

Taking Advantage of Modern Medicine

R. Wood Morgan '18

Today most medicinal treatments employ one of two treatment options: therapeutic drugs or surgery. It is general and does not work in treating or preventing certain individuals and even certain diseases. Many drugs and generic doses of these drugs are not only an impersonal solution but are also ineffective for most users. The future of medicine will be personalized to the individual, by using the own individual's genome, to help their own bodies fight certain ailments. Genomic medicine or as many call it "personalized medicine," works with DNA in order to induce changes in a cells inner workings and in this way combat diseases and other risks autonomously. This newer form of medicine could bring advancements that many may never have thought were possible. The cures for genetic diseases, especially cancer, are making great strides through genomic research and have created many new treatments that provide a patient with more comfort and a higher chance of a cure. Personalized medicine will also bring about new obstacles that have not been faced. "Enormous amounts of new knowledge are barreling down the information highway, but they are not arriving at the doorsteps of our patients." (Annas, 2016). In the future it will be essential to have the basic knowledge of this field to take advantage of the medical advancements and at the same time protect yourself from the uncertainties and implications of your DNA being in large, possibly insecure, databases.

On an ordinary day we see lots of people and besides a few anomalies such as twins we never see a person that looks or acts exactly the same as another. This stark contrast disappears at the DNA level. Compared to your parents or even your best friend you are 99.9% identical. DNA is made up of over three billion organic molecules called nucleotides and similar to code for a computer different nucleotides in a certain order serves as a genetic instruction to carry out every function in the body. Unlike a computer where if one character is out of place then the entire program doesn't work, the human genome contains lots of redundancy and only about 1-2% of your genome actually codes for a function. That leaves the other 98-99% of your genome as non-coding DNA. The name given to this part of the genome should not be misinterpreted as

"unimportant" because we have found that it is essential to DNA structure and preventing mutations from drastically affecting the product of a gene.

I'm sure you're wondering how we even know this about our DNA. How is it possible for us to read through and process billions of nucleotides? Today's technology has breathed new life into the field of genomics and has extended the frontier of medical research. Next-generation sequencing has given us the ability to take a genome like a humans, which would have been previously too big to run in a cost-efficient manner, and sequence it for research or for medical analysis. By sequencing a genome you get the precise order of each nucleotide, producing a long list of A's, G's, T's and C's. These letters are what make up our genes and the way that our body reads them or "expresses" them determines our bodily functions. With our sequence or "code" we can now put it into a multitude of computer programs that will help us find, compare and analyze our genes.

Cancer is one of the most prominent genetic diseases in the modern world. It is one of the many fields of research that have benefitted from genomic research. Over 14 million people worldwide are afflicted with this invisible disease. By invisible I mean, to your own immune system. A mutation of a healthy cells DNA can trigger cells to develop the "hallmarks" cancer which usually leads to benign tumors. Tumors start as benign and over time acquire mutations that lead them to become cancerous. One of the "hallmarks" of cancer is their ability to be immortal. Normal cells eventually cannot replicate anymore and die off. This is because of something called telomeres that are on the tips of your chromosomes. These telomeres degrade slowly each time a cell divides and eventually they become too short for replication. Telomeres in this way can be seen almost like a cannon's fuse. Cancer cells on the other hand have mutated so that they can retain their telomere length resulting in an immortal cell line that can't be detected by our immune system as "dangerous". This type of function can be attributed to certain proteins that are encoded by our genes. By finding the specific gene that runs this process one could potentially stop a cancer from becoming immortal. This would be how a researcher would think about attacking cancer: find a gene or genes of interest, determine its function in cells, and apply an

experiment to show how it regulates cancer growth. In this way Cancer Genomics has dramatically increased the efficacy of treatments as well as helping develop new ones (Annas 2016).

