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Genetics in Healthcare

Manan Shah

The role of genetics in healthcare is expanding – the number of genetic tests available to patients continues to grow and the field of pharmacogenomics offers the enticing prospect of a more personalized approach to medicine. It is important to remember, however, that these new frontiers in medicine bring ethical and social implications along with them.

The field of genetics has flourished since the renewed appreciation for Gregor Mendel's pea plant experiments in the early 1900s and the determination of DNA structure by James D. Watson and Francis Crick in the 1950s. In particular, research developments in genetics have profoundly impacted the medical field. Over the past 50 years, scientists and healthcare professionals have gained insight into the genetic basis of certain diseases, and have since used this knowledge to develop novel therapies, and continued research into diseases with unknown etiology. Today, genetics is revolutionizing healthcare in more ways than one.

The Human Genome Project

The completion of the Human Genome Project (HGP) was a scientific milestone that allowed genetics to have a greater impact on healthcare. In 1990, the HGP set out to determine the sequence of nucleotides on each chromosome and achieved their goal by 2006 (Human Genome Project Information, 2007). The task at hand now is the annotation of the genome, and the identification of functional elements and transcribed regions. The HGP has contributed to the development in the field of genomics, which use various computational techniques to analyze genome sequences of organisms (Russel, 2006).

Genetic Testing

Genetic testing is one area of medical genetics that has matured since the completion of the HGP. Genetic testing examines the DNA of a patient and determines that individual's risk of being a carrier or becoming affected by a genetic disorder (Zimmern, 2007). Table 1 describes the main categories of over 900 genetic tests available today (Genetic Testing, 2008).

Genetic testing can prove to be extremely beneficial to patients. Becoming aware of one's predisposition to disease may permit lifestyle changes to help minimize the likelihood of disease onset. Furthermore, if a high familial risk of disease exists, then all members can take precautionary measures and maintain a healthy lifestyle. Newborn and prenatal genetic testing are especially important, given their ability to predict life-altering conditions for a child. Early detection along with proper treatment can ensure a better chance of normal development.

Table 1 Four major types of genetic testing (Nussbaum, 2007).

TEST	DESCRIPTION
Prenatal	Analyses the DNA of a developing fetus to determine the risk of birth defects or genetic disorders.
Newborn	Carried out after birth to determine the risk of life-threatening or developmental disorders.
Pre-symptomatic	Determines the genetic risk for an adult individual to develop a late-onset disease, such as Alzheimer's.
Confirmational	To obtain confirmation regarding the patient's current status with a specific inherited illness.

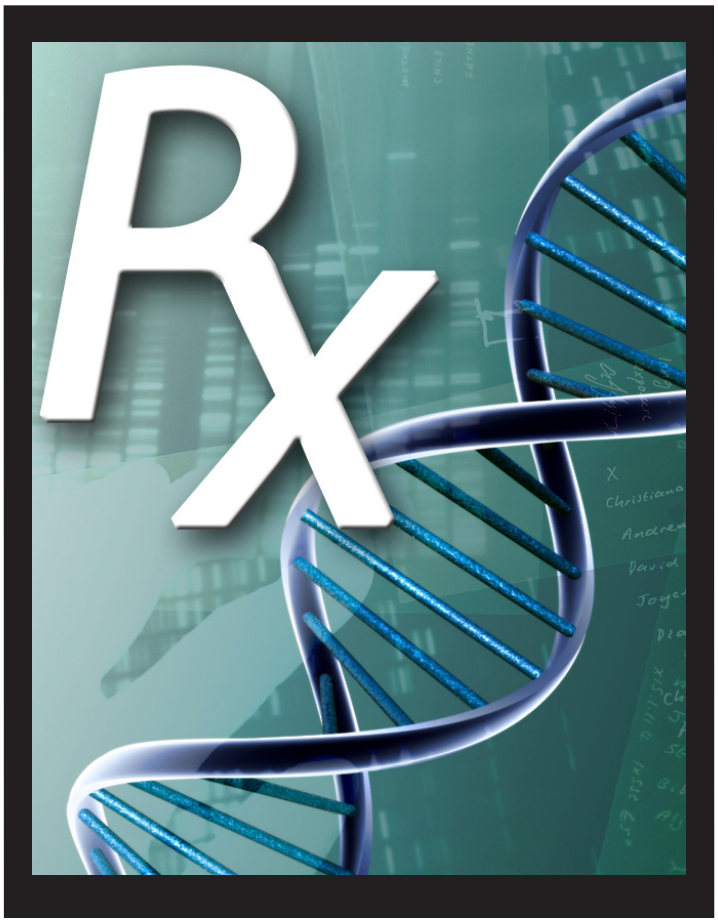
Phenylketonuria (PKU) is an example of a genetic disease that if identified and treated early, can dramatically alter the course of a patient's life. The illness is characterized by a deficiency in the enzyme phenylalanine hydroxylase. In patients with PKU, the amino acid phenylalanine cannot be converted to tyrosine causing a build-up to high concentrations of phenylalanine in the body. Left untreated, the illness can result in mental retardation. Early recognition through genetic testing will result in the patient being placed on a very effective low phenylalanine to control its bodily concentration and high tyrosine diet (Russell, 2006).

