

Virtual Mentor

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Ethical Issues in Screening

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Virtual Mentor

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From the Editor To Screen or Not To Screen?

By the time medical school begins, most physicians-to-be already know a great deal about screening. We have all learned from watching television that mammograms and prostate-specific antigen tests should occur once a year beginning at a certain age. Most of us have had our cholesterol levels checked and the majority of women have had several Pap smears long before their medical careers begin. This issue of *Virtual Mentor* explores some of the attitudes physicians develop regarding screening and why many of us feel obligated to perform these tests, despite a frequent lack of evidence supporting their benefits.

As a urology resident, I have seen many patients whose lives I think were saved by screening. I've been in situations where both the patient and I wished he had undergone screening before the disease progressed. But I've also seen many patients who've had abnormalities detected on screening and had their lives disrupted as they underwent further work-ups and tests, often for what ultimately proved to be a negative result or, sometimes worse, an ambiguous one. Few tasks are as difficult as counseling a patient about a disease that could be fatal or could, in fact, not even become a matter of clinical concern during his lifetime. There have been many days when I've gone home grateful that a patient had undergone a screening, but probably just as many when I've wondered how much we have really helped someone.

In the January 2006 *Virtual Mentor*, we highlight the spectrum of difficulties encountered with screening. We are fortunate to have contributions from many passionate authors who examine important questions about screening. How informed are patients about tests to which they consent and how informed *should* they be? How much do physicians know about the sensitivity and specificity of tests they recommend and the number of screenings it takes to save 1 life? What are the implications of prenatal genetic screening on the diversity of the population and on society's view of disability? How should physicians respond when patients request CT scans in the absence of symptoms, family history, and risk factors for disease? What must we do to ensure that our health care system provides follow-up treatment to all who have positive test results? In addition, the authors provide some basic information about what makes a good screening exam and offer an interpretation of the literature regarding the cost-effectiveness of CT screening for lung cancer.

Screening patients carries tremendous responsibility. I hope this issue gives us all insight into the pitfalls of screening and the necessary preparation a physician must make before ordering a screening test. Understanding these ethical issues should allow us to

more effectively use screening for its intended goals—to provide a benefit to our patients while remembering: first, do no harm.

Adrienne J.K. Carmack, MD

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Clinical Case

Breast Cancer Screening

Commentaries by Heidi Malm, PhD, and Gerald W. Chodak, MD,
and by Antonella Surbone, MD, PhD

Mr Jones, a 49-year-old accountant, visited Dr Seelinn, a urologist, for the first time. Mr Jones's sister had been treated for ovarian cancer and his mother had a history of breast cancer. While his sister was receiving treatment for her cancer, her physician recommended that the entire family be screened for breast cancer 1 and 2 (BRCA1 and 2) mutations, and Mr Jones agreed to have this test.

Several weeks after the test, Mr Jones learned that he tested positive for a mutation and was sent a form letter stating that "men with this mutation have a 6 percent chance of developing breast cancer and a 7 percent chance of developing prostate cancer before age 70." Concerned about his results, he went to see Dr Seelinn to find out what he should do with this new information. As he handed Dr Seelinn the letter, Mr Jones laughed and said, "If I had known the test wasn't going to give me a definite yes or no answer about whether I was going to get cancer, I wouldn't have had it. I don't know anything more than I did before the test."

Dr Seelinn is also unclear about what this reported risk of prostate cancer means. Does it mean biopsy-proven prostate cancer (which may be unlikely to progress if diagnosed in his late 60s)? Or does it refer to the risk of advanced prostate cancer that would present with symptoms? He's not even sure if this letter means that Mr Jones is at a higher or lower risk of prostate cancer than men without the mutation.

Seeking to provide Mr Jones with some guidance and more information, Dr Seelinn performed a urologic history and a thorough physical exam, including a breast exam and a digital rectal exam (DRE) of the prostate. All of these were normal. Dr Seelinn also ordered a prostate-specific antigen test (PSA) even though he didn't expect to find anything abnormal in the results.

After Mr Jones left, Dr Seelinn wondered whether finding out about his genetic alteration held any benefit for Mr Jones. Did Mr Jones really understand what a positive test result would mean? Furthermore, what follow-up schedule is appropriate for Mr Jones, who appears to be a healthy, 49-year-old man?

Commentary 1

by Heidi Malm, PhD, and Gerald W. Chodak, MD

Though Socrates claimed that knowledge is good and ignorance, evil, this case shows that a little bit of knowledge may be worse than none at all. The problems began when Mr Jones's sister's physician (let's call her Dr Protest) encouraged Jones and his siblings to be screened for the BRCA1 and 2 genetic mutations. The encouragement came without counseling (or even an offer to counsel) Jones and his siblings about the outcomes of the test and the various risks and benefits of knowing whether one has the mutations. Instead, by making a blanket recommendation for the screening, Dr Protest led the Jones family to believe that taking the test would be good for them. After all, why else would their sister's trusted physician have recommended it, Mr Jones is likely to have reasoned. So he signed the paperwork for the test believing, among other things, that it would give him a definitive answer as to whether he would get cancer like his sister and mother. If the test were positive, then he could vigilantly watch for the first signs of cancer and start treatment early enough to be cured. If the test were negative, he'd be safe from the disease and free from that worry. However, because BRCA1 and 2 testing will tell him nothing of the sort, it is clear that Jones did not understand what he was doing and thus could not have given truly informed consent to the testing.

The preceding problem of consent would have been minimized had Dr Protest properly counseled Jones about the outcomes, risks, and benefits of BRCA1 and 2 screening. Alternatively, Dr Protest could have merely recommended that Jones speak to his own physician about the possibility of testing and its risks and benefits. As an additional safeguard, the lab that conducted the test might have offered Jones a brochure explaining what the testing would and would not show and asked Jones to read the brochure before signing his consent form.

Of course, neither option would *guarantee* that Jones truly understood what the test could and couldn't do for him. His fear of cancer, heightened by having both his sister and mother suffer from it, coupled with society's general presumption that screening is beneficial, might have created in Jones an irrationally optimistic presumption about the benefits of BRCA1 and 2 screening that standard counseling and a printed brochure would not have overcome. That is, there is a fairly widespread belief in our society that screening is good for people. The incorrect assumption is that simply finding a cancer earlier is worthwhile when in fact screening is only beneficial if it lowers the morbidity or mortality from the disease without causing undue harm. For diseases such as prostate cancer, for example, there is currently no good scientific evidence that morbidity or mortality is affected by screening, hence the inadvisability of strongly recommending routine screening. Still, the counseling and brochure would have gone a long way toward meeting the objective of informing Jones sufficiently to consent to or decline testing, and the counseling itself is a minimum standard that should be met by any physician who recommends a screening test.

But the absence of true informed consent isn't the only problem in the case. Even if Mr Jones had understood that the genetic testing would only tell him about his statistical

risk of getting cancer, the information provided in the form letter response was too cryptic to be of any use and therefore was not beneficial. Did the stated 7 percent chance of developing prostate cancer by the age of 70 mean the chance of developing microscopic autopsy-proven prostate cancer? Or was it a risk of developing potentially aggressive cancer eventually leading to symptoms that would affect Jones's quality of life and possibly his survival? And how does either alternative compare with men who lack the mutation? An answer to this last question is needed in order to determine whether the information gained from the genetic screening will make any meaningful difference to future nongenetic screening—eg, PSA, DRE—and treatment recommendations.

Suppose that the 7 percent risk referred to the first alternative, Jones's risk of developing biopsy-proven but non-life-threatening prostate cancer. In this case, Jones would seem to be at a *lower* risk than similarly aged men who lack the genetic mutation. Autopsy studies have shown that by age 50, approximately 30 percent of all men in the United States have microscopic evidence of prostate cancer, and that this percentage increases to 50 percent by age 80 [1]. Yet the annual mortality rate for this type of cancer is quite low. Thus, given that the majority of these cancers do not progress to the point of adversely affecting the person's life, it isn't clear that learning one is at a comparatively low risk of developing such a cancer is a benefit. Alternatively, suppose the 7 percent refers to Mr Jones's risk of developing potentially aggressive prostate cancer sometime in the future. It still isn't clear where this places him in comparison to other men and thus whether it has any bearing on future screening and treatment decisions. A 7 percent risk of developing prostate cancer before the age of 70 is not the same thing as a 7 percent risk of dying from it: most men with prostate cancer die *with* the disease and not *from* it. Further, if the vigilant search for this potentially aggressive cancer leads to the detection and treatment of the much more statistically likely microscopic cancer that would never progress to the point of adversely affecting Jones's life, then the information may even have made him worse off. In either case, it seems that the information contained in Mr Jones's form letter response did not provide a meaningful benefit.

Furthermore, the minimal information in Mr Jones's form letter led Dr Seelinn to conduct and order other exams and tests, ostensibly to provide Mr Jones with the guidance and information lacking in the letter. But this introduces a new version of the first problem. Dr Seelinn ordered these tests and exams without talking to Jones about their various risks and benefits. Thus Jones was once again denied the opportunity to give true informed consent. In particular, Dr Seelinn ordered a PSA test without telling Jones that the benefits and even usefulness of the test were controversial. To date, no study has proven that asymptomatic men such as Jones live longer if testing is performed, yet the harms of treatments performed in response to suspicious results and diagnosed cancers include impotence and incontinence as known possible side effects. Further, data from a recent study showed that no PSA value can be considered normal, in the sense that some men with the lowest PSA levels will have a positive biopsy for cancer [2]. Therefore, a major goal of PSA screening—definitively informing the patient that he does not have the disease—is not currently possible. Had Mr Jones been told these and other relevant facts, it is at least possible that he would have declined the

screening. But he wasn't given that option. And he was told nothing about the breast cancer screening at all.

Perhaps Dr Seelinn thought that the additional testing was necessary to protect himself from future legal liability. That is, it might be argued that, given our overly litigious society, Dr Seelinn would risk being successfully sued if he failed to order the PSA screen now and Jones were later found to have prostate cancer. But this argument is faulty in a number of ways. First, the argument assumes that Jones would have been better off finding a cancer now rather than later. But that assumption is controversial at best, as discussed above. Second, the question isn't whether Jones should or shouldn't have a PSA test, but whether Jones was given the information and opportunity to decide for himself whether he wanted the test based on known facts about the limitations of screening. Third, Dr Seelinn can limit his medical-legal liability by properly documenting that he did in fact inform Jones of his options and the various risks and benefits and then let Jones decide for himself. Finally, even if a recommendation for screening has become part of the established standard of care in Dr Seelinn's community, the related legal doctrines of the respectable minority and 2 schools of medical thought would protect Dr Seelinn's decision to not recommend the test, as long as he informed Mr Jones of its availability. The details of these doctrines vary among jurisdictions, but each generally serves to protect from malpractice liability physicians who elect to pursue one of several recognized courses of treatment, even if the elected course is not preferred by the majority. As one court stated, "where two or more schools of thought exist among competent members of the medical profession concerning proper medical treatment for a given ailment, each of which is supported by responsible medical authority, it is not malpractice to be among the minority in a given city who follow one of the accepted schools" [3].

In summary, the case at hand involves a series of missed opportunities for clear communication and thus for true informed consent. Jones acted on the first recommendation without knowing what it could and couldn't do for him, and the problems snowballed from there. Such an approach is unlikely to lead to optimal patient care.

