Rose Monahan

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The Effects of Gene Ownership on Personal Genomics

Personal Genomics is an exciting and growing field that may have significant positive impacts on the ability to diagnose and prevent diseases. As research regarding genetics continues, more and more information will be available regarding an individual's health based solely on information garnered from genome tests. While the future of this field holds many potential advantages to the patient and potential advancements in diagnosis, currently the field is flush with legal and ethical debates. The base of the issue is the ownership of both genetic information itself and the interpretation of this genetic information as it pertains to an individual's health. This essay will weigh the possible future advantages of personal genomics against the current downfalls of the field and argue that the decision regarding ownership of genetic information will heavily determine whether the use of personal genomics will ultimately benefit patients.

Background:

Personal Genomics is an evolving field which originated with SNP testing for specific diseases and has the potential to expand into more broad spectrum tests of a whole genome of an individual.[17] As scientists move towards using this diagnostic tool for broad spectrum genome analysis, two major benefits to patients arise: a patient will be able to take precautionary measures in order to prevent a disease for which he or she has a high risk, and the cost of a patient's healthcare will be reduced.

To date the field of personal genomics has not had an influence on diagnostics at a populations level.[1] While the database of known associations between SNPs and diseases is expanding, the total number of associations is low enough that the test may not yet be worth the cost to most patients.[1] In addition the clinical accuracy of these tests is still very low.[1] However, the cost to sequence a single genome has decreased for 100 million dollars in 2001 to just under 10,000 dollars in 2012 according to the Nation Human Genome Research Institute.[2] As this value decreases scientists will be able to fund more studies and scientists will then ideally be able to amass a large amount of data correlating specific genes or SNPs to phenotypic human traits. Once the appropriate correlative data has been recorded, an individual can use the results of a genetic test to determine the *likelihood* of contracting certain diseases. This allows patients to take preventative actions that may allow them to avoid contracting the disease. In addition the genetic screen can act as a more accurate version of the 'family history' normally used by doctors to ascertain predispositions to certain diseases.

Preventative medicine is a cheaper option for the patient and the insurance company, than reactionary medicine. According to the World Health Organization, the United States spends more on healthcare each year than any other country.[21] It spent 2.1 trillions dollars on healthcare last year alone.[21] Preventative care, if applied to every person for every disease would actually increase the overall amount of money spent on health care as opposed to reducing it.[14] However, if there were a way to target the preventative care to only those who need it, this method has the potential to save money and lives.[14] Personal Genomics would be able to do exactly this, and therefore would save both the patient and the government a great deal in healthcare expenditure.

Personal genomics is a field with the potential to save patients the grief associated with illness and to save money for both the government and patients, but it has many ethical and legal issues to sort first. One of the main factors that stands in its way is the determination of ownership of genetic information. The process of interpretation of genetic information encounters three main potential 'owners'. First, there is, of course, the individual who's DNA is being tested. Second, there are the profit-driven sequencing companies and their potential right to patent DNA, and DNA tests. Third, there is the doctor or genetic advisor who will inform the patient of the outcome of their test. All three of these entities could hold a potential claim on either a portion of the DNA or its interpreted consequences for patient health.

The Sequencing Company:

Sequencing companies often work as for-profit institutions. The United States has already experienced some of the issues that arise when combining for-profit companies and patient healthcare in the form of insurance companies. The field of personal genomics has the potential to encounter some of the same issues. There is one such company whose profit-based motivations have been called into question recently.

Myriad is a company whose patent on two genes has sparked enormous controversy over the ownership of DNA. Myriad patented the BRCA1 and BRCA2 genes which, when sequenced, can determine a person's risk for breast and ovarian cancer.[13] The information garnered from these genes can indicate up to five fold increased risk of breast cancer, and up to a ~40 fold increased risk for ovarian cancer.[18] Because Breast cancer is highly prevalent, with up to 1 in 8 women diagnosed in their lifetime, it is no

surprise that this monopoly held by Myriad would spark turmoil.[10] Contenders cite two main issues with Myriad's patent. First, this patent prevents patients from seeking a second opinion regarding their results, a practice that has long been supported and upheld in medicine.[6,11,16] Second, the patent sets a precedent for the practice of patenting a strip of DNA.[11,12,16] Myriad did not create this strip of DNA; it was created by life and evolution. Does discovery alone, when the genes in question are a part of every cell in every human on the planet, validate the right to patent them?