Genomics and its application to drugs has also shown significant promise and will help prevent adverse reactions and improve the efficacy of drugs. The drug warfarin is a great example of how genomics has helped hundreds of thousands of people suffering from blood clotting issues to properly use their medicine. Warfarin is an anticoagulant, meaning it prevents blood from clotting. It is a common misconception that anticoagulants “thin” the blood, however they do nothing in changing the viscosity of your blood. If a person who is taking warfarin doesn't take enough they run the risk of having a stroke or other blood clot related problems. On the other hand if the user takes too much then a hemorrhage could occur. It has been found that two genes affect how an individual will react to the drug warfarin and by having a precedent of how certain genetic groups react, fatal reactions can be prevented. This kind of genomics is known as Pharmacogenomics. Pharmacogenomics is becoming an important tool for a prescribing doctor since knowing about the genetic makeup of a patient can help prevent overdose and help improve the efficacy of a drug. (Annas, 2016),

With the breakthroughs that science is experiencing due to the ever growing knowledge of the human genome comes many of the new treatments and advances in healthcare that are previously mentioned and since good health care is at the top of the list of many families concerns it has become a very controversial topic in modern politics. Many debates in politics also involve race and it is often mixed in with health care. It is important not to misconstrue any genetic research or other studies with having anything to do with race. A particular gene does not encode race, and it is more of a social construct than a biological one. Where we see genetic differences is between ethnic groups and in this since it has more to do with any given groups environment than with the color of their skin. In 2014 a study was done across fifty major cities in the U.S., testing the mortality disparities of breast cancer patients between African-Americans and Caucasians. It was found that African-Americans were 40% more likely to die from breast cancer. A headline of this nature could be very misleading and lead to political and social arguments that a new field like genomics

does not intend to create. The disparity does not lie solely within your own genetic code but also the environment someone is living in and their access to healthcare. The question that goes hand in hand with this notion would be, is it nature or nurture that is the driving factor of this disparity? (Annas 2016).

The nature of something is based off its surrounding environment. In a desert you wouldn't expect to find much leafy green vegetation because that environment does not have the water or soil necessary to support such growth. Similarly, it would make no sense for someone to wonder why a child misbehaves if their parents were known to be poor examples. This is not to say that a parent by raising a child a certain way could affect their genes and nurture better ones to arise but there are many events that can occur especially during gestation that could lead to irreversible gene alterations. A commonly observed event in nature is imprinting. We always love to see the cute little ducklings following their mother in almost a blind manner. In 1909 a boy, at the time, Konrad Lorenz hatched a couple of ducklings that became imprinted on him and would follow him everywhere. Later in life this would inspire his research of imprinting but he would not achieve the imprinting event as easily as he did as a boy. It wasn't till he tried making mallard-like sounds that the ducklings finally began imprinting on him again. Another scientist who studied imprinting was Gilbert Gottlieb who found that mallard and wood ducks were drawn toward the call of their species. Gottlieb did not stop there with his research and to further test this specific imprinting he muted the ducklings while they were still in their egg by operating on their vocal cords. The ducklings that had been muted once hatched had no preference to their species. By introducing an environmental trigger before birth he was able to determine that instinct did not play a role in the nature of the ducklings. Relating similar prenatal traumas or experiences to humans has also been researched, like the “thrifty phenotype” hypothesis. A fetus who is not well nourished in the womb can result in an adaptation in the fetus' metabolism to be ready to face a world where food is limited. If then that same fetus once born finds itself with an excess of food it will compensate by growing fast and taking advantage of the excess. This quick growth can put strain on the heart and lead to a higher chance of heart disease for the baby. These types of studies are quickly determining that prenatal events may have a stronger influence on the

longevity of offspring than the genes of their parents. (Ridley 2003).