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Commentary 2

by Antonella Surbone, MD, PhD

*“Future things: not our domain.
But in this today which unravels in front of us,
what shall we do?”*

—Sophocles, *Antigone*

The case of Mr Jones illustrates how the many medical, psychological, ethical, and social dimensions of genetic testing for cancer susceptibility are intertwined. This commentary addresses the controversial medical aspects, summarizes briefly the main ethical considerations raised by breast cancer (BRCA) testing in general, and looks at those concerns as they relate to this case.

BRCA-Associated Risk of Prostate Cancer

Prostate cancer is the most common cancer in men and the second leading cause of cancer mortality in men. In 2003, 220 900 new cases were diagnosed in the US, with 28 900 estimated deaths [1]. Approximately 40 percent of aggressive early onset prostate cancers are linked to inherited factors and 5 percent of them to BRCA germline mutations. BRCA1 and 2 mutations, first identified in association with hereditary breast and ovarian cancer, disrupt DNA repair, which results in increased cancer susceptibility in both women and men [2, 3].

Carriers of BRCA2 mutations seem to have a 5-fold increase in the risk of prostate cancer, while BRCA1 carriers seem to have about half that risk [4]. The relative penetrance of different BRCA mutations is still unknown, and contradictory clinical findings have been reported, including a recent suggestion that only BRCA2 mutations are associated with an elevated risk of prostate cancer [5]. Not all studies support the association between early onset prostate cancer and BRCA mutations.

Screening and Follow-Up: Recommendations and Controversies

Despite the high incidence and mortality of prostate cancer and the availability of different screening modalities, the efficacy of screening has been questioned. First, empirical evidence is lacking from prospective randomized studies to prove that screening for the mutation translates into a reduction of mortality from prostate cancer. The morbidity and costs of overdiagnosis and overtreatment of well-differentiated or localized tumors are cause for concern, as are the psychosocial “costs” [6, 7].

Currently, screening is recommended for men beginning at age 50 and consisting of yearly DRE and measurement of serum PSA concentration, followed by biopsy if necessary. These recommendations also apply to known male carriers of BRCA mutations with screening possibly starting, instead, between the ages of 40 and 45 [8, 9]. A large ongoing trial known as IMPACT enrolls men aged 45-69 with a known germline BRCA mutation in a screening program. This study aims to identify men with a high risk of aggressive disease [4].

Ethical Considerations Raised by BRCA Testing

The ethical implications of BRCA testing relate to (1) information and informed consent; (2) rights and responsibilities of the individual, the family, the social community, and the scientific community; (3) confidentiality and privacy; (4) possible discrimination in life and health insurance, the work place, the process of adoption, and access to education; (5) prenatal diagnosis and the risk of eugenics; (6) specific ramifications of BRCA testing in minority and underprivileged populations; and (7) justice and fairness in allocation of genetic resources [10, 11]. Not all of these concerns come into play in the case of Mr Jones, but several do.

Information and genetic responsibility

Mr Jones's sister's physician requested that he undergo BRCA testing, most likely for his and his family's benefit. Mr Jones agreed to be tested, but subsequently expressed doubts about his perceived lack of personal benefit from the testing, since his risk of developing cancer remained uncertain. Two main ethical problems are involved in this case: it seems that Mr Jones was not adequately informed of the limited predictive power of BRCA testing, and he did not receive proper counseling before and after testing.

Genetic information is complex and can be difficult to convey in lay terms. High-risk subjects who are anxious and vulnerable may not fully absorb or understand the process of genetic risk assessment in a single encounter and often overestimate the predictive power of genetic testing. Informed consent should be part of an iterative process of communication between the patient and the doctor and the other health care professionals involved. In the case of genetic testing for cancer susceptibility, additional pre- and post-test counseling is needed, given the complex repercussions of any decision and of any result for the patient and his or her family. The literature on BRCA testing shows that subjects who test negative in a high-risk family may suffer significant psychosocial repercussions related to feelings of guilt and isolation that may also require counseling [12].

Mr Jones's case also raises the question of whether or not the healthy members of a high-risk family should be encouraged to be tested for the good of other family members. Most would agree that members of a high-risk family or community have moral responsibilities toward other members that extend beyond their own personal interest [13]. In clinical practice, we now often see scattered families coming together to face the possible risk of being BRCA carriers, to help other members interested in their family history gain more information, or just to "be there" for each other.

A high-risk individual may, however, refuse to be tested or refuse to reveal genetic information—a shirking of individual genetic responsibility in the eyes of some [14]. Indeed, for almost any person at risk, the decision-making process is extremely complex, and the physician's role is to be nonjudgmental and to facilitate understanding and communication among all family members. The physician's responsibility vis-à-vis genetic testing is, in fact, also expanding beyond duty to the individual patient to include a duty toward his or her extended community. In the clinical setting it is often difficult

to strike a balance between the rights of one person and the rights of other community members.

Uncertainty and trust

As Mr Jones and Dr Seelinn realized with great concern, genetic testing precedes, in most instances, the development of effective preventive and therapeutic measures. After testing positive for BRCA, Mr Jones is left with many uncertainties about his future risks and especially about what to do. His physician correctly chooses a strict clinical and laboratory follow-up for Mr Jones. Yet Dr Seelinn does so in the face of major medical uncertainties, which he seems to convey honestly to Mr Jones.

The uncertainty that follows many instances of genetic testing, as well as the concerns related to potential social and ethical abuses of genetic knowledge, can be very challenging for the patient-doctor relationship. This is especially true in the climate of a patient-doctor relationship that has suffered from growing economic and legal pressures and has come to resemble a marketplace exchange between provider and consumer. As a result the role of trust in medicine has come under scrutiny [15, 16]. In my clinical experience, persons involved in genetic testing often express a strong need to trust that the experts are being truthful and also that they are willing and able to advocate on their patient's behalf. This need for trust extends beyond individual relationships to institutions, policy makers, and the media [17].

Conclusion

Genetic knowledge may increase the sense of control over one's life, but it may also shed a dim light on one's future, thus paralyzing the decision-making process. This is the real quandary of genetic testing. We should listen to and respect our patients' different perceptions of whether genetic information provides empowering knowledge or is accepted as a sign of predestination [17].

The influence of genetics on our lives is likely to be much more limited than we tend to believe [18]. Yet, knowing that one is a carrier of a genetic predisposition to cancer involves risks that have a dramatic impact on a person's life. Genetic risk, in fact, entails not only the possibility of developing a future serious illness but also of being forever "asymptotically ill" in the absence of disease [19]. Mr Jones, for example, will be subjected from now on to medical tests and possible interventions that may carry substantial economic and emotional costs for him and for the community. He may also experience the psychological and social consequences of being a BRCA mutation carrier that may deeply affect the dynamics of his relationships.

The fundamental question posed by genetic testing is thus whether some degree of knowledge about possible future events helps us or limits us. Genetic information does not come at present with clear answers about what we should do. The worth of genetic testing needs to be evaluated at the individual and community levels and to be balanced against broader medical, psychological, social, and ethical considerations.

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Clinical Case

Presymptomatic Genetic Testing for ALS

Commentary by Leo McCluskey, MD, MBe

Mr Smith is a fit 35-year-old without any major health complaints. He recently moved, and, during his first visit with his new physician, Dr Sanders, he revealed that his older brother had died of amyotrophic lateral sclerosis (ALS, also called Lou Gehrig's disease) at age 47. Dr Sanders acknowledged that this must have been a staggering loss for Mr Smith, who disclosed that what his brother went through was terrible and quite traumatic. He told Dr Sanders that he could not imagine going through that disease course and doesn't know how his brother coped.

Dr Sanders remembered having read that about 10-15 percent of ALS cases are familial. She inquired whether any other family members had developed the disease, and Mr Smith said that a great-uncle had died of an unknown disease at a young age, but no one ever thought it might have been ALS. Other than that, there was no reason to think ALS ran in his family.

Dr Sanders thought about offering genetic screening to Mr Smith but wanted to consider further the risks and benefits to her patient. She wondered whether telling Mr Smith that he had the genetic markers for this deadly disease before he became symptomatic would only distress him and not yield an offsetting benefit, since the age of onset was unpredictable.

Commentary

ALS is a rare, presently incurable neurodegenerative disorder that annually affects 2-2.5 persons per 100 000. Ninety percent of ALS, known as sporadic ALS, is not inherited. Five to 10 percent of ALS is familial, and only 20 percent of familial ALS (FALS) is caused by a recognized dominant mutation in the so-called SOD1 gene for which testing is currently available. Only one drug, Riluzole, has been found to affect the progressive clinical course of ALS, but there is no evidence to support the use of this drug to prevent or to delay the onset of clinical symptoms in individuals with FALS. Palliation of symptoms is currently the main focus of ALS clinical care, and clinical management is the same, regardless of the patient's genetic status [[1](#), [2](#)].

Dr Sanders has much to contemplate about whether to discuss the potential for familial ALS with Mr Smith. Many ethical principles are in tension in this case—autonomy (respecting the patients' rights), truth-telling, beneficence (helping patients), and nonmaleficence (not harming patients). Let's consider the possible outcomes.

It is ultimately Mr Smith's right to decide whether he wants to discuss with Dr Sanders the inheritance of ALS and the availability of genetic testing

Mr Smith is a new patient for Dr Sanders. Even though he told her that “what his brother went through was terrible and quite traumatic,” and “he could not imagine going through that disease course,” and “he doesn't know how his brother coped,” it will be difficult for Dr Sanders to accurately predict the reactions this new patient will have to discussing the pros and cons of genetic testing, undergoing testing, and learning of the (potentially positive) results. Lying by omission, that is, withholding information about the availability of genetic testing from Mr Smith, is justified only if Dr Sanders concludes that Mr Smith will be significantly and irreparably harmed by the information he receives. Absent this conclusion, Dr Sanders' paternalistic response may harm the trust necessary for a good patient-physician relationship. While Dr Sanders may be justified in delaying the discussion until she can assess its probable effects on her patient, it is most likely that she or a genetics counselor will eventually have to have this conversation with Mr Smith.

Mr Smith may benefit from a discussion of the genetics of ALS and the availability of genetic testing whether or not he decides to be tested.

Dr Sanders should consider the benefits Mr Smith could derive from talking with her about FALS. For example, he may not have known of his potential risk. A frank conversation about the realities of genetic testing for the SOD1 gene would almost certainly help Mr Smith weigh the pros and cons of going ahead with the test for himself. At the same time, Dr Sanders could inform him about the current status of ALS care, the potential for disease-modifying therapy via Riluzole, and the palliation of even the most distressing symptoms via medical therapy. If she wants to go beyond the topic of treatment, Dr Sanders can inform Mr Smith of ongoing ALS research, the potential for clinical trials, the benefits of disease-specific advocacy, and the potential benefits of organizations, such as the ALS Association and the Muscular Dystrophy Association, that provide specialty care. Such a discussion may provide Mr Smith with some measure of hope despite the serious and life-threatening reality of the disease.

Mr Smith may benefit from being informed even if he decides to forgo testing. The possibility that he may have the harmful mutation might influence Mr Smith's choice of health care insurance coverage. For example, he may elect insurance that has ample coverage for pharmaceuticals (the current cost of Riluzole is about \$900 a month), durable medical equipment, and home care. He may also decide to obtain long-term care insurance and alter his current life and disability insurance status.

