The Movement of Genetic Disease through Human Migration

Amanda Hughett

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GSTR 410: Human Migration

When most of the population thinks of migration they think economic impacts, job placement, and freedom from oppression. However, there is a largely overlooked aspect of the movement of humans, and that is the diseases humans bring with them. According to Cummings, "more than 10 million children or adults in the United States suffer from some form of genetic disorder, and every childbearing couple stands an approximately 3 percent risk of having a child with some form of genetic anomaly" (Cummings 10). Thus genetic disease plays no small role in the daily life of Americans, or in any other country.

Genetic disease, also known as hereditary disease or genetic disorder, is any mutation in the genetic code that leads to a malfunction of the element for which it codes (Cummings 10). Tay-Sachs disease, for instance, causes a malfunction in each cell's lysosomes. This can happen either pre-fertilization or after the union of the sperm and egg during gestation. This individual, once born, now carries a copy of that corrupted DNA strand in every cell of their body. During meiosis, or sex cell division, the DNA strands separate into sister chromatids. Each cell gets half of the parent cell's genetic coding, and the diseased DNA is passed onto the sex cell, which combines with that of their mate, thus making a carrier of the trait. A carrier, referred to as a heterozygous individual, usually does not show any symptoms of the disease, but carries the ability to pass on the disease gene to offspring. If two individuals who are carriers breed, they typically make more carriers or even an individual who expresses the disease trait, which in most cases, means the individual is carrying two copies of the mutated gene. These individuals with two copies of the disease gene are homozygous for the disease trait. When generations pass, and these small populations breed, more carriers are produced, thus increasing the likelihood that more homozygous individuals will be born. Consequently, as the genetic code is circulated

amongst the population, the frequency of infected individuals rises, causing a gradual amplification of the gene in the community.

Genetic diseases usually begin in small populations of less than 1,000 individuals and are the result of a random mutation during cell division. The mutations survive and are multiplied if 1) they are non-harmful or beneficial or 2) they are magnified within a small ethnic group. In a natural environment in which a disorder is not being treated, individuals that express a disease trait are less likely to survive to maturity and pass on their genes through their offspring, or they are deemed an undesirable partner and thus never mate. However, if a disease trait is being carried, but not expressed in a noticeable manner, then it is likely the carrier will mate and pass on their genetic code to a new generation. There is also the alternative of breeding crossovers occurring in small ethnic groups that multiplies the disease trait. These small groups tend to be small immigrant populations that live in close proximity and intermarry. Most of these individuals tend to pair with each other due to some social or religious boundary between them and their host country.

Once it has begun, the survival of a disease depends heavily on the population in which it exists. The disease can only grow if individuals who carry the trait have viable offspring capable of reproducing. Thus, the disease usually weeds out those who are homozygous for the trait. Many genetic diseases have survived because they are harmless or not as harmful in the heterozygous state in which the individual carries only one copy of the disease trait. For the two diseases that were chosen to be included in the project, Tay-Sachs Disease and Sickle Cell Disease, the heterozygous individuals experience little to no impact on their health from carrying the disease, but experience catastrophic consequences for carrying two copies of the gene

(Cummings 61). These two diseases were chosen in particular due to their widely different mutations they cause, and their particular method of survival differ.

Tay-Sachs disease is but one of the 50 currently identified lysosomal storage diseases ("Treating Tay-Sachs Disease"). Lysosomal storage diseases are characterized by a mutation in some element of the lysosome, an internal organelle in cells. Tay-Sachs disease is caused by one or several of 50 different mutations on the gene responsible for coding part of the Hex-A enzyme on chromosome 15(Cummings 61). The cells of these individuals lack a particular enzyme necessary to break down waste products inside the cell, such as Hex-A in Tay-Sachs patients (Cummings, 61). These waste products build up in the body, usually in nerve cells, which irreparably damages the cell so that it is incapable of functioning ("Treating Tay-Sachs"). This disease in its heterozygous form is almost benign, but has a very high fatality rate in individuals who are unfortunate enough to be born with two copies of the gene. The most dangerous form of Tay-Sachs disease begins degrading the neuro-pathways of the individual from birth, with signs appearing around the time the child is six months old, and death usually occurring before 5 years of age ("Treating Tay-Sachs"). The neuro-degeneration caused by the disease causes the individual to go blind, deaf, suffer mental retardation and eventually lose the ability to breathe causing death ("Treating Tay-Sachs").

