

Genetic Testing

With the near completion of the Human Genome Project comes a new technology that will inevitably change the lives of thousands of human beings. Today it is possible to test a human for more than 400 genes that are related to serious diseases. One can go to the doctor and discover whether or not they will develop Huntington's Disease sometime in their mid-life, a fatal disease for which there is still no cure. As part of the Human Genome Project, an enormous effort is being made to examine the social, ethical, and legal implications of this new technology. The creation of such a tool engenders just as many questions as solutions. In order to see the whole picture, one must examine the current state of genetic testing, its potential benefits and consequences, the steps that are already in place to ensure that this technology is more beneficial than harmful, and the steps that must be put into place in the future in order to ensure that patients are protected and aided by this discovery.

According to GeneTests, a Seattle consortium that monitors genetic testing, there are now tests for more than 400 gene mutations that are linked to hereditary disease (Willing p.2). In a USA Today article from April 23, 2001, Richard Willing discusses the current explosion of this technology and some of the questions that it raises. Genzyme Genetics, for example, has seen a 40% increase in work since 1995. Likewise, a lab in Salt Lake City performed 250 breast cancer tests in 1997 and 10,000 in 2001. These companies compare the DNA of a patient to the discovered genetic code for a particular gene. Scientists then interpret the data (whether or not there is a mutation and what a particular mutation means) and return a positive or negative result. However, these tests alone do not determine whether or not a patient will inevitably contract the disease—the penetrance of some genetic mutations is stronger than others.

The potential benefits of this technology are numerous and extraordinary. First of all, there are many genetic diseases for which cures or helpful therapies exist. If a patient discovers that they will inevitably contract a certain disease, their doctor can keep a more watchful eye on them and schedule regular exams. In addition, regardless of the nature of the disease, the patient can avoid certain risks and look for certain signs on their own. Also, certain diseases can be rendered insignificant with certain pharmaceuticals or enzymatic therapies. My brother, for example, suffers from Wilson's Disease. Wilson's patients do not develop the proper enzymes necessary for removing copper from their blood. This disease can be fatal if not discovered early enough, however with certain enzymatic supplements, a Wilson's patient can live a normal life and even eat many foods with high copper content. I am not certain whether or not a genetic test exists for this disease, but it is clear that with this new technology my brother and myself will be able to learn whether or not our future spouses are carriers of this disease, and we can even guarantee that our children will not have the disease by using IVF techniques. For diseases such as Huntington's, where certain people may be at risk but do not want to know their fate, a company in Fairfax, VA will actually perform IVF to ensure that the patient's children will not have Huntington's without telling the patient whether or not they will contract the disease themselves (Willing, p.2).

For an example such as Huntington's Disease, it is important to consider the fact that some people will choose to know their fate whereas others may choose not to know. By taking the test and discovering that they do in fact carry the HD gene, the patient can then fashion their life with the knowledge that it will end sometime in their forties or fifties (most likely). With this knowledge the patient can decide whether or not they would like to have children (by using IVF perhaps) or whether or not they would like to work a full-time job until they meet their end. However, it is very important to consider those who choose not to take the test and their right to ask a family member who takes the test not to reveal their

results. This problem is discussed in Willing's article as he takes us through the scenario of a 12-year old girl who asked her mother not to take a test for breast cancer related genetic mutations. Claire Smith, the girl mentioned in the article, did not want to be a teenager with the knowledge that she had a 50% chance of contracting a disease in her adulthood because her mother tested positive for certain mutations.

Certain drawbacks to this technology could become more apparent if they are not handled properly. For example, errors in genetic tests or in their interpretations could be extremely serious. The results of this sort of test will certainly change the patient's life; if the result is incorrect the consequences could be tragic. Also, a positive result does not guarantee that the patient will contract the disease, and a negative result does not guarantee that they will not contract the disease. In Claire Smith's case, her mother took the test, received a negative result, and eventually contracted a different type of breast cancer entirely. The fact that these tests are not perfect definitely diminishes their utility. In fact, the Stanford Program in Genomics, Ethics, and Society convened a working group to measure the utility of a test for certain genetic mutations related to breast cancer (BRCA1 and BRCA2) (Koenig et al). This working group concluded that testing for these mutations was only worthwhile in very specific circumstances because the potential negative consequences often outweighed the potential benefits.

Issues of privacy and confidentiality must also be examined as this new technology becomes more and more common. Will health insurance providers or employers have access to the results of a genetic test? In order to avoid genetic discrimination, it is imperative that the privacy and confidentiality of patients be protected. However, what does a doctor do with information that an entire family has a very high risk of contracting a certain disease, but their patient refuses to share that information with their relatives. Does the doctor protect the

privacy of his or her patient, or is he or she required by law to disclose certain pieces of information if they will potentially save lives?

It is clear that by providing a solution to a great problem, many questions must be asked and multiple scenarios must be considered. On a legal level, certain legislation must be implemented to ensure that society and individuals will benefit from this technology rather than suffer as a result of its implementation. The Estonian model of legal protection of gene donors is an exceptional one. By requesting the DNA of a vast majority of its population, the European nation of Estonia was forced to answer some of the questions raised here through anti-discriminatory legislation. In fact, the overall theme of the law is that the use of this knowledge must benefit the patient. By using that particular intention as a framework, the law itself was fashioned in a way that this technology, though new and potentially frightening, can only be used to help the citizens of Estonia. I believe that this law should be used as a model for other nations, and that anti-genetic discrimination laws are necessary if this technology is going to be effective and beneficial.

Many journalists and scientists have referred to recent genetic discoveries as a sort of modern-day "Pandora's Box." In Pandora's case, because of her overwhelming curiosity she unleashed many atrocities and only closed the lid in time to trap hope inside. I truly believe that genetic scientists have re-opened the box in order to let hope out, but it is up to law-makers and other important members of society to ensure that we use this technology solely to improve the lives of those who suffer. The potential for both amazing and terrible things exists within these new discoveries, it is up to us to determine which are let out and which will remain locked inside.