct them, and even serving in many instances as a marketing strategy after the fact. This was already evident in the decades-old crusade against high blood pressure, beginning with the marketing and widespread use of thiazides in the 1950's. As several new generations of drugs were introduced, the disease itself has been redefined to include lower and lower thresholds of diastolic pressure that were once considered harmless, and even systolic hypertension in the elderly, which recent studies have also targeted as a risk factor for the first time, as we saw. New, aggressive treatment protocols directed against even mildly elevated pressures have required subjecting ever-larger segments of the population to lifetime drug maintenance, and the result, according to some studies, has been some lowering of the incidence and morbidity of heart attacks and strokes, but also the exact opposite in many cases where the drop in pressure proved excessive, and without any clear or fixed boundary between therapeutic and dangerously low levels.[22] Once again, the felt need for more and more aggressive treatment, in this case largely generated by the drug industry, subliminally persuaded the profession to broaden the definition of the disease itself.

Despite the measured scruples and conscientious warnings of senior clinicians, the impetus for preventive screening today recognizes few restrictions beyond the limits of the drug industry's own feverish and well-funded imagination. Just how far afield it can lead is well illustrated by the current fashion of routine cholesterol screening in young *children,* partly inspired by a legitimate concern about obesity both before and after puberty, and designed and marketed to lower the risk of atherosclerosis, heart attack, and stroke in mature years. In a disclaimer issued by a joint task force of the AMA, the American Academy of Pediatrics, and the American Academy of Family Practice, the United States medical establishment stopped just short of mandating such tests for all children, but did recommend targeted screening of tens of millions with "high-risk parents," according to the same criteria that have long been part of the conventional wisdom for adults.

> "Of the leading risk factors for coronary heart disease, 25% of children have total serum cholesterol levels above 170 mg./dl. Autopsies of children dying from other causes have found that aortic fatty streaks correlate with total cholesterol and LDL levels. Children with high cholesterol are also at risk of having high levels as adults. Many children with high cholesterol do not maintain high levels as adults, and the safety and effectiveness of treating high cholesterol has not been established. We therefore do not recommend universal cholesterol screening in children. But *children older than 2 years with a parent whose total cholesterol is 240*

or higher should be screened, as should those with a parent or grandparent who had documented MI, angina, peripheral or cerebrovascular disease, sudden cardiac death, or atherosclerosis at 55 or younger, or had coronary bypass surgery or angioplasty. Screening may also be indicated if the family history is unobtainable and risk factors are present. The Canadian Task Force holds that there is insufficient evidence to recommend routine screening in children and adolescents, and that individual clinical judgment should be exercised."[23] [Italics mine: R.M.]

Although further doubts continue to be raised about the validity of cholesterol screening even for adults, statins and other cholesterol-lowering drugs still hit the jackpot as some of the top-selling drugs in history. Yet *Medical World News* warned many years ago of serious and even fatal consequences from lowering the cholesterol below 160 mg./dl, a level previously thought optimal.

"Some leading heart researchers are calling for a change in the nation's cholesterol policy, including a retreat from universal screening and treatment of high cholesterol for prevention of heart disease. In an editorial in the journal *Circulation,* Dr. Stephen Hulley and his colleagues at UCSF wrote that the 6% of middle-aged adults with cholesterol *below* 160 are at increased risk of dying from lung and other non-colon cancers, respiratory and digestive diseases, trauma, hemorrhagic stroke, and other causes, and concluded, "The overriding ethical obligation is to do no harm. Especially when considering the long-term use of drugs for people in good health, the burden of proof falls on the proponents of intervention." [22]

As diseases are broadened, multiplied, and redefined in accordance with the drug industry's commercial priorities, even normal physiological processes become fair game for manipulation, as new abnormalities are identified, new drugs developed to correct them, and new "diseases" created as a marketing strategy for them. A familiar example is the exploitation of menopause, beginning innocuously enough with the use of estrogens to relieve hot flashes and other symptoms for a limited period, but quickly progressing to X-ray detection of osteoporosis and "osteopenia" or low bone density at an early stage, and ultimately to prescribing synthetic hormones routinely and for many years to millions of women in order to prevent hip and spinal fractures, cardiovascular disease, dementia, and a host of other common ailments of middle and later life.