Testing services can be obtained through a healthcare professional or from life science companies. Companies generally offer direct-to-consumer (DTC) genetic testing, in which consumers can purchase and undergo tests independently, rather than under the supervision of a healthcare professional (Wade, 2006).

Advantages of offering this testing approach include increased access to genetic testing, greater confidentiality, and reduced testing costs for the patient (Wade, 2006). However, there are several negative aspects to offering genetic tests in this manner that may outweigh its benefits. For example, the methods used by life science companies to advertise their testing services are often criticized. Critics argue that these companies provide "deterministic and simplistic explanations of genetics" to the public, and by exploiting public anxiety, convince patients to order expensive genetic tests that may not necessarily provide them with useful information (Williams-Jones, 2006). Furthermore, given that DTC marketing of genetic tests is relatively new, few governments have stipulations in place that regulate this service (Williams-Jones, 2006). Lastly, allowing consumers to obtain their personal genetic information without the supervision of a healthcare professional can lead to the possibility of patients misinterpreting information that they purchased. For these reasons, it may be necessary to involve a healthcare professional when considering a genetic test, or attempting to interpret its results.

Pharmacogenomics

Perhaps one of the most intriguing proceedings in medical genetics today is the field of pharmacogenomics, originating from genomics and pharmacogenetics – the study of how a single gene can affect the body's response to a drug. Pharmacogenomics focuses on differences in drug response from various polymorphisms, differences in a specific phenotype, between individuals. The aim to search for the optimal drug and dosage for patients based on single genetic differences (Lindpainter, 2002).



As many illnesses involve more than one gene, pharmacogenomics can examine the impact of an individual's genome on the body's response to a drug, allowing an elucidation of a tailored treatment (Russehal, 2006). The differences in patients' genomic response to a medication are often caused by single nucleotide polymorphisms (SNPs) in a multiple number of genes. SNPs can alter genes which encode proteins essential for drug metabolism. For example, Patient A may have a gene with a nucleotide sequence of A-T-G-C which produces a fully functioning protein that facilitates excretion of the drug from the body. Patient B, on the other hand, may have a SNP on the gene that results in a nucleotide sequence, A-G-G-C. This patient does not produce the functioning protein, and thus cannot excrete the drug from the body (University of Utah, 2008). Such an instance would lead to an adverse drug reaction (ADR). Many individuals experience ADRs when taking medication due to genetic predispositions to poor drug metabolism (Hood, 2007).

Utilizing a genetic profile, pharmacogenomics is used as a tool in drug discovery to examine the different effects of several medications. It identifies the best candidate from a variety of drugs based on maximum efficacy and the lowest potentials for toxicity and complications (Lindpaintner et al., 2002).

Pharmacogenomics and pharmacogenetics offer a more personalized approach to treatment. This is significant as the current 'one-size-fits-all' approach to treatment has its drawbacks. Both pharmacogenetics and pharmacogenomics offer the possibility of reducing ADRs, and saving crucial time by preventing the use of a drug that would be ineffective for certain individuals. Time is an especially important factor, given that delays in treating a disease could have serious implications (Hood, 2007).

However, there are obstacles that stand in the way of implementing pharmacogenomics into standard healthcare practice. First, it has been difficult for scientists to demonstrate an improvement in the quality of patient care stemming from pharmacogenomics. Many pharmacogenomic research studies have utilized small sample sizes, healthy participants as opposed to patients, and failed to consider dose-dependent effects of the studied drugs therefore decreasing the studies' validity. Furthermore, most pharmacogenomic studies that display positive results in a laboratory setting lose their generalizability when tested against an independent patient population (Swen, 2007).

Ethical questions concerning pharmacogenomics have also arisen. The primary ethical concerns involve the wealth and availability of sensitive genetic information and ensuring patient confidentiality. Information regarding ethnic differences in polymorpshisms is also a concern. For example, there are significant differences in the frequency of P-glycoprotein (PGP) polymorphisms among certain races. PGP is a transporting protein employed by many drugs to enter a cell, a polymorphism in a PGP gene could thus significantly affect drug efficacy in that individual (Kim et al., 2001). The issue here is the use of the patient's ethnicity as a variable in the prescribing process.

Conclusion

Although "personalized medicine" is a long way from being integrated into everyday healthcare, the potential for the field to significantly improve healthcare holds promise. Genetic testing has already improved healthcare and will continue to do so, as the number and quality of tests available to patients increases with further genomic research. It is important to recognize; however, that the growth of medical genetics will bring new ethical, legal, and social implications into the healthcare forum. While genetics continue to revolutionize healthcare, it is imperative to maintain the patient's privacy rights and best interests in mind.

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