Tay-Sachs disease came about as a random mutation and was perpetuated by intermarrying and mating amongst small populations. The Ashkenazi Jews are the most affected ethnic group recorded with 1 in 27 being carriers, as compared to the 1 in 250 individuals in the general population (Cummings 61). Other affected ethnic groups include "non-Jewish people of French-Canadian ancestry, members of the Cajun population of Louisiana, and Irish Americans"

(“Treating Tay-Sachs). The Cajun and French-Canadian populations share common ancestors which may have contributed to both lines possessing heightened frequency of the disease.

The other disease to be documented in the project is Sickle Cell Disease. Sickle Cell Disease, or SCD, is characterized by a “sickle” shape to red blood cells. The bending of the red blood cell into the sickle shape is caused by a mutation in the hemoglobin contained within the cell which causes it to bend in low oxygen environments, bending the entire cell into a sickle shape and making the outside of the cell sticky (College of the Siskiyous). These sickle shaped cells do not carry oxygen well, and most importantly they hook on one another in small blood vessels. The cells create blockages in smaller blood vessels, and thus restrict oxygen flow (College of the Siskiyous). The organs that do not receive the oxygen they need begin to lose functionality and eventually die from suffocation. While milder forms of the disease caused by having only one copy of the gene may only lead to the formation of small blood clots, the most extreme version which is encountered in homozygous individuals, usually causes lethal complications before young adulthood (College of the Siskiyous). Unlike Tay-Sachs disease, the heterozygous form of the disease causes health complications that may not be immediately problematic, but cause a gradual shortening of the individual’s lifespan.

Unlike Tay-Sachs disease, the amplification of the Sickle Cell trait did not arise from breeding of small populations due to religious and ethnic restriction. While SCD did arise as a random mutation in the gene that codes for hemoglobin, it survived because it was actually beneficial in the regions it evolved. Sickle cell originates from a region in Africa along the equator where *Falciparum malaria* is prevalent (College of the Siskiyous). Malaria is a blood-borne parasite that infects the red blood cells of an individual, causing them to burst (Sabo). The disease is thought to have been so successful because it provided partial immunity to the parasite

responsible for malaria (College of the Siskiyous). Those who were carriers for the disease prospered from near immunity to the disease while those who were homozygous for the disease usually died from health complications from sickle cell itself.

The research project did hold several obstacles that delayed its completion on several occasions. The greatest obstacle to overcome was the lack of reliable information on Tay-Sachs disease. While I prefer to use journals and books to do scientific research, I found that the local libraries held no information at all on this disease. I later came to realize that the lack of information was no fault of the library, but was in fact due to the method used to observe and document disease in modern society. To properly research and publish a finding on any disease, a researcher must thoroughly observe and test many individuals with the disease. However, due to the relatively recent discovery of the disease, and a means to view the damage through complex microscopes, there has not been a large span of time for scientific documents to be reviewed, published, and released to the public for educational use. Most of the knowledge I did find on the subject were in text books written in the last few years and in digital sources that had been released within the past two years. This lack of reliable, credible information caused many delays in both the writing of the paper, and the development of the website.

The second greatest obstacle was to build and publish the website once I had the information ready. It took many attempts, and a change of how I approached the web publishing to finally get the finished product published. First, I attempted to build the website out of raw HTML code. The first website, built out of the raw code, was unimpressive, and riddled with problems when viewed on web explorers such as Internet Explorer and Google Chrome. The second attempt was made while using a free site publisher, Yula. While this is the publisher that I managed to use in the end, the second website attempt was completely deleted due to a glitch in

web widgets that allowed video to stream from another site for the purpose of explaining sickle cell anemia through computer animation.