The unprecedented scale of this sales pitch, and the new diagnosis that appeared to justify it, goaded Susan Love into writing another eloquent and timely dissent, "Sometimes Mother Nature Knows Best," in which she spoke out clearly and forcefully against the practice of long-term hormone-replacement therapy, and the commodification of the female life cycle itself as a fit target for the drug industry and its self-serving redefinition of menopause as a disease requiring treatment:

"The pharmaceutical industry and the medical profession have discovered a new disease: menopause, or estrogen deficiency disease. Women with hysterectomies may want to take hormones until the natural age of menopause, while others have troubling symptoms like hot flashes and insomnia that warrant treatment. No one argues that short-term use is dangerous. But the push is on to use these drugs long-term in the name of "disease prevention." *The American College of Obstetrics and Gynecology has recommended that every post-menopausal woman should be on hormones for life unless she has a compelling reason not to be.*

"This sweeping recommendation was based on inadequate evidence. *Menopause is no disease, but a normal part of life. A woman's ovaries don't shut down: they continue to produce hormones well into her 80's. Synthetic hormones don't replace something that is missing, but add something that is not naturally there.*

"Pharmaceutical companies realize that for marketing it is smarter to emphasize diseases than hormones. In this they are helped by the medical profession, which has recently redefined osteoporosis. This term once referred only to actual fractures caused by the thin bones of old women, but now it is defined as low bone density. This is like telling someone with high cholesterol that they have heart disease.

"Women are encouraged to have bone density tests just the same as mammograms or Pap smears. The result is an epidemic of healthy 50-year-old women being diagnosed with osteoporosis, even though women on average don't have hip fractures until they turn 79. Someone once said that if you're healthy, you haven't had enough tests done yet. We need accurate information and must be on guard lest vested interests sell us a bill of goods."[25] [Italics mine: R. M.]

It is notable that Dr. Love wrote this article solely on methodological grounds, years before the extra cases of breast cancer finally discredited long-term estrogen therapy and confirmed what many had always suspected but dared not oppose publicly. Unfortunately, her arguments went unheeded for so long that the drug company's vast profits were unaffected, and when hormone-replacement therapy did go out of fashion, the conquest of menopause continued uninterruptedly, according to a thoroughly predictable sequence. By then the industry had already produced and marketed a new generation of even more potent and dangerous drugs, the biphosphonates, which similarly claim to prevent fractures by increasing the bone density, but this time

by playing with the critical and still poorly-understood mechanism of bone formation, and with calcium and phosphorus metabolism in particular, which influences and therefore has the power to threaten the structural integrity of the body as a whole.

The wholly predictable recycling of such patterns impelled Bernard Lown, the Nobel Prize-winning cardiologist and humanitarian, to question the need for early diagnosis in all its forms, and the virtually limitless multiplication of diseases that generally accompanies it.

> "Medicine has expanded into almost all facets of human existence, including conditions that do not cause symptoms or impair life, but indicate potential illness in the future, such as high blood pressure, blood sugar, cholesterol, osteoporosis, colon polyps, heart murmurs, carotid artery narrowing, memory loss, and sun exposure: the list is constantly expanding. One may reasonably anticipate that such risk factors will be recognized even earlier in life, at birth, *in utero*, even before conception. Should everyone be screened? Everyone is tied to the medical establishment from birth, resulting in increased preoccupation with survival rather than the challenge of creative living."[25]

One product of this tendency is some kind of compendium of officially recognized disease entities and subtypes, such as the prodigious ICD-9 classification, a book and software database which runs to hundreds of densely-packed pages, lists tens of thousands of pathological diagnoses, each one numerically coded, and is updated annually as new diseases are identified and old ones renamed or discarded. Even a casual glance at this monstrosity leaves little doubt that the vast majority of entries are merely technical abnormalities, adopted for taxonomic and administrative purposes only, and no longer make the slightest pretense of referring to dynamic physiopathological processes that could help us understand the lived experience of our patients.

