Honors Case Study Challenge Entry Form

Individual Member Entry Information

Chapter Entry Information

Title of Case Study	**Personalized Medicine: Power, Peril, and Promise**
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Case Study Title:

Personalized Medicine: Power, Peril, and Promise

Article Information:

A treatment just for you? Genetic testing may help
Rita Rubin
USA TODAY February 25, 2010
News
1A

Gene test cuts warfarin risk
Steve Sternberg
USA TODAY March 17, 2010
Life
5D

New pill shrinks tumors better than chemotherapy
Liz Szabo
USA TODAY June 7, 2010
Life
4D

Gene research is a 'new biology'
Steve Sternberg
USA TODAY July 8, 2010:
News
1A

Genes as part of the puzzle
Rita Rubin
USA TODAY October 25, 2010
1D

Summary Statement:

Imagine the power of using genetic information to establish a world free of disease and to develop medicine tailored to individuals. The key lies within the genome. In 1866, Gregor Mendel discovered that pea plants pass certain traits to offspring. In 1944, Oswald Avery proved this hereditary information is stored in deoxyribonucleic acid (DNA). DNA in all organisms is organized into chromosomes resembling a spiral ladder; the ladder's steps consist of four chemicals called nucleotides (abbreviated A, T, C, G). The order of nucleotides, like letters in a word, comprises the genes. Each gene contains the instructions that cells use to build the whole organism. An individual's genome consists of all its chromosomes. The human genome has about three billion nucleotides.

Errors can occur in DNA. One improper nucleotide in DNA creates a mutation, which could alter the function of the gene. Inherited mutations that occur in more than 1% of the population are called single nucleotide polymorphisms (SNPs, pronounced "snips"). Most DNA in humans does not encode proteins so the great majority of SNPs are silent, i.e., they have no effect. However, some SNPs cause single-gene diseases or affect how individuals develop diseases or respond to infections and drugs. Thus, studying SNPs can identify areas within the human genome associated with various diseases.

In 1989, the Human Genome Project began to determine the sequences of nucleotides and to identify the 20,000-25,000 genes in the human genome. The completed nucleotide sequence published in 2003 is a reference genome because all humans have unique gene sequences. With the sequence in hand, SNPs that increase the risk for common diseases are being identified. This has ushered in the new era of personalized medicine to diagnose, treat, and possibly prevent disease.

Approximately 100,000 people die annually in the United States from adverse drug reactions. Recent research in pharmacogenetics, the study of genetically determined responses to drugs, has shown that differing genetic composition is a major factor in patients' responses to conventional treatments. The drug herceptin is only effective in breast cancer patients with a specific SNP in the HER2 gene. Since the 1950s, patients taking the blood-clot prevention drug coumadin had to be closely monitored to prevent fatal bleeding. Following the discovery of a gene for vitamin K metabolism in 2005, a genetic test for SNPs in this gene is now used to provide appropriate dosages.

Personalized medicine uses information about an individual's genes and environment to prevent, diagnose, and treat disease. Its goal is to personalize treatments by catering to individual genomes enabling medications to have more chance of having the desired positive effect. Individuals can now purchase the genetic tests used by physicians. The benefits are undeniable but may be limited by the quality and appropriate usage of the information. Additionally, the effect of environmental factors on SNPs and the interaction among SNPs are, as yet, unknown. A critical examination of the legal and ethical perils associated with personalized medicine is necessary to ensure that its promise can become reality.

Discussion Questions:

1. In 1950, the United Nations Educational, Scientific, and Cultural Organization issued a condemnation of racism stating that "*race* is not so much a biological phenomenon as a social myth" and suggested to "drop the term 'race' altogether." Genetic evidence shows that 90% of the DNA in all humans is the same but about 10% of the human genome is more common in certain ancestral groups. For example Jews are offered prenatal tests for the inherited Tay-Sachs disease gene. The heart-disease drug BiDil is marketed exclusively to African-Americans, in which a modification of the ABCA1 gene is common. Some asthma medications can shut down the lungs of people with a certain SNP in the beta2AR gene that is more common in African Americans. Han Chinese are more likely to have adverse reactions to methotrexate, a drug commonly used to treat rheumatoid arthritis.
   a. Can these similarities within populations be used to define races?
   b. Should races be defined in order for drug therapy to be useful? Will this lead to discrimination?

2. Daniel's and Monica's 5-year-old son, Matthew, needs a bone marrow transplant to treat aplastic anemia. Neither parent is a donor match and the boy will die without the transplant. Daniel and Monica want to use in vitro fertilization to have a child that will match Matthew's HLA type. The couple plans to have embryos genetically tested for the HLA gene and implant the desired embryo into Monica's uterus for gestation.
   a. Should couples in this situation select embryos?
   b. Later in life, the younger son feels he did not get the same care as his older brother. Unlike Matthew, the younger son worked through high school and paid for his own college education. As an adult, he sues his parents for wrongful life. Does he have a basis for his complaint?
   c. Jonathan and Tina want to use in vitro fertilization to have a child who will grow up free of diabetes, high cholesterol, cancer, high blood pressure, and alcoholism. The couple plans to have embryos genetically tested for these disease-related genes and implant the desired embryo into Tina's uterus for gestation. Should Jonathan and Tina be able to select an embryo with their desired traits? Is their case any different from that of Daniel and Monica?