Regarding the first issue, if an individual cannot obtain a "second opinion," they may undergo unnecessary and drastic preventative medical procedures. There is an innate amount of error in any scientific test. For example, one of the current preventative measures for breast cancer is a mastectomy. It is a drastic body change and serious surgery to consider, especially without a certainty regarding the diagnosis. Therefore second opinions are imperative to a company operating under a title of 'healthcare. This points to the larger overarching problem with the upcoming personal genomics field. Currently this field is being run by companies whose regulations have yet to take into account that these companies are now part of a medical field, as opposed to simply a consumer field. One could argue that in the same way that different restrictions apply to a hospital than a hotel, so should different restrictions apply to a genetics company whose products are required for health assessment, and one whose products affect only a consumer market. A counter argument could state that drug companies also hold human lives in their hands but only sell to those who can afford it. The constant struggle between profit driven companies, and individual human health is one that this country is dealing

with on a much larger scale in terms of healthcare, but personal genomics is likely to face some of the same issues.

The second issue deals more directly with the question of ownership. In order for an invention to be patentable it must be non-obvious, novel and useful.[20] Suppose for example, that a company developed a test in order to determine the risk of breast cancer. This test would certainly fall under these three criteria. However, does a gene itself fit these criteria? A newly discovered gene is useful, in the case of the BRCAs, since they can be used as a diagnostic tool. Is the gene novel? This gene is certainly not novel to the human species, and is contained in every human on the planet. However, the patent office considered it novel because it had to be significantly modified/purified in order for Myriad to extract the gene.[20] But whether this distinction alone should be a reason to allow the patent is a highly contentious issue. The patenting of genes under the restriction of novelty is therefore a complex issue. The last criterion is that the subject must be non-obvious. Again the presence of the gene in every human makes it seem somewhat 'obvious'. Because Myriad extracted this gene, it was non-obvious prior to their work, and further the implications it would have for breast cancer were non-obvious prior to Myriad's discovery. Therefore this patent may only fit two of the three patent criteria.

Myriad's trial will likely move to the supreme court in the coming year.[6] If patents for specific sequences of DNA is approved the field of personal genomics will become complex and expensive. By patenting BRCAs Myriad would set a precedent that would restrict the ease of industry-based research on genes. Multiple companies would hold patents for different disease-related genes. In order to get a full scan of all of an

individual's potential risks, the individual might need to go through multiple different companies each demanding their own fees. Further, a second opinion might be difficult to procure. Therefore, if for-profit companies are allowed to 'own' DNA the field of personal genomics may fail to best fulfill its main purpose: encouraging prevention/early management of diseases in a patient.

The Doctor or Genetic Advisor:

Once the sequencing company has assembled the results, someone, most likely a doctor trained to be a genetic advisor, will present these results to the patient. If this role of genetic advisor does fall to medical doctors, then it follows that this information will be recorded in a patient's medical file. This action would be taken with the best interest of the patient in mind. A DNA test is a far better version of a family history, which is one of the primary items of information emergency medical personnel need to know in order to treat a patient.

However, while the interpretation of a DNA sequence (or SNPs) can sometimes lead to definitive diagnostics, more often the data indicate the patient's risk for certain diseases. In the case of the BRCAs the assay performed by Myriad gives the likelihood that this individual will contract breast cancer, or more basically it measures how many mutation events would be required to induce a cancerous growth in an individual.[18] Therefore, this information does not definitively state whether this person has a disease. Looking into the future of personal genomics, there is great potential for the information to be misapplied in a legal sense to the detriment of the patient.

Legally, a court of law can subpoena medical documents. In general medical documents are only allowed in court under specific circumstances, for example, if a party is accused of inflicting physical or mental harm on the defendant.[7] The medical records, while normally protected by doctor-patient confidentiality are released in these instances. This is an accepted procedure because the medical records, are a compilation of facts regarding patient health.[7] However, the results of a DNA test show the *likelihood* the patient will get the disease, and is not a definitive diagnosis. Looking into the future of personal genetics, it is possible that scientists will have methods to assay many more disorders with indefinitive diagnosis results. Psychological disorders come to mind. It does not seem out of the realm of possibility for a court were asked to use the knowledge that an individual had a fifteen fold higher risk of a certain psychological disease as a piece of evidence against them in a court of law.[8.15] Preventative medicine has the potential help the entire population, while the number of people whose medical records are called to court each year is only a minor subset of the total population.[8] Therefore the potential negative aspect of entering the data into the patient's file does not outweigh the potential positive aspects of giving doctors access to this information.