6. The Patient as Specimen

Reducing illness to disease also harms patients in an even more fundamental way that I have already mentioned. Studying disease processes in the abstract, apart from the individuals who suffer from them, tends to reduce the patient to a specimen, and his or her actual experience of illness to an automatism, a self-propelling chain of necessary causes that appears pre-programmed to *worsen,* because the corresponding tendency of every patient with any illness to *recover* depends upon idiosyncratic variables within each individual that are rendered invisible or mysterious by the disease concept, and simply fall through the cracks, rarely if ever to be seen or thought of again.

By thus obscuring and ignoring the self-healing capacity of the patient, modern medicine effectively undermines and defeats the integrating power of the living organism as a whole, simply because it is global, indivisible, and resistant to definition, measurement, or analysis into separate parts, even if we remember to look for it. Once we fall ill or are labeled as a specimen of disease, actual or potential, it is difficult not to feel overawed, intimidated, and even compelled to accept such pathology as *given,* to surrender ourselves to what we suppose or are told to be *its* laws, and thereby to forget that every illness must also be *received* and expressed by each of us in our own way, and that whatever factors may have influenced our falling ill in the first place are just as likely to help us recover in the future. Because both the concept and the reality of self-healing have been hidden from view and are unfamiliar to most people in most situations, the idea of a disease process clarifies nothing but its tendency to persist and *worsen,* and therefore seems to require an aggressive strategy of treatment to control it insofar as possible.

The concept of objectified or reified disease processes likewise distorts and trivializes both case management and prognosis, by reducing the fine art of treatment to the technology of forcibly correcting the abnormalities used to define them; i.e., killing the tuberculosis bacilli, selectively destroying the cancer cells, normalizing the blood pressure, and so forth. In the same sense that histologically "cured" cancer patients who develop cachexia and aplastic anemia as a result of their chemotherapy are at least as sick as before, these crude oversimplifications tend to leave a dangerous ambiguity in the assessment of improvement and worsening, and even in the definition of the disease itself.

Thus patients who develop a severe or intractable illness following an apparently successful treatment of another illness pose an important dilemma that cannot be solved or even meaningfully stated within the concept of the disease process. For whether or not we say that the two conditions are *related,* the patient will still require separate diagnosis and treatment for each of them, and there is still no meaningful way to address the condition of the individual patient as a whole, as a fully-integrated bioenergetic system evolving and developing through time. In either case, the net effect of medical science as a philosophy or conceptual system remains largely to multiply exponentially and across the board the aggregate total of diseases, abnormalities, and diagnostic and treatment procedures to be comprehended within it, through its awesome power to subdivide living patients into ever more numerous identifiable and potentially controllable phenomena.

7. Diagnosis as Truth and Falsehood

On the other hand, it would be unjust to dwell on the authentic defects and limitations of routine screening tests without also acknowledging the many instances where our patients would die or suffer far worse if they

were *not* done. I'm thinking of an attractive woman of fifty, newly divorced with two young children, who came to my office recently, distraught and sobbing uncontrollably after a routine Pap smear found a large number of what her gynecologist described as "highly abnormal and suspicious cells," which prompted him to recommend total hysterectomy; i.e., removal of the uterus, ovaries, Fallopian tubes, and even the vagina and regional lymph nodes, even though and in a sense precisely because she felt entirely well, had no symptoms or complaints of any kind, and ultrasound, colposcopy, and endometrial biopsy had all failed to detect a localized tumor mass to account for them. Consulting another gynecologist at my request, she learned that her pathology report actually read "adenocarcinoma, type undetermined," which explained why immediate action was called for, and also why her own doctor had lied that she *didn't* have cancer, and touted the surgery as a 100% effective way of preventing the near certainty of it in the future.