If Mr Smith proceeds with testing he may discover that, although he is presently asymptomatic, he does have the mutant SOD1 gene. While this would certainly be a devastating result, he may view even this knowledge as having some benefit for him. For example, while Mr Smith's marital status or his plans for having a family are not discussed in the case, knowledge of his genetic status would almost certainly influence his family planning. He may choose not to conceive children but to adopt or pursue other options such as artificial insemination from an anonymous donor. He may opt to

use in vitro fertilization with preimplantation screening of the embryos for SOD1 and implantation of only those embryos that do not carry the mutant gene.

Mr Smith may be harmed by a discussion of the genetics of ALS, the availability of genetic testing, and by proceeding with the testing.

Dr Sanders should consider that Mr Smith might become distressed and suffer significant psychological harm as a result of even a discussion of FALS. While Mr Smith's concern may be limited to himself, Dr Sanders must also consider that he may have genetic guilt or worry about the possibility of transmitting the SOD1 mutation to his offspring. It is not possible to calculate the likelihood that Mr Smith carries the mutation, but Dr Sanders can assure Mr Smith that ALS is rare (only 2 or 2.5 cases per 100 000), and familial or inherited ALS is an even more unusual disorder (with an average of 2 or 2.5 cases per million). The variety of familial ALS for which testing is available is more rare still (4 or 5 cases per 10 million). But the instance of familial ALS for which testing is *not* available is 8-12 cases per 10 million. Thus, while Dr Sanders may introduce the specter of FALS with Mr Smith, genetic testing is unlikely to predict definitively whether Mr Smith will or will not get ALS. A negative test may, in fact, provide Mr Smith little solace.

If Mr Smith decides to be tested, he may also be adversely affected by the month-long waiting period before the results become available. While he may eventually be relieved by a negative result, he may be dejected by a positive test result.

If he tests positive, it is very likely that Mr Smith will experience significant fear, anxiety, and, potentially, depression triggered by concern for both himself and his offspring. He may even contemplate suicide. A positive test result status may adversely affect Mr Smith's ability to maintain his present health insurance or procure new coverage; if he retains coverage, his carrier may raise his rates. The Health Insurance Portability and Accountability Act (HIPAA) of 1996 provides some protection for people who have employer-based health insurance by prohibiting group health plans from using genetic information as a basis for denying coverage if a person does not currently have a disease. However, the act does not prohibit employers from refusing to offer health coverage as part of their benefits, nor does it prevent insurance companies from requesting genetic information from potential buyers. Moreover, HIPAA does not provide protections for those who are self-employed.

If Mr Smith's employer learns about the positive test result, Mr Smith may experience genetic discrimination in the workplace. Although there are currently no federal laws specific to genetic nondiscrimination, some protection from discrimination by employers is offered through the Americans with Disabilities Act of 1990 (ADA). In 1995, the Equal Employment Opportunity Commission expanded the ADA definition of "disabled" to include individuals who carry genes that put them at higher risk for genetic disorders. The extent of this protection, however, has not yet been tested in the courts. Several states have laws that address genetic discrimination by employers and health insurance companies. The degree of discrimination protection varies from state

to state. Therefore, the decisions that Mr Smith makes about genetic testing while living in one state may have repercussions in the future if he moves to another area.

If Mr Smith tests positive for a SOD1 gene mutation, he may not be able to obtain private life, disability, or long-term care insurance. He is likely to be more successful in obtaining such coverage if it is offered by his employer, but it is possible that the employer may refuse to offer such benefits to him.

Conclusion

Weighing the potential benefits and harms of testing for FALS in this way, it is safe to conclude that Mr Smith would benefit from a discussion of the genetics of ALS through which he would become better informed and therefore empowered to make decisions regarding insurance coverage, family planning, and the pros and cons of proceeding with testing. While he may encounter some psychological stress and anxiety, it is unlikely that he would suffer depression or even contemplate suicide as a result of the discussion. Dr Sanders, therefore, is ethically responsible for initiating this conversation with Mr Smith. Since the discussion is likely to take a considerable amount of time and has many facets that may well be beyond the expertise of Dr Sanders, it would be appropriate for her to refer Mr Smith to a genetic counselor or to a neurologist with expertise in the genetics of ALS. Like most patients who weigh the benefits and burdens of presymptomatic testing for FALS, Mr Smith may elect to forgo genetic testing. Nonetheless, an informed Mr Smith is better prepared to make life choices that might be influenced by the possibility of FALS.

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Clinical Case

Patient-Requested, Non-Recommended Screening

Commentary by Mark T. Hughes, MD, MA, and Bimal H. Ashar, MD, MBA

Mrs Ackerman went to visit Dr Noell for an annual gynecological exam that included a Pap smear. She sees Dr Noell regularly for check-ups and the recommended screening tests for women in her risk and age groups. So far, she has been remarkably healthy—she exercises regularly, does not smoke, and is a vegetarian—and no screening test has ever shown cause for concern.

Following the exam, Mrs Ackerman told Dr Noell that she had seen an ad in a newspaper for a whole body CT scan for \$600. “I’d like to have this done to make sure there’s not a treatable problem in there,” Mrs Ackerman said pointing to her stomach. “The problem, Dr Noell, is that it’s a pretty expensive test. But imagine how much *more* expensive it would be if they didn’t find something until it was too late.” Dr Noell gently told Mrs Ackerman that there was no reason to think that there was a disease “lurking” inside her and that, after having a scan, many patients believed that it was a “waste of time and money” as it did not show anything that wasn’t already known. As a compromise, Mrs Ackerman suggested ordering a CT scan of only the abdomen and pelvis “just to check.”

Commentary

Dr Noell is faced with a situation seen much more frequently today than in the past. Patients now have a greater role in deciding what medical services they receive. Viewing the patient as a consumer of services has led to the growth of direct-to-consumer (DTC) advertising for both pharmaceuticals and diagnostic tests. DTC advertising often preys on patients’ fear of the unknown and may suggest that a particular test is necessary for “peace of mind.” This advertising tack is taken despite the fact that no clear data currently support the use of many of these new technologies and, specific to this case, whole body CT scanning for disease screening [\[1\]](#).

Dr Noell’s first step in addressing Mrs Ackerman’s request should be to get a better understanding of the reasons for her inquiry. Patients often withhold symptoms from a busy practitioner despite being worried that they may have a serious problem. Mrs Ackerman may be using the discussion about the CT scan as a way to talk about abdominal symptoms that she is experiencing. She may have opened up the subject of the abdominal scan to prompt further questioning by Dr Noell. Then, rather than appearing as if she is complaining about her health, Mrs Ackerman can concentrate on responding to pointed questions posed by her physician. Or it might be the case that a family member or a friend had an illness in the past that was distressing for Mrs

Ackerman to observe, and her desire for “peace of mind” arises from apprehension that a similar fate awaits her. It is Dr Noell’s responsibility to try to uncover unspoken reasons for the patient’s request.

If specific symptoms or other concerns are not uncovered after a thorough history, and the patient’s physical exam does not reveal any worrisome signs, then the scan would be solely for screening purposes (that is, not diagnostic). In such a case, Dr Noell should engage in further discussion with Mrs Ackerman (remembering that *docere*, the Latin root for doctor, means “to teach”). In doing so, he must be guided by certain underlying ethical principles [2].

First, medicine is a scientific discipline. While much of medical practice has been passed down without the rigors of “gold standard” clinical trials, there is nonetheless a scientific basis for the recommendations that physicians make. This is demonstrated by the fact that evidence-based medicine is emphasized in all disciplines. Emerging diagnostic technologies should be held to the same scrutiny, so that test performance, bias, and cost-effectiveness all figure into the recommendations about these modalities. While it may not be necessary for Dr Noell to go into lead-time bias, length-time bias, sensitivity, specificity, and other highly technical details of a test, he should convey the real possibility of false positives and false negatives in scanning. The physician has the responsibility of communicating to the patient a basic understanding of science and technology and their proper applications in each individual case.

Second, the physician should be guided by the patient’s best interests. The first step in acting beneficently in this case is, as already mentioned, that Dr Noell explore the patient’s motivation for testing. If Mrs Ackerman wants to pursue the test out of fear, it is more appropriate for the physician to address her fear and see what is behind it than to simply order the scan. The ethic of care directs the physician to understand the individual patient in a particular context, recognizing that emotions factor into decisions but should not necessarily be directive. Acting in the patient’s best interest also means that Dr Noell will use his expertise to uphold his duty to warn patients about the risks and benefits of the scan and to protect them from harm. Despite the appeal of whole body CT scanning to diagnose problems in the early stages, existing medical evidence does not support this anticipated benefit. In fact, some clinicians are concerned that harm may result if, for example, an ill-defined abnormality is detected on the scan and leads to invasive follow-up testing that increases the physical risks and the financial costs to the patient. The patient also risks being labeled by insurers as having a pre-existing condition, a label that could affect the insurer’s coverage of the condition for which the patient was tested. So Dr Noell may have grounds in the interest of both beneficence and nonmaleficence for dissuading the patient from pursuing the scan.

Third, respect for autonomy means that Dr Noell should act as teacher and counselor to guide Mrs Ackerman to a thoughtful decision. Respect for autonomy also dictates that Dr Noell engage Mrs Ackerman in the process of informed consent. He should feel confident that Mrs Ackerman is making a voluntary decision, free of coercion or undue influence (such as the emotional impact of fear). He should disclose to her the risks and benefits of a whole body CT scan, giving special attention to their unproven nature. As

a consumer of medical care, Mrs Ackerman may decide, in the end, to pursue the whole body scan at a stand-alone facility without a physician order, should Dr Noell refuse to write her one.

But respect for autonomy applies to the physician as well. Respecting Mrs Ackerman's autonomy does not mean that Dr Noell simply has to acquiesce to her personal preferences. When a patient seeks a physician's opinion, the physician is not obligated to order a test or supply a service that he or she does not think is medically indicated simply because the patient requests it. The Charter on Medical Professionalism promulgated by the American Board of Internal Medicine and other professional organizations states that the physician strives for "scrupulous avoidance of superfluous tests and procedures" [3]. If Mrs Ackerman proceeds with testing without Dr Noell's involvement, even if he disagrees with the decision, he should remain available to her for counsel if an abnormality is found.

Next, physicians must be mindful of social justice. A physician's professional duty encompasses responsiveness to social concerns. Physicians have an obligation to contain costs and improve access to health care for all. If Mrs Ackerman wants her insurer to pay for the abdominal CT scan, then Dr Noell would have to say that there is some indication for the procedure, so that the health plan will cover the expense. If the only way to achieve this is for the physician to put erroneous information on the requisition (ie, "game the system"), then this goes against the physician's duty of truth telling and honesty. Dr Noell has a contractual obligation to the insurer to order only those tests that are medically necessary. Moreover, he has to have an eye toward proper utilization of society's resources. When physicians order additional tests and discover that ill-defined abnormalities noted in public screening reports are of no clinical consequence, society bears the costs of those downstream tests through higher insurance premiums and more limited access.

Finally, medicine is an art. The physician should be adept at patient-doctor communication in order to put the above-named principles into action. In discussing new technologies with a patient, the physician should explore the patient's concerns and motivation for pursuing testing and convey his or her desire to act in the patient's best interests. Sometimes, the physician's objective will be the likeminded goal of preventing disease, but in other circumstances it may be to protect the patient from harm by recommending against unproven tests. Science can serve as the arbiter in responding to patients' demands as a physician practices the art of medicine. In the end, the physician should bear the words of William Osler in mind, "Let us remember that we are the teachers, not the servants, of our patients."