The website itself contained several small choices that gave rise to its style. I wished for the site to appeal to intellectual individuals with a genuine interest in the science of human migration, not simply in a fun and witty website for entertainment purposes during one's spare time. The site was designed like a scholarly, scientific website. The background and theme were chosen to minimize distraction from the topic at hand, utilizing a neutral color scheme for the background to emphasize the text and foreground of the site. The colors chosen were black and white for simplicity, and ease of reading. One of the easiest texts to read is a black text on a white background. It offers contrast, and made the text easy to decipher. Unnecessary images were also avoided as much as possible to prevent distraction and disruption of the flow of the piece. The images were kept professional, with no cartoon-like images for comic relief. The images used came primarily from text books, and academic websites that were affiliated with the government or universities to ensure credibility. The illustrations that were used were also usually colored. This drew emphasis to the images, bringing them out of the background and into the foreground of the reader's vision, allowing them to view the images as part of the text, not in addition to it.

Due to the content and the level of comprehension necessary to completely understand the meaning of said content, the chosen audience was composed of individuals with a high school or higher level education, preferably with a basic understanding of biological terms. I attempted to simplify the information as much as possible without making it so easy as to be insulting to the intelligence of the individuals visiting the website. The topic seemed too inappropriate to address a younger audience. It was unlikely that a younger audience would

possess the maturity to take the project seriously. The topic, which often refers to the pairing of humans for sexually reproductive purposes, would prove to be unfit material to be taught to a young audience, especially one that may have not learned about reproduction yet.

The audience is further narrowed down by interest groups. Individuals best suited to being viewers of the site would be significantly narrowed based on personal bias and beliefs. The site is heavy in scientific references, and often refers to different traits and genes. The site also goes into detail on two genetic diseases, and mentions how these diseases have evolved into what they have become today. This would shift the audience spectrum towards individuals with at least a passing curiosity in the field of genetics. The audience is then narrowed even more by the mention of evolution in a description of the creation of disease. While evolution is not specifically forced onto the reader, it is mentioned multiple times, along with the Red Queen hypothesis of evolution. The basic concepts that are used to describe the the details of how genetic disease came into being are, in of themselves, a description of evolution on a small scale. Micro-evolution, as it is commonly called, is a gradual change within a species that gives rise to miniscule changes in the genetic coding for a species, such as skin color, diseases, and physical traits. However, micro-evolution gives rise to macro-evolution, a multitude of changes that gives rise to a new species from a previously existing, viable species. This series of evolutionary concepts would prevent many devote religious followers from comprehending and acknowledging the information contained within the site. While the site definitely does not attempt to prove or disprove evolution or creationism as a fact, it does contain the controversial message of change through evolution as opposed to through a higher, divine power. While the knowledge contained within the site is sound, it would be viewed with much bias by some individuals who adamantly refuse to acknowledge any information that is reported in conjunction

with evolutionary views. Therefore, the site is best geared towards individuals with an interest in science, and a view of evolution as a perfectly viable theory for how gradual change comes about in the natural world through micro-evolution.

The decision to present the project in the form of an internet based website was approached in the same way that I chose my audience. I had to consider the needs of my audience, and how they might wish to access my research. In today's modern era of technology, the most accessible forms of media are found in internet-based, web sources that can be utilized from nearly every continent. The majority of the young minds that are now reaching adult hood are highly attuned to the digital heartbeat of the wireless world. One constantly sees individuals using computers, personal data assistants, and internet-capable cellular phones. Media has made leaps and bounds in its ability to reach a wide audience, and this has been partly in thanks to the popular usage of the internet. Books and other physical media are being transcribed onto disks and other digital forms of data for mass transmittal across the globe. The scientific community is no different. With scientific findings now being published on subscription websites, as opposed to the outdated journal method, the scientific community is attempting to stay up to date with their audience. In an attempt to do the same, I decided that a website was the best course of action for making a scientific research project accessible to a wide range of individuals from the convenience of their homes, offices, or nearest internet café. I wanted the research to be available to any individual who was capable of accessing the nearest internet communications hub and reading it.

Works Cited:

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