In short, hers seemed to be precisely the test case I had envisioned earlier, a life-changing decision made solely on the basis of what cells look like under a microscope, with no symptoms or illness for them to explain. Yet whatever the differences among us as to the exact interpretation of these findings, or what we thought the wisest course to follow, all of us, her two specialists, the patient herself, and even I, seemingly contrary to what I had just written, accepted the *truth* of this report as signifying a high probability of life-threatening illness in the near future, and agreed on the urgency for some kind of active and timely intervention to try to save her life, or at least to buy her as much time as possible for continuing to parent her children as she sees fit. By that calculus, neither she nor I had the slightest doubt that her diagnosis was accurate, useful, and even supremely valuable to her, however grim and terrible the news it brought, and although the action it called for will almost certainly be disfiguring or worse.

As this by no means rare example makes clear, preventive screening tests and procedures continue to earn their central place in modern diagnosis and cannot be dismissed *a priori* on purely ideological grounds, even when no illness is visible or complained of. As to when they are likely to be helpful, harmful, or merely distracting, the doubts and scruples I have already cited may help to redefine the circumstances under which the additional information they provide may be regarded as "true," "false," or uncertain, and even what we mean by these heavily loaded words.

The example of my patient is also instructive in that sense, because the Pap smear is widely regarded as inadequate for detecting frank or invasive cancer, so that the presence of unmistakably malignant cells was already sufficiently unusual to alarm every doctor and technician who saw them, while the absence of well-differentiated cell types left open the possibility of ovarian cancer, the most dangerous variety, and called for immediate action just to rule it out. Under these ominous circumstances, not least because she felt well and had no symptoms, the test helped to convince my patient that she had or would soon develop a serious and life-threatening illness, and that further action was imperative to forestall it. The integrity of the various possible disease processes, that is, of cervical, endometrial, vaginal, tubal, ovarian, and bladder cancer, was also tied up in the apparent urgency of further diagnostic procedures to differentiate them, and the only disagreements between the various players involved the ever-present issue of how best to balance the need for effective action with the patient's comfort and quality of life. In short, there was no ambiguity or uncertainty to speak of in her case, apart from the standard policy of screening all adult women.

But that is the whole point. Pap smears are done routinely to screen the entire adult female population, involving the detection of cervical dysplasia and other abnormalities as risk factors for developing cancer of the cervix in the more or less distant future. Even its most zealous proponents would hesitate to advocate the procedure solely to catch the few exceptional cases like my patient, and it is now widely known, as we have seen, that mild and moderate dysplasia, which sometimes progress to cancer after several months or years, most often revert to normal if simply left alone and allowed to do so. Charting a path somewhere between these two essentially uncontested truths suggests the sensible and increasingly popular middle ground of *doing* the test, yes, but then simply watching, waiting, and taking no further action unless the morphology spreads and worsens.

Much the same strategy seems equally applicable to mass screening for cholesterol, blood pressure, bone density, and many other such variables, which are similarly designed to assess risk factors for serious diseases and illnesses in the relatively distant future. As with the Pap smear, the main problem with most routine screening is not so much the burden of the procedures themselves as the unnecessary pain, suffering, disability, and expense incurred by the additional diagnostic tests and treatment regimes that they appear to make necessary. No competent physician would hesitate to recommend further testing for those with abnormalities lying well outside the normal range and thus indicative of something in the *present,* some illness as yet unsuspected and unperceived by either the patient or the doctor, but which a more extensive diagnostic workup would likely reveal, and which the patient would want, expect, and indeed be grateful to know about. Given a blood pressure of 160/110, a cholesterol of 300 or more, a TSH of 10 or 15, a liver transaminase in the 400's, or a PSA of 20, few doctors would be content

to dole out the usual drug without first trying to get to the bottom of these findings, to establish a more definite and comprehensive diagnosis, because all of them indicate authentic disease processes and often actual illnesses that are already under way, such that the pathology is highly likely to illuminate, explain, and even at best to *predict* them. In this situation, such striking abnormalities and the diseases that they represent may both be considered real and *true* to that extent, in the same sense as the acute, subacute, and intermittent diseases we discussed earlier.