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Clinical Case Informed Refusal

Commentaries by Howard Brody, MD, PhD, and Ruth Jepson, PhD

Dr Michaels looked at his clinic schedule for the day and immediately felt uneasy. The first person on his roster was Frank Elgie, a 56-year-old man coming in for his annual physical. Mr Elgie is generally healthy and takes one medication for hypertension and several vitamins.

Dr Michaels keeps up with the medical literature, has an MPH, and thinks that screening tests help to improve outcomes for individual patients as well as society by decreasing costs and the burden of disease. Dr Michaels recommends screening tests to his patients based on a combination of their medical history, age, risk factors, and clinical indication. As he stares at his patient list, he remembers his last visit with Mr Elgie a year ago because of their heated argument that upset Dr Michaels for days.

Last year, Dr Michaels told Mr Elgie that he needed a colonoscopy, and several other screening tests. Mr Elgie responded by saying, "I'm not going through that. Besides, I'm not at risk. No one in my family's ever had cancer." Dr Michaels tried to explain to Mr Elgie that the colonoscopy would be done with sedation so that the discomfort would be minimal. More importantly, Dr Michaels stressed that colon cancer was common enough in those without a family history to warrant screening. Mr Elgie said "Look, doc, I know my body and I'll know if I have a problem." Frustrated and running out of patience, Dr Michaels reprimanded Mr Elgie for not taking his health seriously enough. He even went so far as to ask Mr Elgie why he came to the doctor if he didn't intend to follow professional advice. Mr Elgie had not returned the rest of the year, but now he was back for his annual visit.

Dr Michaels does not want to get into another debate, but he strongly believes that screening is important. As he enters the exam room, he is still contemplating whether or not to mention any screening tests to Mr Elgie.

Commentary 1 by Howard Brody, MD, PhD

Dr Michaels should take 2 aspirin, lie down, and call me in the morning.

Dr Michaels has experienced the unfortunate shift that has occurred for many in our society (both physicians and the general public)—the turning of "preventive screening" from science into religion. The shift from science to religion may have resulted in part

from the zealous overselling of screening by patient advocacy groups, but I suspect it is due, at least in part, to the pervasive death phobia in our culture and the desire to convince ourselves that we can become immortal through the proper application of medical technology. Dr Michaels is now concerned because he cannot convert Mr Elgie to his own religious sect. But that is not his job. As a physician he should ensure that Mr Elgie is well informed about the pros and cons of all screening tests. He should also attempt to dissuade Mr Elgie whenever it appears that ill-founded fears or concerns might be swaying him toward a decision that he would later regret. Once Mr Elgie has understood Dr Michaels' point of view and has made up his mind, and Dr Michaels has documented the conversation in the record, Dr Michaels' job is done until the following year when he can ask Mr Elgie if he would like to reopen the conversation. Perhaps in the intervening year a good friend of Mr Elgie's will have been diagnosed with colon cancer, and he will then be in a totally different frame of mind. An important "law" from the novel *The House of God* states: "The patient is the one with the disease" [1]. The patient is also the one with the risks. Dr Michaels should never allow Mr Elgie's risks or decisions to make him, Dr Michaels, feel ill.

Since Dr Michaels has both a medical and an MPH degree, he presumably knows that it is now common to view a medical journal article as seriously lacking unless it reports its findings in terms of number needed to treat (NNT). Reporting the statistics as NNT is the best way to introduce healthy skepticism among readers when a new therapy is being recommended on relatively weak grounds. For example, imagine that after 10 years, 2 percent of subjects die in the control group, while 1 percent dies in the treatment group. These results would often be reported as a "50 percent reduction in mortality." It is much less impressive to report the NNT—that 100 patients would have to be treated with this drug for 10 years to prevent 1 death.

One could logically argue that information that helps physicians is also good for patients [2]. One systematic review concluded that we would have to screen 1173 people a year for colon cancer for 10 years to prevent 1 death [3]. If we told Mr Elgie these statistics, would he be more or less likely to accept the recommended screening? If we do not tell him these statistics, are we adequately informing him? The unfortunate fact is that the number of people who need to be screened for many commonly recommended tests in order to save 1 life runs into the thousands and tens of thousands. It is very likely that if patients were informed and truly understood the meaning of these statistics, enthusiasm for screening would wane rather than grow.

Being adequately informed about preventive screening requires that one know the disadvantages as well as the advantages of the tests. Did Dr Michaels, in his enthusiasm to convince Mr Elgie to have a colonoscopy, frankly discuss the risks of perforation and death from the procedure? Did he disclose the rate of false positive and false negative results?

It is also rather odd that Dr Michaels is ready to go to the mat with Mr Elgie over a colonoscopy, when he ought to know that the US Preventive Services Task Force (USPSTF) has been unable to discover compelling evidence that colonoscopy is superior to other alternatives for colon cancer screening. Indeed, the USPSTF found "good"

evidence that fecal occult blood testing is effective, but “did not find direct evidence” that screening colonoscopy is effective [4]. Did Dr Michaels offer Mr Elgie an annual fecal occult blood test instead of demanding the colonoscopy? It is possible that Dr Michaels may have become confused when the different specialty societies produced practice guidelines with different recommendations, making it very difficult for the well-intentioned physician to sort out the evidence.

The ethical model for preventive screening, as for most other encounters in medical practice, ought to be shared decision making. According to this model, Mr Elgie and Dr Michaels should be partners in deciding whether and how to screen for colon cancer. Different partnerships work differently; some are 50-50 and some are 80-20. Mr Elgie should have a say in the extent to which he wishes to meet Dr Michaels; will it be half way? Will he defer to Dr Michaels’ well-informed clinical recommendations? Or will Mr Elgie demand veto rights over any and all decisions? Whatever level of participation Mr Elgie chooses, he should emerge from the encounter feeling that he has been as involved as he wished to be in whatever decisions have been made. Dr Michaels should also recall that there is nothing about “shared decision making” that makes it wrong for him to try to persuade Mr Elgie that he might be making a mistake. This is especially true if Mr Elgie’s refusal seems to be based on a misunderstanding of his actual level of risk because he has had no relatives with colon cancer. The persuasion should be grounded, however, in genuine respect for Mr Elgie and his right to make his own decision and not in fervor to “tick off” another colonoscopy referral on the scoreboard.

If Dr Michaels remembers that the goal of this encounter ought to be shared decision making and not religious conversion, it is much less likely that either he or Mr Elgie will emerge from the visit with dyspepsia.

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Commentary 2

by Ruth Jepson, PhD

Doctors have an obligation to disclose relevant information (particularly with regard to risk), so that patients can make autonomous decisions; that is, decisions that are neither controlled nor coerced. Because of Dr Michaels’ enthusiasm for screening, he is

(wittingly or unwittingly) failing to disclose all of the known limitations and negative consequences of screening. Current clinical practice emphasizes shared decision making in which doctor and patient reveal treatment (or screening) preferences and agree on how to proceed [1]. In this case, it appears that Dr Michaels is not adhering to the principles of shared decision making and is failing to respect both Mr Elgie's wishes and his right to make an autonomous choice.

Dr Michaels and Mr Elgie need to find a way to move forward so that each believes his views are respected, patient autonomy is protected, and the patient-physician relationship remains strong. Dr Michaels may wish to consider alternative ways of improving Mr Elgie's health outcomes that are acceptable to both.

Benefits and Risk of Screening

Screening stands apart from traditional medicine in that it seeks to detect disease in individuals before they present with symptoms. Benefits of screening include improved prognosis for some illnesses because of early diagnosis, the possibility that less radical treatment is needed to cure the early-stage case, resource savings, and reassurance for those with negative test results. Unintended adverse effects of screening include longer morbidity for cases where the prognosis is unaltered by the early diagnosis, overtreatment of questionable abnormalities, resource costs, false reassurance for those with false-negative results, anxiety and sometimes morbidity for those with false-positive results, and the process hazards of screening tests [2].

Information Needed for Informed Decisions about Colorectal Screening

Dr Michaels is convinced of the benefits of screening, but has failed to disclose all of the unintended consequences and limitations of colonoscopy. A *Cochrane* review concluded that harmful effects of colorectal screening include the physical complications of colonoscopy such as perforation and haemorrhage, disruption to lifestyle, and stress and discomfort from testing and follow-up investigations [3–5]. In addition, whilst colonoscopies will only detect a few cancers, they will detect and remove a large number of polyps. This detection can be seen as a part of the benefit of screening or part of the harm. Part of the benefit of screening will come from removal of the small proportion of polyps that would have progressed to invasive cancer. Part of the harm of screening will come from regular colonoscopies that are recommended for people who have benign or inconsequential polyps removed [6].

Patient Autonomy within the Medical Encounter

Over the last few decades, the public in general, and bioethicists in particular, have become concerned about the rights of patients, including the right to give informed consent and the right to control one's health care choices [7]. It has been argued that whether or not the benefits of screening outweigh harmful consequences is essentially a value judgment, and one which until now has been made by "paternalistic agents of the state" (physicians) rather than by those invited for screening (the patients) [8]. The patients' rights model seeks to give patients information about the risks and consequences so that they can then make informed choices and judgments themselves.

The goal of enhancing choice—eg, by providing evidence-based information—should not be to encourage a *specific* choice [9]. Approaches to communication of risk information are based on the assumption that individuals will review the evidence rationally and choose the course of action that will maximise benefit to their health. However, rationality is not the only component in decision making; irrational influences and considerations can also exert strong pressures [10]. Whilst information provided by the physician may contribute to more rational decision making, its primary aim is to enhance patient choice and autonomy. In this case, Dr Michaels is convinced that the “right” choice is for Mr Elgie to be screened, but he is not taking into account Mr Elgie’s preferences and what the “right” choice is for him based on these preferences.

The doctrine of informed consent emerged in response to the perception that patients were not being given sufficient information and were thus powerless in health care (ie, without autonomy). One way to redress this imbalance was to better inform patients. Alongside the doctrine of informed consent evolved the complementary patient right to refuse treatment. The right to refuse, combined with the ethos of informed consent, enables patients to retain control over their lives and their health care [11]. Thus a shift took place from paternalism and beneficence in medicine (however benign) towards a partnership between patient and physician. In our case, Dr Michaels is angry because he feels that his professional opinion is not respected by Mr Elgie. But Dr Michaels is not respecting Mr Elgie’s attitudes, beliefs and values, and right to autonomy. Patient autonomy is a relatively new concept and, as such, may be uncomfortable for physicians like Dr Michaels who are used to having their professional views followed unquestioningly.

In shared decision making—“decisions that are shared by doctor and patient and informed by best evidence, not only about risks and benefits but also patient-specific characteristics and values” [12]—both the health professional and the patient are assumed to have a legitimate investment in the treatment decisions [13]. It is this model that Dr Michaels needs to think about and adopt in his encounter with Mr Elgie.

Conclusion

If Dr Michaels decides to talk to Mr Elgie about screening, he needs to give him more complete information, including the limitations and possible consequences of colonoscopy. It appears unlikely that Mr Elgie will change his mind, but at least he will have made a more educated choice. If he continues to refuse screening, both men may wish to discuss other ways of improving Mr Elgie’s health outcomes. For example, Dr Michaels could offer advice on the risk factors for colorectal cancer and provide information on how to modify such risk factors (eg, by diet and exercise). He will also need to provide information on the signs and symptoms of bowel cancer and encourage Mr Elgie to come and see him if he is worried or changes his mind about having a colonoscopy. Mr Elgie may choose to ignore this advice, but Dr Michaels can be assured that he has performed his obligations to disclose, that there has been some element of shared decision making, and that Mr Elgie has made an autonomous, informed choice.