In addition to the risk of entering this medical data into a government system, there is a natural risk associated with entrusting private information with anyone other than the owner. Hospital and patient data are not 100% secure as seen by Stanford Hospital's accidental release of patient data in 2010.[5] Sufficient protective security measures could minimize the risk of release of this information.

In general, doctor access to these files will likely be extremely helpful to the patient and to society in general. Therefore 'doctor' ownership of this information would, in most cases have a beneficial effect on the field of personal genomics because it would aid in the ultimate goal of preventing and managing potential patient ailments. However, doctor ownership of DNA information is not without its downfalls. Because this information provides the probability of a diagnosis, not the diagnosis itself, it has the potential to be misused if called to court.[8,15] There is also the potential for a breach of confidentiality.

The Individual:

The person for whom the DNA is the blueprint of life has the strongest claim to any information presented. They should have the ability to receive all information that can be gathered from the analysis of their genome, whether or not they choose to do so. The question is whether this person has sole ownership over this information. In the case of the sequencing companies and the doctor/genetic advisor, the patient had no choice but to release the information to these parties in order to acquire test results. Does the individual have the right to withhold this information from other interested parties who are not part of the testing process? Examples include insurance companies, school officials, and employers.

Should insurance companies have access to patient disease risk? There is a tradeoff in giving this information to insurance companies. For a person with high risk for many diseases, an insurance company might increase their rates. There is already evidence suggesting that insurance companies will take genetic information into

consideration when raising rates.[4] The issue with using DNA results as a measure of 'healthiness' is that for many diseases, the genetic information merely gives the odds that a patient will get a disease. A person might have a 40 times normal risk of ovarian cancer, but without certain mutation events they could lead completely healthy lives. Contrarily, a low-risk person could contract a disease that a high-risk person avoids. A fraction of the individuals would have higher or lower rates than they should based on their 'actual' health. On the other hand, insurance companies might also use this information in a way that achieves a positive result for the patient. Insurance companies cover more of the preventative medical measures of a high-risk patient in an effort to their lower overall costs for that individual. If the insurance companies were to provide coverage for preventative medicine, informing the company might lead to less grief for the patient and less money spent for the insurance company and the patient. There is as of yet no evidence to support that insurance companies would choose this route. Therefore, the patient should be given a choice regarding the release of this information to their insurance company based on the company's policy.

Should employers be allowed full access to a patients DNA records? This question can be answered with an assured 'no' based on precedent. Employers are not currently allowed access to all of an employee's health history, so they should not be allowed to examine the results of DNA analysis.[3] On the other hand, employers are allowed information regarding vaccinations, pregnancies and in some cases allergies with employee consent, in order to create a more safe work place for employees.[3] This same argument could be applied to some information in a genetic screen. In the same way that

a workplace might have an epi-pen for a person allergic to bees, and prevent a pregnant woman from entering a toxic laboratory environment, it would make sense to have an AED handy for a person at great risk for heart disease. Further, if the results of a DNA test indicated that a person might directly or indirectly cause danger to the workplace, one could argue that this should be brought to the employer's attention. This sort of information, if given to the employer, might benefit the individual. However, it could also lead to genetic discrimination in the workplace[4]. In the same way that disclosure of allergies is up to patient discretion, disclosure of, for example, high risk for heart disease should also be left up to patient disclosure. An employer might require certain medical procedures to be accomplished before work, in the same way that certain vaccines are required. But these procedures should be standardized and not based on medical disclosure by the patient. Therefore, the patient should again maintain full rights to the ability to disclose or retain this medical information.

Even in the case when releasing the information is in the patient's best interest, precedent states that this release can only be made voluntarily by the patient. Ultimately, giving the patient the decision regarding whether or not this information is released and to whom allows each individual to choose whether the advantage outweighs the possible risks or disadvantages.

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The field of personal genomics is moored in controversy. Instead of actual 'positive' or 'negative' results regarding a disease, patients are instead given the odds that they will get a disease, which, if taken as an assurance of disease contraction, may lead to

misuse of this information. Incomplete information, due to inability to access second opinions, can lead patients to choose routes that are not necessarily best for their health. Sensitive genetic information may be released to the public by other parties. Despite all of these potential issues personal genomics still has an enormous amount of potential positive impacts on patients. DNA screens are a much more accurate version of a family history and can be used to enable preventative medicine. Whether personal genomics, as a field, has a more positive or negative effect on a patient will depend largely on who owns what information about the genetic screen.

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