3. Researchers at CaroGene Inc. developed a coronary artery disease (CAD) drug that can be sold to millions of people with CAD. The data from their clinical trials on patients with CAD are shown below:

	No heart attack within 5 years	Number having heart attack within 5 years
Control (no drug)	543	1957
Test (received new drug)	648	1852

   a. Should CaroGene market this drug to everyone with CAD?

b. During the clinical trials, a CaroGene geneticist read an article about SNPs linked to CAD and looked for these CAD-related SNPs in the trial participants. In light of these new data, should they market the new drug to everyone with CAD?

SNP*	Control (no drug)		Test (received new drug)	
	No heart attack within 5 years	Number having heart attack within 5 years	No heart attack within 5 years	Number having heart attack within 5 years
rs17672135	74	415	67	376
rs383830	84	286	81	282
rs1333049	57	145	177	41
rs6922269	141	658	138	646
rs8055236	187	453	185	507
*SNPs are identified by number. rs = Reference SNP.				

4. Katrina saw an advertisement about genetic testing for a breast cancer gene. She ordered the kit online and sent her cheek cells.
   a. Do you agree with Katrina that if the test is negative you know you're safe?
   b. If the test is positive, should Katrina tell family members that they might have the mutated gene?
   c. How might an individual's self-perception be affected by knowledge of one's genes?
   d. Should testing be performed if no treatment is available?
   e. Should children be tested for diseases that don't show up until middle age?

5. While working on a project for her Genetics class, Marisa found that direct-to-consumer (DTC) genetic tests are categorized as services, and as such there are no U. S. Food and Drug Administration (FDA) regulations for evaluating the accuracy and reliability of genetic testing. Some of these companies make dubious claims about how the kits not only test for disease but also serve as tools for customizing medicine, vitamins, and foods to each individual's genetic makeup. Marisa purchased two DTC genetic tests from Internet advertisements. The companies are not necessarily using the same SNPs and don't calculate risk using the same data. Marisa's results are shown below:

	Relative risk of getting disease reported by	
Disease	Company A	Company B
Heart attack	Average	Decreased
Lupus	Decreased	Decreased
Macular degeneration	Average	Increased
Psoriasis	Increased	Decreased
Restless legs syndrome	Decreased	Increased
Type 2 diabetes	Average	Decreased
Multiple sclerosis	Decreased	Decreased
Colon cancer	Average	Average
Breast cancer	Decreased	Decreased

   a. Discuss whether the FDA should regulate these companies. Will regulation prevent consumers from getting information? Will regulation benefit consumers?
   b. Should DTC results include an evaluation and recommendations by a genetic counselor?

Future Implications:

Personalized medicine holds enormous promise, yet that promise may not be realized without some peril. Fortunately, the Genetic Information Nondiscrimination Act prohibits discrimination by employers and health insurers based on genetics. But, will it be possible to keep personal genetic information private once a genomics company has acquired it? Will some individuals refuse genetic testing because the results may impact their ability to get adequate, affordable life insurance? Discovery of specific SNPs in an individual can be indicators of future disease and generate health and ethical issues in genetically-related people. Information on specific genes increases the possibility of using selective breeding for eugenics. Genetic diversity in a population is critical to survival because loss of diversity increases the possibility of extinction due to increased vulnerability to disease, reduced ability to adapt to environmental change and, other as yet unknown factors. Biologists are only beginning to unravel the effects of SNPs and the interaction between SNPs. Will our desire for health and beauty lead to a dangerous loss of genetic diversity? Though much research is still needed, personalized medicine will eventually lead to treatments that can target specific genetic make-ups in treating various diseases including cancers, schizophrenia, and autism.

Additional Resources:

Barry, P. (July 4, 2009). "Seeking Genetic Fate." *Science News* 176(1), 16-21.

Collins, F. S. (2010). *The Language of Life: DNA And The Revolution In Personalized Medicine*. New York: HarperCollins.

Hall, S. (2010). "Revolution Postponed." *Scientific American* 303(4), 80-87.

Lynch, A. and V. Venne. (2009). *The Genome Book: A Must-Have Guide To Your DNA For Maximum Health*. North Branch, MN: Sunrise River Press.

National Human Genome Research Institute. National Institute of Health. <http://www.genome.gov/>

Public Health Genomics. Centers for Disease Control and Prevention. <http://www.cdc.gov/genomics/>