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Journal Discussion

Screening for Lung Cancer: Too Much for Too Little?

by Christopher Kyle, MD, MPH

Mahadevia PJ, Fleisher LA, Frick KD, Eng J, Goodman SN, Powe NR. Lung cancer screening with helical computed tomography in older adult smokers: a decision and cost-effectiveness analysis. *JAMA*. 2003;289:313-322.

Background

Lung cancer is the leading cause of cancer-related deaths for both men and women in the United States. It accounts for 163 510 deaths a year, which is 29 percent of all annual cancer deaths. An estimated 172 570 cases of lung cancer will be diagnosed in 2005 [1], and three-fourths of those patients will have metastases beyond the lung at the time of their diagnosis. The average 5-year survival rate is 15 percent if metastatic disease is present [2] whereas patients who are diagnosed with stage I lung cancer have a 5-year survival rate of more than 60 percent [3]. Hence, the benefit of early diagnosis and treatment is evident and compelling.

The goal of screening asymptomatic populations is to diagnose a disease at a stage when early diagnosis and treatment makes a clinical difference. Lung cancer, with its prevalence, mortality, and known risk factors, is an excellent candidate for screening. But multiple large scale screening studies using chest radiographs and sputum have shown no reduction in lung cancer mortality [3, 4].

Technological advancements in medicine, notably the widespread use of computed tomography (CT), have reopened possibilities and renewed interest for effective screening. Low dose helical CT scanning of the chest can pick up small pulmonary lesions and may be useful diagnostically. Furthermore, extensive advertising to consumers for screening CT scans has increased the demand for these studies [5]. On the one hand, the benefits of screening are obvious: early detection, early treatment, and improved life expectancy. On the other hand, there are risks associated with screening. Cancerous lung lesions can appear as noncalcified nodules on CT, but most noncalcified nodules are benign. So, screening necessarily subjects many people who don't have lung cancer to invasive follow-up tests, significant costs, and increased anxiety.

At present, 2 large randomized controlled studies are in the process of evaluating the efficacy of CT scanning for lung cancer. The National Lung Cancer Screening Trial (NLCST) was started in 2002 by the National Cancer Institute (NCI). Full subject accrual was completed in February 2004 with 50 000 individuals randomly assigned to

either CT or chest radiograph; the subjects will be followed through 2009. There is also a European study involving 20 000 former smokers that will finish around 2010. Until these trials are completed and analyzed, clinicians must rely on projections of smaller studies to determine what is best for their patients.

One such projection using a computer model was reported in *Journal of the American Medical Association* in 2003. Mahadevia and colleagues presented a computer-simulated model that assessed the cost-effectiveness of CT scanning for lung cancer screening in smokers, as well as the mortality rates and potential harm under a variety of assumptions [6].

Methods

To analyze the cost-effectiveness of screening, Mahadevia and colleagues created a hypothetical study population of 100 000 heavy smokers, all 60 years old. The demographics of the population were adjusted to mirror participants in previously published screening trials. To account for smoking cessation among participants, the population was divided into 3 cohorts: current smokers, quitting smokers (those who had stopped by the time of initial screening), and former smokers (those who had not smoked for more than 5 years). Participants in each group were randomly chosen to receive the CT screening or the chest radiograph. The annual screening was modeled for 20 years, with a 40-year follow-up.

The computer model performed cost-effectiveness analyses at each step and for each parameter in the clinical pathway. Each unscreened participant faced the probability of staying alive without clinically apparent lung cancer, developing lung cancer and dying from it, or developing lung cancer but dying from other causes. Screened participants were given the same overall risks of developing lung cancer, with additional pathways developed for those diagnosed with indeterminate nodules. Participants in groups with suspicious lesions and indeterminate nodules underwent a series of tests and interventions. Those ultimately diagnosed with lung cancer were treated with various management strategies (ie, chemotherapy, radiation, surgery).

Widespread screening of asymptomatic populations has inherent biases—eg, overdiagnosis, and lead-time bias (the perception that screened individuals live longer with the disease than unscreened people when, in fact, their lives are not extended but the disease is simply known about longer)—and the authors adjusted the model to account for these and other biases using rates from other published studies. False negative and false positive rates were factored into the model as were rates of patient nonadherence to clinical advice. A histologic bias was even considered, since cancers detected by helical CT tend to be peripheral tumors, which are more likely to be adenocarcinoma. Endobronchial lesions are more often missed by CT (false-negative) and are more likely to be squamous cell carcinoma [7].

Analysis was conducted for a base-case scenario in which the most accurate estimate for each parameter was used. Next, one-way sensitivity analyses were performed under different extremes for each parameter to assess which were most influential for cost-

effectiveness. Finally, a multivariate analysis taking into account changes across multiple parameters was performed using favorable and unfavorable conditions.

Several outcomes were measured to determine cost-effectiveness. The absolute and relative differences in lung cancer-specific deaths were calculated. The number of unnecessary (false-positive) screening tests performed was estimated, as well as the harm from these tests. Finally, the effect of screening on quality-adjusted life-years (QALYs) was determined.

Results

In the base-case scenario of the current smoker cohort, there were 4168 lung cancer deaths per 100 000 persons in the unscreened population and 3615 per 100 000 in the screened group. The absolute mortality reduction was 553 deaths per 100 000 persons, or 13 percent. Those in the screened group underwent 1186 invasive tests or surgeries for benign lesions. The calculated cost-effectiveness of screening was \$116 300 per QALY gained. For the quitting and former smokers, the cost-effectiveness was \$558 600 and \$2 322 700, respectively, per additional QALY.

The authors were also able to change the parameters to create a *best-case* scenario. This model used current smokers only, decreased nonadherence, decreased cost of CT, increased quality-of-life improvement for detection of small lesions, decreased the length-time and overdiagnosis biases, and eliminated consideration of anxiety over unclear diagnosis from the QALY formula. Under these ideal circumstances, the absolute reduction in lung cancer mortality was 900 people per 100 000, a 16 percent relative difference. The number harmed by unnecessary tests increased to 1520 per 100 000, and the cost per QALY gained was \$42 500. Quitting smokers and former smokers had adjusted costs of \$75 300 and \$94 400 per QALY gained, respectively.

Conclusions

Even under the most favorable of circumstances, CT scanning at current cost per scan seems unlikely to be highly cost-effective as a screening test for lung cancer. Widespread screening causes harm in individuals with benign lesions who undergo invasive follow-up testing.

Critique

The gold standard for determining the actual cost-effectiveness of CT scanning for lung cancer screening is a large-scale randomized controlled trial. Such studies are currently under way, and, until those results are available, pilot studies and computer models will be relied upon to predict the utility of CT-based screening. Mahadevia and colleagues have performed a thoughtful, well-designed computer-simulated model analysis to answer this question, accounting for many variables, from adherence rates to costs of CT scans. The authors made many adjustments in the model and analysis to favor screening efficacy. Indeed, one criticism is that their model is too optimistic and that the actual cost is even greater than they project. An additional limitation of the study is that it did not account for other incidental findings. As more people undergo whole-body (rather than chest-only) CT scanning, diagnoses of incidental renal and adrenal masses, aneurysms, and other abnormalities increase. The authors discussed this limitation in the

paper but estimated that it would have little impact on the cost-effectiveness of screening for lung cancer. Furthermore, a computer model is only as good as its assumptions. Changes in technology and advancements in diagnostic and therapeutic technique will also impact the calculations.

As the consumer demand for screening CT scans increases, health care providers are placed in the awkward position of weighing the individual versus societal costs and benefits of screening. The ethical responsibility of conscientious physicians is to educate patients and colleagues and avoid unnecessary screening. Until the results of large scale randomized controlled studies are available, the best evidence (including this article) suggests that CT-based screening for lung cancer is too much for too little.

Question for Discussion

The journal article authors report that, in their computer model, there were 553 fewer deaths from lung cancer per 100 000 current smokers who received CT screening than among the 100 000 current smokers who did not receive scanning—a 13 percent reduction. The authors calculated that the cost-effectiveness of the screening was \$116 300 per quality-adjusted life year and concluded that CT scanning for lung cancer is not highly cost-effective. Does this information alter the advice you would give a patient who smokes and wants to have a CT scan of his or her lungs to see whether he or she has lung cancer? Does, or should, this information affect government health policy about reimbursement for lung CT screening?

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Clinical Pearl

What Makes a Screening Exam “Good”?

by Cheryl Herman, MD

Screening tests are used to determine whether an asymptomatic individual has an undetected disease or condition. Screening is currently used in many contexts, including blood pressure monitoring for identifying hypertension, prostate-specific antigen measurement for signs of prostate cancer, colonoscopy for detection of colorectal carcinoma, and mammography for evidence of breast cancer. Unfortunately some screening tests lack credible scientific bases, and the risks and benefits of testing are frequently misrepresented to the patient. Many of the tests are marketed directly to the patient [1], so it is important for people to know what makes a screening exam “good.” How do we know that a screening study accurately determines the likelihood that a patient does or does not have the disease in question?

The 2 major objectives of a good screening program are: (1) detection of disease at a stage when treatment can be more effective than it would be after the patient develops signs and symptoms, and (2) identification of risk factors that increase the likelihood of developing the disease and use of this knowledge to prevent or lessen the disease by modifying the risk factors [2]. To fulfill these objectives, a screening test and the disease it screens for must meet the following criteria.

The disease in question should:

- constitute a significant public health problem, meaning that it is a common condition with significant morbidity and mortality.
- have a readily available treatment with a potential for cure that increases with early detection.

The test for the disease must:

- be capable of detecting a high proportion of disease in its preclinical state.
- be safe to administer.
- be reasonable in cost.
- lead to demonstrated improved health outcomes.
- be widely available, as must the interventions that follow a positive result [1].

These criteria bear a closer look.

The Screened-for Disease or Condition

The preclinical phase of a disease starts with the onset of the disease process and lasts until signs and symptoms appear, which is when the clinical phase begins. The

detectable preclinical phase is the interval during which the disease is detectable by screening, but the patient is still asymptomatic. During this period, there is a critical point at which intervention is more effective than if started after the clinical phase begins.

The disease being screened for must be serious enough to warrant testing asymptomatic people. The disease should be one that, if not found in its detectable preclinical phase before the critical point, will become life-threatening or cause significant morbidity. If the critical point occurs soon after the start of the detectable preclinical phase, screening may be too late to be helpful.

Pseudodisease is a condition detected by screening that does not require treatment because it will not adversely affect the patient's life. Type I pseudodisease refers to conditions that might not progress to symptomatic disease and may even regress. A commonly used example of Type 1 pseudodisease is ductal carcinoma in situ of the breast, which may remain in an intraductal state and not progress to invasive carcinoma and may even regress to atypical ductal hyperplasia (ADH). Type II pseudodisease is an indolent, slowly progressive disease found in conditions with a long detectable preclinical phase. Often, this type of pseudodisease cannot be diagnosed until after the patient has died from other causes, when autopsy results reveal histologic evidence of, for example, prostate, breast, or lung cancer that was previously unknown. If pseudodisease conditions such as these are treated, the patient may be considered "cured" because he or she died from a cause other than cancer. But designating such outcomes as "cures" is erroneous because the cancer—even if *untreated*—would not have killed the patient before the time that he or she actually died of other causes.

To justify their cost, screening tests must be able to detect a high number of cases of preclinical disease in the screened population. If prevalence of the condition or disease is low, screening will not identify many cases, rendering the test less cost-effective. In addition to cost considerations, some tests are not without risks of their own (eg, radiation) or discomfort. To justify administering these tests to the population, the potential harm to the patient if the disease is not diagnosed must outweigh the distress or pain of the test.