With a "borderline" result, on the other hand, a slight deviation with at most purely actuarial significance, it seems excessive, exorbitant, and amounts to cruel and unusual punishment to inflict these largely arbitrary and ever-changing standards on patients for years and decades of their lives, artificially forcing their test results back into line and keeping them there without compelling evidence of benefit to the general health and well-being from doing so, let alone due regard for the predictably adverse effects of the coercion itself. Merely being less zealous about *enforcing* such dubious diagnoses would save enormous sums in follow-up visits, drug costs, and tests to monitor them, to say nothing of the anxiety that sustains them all, and would thereby help to reduce the size and weight of our famously bloated medical enterprise to a more nearly human scale.

Meanwhile, on the receiving end, so to speak, confronted by their own fear and hesitation on the one hand, and the vast knowledge and formidable power of the medical system on the other, our patients themselves provide a fitting epilogue in their profound desire, curiosity, need, and endless fascination to match up and integrate their own unique lived experience with the independent and profoundly alien version of the body as a machine, so blissfully or horribly anonymous and neutral.

That is why it makes perfect sense, for medical as well as social, political, and ethical reasons, to offer these same tests to those patients who *want* them, whether we recommend them or not, and to build a patient-centered criterion into the system, even if it costs a little more, to allow patients to screen themselves, to allow their physicians to attempt to dissuade them when appropriate, and thus to facilitate an ongoing negotiation to fine-tune their care.

The value of diagnosis to the individual patient and its awesome social power are nowhere more evident than in the remarkable growth and multiplicity of charities and support groups for patients bearing diseases of every type, many recruiting informally through the Internet and achieving major status and political clout through periodic conferences, fund drives, and research sponsorship. Although many individuals find it distasteful to be lumped together with people who have little else in common, or to dwell on their disease to the exclusion of pleasanter topics, those who suffer most intensely often find comfort, solidarity, and even personal truth in the company of others similarly afflicted, at times attaining a level of peace and equanimity that would have hardly been possible otherwise. Still others are moved to social action, such as lobbying Congress to call attention to diseases that are new, controversial, or not well understood, like Autism and Asperger's Syndrome, or even to right a perceived wrong, such as the side effects of drugs, vaccines, toxins, chemicals, and additives. Precisely because they empower patients to speak their truth, however unreasonable or unpalatable it may seem to others, and thereby to participate more actively in a health care system increasingly disinclined to listen, such groups deserve encouragement and support from physicians no less than the public at large.

For all of these reasons, I can think of no wiser rule for our profession to live by than the one set forth by Hippocrates, our great teacher and patriarch, almost twenty-five hundred years ago:

> "Declare the past, diagnose the present, and foretell the future;
> But as to diseases, let us try to help, or at least to do no harm."[27]

NOTES

22. *Medical World News,* August 1988, p. 51.
23. "Cholesterol Screening in Children," U. S. Public Health Service, *American Family Physician,* June 1995, p. 1923.
24. "Lipid Controversy Builds Up," *Medical World News,* October 1992, p. 15.
25. Love, S., "Sometimes Mother Nature Knows Best," *New York Times,* March 20, 1997, reprinted in the *Public Citizen Health Letter,* **13:1**, 1997.
26. Lown, B., "The Commodification of Health Care," *Hastings Center Report,* September-October 2006, p. 42.
27. Hippocrates, *Epidemics,* Book I, Section 2, par. xi.

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