As DNA sequencing has become more and more accessible the storage and privacy of this data has become a topic of ethical discussion. It is important when getting your DNA tested to know the implications and risks of doing so. Currently in Australia life insurance companies can require applicants to disclose all genetic tests. This includes those done for medical analysis or research as well as things such as "AncestryDNA" and "23 and Me." This would mean that when applying for life insurance or any insurance for that matter you could be required to release your genetic test results that could in turn make you completely ineligible to receive coverage or make it extremely expensive. That particular risk can also be seen as a good thing for someone in good health. Someone with good genes and who are less likely to develop a fatal ailment could receive better or cheaper coverage. (Tiller 2017). Another controversial topic in the use of DNA is the ability to patent DNA processes or sequences, which is legal in the U.S. and Europe. It is a touchy subject since it involves many moral dilemmas as well as economic and social issues. Laws in the U.S. on patenting DNA can also get very confusing. Under U.S. law you cannot patent a "product of nature", so the question now is, where does DNA fall as a patentable entity? The United States Patent and Trademark Office (PTO) says that if someone isolates and purifies or alters DNA in any way can patent that particular sequence or process. Although DNA is a natural compound and would seem to be something that cannot be patentable as long as it is not considered "natural DNA" then it can receive a patent. Many of the social and moral arguments against DNA patenting is that you would be saying that humans created or invented something that was made by God. Another moral argument is that it can be deduced that by patenting DNA or DNA processes you could potentially slow the progress of science by allowing a newly discovered species or DNA sequence to not be used by anyone but the one who holds the patent. On the other hand DNA patenting could bring lots of money into the field of science by allowing more private companies to invest money into bringing newly found processes and technology to the market. This is key in science since there is a limited amount of publicly funded research. As a result without private companies investing into research many promising

research projects could go unfunded and never get off the ground. DNA patenting is a tricky subject matter and many people still aren't sure of the long-term effects DNA patenting can have. (Resnik 2004). A great example of the promise of genomics is the very research that is being done right now. In a 2013 publication it was found that two genes, TMC1 and TMC2, that function as components of hair cells. Hair cells have the ability to take sound and transform it into an electrical impulse that can be read by the brain. One form of hearing loss can result as a malfunctioning TMC1 gene and at this moment there are no ways to treat this form of hearing loss. In comes genomics and gene therapy. Through gene therapy a functioning form of the malfunctioning gene can be integrated into a cell's DNA through a virus. The way a virus causes disease is by invading a cell and integrating its own DNA into that cell. The viruses' negative effects are removed from its DNA and the functioning TMC1 gene is integrated into the viruses DNA so that it will integrate that gene into a target cell. When the modified virus is injected into the ear cells of a mouse, whose DNA had been modified to contain a malfunctioning TMC1 gene, the mice were found not to develop late onset deafness. This suggests that the TMC1 gene has a direct mechanism to prevent hearing loss. (Askew 2015). This is just one of hundreds of studies that are being done right now. These are very exciting findings that show how genomics is quickly changing the way medicine and treatments are looked at.

The future of medicine holds a lot of promise and it is exciting to know that it will be our own bodies helping make the discoveries. The possibilities are endless for the results from genomic research. It could provide more accurate and affordable healthcare, prevent overdoses and increase drug efficacy and even help a patient prevent ailments they may be at higher risk for. With the excitement of the amelioration of health care through genomic research also comes new concerns. The threat of insurance companies being able to attain your DNA and base you premium or deductible on such means is definitely a worry and the laws surrounding this should be closely followed. The key to taking advantage of this new field is to inform yourself and to be ahead of the game when it comes to others catching on. The topics covered in this paper will give you a good understanding and grasp on genomics but it is up to the individual to completely inform themselves. The sooner one can learn how to best

utilize and prevent downsides, the sooner the worry of your health and wellbeing can be lifted.

REFERENCES

- Annas, George J., and Sherman Elias. Genomic messages: how the evolving science of genetics affects our health, families, and future. HarperOne, an imprint of HarperCollins Publishers, 2016.
- Askew C, Rochat C, Pan B, Asai Y, Ahmed H, Child E, Schneider BL, Aebischer P, Holt JR. Tmc gene therapy restores auditory function in deaf mice. *Sci Transl Med*, 7(295):295ra108. 2015.
- Resnik, David B. *Owning the genome: a moral analysis of DNA patenting*. State University of New York Press, 2004.
- Ridley, Matt. *Nature via nurture: genes, experience, and what makes us human*. Harpercooins, 2003.
- Tiller, Jane, et al. "Should Australia Ban the Use of Genetic Test Results in Life Insurance?" *Frontiers*, *Frontiers*, 22 Nov. 2017, www.frontiersin.org/articles/10.3389/fpubh.2017.00330/full.