The Screening Test

In an effective screening program, the test must be inexpensive and easy to administer, with minimal discomfort and morbidity to the participant. The results must be reproducible, valid, and able to detect the disease before its critical point.

Screening tests must be widely available to the population for which they are intended. They cannot be available only at academic or other large medical centers. The tests must not have associated morbidity or mortality—even minor side effects may offset the benefits of screening. The test must also be reasonably priced, otherwise insurers may not provide coverage, and patients may be unable or unwilling to pay for the tests themselves.

The usefulness of the screening test is evaluated by its *sensitivity* and *specificity*. Sensitivity is the true positive rate; that is, the probability that a patient with a positive test result has the disease. As sensitivity increases, the number of patients with preclinical disease not diagnosed by the test decreases. Specificity is the true negative rate; the probability that a patient with a negative test result does not have the disease. A highly specific test produces a small percentage of erroneously positive results. Sensitivity is usually increased at the expense of specificity when the disease is serious and curable in its preclinical phase. However, high specificity may be desired over sensitivity when the costs or risks of further testing are significant, as they are, for example, with surgical biopsy. Patients must be informed that a negative screening result does not mean disease is not present, but rather the likelihood of disease is low. Since few tests have both high sensitivity and high specificity, multiple tests are often used to aid in detection of disease in the preclinical phase.

Screening test results must be reproducible. There are 4 frequent causes of variability: (1) Patient-related variation seen with cardiac motion or changes in patient size; (2) test-related variation, seen in patient positioning changes or technical factors in film development (such as in mammography); (3) intra-observer variability due to the differences in interpretation of a test at different times by the same clinician; and (4) interobserver variability due to variation of interpretation of a test by 2 or more clinicians. The last 2 often occur in interpretations of radiologic screening exams such as mammography. Interobserver variation may be minimized by use of strict criteria during interpretation.

Evaluation of Screening Tests

Comparing the outcomes of screened and unscreened groups can be challenging due to several biases. Lead-time bias refers to the fact that patients whose diseases are detected by screening before they experience symptoms have a longer survival time from diagnosis to death. But this seemingly increased life span is not due to the screening, it is merely the added time interval between the diagnosis of disease at screening and the time at which it would have been detected had the patient waited until the onset of signs and symptoms. Although overall survival—from onset of disease to death—may be the same for both screened and unscreened patients, the cause-specific survival, which is the time from diagnosis to death, may seem longer for screened patients because of their earlier diagnosis. In such instances, there is no advantage for the patient, and there may even be a disadvantage, since the screened patient has knowledge of the diagnosis for a longer period of time, which may increase emotional or psychological stress.

Not all diseases advance at the same rate. Those diseases with a long preclinical phase have more favorable prognoses, regardless of when they are diagnosed. When patients with these diseases are overrepresented among screen-detected cases, length-time bias occurs. Length-time bias could lead to the mistaken conclusion that screening is valuable, when the differences in mortality rate are actually due to the detection of less rapidly fatal diseases, while diseases that are more rapidly fatal were diagnosed after symptoms began.

Comparison of cause-specific mortality rates (the number of deaths in a population due to a specific cause divided by the total population) for screened patients versus rates for those patients whose diagnosis was made after the onset of signs and symptoms offers the best measure of the effectiveness of a screening program. Lead-time and length-time biases are canceled, and, while it is not possible to attribute all differences in mortality rates to screening programs, it is highly likely that at least some of the difference is due to them.

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Health Law

When Does the Malpractice Tort Clock Start Ticking?

by Faith Lagay, PhD

When it reached the Massachusetts Supreme Court, the case of *Franklin v Albert* [1] asked a specific question: how long after an alleged medical error can the person who claims injury file and pursue a malpractice suit? The question and the Massachusetts court's answer have gained importance in an era of widespread screening for asymptomatic disease. If a patient suffers harm as the result of an error in prescribing or a mistake during surgery, that harm is known to the patient or others soon afterward, certainly well before the time limit expires for filing personal injury claims in most jurisdictions. But if a mammogram, say, or a lung CT is misread as negative for cancer, that person has no way of knowing about the mistake, perhaps not until he or she develops symptoms. If the individual who was tested develops the disease years later—as happened to Peter Franklin—can he or she still sue the radiologist who misread the image? Within how many years after the x-ray or other scan took place must the claim be brought? That is the question that *Franklin v Albert* asked the court.

Peter Franklin's Case

Peter Franklin was a second-year medical student when he checked into Massachusetts General Hospital in January 1974 to have his wisdom teeth extracted under general anesthesia. He was experiencing some chest pain at the time, so a chest x-ray was ordered. Franklin underwent the oral surgery and was discharged 2 days after his admission by Thomas Albert, a resident, who noted on the discharge summary that Franklin's presurgery chest x-ray had been normal.

In January 1978, Franklin returned to Mass General for a chest x-ray, this time because he had flu-like symptoms. On this occasion, the x-ray showed "an enormous tumor filling Peter's chest, compressing his lungs from the middle and pushing outward" [2]. Franklin was diagnosed with Hodgkins disease. Surprised that the disease had progressed to such an advanced stage without detection, Franklin's father, a physician who was also on staff at Mass General, had 1 of the radiologists pull his son's 1974 x-ray. Not only did this radiologist find evidence of a mass in the earlier film, he found that the radiologist who had read it in 1974 had also noted "an apparent left superior mediastinal widening" and had recommended further evaluation of the abnormality [3].

Peter Franklin's disease, which might have been cured by radiation had it been diagnosed 4 years earlier, required months of chemotherapy and high doses of

radiation. A new chemotherapy regimen was employed to combat the resistant malignancy and so weakened Franklin's immune system that he suffered a severe viral infection of the lungs, forcing him to take a leave of absence from medical school [4]. Franklin brought suit against Dr Albert and Mass General 6 months after discovering the 1974 radiology report. The attorneys for defendants Albert and Massachusetts General Hospital asked the court for summary judgment—that is, a decision in their favor that precluded the need for trial—because, under Massachusetts General Law, suits for medical harm had to be brought within 3 years of the injury. Inasmuch as 4 years and 6 months had elapsed between the 1974 x-ray and the 1978 suit, the trial court granted Albert and the hospital the summary judgment they requested.

When Does the Cause-of-Action “Accrue”?

In the words of chapter 206, section 4 of Massachusetts General Law, as amended in 1965, “actions of...tort for malpractice, error or mistake against physicians, surgeons, ...hospitals...shall be commenced only within 3 years next after the cause of action accrues” [5]. The trial court that first heard the Franklin case had relied upon that section of the General Law and also upon a precedent case, *Pasquale v Chandler* [6], to determine at what point that cause-of-action clock began to tick away the 3 years. The *Pasquale* court had ruled in 1966 that the cause of action “accrues” at the time the malpractice *takes place* and “not when the actual damage results or is ascertained” [7]. In Franklin's case that meant that the statute of limitations clock had begun ticking when the January 1974 x-ray was taken and had expired 3 years later in January 1977, 1 year before Peter's symptoms led to the second x-ray and the Hodgkins diagnosis.

Looking at these facts, the Massachusetts Supreme Court recognized that the *Pasquale* decision could deprive injured parties of access to remedy before they were even aware that they had been harmed. Such a ruling was unjust in the view of that court, which decided instead that “a cause of action for medical malpractice accrues when the plaintiff learns, or reasonably should have learned, that he has been harmed by the defendant's conduct” [8]. There was no “reasonable” way that Peter Franklin could have learned, upon leaving the hospital after oral surgery in 1974, that he had been harmed by Dr Albert's inaccurate discharge summary of the chest x-ray.

Implications of *Franklin v Albert* in an Era of Widespread Screening

The 1980 decision of the Massachusetts Supreme Court that the cause of action in medical malpractice “accrues” when the plaintiff learns of the harm still stands and is in line with the discovery rules in the vast majority of states. As asymptomatic screening becomes more popular in our increasingly health- and mortality-conscious society, the ruling has a message for patients and physicians. The decision warns, or should warn, patients and other members of the public to be certain that all screens and tests they undergo for medical conditions will be interpreted by physicians. Physicians are accountable for their interpretations of screening and diagnostic tests, the Massachusetts decision tells us, long after those interpretations are recorded. Should an error occur, as in the case of Peter Franklin, the patient can recover

economic harms. When physicians do not order screening or interpret the results—as may occur in some commercial screening contexts—patients may find it more difficult to obtain timely recourse for harmful oversights.

It is in patients' best interest to take the results of unordered, commercial screens to their own physicians immediately. While *Franklin v Albert* says patients have 3 years from the time they discover a harm until they can file a claim, certainly no prudent person would allow so much time to elapse before having screening results interpreted and receiving proper recommendations and, if necessary, treatment.

For physicians, *Franklin v Albert* underscores once again the critical importance of communication and follow-up among all members of a patient's care team. One must wonder how it came about that neither Peter Franklin's oral surgeon nor his anesthesiologist discussed his x-ray findings and recommendations with him.

Because of the increased marketing of screening exams to the public, there are some indirect implications of *Franklin v Albert* for physicians. More patients are requesting screening exams in the absence of symptoms and bringing reports from tests and scans done in nonclinical settings to their physicians, asking what the reports mean. In response to this trend, the American Medical Association recently developed policy on the responsibilities of physicians who perform tests they do not deem medically necessary at the request of the patients. This policy, Opinion 8.045 Direct-to-Consumer Diagnostic Imaging Tests, states that "once a physician agrees to perform the test, a patient-physician relationship is established, with all the obligations such a relationship entails" [9]. Further, "in the absence of a referring physician who orders the test, the testing physician assumes responsibility for relevant clinical evaluation, as well as pre-test and post-test counseling" [9]. Hence, physicians who test, or interpret tests for, patients in the absence of medical indication assume the responsibility for harms that accrue to the patient as a result of misinterpretation of results or failure to recommend appropriate follow-up.

Follow-up tests can themselves expose patients to risk and discomfort, sometimes unnecessarily. Physicians must discuss the risks of invasive follow-up tests with patients and be willing to help them decide whether those risks are warranted and acceptable. And, of course, the screen results, the discussions, and the patient's decision must be documented.

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Policy Forum

What Good Is Hypertension Screening If You Don't Do Anything about It?

by Christian J. Krautkramer

Introduction

Hypertension, or high blood pressure, one of the most common diseases worldwide, has special significance in the United States. Nearly one-third of Americans are hypertensive, and approximately half of them don't realize they should seek medical intervention [1]. Because it affects so many individuals and frequently contributes to other morbidities (and potential mortalities), hypertension represents a high cost to society and a major public health challenge. Research has shown that hypertension is the most significant—and modifiable—risk factor for coronary heart disease (the leading cause of death in North America), stroke (the third leading cause), congestive heart failure, end-stage renal disease, and peripheral vascular disease [2].

So-called “primary” hypertension is generally caused by lifestyle factors, such as excess weight; lack of exercise; poor diet with an excess of fats and deficiency of grains, fruits, and vegetables; stress; and use of tobacco products. “Secondary” hypertension is often the result of comorbidities such as kidney disease, underproduction or overproduction of adrenal hormones (including epinephrine, norepinephrine, aldosterone, and corticosteroids), and diseases of the heart and aorta [3]. There is also a growing body of evidence that many people are genetically predisposed to hypertension, regardless of healthy diet and lifestyle, and researchers are working to develop drugs specific to these predispositions [4, 5].

