Genetic Testing Without Treatment
The Value of Knowledge for Its Own Sake

By Rebecca Leibowitz

As genetic testing comes of age in the practice of medicine, patients may be provided with greater insight into the form and timing of diseases that will impact their lives. Unfortunately, developments in disease treatment do not always keep pace with diagnostic ability. The result of this disparity is that physicians and patients together will have to face the difficult choice of whether or not to pursue knowledge that is unaccompanied by the hope of effective therapies. For some patients, learning of a predisposition for a disease may force them to confront their own mortality for the first time. This may compound the already steep challenge of dealing with an unfavorable prognosis.

Genetic testing has the unique capability of allowing a currently healthy person a glimpse of future illness. Beyond the obvious personal impact of this information, the cultural attitude in the United States towards disability and death reinforces the distress of becoming aware of a predisposition for a disease. In today’s society, emphasis is given to the preservation of youth, beauty, and health at all costs. A common criticism of our social system is the neglect and disrespect experienced by the elderly and the disabled. The implication of loss of health in our society is a loss of perceived worth. Expectations from employers, fear that insurance will be denied, and feelings of love and responsibility for family and friends are all pressures that leave little room for the acknowledgment that physical and mental capacity are not eternal. In addition, these pressures may cause us to view misfortune and illness as contagious and to therefore further isolate those who are affected, or simply “predisposed”. In such an environment, the potentially destructive consequences of genetic information are magnified. The question emerges, how does one integrate an acknowledgment of disease into a healthy view of life? For some people, insight into the future offered by genetic testing may take the form of a gift; for others, a death sentence. Patients in the latter category may perceive a report of a genetic propensity to be an oppressive force that overshadows everything else. They may live in constant anticipation of symptoms indicating that a disease process has begun. Some of these patients may feel that, indeed, ignorance is bliss. At the opposite extreme is the category of patients who require the full, undisguised truth for their own piece of mind, no matter how grave it may be. In these two scenarios, the fact of the genetic predisposition for a disease may be exactly the same; the important difference is the patient’s perception of the benefit that knowledge for its own sake would bring.

Choosing to remain ignorant of one’s genomic particularities is not simply a desire to avoid reality. Selective ignorance can be a very adaptive quality when coping with the stresses of living and dying. If it were possible to envision every event that would take place within one’s lifetime, including the lives and deaths of loved ones, the tremendous weight of compounded grief would fall upon one’s shoulders all at once. Time is an essential factor in the rhythm of life. The ups and downs with which we are all so familiar generally transpire in a cyclical pattern, allowing for appropriate healing to occur. With genetic testing, however, one would have the opportunity to learn of a predisposition for a disease that could define one’s future, with rippling repercussions for relations as well. The weight of this information may far exceed what a patient expects when the decision is made to bring the unknown to light.

As a physician advising a patient on the value of genetic testing, it is important to ensure beforehand that the patient has an understanding of the implications, both dramatic and subtle, of the results. For example, the patient may not have fully considered how a genetic profile may reveal the chances that children and other relatives are carrying the same genes. In the face of this knowledge, the patient may feel compelled to encourage other family members to be tested, or the children themselves may feel this need. Guilt may play a prominent role in the range of emotions for patients with children at risk of developing the disease. It should be remembered that parents have been passing on genes, both
good and bad, from the beginning of time. The capability to track the transmission of genes only reveals the fact of what has always been true.

An important determinant of the utility of genetic testing is the perspective of the individual. The distress of knowing about future illness can itself impact upon present well-being. Although the effects of attitudes and emotions are still controversial, there is some evidence that suggests an interaction between emotional state, immune function and health. A book entitled Healing and the Mind documents Bill Moyers’ interviews with various health professionals on the connection between the mind and the body’s ability to heal. In Bill Moyers’ interview with Dr. Dean Ornish, President and Director of the Preventive Medicine Research Institute at the School of Medicine, University of California, San Francisco, Dr. Ornish states:

“I think there is a scientific basis to the idea that beliefs are powerful. If you believe that you have some control over your life, and that you have the ability to make choices instead of being the passive recipient of medical care, or the victim of bad luck or bad genes, then you are more likely to make changes that are going to do you good in terms both of your behavior and of the direct effects of your mind on your body.” (Dean Ornish, M.D.: in Healing and the Mind, Bill Moyers, 1993, Doubleday, p. 97)

For some patients, the relative certainty afforded by test results, even if it is an undesirable certainty, may impart a sense of control. It is precisely the experience of losing control of one’s body and of one’s life that is so distressing about most disease processes. An active response to a diagnosis may take many forms. Examples include researching the disease in question, being aware of possible future avenues of treatment, and donating time or money to organizations that help people suffering from the disease.

Another aspect of genetic testing that is largely determined by the individual is an interpretation of the statistical probability implied by a genetic predisposition. Rarely is a genetic test 100% predictive; therefore, a patient may be given a percentage, or an estimate of the “chance” that they will develop the disease. The translation of a number into an emotional and psychological reaction opens up a chasm of variability. Some patients may react to a 45% chance of developing a disease by crying with joy, believing that it is less than likely that they will be afflicted. For other patients, a 3% chance may be threatening enough that they respond as if it were a definitive diagnosis. The potential impact of the perception of health on the actuality of health reinforces the need to assist the patient in arriving at a realistic understanding of test results.

In general, genetic testing for diseases with existing therapies may be advisable, especially when early detection may alter the prognosis. However, when there is no effective treatment for the disease in question, wherein lies the benefit of knowledge? If there is no obvious answer to this question from a purely medical perspective, perhaps it is because there is another level of impact. A patient may find it possible to incorporate the expectation of future disease into a greater appreciation of present life. This may be simultaneously the most difficult and the most rewarding response. In his book entitled Who Dies, Stephen Levine describes the gift of awareness that comes with a recognition of the role that death, even in the form of tragedy, plays in life. He states:

“If we can say that grace is a sense of connectedness, that it is the experience of your underlying nature, then we may see how what is often called tragedy holds the seeds of grace. We see that what brings us to grace is not always pleasant, though it seems always to take us to something essential in ourselves.” (Who Dies, Stephen Levine, 1982, Anchor Press/Doubleday, p. 87)

Future physicians may have the capacity to reveal to patients the most likely guise of their own death, and of the disease process that will define their lives for the time preceding death. With this power comes the responsibility of guiding patients through the inevitable psychological, emotional, and social struggles they will encounter. The goal is to have patients choose genetic testing as a component of their health care with their eyes open to all its implications and an appreciation of their role in determining its ultimate value. Genetic information on an individual level has the potential to change people’s lives through the infinitely powerful mechanisms of the mind. The danger lies in distortion and despair; the hope lies in a realistic understanding and appreciation of life.

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