Screening Guidelines

Early detection of hypertension is key to effective disease management. Educational efforts by government agencies, health promotion foundations, and specialty medical societies have urged both patients and physicians to start screening early and to formulate preventive lifestyle and treatment strategies. These educational efforts have led to a wide availability of blood pressure testing, often at little or no cost outside a physician's office or clinic. Both screening and treatment guidelines in the United States are issued by the Joint National Committee on Detection, Evaluation, and Treatment of High Blood Pressure (JNC), an independent group organized by the National Heart, Lung, and Blood Institute. According to the JNC, everyone should have a blood pressure check at least every 2 years. People at increased risk for hypertension may need more frequent readings. Elderly people should be screened for hypertension at every health care visit and at least annually. Those with certain risk factors, including being overweight, having a family history of hypertension or heart disease, or being of African American or Hispanic heritage,

should be screened more frequently. Before a diagnosis of hypertension is determined, an individual should have a high reading on at least 2 separate occasions with at least 2 separate measurements on each occasion [6].

The Gap between Screening and Treatment

A sizable gap remains between recommendations for screening and the ability to offer subsequent treatment to many of those whose results indicate that they should have follow-up care. In many cases lifestyle changes are not sufficient to reduce hypertension and its associated comorbidities, and physicians must prescribe 1 or more of the several classes of antihypertensive medications. These include diuretics; beta blockers and alpha blockers; calcium channel blockers; angiotensin-converting enzyme (ACE) inhibitors; and angiotensin II receptor blockers. Since hypertension screening is simple to conduct and inexpensive, medical authorities feel justified in recommending universal screening. But the cost of follow-up care—in particular, antihypertension medications—makes it difficult for many to make the transition from screening to treatment. Uninsured adults, particularly African Americans and Hispanics, with common chronic conditions such as hypertension, suffer serious, identifiable gaps in needed medical care. Among the key messages in the JNC report: “The most effective therapy prescribed by the most careful clinician will control hypertension only if patients are motivated. Motivation improves when patients have positive experiences with, and trust in, the clinician” [6]. Many of the populations most at risk are also populations least likely to be able to afford therapy, regardless of “motivation” due to lack of health insurance and, therefore, lack of access to treatment. In essence, health policy makers need to ask the question, “What good is hypertension screening if you don’t do anything about it?”

Limited access for the uninsured and minority populations

Regardless of what the guidelines suggest for postscreening follow-up, lack of insurance puts a damper on patients’ ability to purchase needed medications. It is well known that those without health insurance or those with coverage inadequate for necessary care will be far less likely than those with sufficient insurance coverage to seek out medical services and purchase prescription drugs [7]. According to the National Center for Health Statistics, nearly half of all uninsured adults with chronic conditions have reported forgoing needed medical care or prescription drugs due to cost; one-third reported unmet need for medical care, and 1 of 3 reported an unmet need for prescription drugs. These individuals are also far less likely to take advantage of low-cost means to reduce their risk for chronic health conditions through better nutrition, higher rates of exercise, lower alcohol consumption, and tobacco-use cessation [7]. For example, smokers in the lowest income brackets are less likely to quit than those in higher income brackets, in part because higher income is correlated with greater health knowledge, a receptivity to new health information, and ability to take advantage of health-enhancing opportunities [8].

A recent study notes that annual deaths from 3 leading causes—heart disease, cancer, and stroke—are significantly greater in minority populations. These illnesses and related chronic conditions—hypertension, diabetes, and obesity—are the key

contributors to excess levels of ill health, premature mortality, and disability among African Americans and Hispanics [9]. In addition, the National Health Interview Survey, a program sponsored by the Centers for Disease Control and Prevention, estimated that approximately 30 percent of Hispanic persons and 20 percent of African Americans in the US are uninsured [7]. It's not difficult to see from these findings that many in the most susceptible population will be unable to afford treatment should screening reveal hypertension.

Although whites make up the largest group (59 percent) of uninsured adults with chronic conditions, a significantly larger proportion of African Americans and Hispanics with chronic conditions are uninsured [10]. These economically disadvantaged African Americans and Hispanics are, on the whole, less likely to reduce high-risk behavior or to initiate new health-enhancing practices that would help reduce hypertension and its associated comorbidities. About a third of uninsured African American adults and a slightly higher percentage of Hispanics with chronic conditions lack a consistent source for health care [7]. About half of uninsured African American adults who had a chronic condition also had an unmet need for either medical care or prescription drugs; 35 percent reported an unmet need for medical care; 36 percent reported an unmet need for prescription drugs.

The underinsured and prescription drug formularies

Even insured individuals face restrictions in the classes of medications for which their insurers will pay. Most physicians adhere to the JNC when prescribing first-line therapies to patients with uncomplicated hypertension. In their most recent report, the JNC recommends that, "because diuretics and beta blockers are the only classes of drugs that have been used in long-term controlled clinical trials and [have been] shown to reduce morbidity and mortality, they are recommended as first-choice agents unless they are contraindicated or unacceptable, or unless there are special indications for other agents" [6]. In individuals with several coexisting diseases including type 1 diabetes, some kidney diseases, heart failure, and a history of myocardial infarction, newer, more expensive classes of antihypertensive drugs (including ACE inhibitors and angiotensin-receptor blockers) may be more effective [4]. Other evidence suggests that the newer classes are highly effective in persons of a certain age or racial or ethnic background [11, 12]. Sometimes insurers do not designate the newest or more expensive medications as part of their "formulary." More frequently, insurers will create tiers of several copayments, where newer antihypertensive medications cost more out-of-pocket for patients [13]. This can lead patients to choose drugs that their physicians believe are less effective. While having insurance dramatically reduces the problem of unmet need for services, it does not eliminate it entirely.

Dovetailing Screening and Treatment

Straightforward screening and treatment guidelines ignore the inconsistencies in the ability for uninsured populations to follow-up on postscreening recommendations. Certainly it is outside the purview of a clinical practice oversight body such as the JNC to make policy recommendations. Any clinician or public health improvement

group should be troubled, however, if follow-up care is not available to the populations that need it most. Barring substantive reforms to the American health insurance system, piecemeal and politically feasible policies could be implemented to address hypertension in the most at-risk groups. Some studies have suggested that expanding government-sponsored health coverage to nonelderly, low-income persons with hypertension and associated multiple comorbidities (eg, diabetes and heart disease) will not only greatly improve their health past age 50, but will also save money in the long term by paying for preventive therapies “up front” rather than for costly long-term care near the end of life [14]. Patient-assistance programs (PAPs) sponsored by major pharmaceutical companies have been another helpful way to provide prescription drugs free of charge to low-income patients who meet certain requirements. But most uninsured patients—and many medical professionals—are not aware that such programs exist. Further, because PAPs are administered at health care facilities, it can be difficult for uninsured individuals who don't have a consistent source for health care to stay on those programs. National pharmacy chains, pharmaceutical manufacturers, and the government could work together to set up a program whereby individuals, once registered for a PAP, could pick up their medications at any participating pharmacy. Patients would be required, as they are currently, to renew their medication each year at an office visit with a physician or other qualified medical professional.

Lifestyle education, though, remains the best and least expensive way to control hypertension. In the case of diabetes, directed, comprehensive patient education not only improves health outcomes but reduces overall costs associated with the disease, including medications [15, 16]. Such programs are now available in clinics, hospitals, and through nonprofit educators, either free or at low cost to patients. Creating incentives for clinicians to direct their hypertensive patients to these programs—and follow-up to make sure they've attended—is a sound way to promote individual health, especially for those unable to afford medications on a regular basis.

Conclusion

Clear guidelines for screening chronic health conditions are important. Such guidelines, especially when crafted by consensus of top advocates for prevention and treatment of a particular disease, are powerful ways to publicize the need for early and persistent care. Hypertension guidelines developed by the JNC have set a standard of care that aims to provide the best prognosis for all patients. But these guidelines become moot when populations such as the uninsured or underinsured lack access to the treatments recommended within them. Hypertension, in particular, remains a disease that is disproportionately prevalent among the uninsured and underinsured. Because screening for hypertension is widely available at little or no cost, many hypertensive individuals know they are at risk for worse disease conditions later in life. But those without insurance generally lack the ability to follow up on physician-recommended treatments that would reduce the instance of dangerous comorbidities because of the costs involved, predominantly the expense of antihypertension medications. Some underinsured may find that formularies set by

insurers restrict the class of medications available to them, even when their physician suggests a restricted class as a first-line therapy. While clinical practice oversight bodies, such as the JNC, are not socioeconomic policy makers, there must be a better connection between making clinical policy and providing a means to get care to at-risk populations. This serious gap between screening for and treating hypertension leaves the populations most at risk without a way to improve their health and live their life to a fuller potential.

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Medicine and Society

Recognizing Our Responsibilities

by Adrienne J. K. Carmack, MD

Public screening programs are commonplace. Medical students are often encouraged to participate in providing such programs and many continue to contribute to this type of activity throughout their medical careers. Programs such as cholesterol checks at the mall, breast exams and mobile mammography units, and prostate cancer screening with digital rectal exams on site and laboratory testing for prostate-specific antigen levels (with results mailed to the screened individual) are examples of public screening programs. Unfortunately, following up with the patients who undergo public screening programs is extremely difficult, and no evidence-based guidelines for these types of programs exist. Given this lack of data, and the implication of screening results, those who participate in public screening should give thorough consideration to several points. Screening outside of a clinic setting poses unique challenges; the responsibilities incurred in this setting differ significantly from those physicians commonly encounter.

The Goals

One goal of physicians is to prevent disease. Screening may allow us to do so, or at least to identify disease at a stage in which it can be treated more effectively. The goal of a public screening program is to search for disease in populations that are at high risk by using minimally invasive tests to detect disease or risk factors before symptoms develop. The intention is, of course, that once the patient is diagnosed, he or she will seek medical care elsewhere to treat whatever abnormality or risk factor is discovered. Screening is a short-term clinical encounter for what will prove to be a long-term intervention for any patient with positive results.

The Challenges

The most obvious challenge of public screening lies in the nature of the patient-physician relationship. The interaction with the physicians, medical students, or other health care professionals who conduct the screening is necessarily brief. A full history and physical is rarely, if ever, done, limiting the physician's ability to counsel patients in a personalized manner. Often, test results are not available on-site. With prostate cancer screening programs, for example, patients undergo one on-site test (a digital rectal exam) and then wait for the serum prostate-specific antigen measurement results to come in the mail. It would be rare, then, for the physician who conducts the screening exam to identify potential patients; it is more likely that individuals being screened already have physicians to whom they will report their results *or* that they are un- or under-insured and hence unable to get routine care. If a potentially serious diagnosis is

uncovered during a screening test, a person whom the individual has just met must deliver the diagnosis and somehow ensure that the recipient of the news will have follow-up care, usually from another physician in another setting.

It is also necessary to step back and look at screening in the context of the limited access to health care that characterizes our system in the US. Those individuals who cannot afford health care may be unable to seek care for any condition that the screening uncovers. It is critical that those organizing screening programs make sure *before* screening takes place that the appropriate follow-up tests and physician visits will be available to those who need them. Screening in and of itself does not serve to help any one patient; the benefit comes from receiving effective care after results are known.

Another ethical challenge in public screening is ensuring informed consent. Often a person is shopping in a public place when he or she sees an offer for a free medical test and decides to participate. Even though performing the test is easy, the person tested is done a disservice if he or she is not fully counseled about the intentions of the test (what it really can show), the implications of positive or negative results, and the need for follow-up studies in the case of certain findings. Anyone who expresses an interest in a public screening program should be given this information and required to sign a written consent form that outlines them.

Ultimately, our intentions are to help the public while respecting the principles of beneficence and nonmaleficence. Anyone who does not understand the test, cannot obtain follow-up care, or is inappropriately diagnosed or counseled because of the lack of a patient-physician relationship does not benefit from the test.

The Reality

The major, sustainable benefit of public screening lies in education. Only by educating our patients can we ensure that the real goal of screening will be met after the brief testing encounter. It is more important to inform patients about the existence of a test and ways that they can obtain medical care than it is to actually perform the test in the restricted environment with the myriad challenges just described. Patient education should be the primary goal of all public screening programs if we wish to maximize our potential to improve the lives of our patients and the public.

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Op-Ed

Is Prenatal Genetic Screening Unjustly Discriminatory?

by Jeff McMahan, PhD

Occasionally when an infant is born with terrible but unforeseen afflictions, the parents wish to allow it to die, but their decision is contested in the courts by medical personnel. In other instances, such as the recent case of the infant Charlotte Wyatt in the United Kingdom, doctors object to devoting scarce medical resources to keeping a severely afflicted infant alive, judging that death would be in its best interest, while the parents fight through the courts to obtain the necessary treatments. These tragic conflicts might be avoided if we practiced more extensive screening that would allow potential problems to be detected prenatally or, ideally, prior to conception. It's usually true at present, of course, that prenatal screening can obviate later problems only if it's followed, in relevant cases, by abortion, and many people object to abortion on moral grounds. But even those who are opposed to abortion tend to believe that an early abortion is less objectionable than allowing a newborn infant to die after experiencing a brief and perhaps painful life. And even though an early abortion is seldom morally or emotionally unproblematic, it is considerably less wrenching for the parents than having to acquiesce to the death of a tiny being to whom they may have become deeply attached.

There are various means that people may employ to determine whether their possible child would have serious impairments. These include preconception screening of potential parents, screening of embryos prior to implantation when in vitro fertilization is employed, and screening of fetuses in utero. The last 2 methods, as I noted, may result in the killing of a being that many people believe it would be wrong to kill. But some people object to screening for impairments for reasons that are independent of any objections they may have to the killing of embryos or fetuses. They claim that screening is perniciously discriminatory in that it seeks to rid the world of certain types of people, that it reduces the number of disabled people, thereby diminishing human diversity and increasing the isolation of existing disabled people, and that it is hurtful to the disabled because it implies, in effect, that it's bad if people like them exist or that the lives of the disabled are worse than the lives of others.

These are legitimate concerns. But they're insufficient to show that screening is wrong or that it should be prohibited. For if these reasons were strong enough to show that screening is wrong, it would also follow that it would be permissible deliberately to cause oneself to have a disabled child. For if it's *mandatory* to *allow*

oneself to have a disabled child rather than to try—by the use of screening—to have a child who would not be disabled, then it should be at least *permissible* to *cause* oneself to have a disabled rather than a nondisabled child. Indeed, to deny that it would be permissible to cause oneself to have a disabled rather than a nondisabled child would seem to express a negative view of the disabled and could contribute to a reduction in the number of disabled people, something that opponents of screening fear. So, if you think it would be wrong for someone to *cause* herself to conceive a disabled rather than a nondisabled child—for example, by taking a mutagenic drug prior to conception or by selecting and implanting a genetically defective rather than a genetically healthy embryo—then you can't consistently believe that it's wrong to screen for disabilities.

Many people, myself included, believe that it's permissible to conceive a disabled child when the alternative is to have no child at all. And some people are willing to accept that it's permissible to conceive a disabled child even when it would be possible to conceive a *different* nondisabled child instead. These views can be defended by noting that in neither case would there be a *victim*, for causing a disabled child to exist isn't worse for that child than never existing at all, provided that its life would be worth living.

By contrast, virtually no one thinks that it would be permissible to cause an already existing individual to be disabled when he or she would otherwise not have been disabled—for example, through the infliction of prenatal injury. For here there would be a victim. Yet some advocates for the disabled seem to be committed to accepting even this—that the infliction of disabling prenatal injury is permissible. These are people who object to screening for the reasons I have cited *and* deny that it's worse in itself to be disabled than not to be. On their view, the injury would not be bad for the fetus at any point in its life. They could accept that causing an older child or adult to become disabled would be wrong because it would violate her autonomy or force her to endure a period of adaptation to her disability. But these objections don't apply to the infliction of a disabling injury *prenatally*, because a fetus has no autonomy that could be violated and would never suffer the transition from “fully abled” to disabled. So to cause a fetus to be disabled would not, on these people's view, be worse for the individual it would become. And it would increase rather than decrease human diversity and would expand the ranks of the disabled. Moreover, if screening expresses a negative view of disability or of the disabled, then it seems that the same view would be expressed by publicly objecting to or trying to prevent the infliction of disabling prenatal injury. If, therefore, we *do* think it's in general wrong to inflict prenatal injury and that we should try to prevent it, we can't accept the position of those who object to screening in part by claiming that disability is a neutral condition.

While many people use screening to try to avoid having a disabled child, most people who have had a disabled child don't regret it and indeed tend to find special meaning, satisfaction, and even wisdom in their relations with the disabled person. Finding ways to give public expression to the view of the disabled held by those who

know them best could help to offset any negative effects of the practice of screening. This would be better for disabled people than to alienate those who value screening by stigmatizing or attempting to suppress it.

Jeff McMahan, PhD, is professor of philosophy at Rutgers University in New Jersey and the author of The Ethics of Killing: Problems at the Margins of Life.

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Op-Ed

The Uncertain Rationale for Prenatal Disability Screening

by David Wasserman, JD, MA, and Adrienne Asch, PhD

On November 10, 2005, an article in *The New England Journal of Medicine* reported the increasing accuracy of first trimester screening for Down syndrome. The introduction of first trimester tests for the condition was heralded in 1998 by the National Institute of Child Health and Human Development (NICHD), as reducing complications for women who choose abortion. NICHD reportedly spent \$15 million on the study—presumably to fulfill its mission “to ensure that every person is born healthy and wanted.” Of course, few children with trisomy 21 detected in the first trimester are likely to be born at all. NICHD’s mission is also “to ensure that women suffer no harmful effects from reproductive processes,” and that goal may also have provided a rationale for funding the research—many women might see the birth of a child with Down syndrome as a “harmful effect” of their pregnancy. We suggest that *it is difficult to justify prenatal screening for disability on either of these grounds, as protecting the health of the fetus or child or as protecting women from harmful effects of reproduction.*

Prenatal diagnosis—through amniocentesis, chorionic villus sampling, or preimplantation genetic diagnosis (PGD); for Down syndrome, cystic fibrosis, female gender, or blue eyes—needs to be seen for what it is, or more importantly, what it is not. It is not a medical procedure—that is, a procedure intended to protect or restore an individual’s physical or mental health. Rather, it is typically a procedure to identify unwanted organisms. Occasionally, testing is sought to guide the management of delivery and labor. But far more often its purpose is to provide information about fetal characteristics so a woman can decide whether or not to continue her pregnancy.

To say that prenatal testing and any resulting abortion are not medical procedures is not to say that they are wrong or that a doctor is wrong to perform them. A pregnancy test for an unmarried adolescent who does not want a child is not a medical procedure either, nor is the abortion that may follow positive pregnancy test results. We may regard that test and abortion as justifiable, and regard a doctor as the appropriate agent to carry them out, without believing that they serve to protect or restore the health of an individual patient. If doctors can properly perform a non-healing intervention in aborting the unwanted fetus carried by a teenager, can they do so in enabling parents to prevent the birth of a child with Down syndrome?

The answer will depend on whether there is a distinct justification for the intervention that is not based on protecting or restoring the health of individual patients. Two

rationales are often given for the use of prenatal testing, and both gain spurious strength from their conflation with stronger rationales for different practices. The first is the public health rationale of reducing the incidence of genetic disease and “defects.” This rationale elides the striking difference between prenatal testing and true medical preventive measures: for the foreseeable future, prenatal testing can prevent disease and disabilities only by preventing the existence of people who would bear them. Prevention by prenatal screening lacks the obvious justification of most public health measures: preventing medical harm to existing people. While it may be reasonable to treat the incidence of disability among existing people as, in part, a public health problem, it is problematic to treat the existence of future people with disabilities that way. A policy of prevention-by-screening appears to reflect the judgment that lives with disabilities are so burdensome to the disabled child, her family, and society that their avoidance is a health care priority—a judgment that exaggerates and misattributes many or most of the difficulties associated with disability.

We believe the principal difficulties faced by people with disabilities and their families are caused or exacerbated by discriminatory attitudes and practices that are potentially remediable by social, legal, and institutional change—in much the same way that many of the difficulties associated with being African American or female in America have been ameliorated. A policy that promotes selection against embryos and fetuses with disabling traits conveys the strong impression that the problem is the disability itself rather than the society that could do so much more to welcome and include all its members.

The second rationale offered in support of prenatal screening is the enhancement of parental autonomy. The justification for enabling a woman to decide *whether* to have a child is stronger than the justification for enabling her to decide *what kind* of child she will have. Pregnancy makes massive demands on a woman’s body; parenthood involves an enormous, open-ended commitment. To treat the difference between having a disabled and a nondisabled child as being of a similar magnitude as the difference between having and not having a child greatly exaggerates the burden of disability and ignores the source of so much of that burden.

We recognize that people with disabilities and their families face difficulties in our present society and that perhaps some of those difficulties would remain even after comprehensive social reform. But we maintain that few disabilities are so undesirable that they provide good reason for abandoning a parental project, for declining to become a parent to the child who would develop from the diagnosed fetus. Given the difficulties that a disabled child is likely to face in our present society, a prospective parent may have good reason not to *cause* disability, but that is not reason enough to *select against* a fetus with a disability. In creating families, prospective parents should aspire to an ideal of unconditional welcome; an ideal opposed to the exercise of selectivity through prenatal testing. If a child develops a disease or disability—diabetes or attention deficit disorder—loving parents incorporate the challenges posed by that condition into the project of raising and nurturing him. We do not believe that parents should reject those challenges in bringing future children into their families. (It is

important to recognize that most disabilities are caused by accidents or disease, not by genetic variations.)

If, however, we accept the use of biomedical technology to give parents greater choice in the kind of children they have, we should not limit that choice to the avoidance of genetic impairment; we should facilitate testing for any conditions parents might find burdensome or desirable. And even if we are comfortable with such parental selectivity, enhancing it clearly should not enjoy the priority given to measures that protect the choice about whether to become a parent in the first place.

On the other hand, if we object to such unfettered choice as corrupting or debasing the parental role, we should not make an exception for disability. To do so is to treat disabilities as uniquely burdensome, in the face of strong contrary evidence from research on families with disabled children [1-5]. To assume that most genetically detectable disabilities impair the prospects for individual and family flourishing in a way that other potentially detectable characteristics do not is truly to stigmatize disability. While such stigmatization is understandable when it is displayed by anxious couples awaiting a life-transforming event, it should not guide the public funding of reproductive research or the formulation of reproductive policy.

Given the difficulties in justifying the public funding of research and development in prenatal screening, the money spent for that purpose might be better used for research on improving the health, functioning, and longevity of children with genetically based disabilities.

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Suggested Reading

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