8th Annual DNA Day Essay Contest Submission

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The following are the details of essay(s) submitted on 2013-03-15 01:38 US ET

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Essay 131482

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Essay
Histones and Cancer

One of, if not the most, significant breakthroughs in the field of genetics is the discovery of the double helix by James Watson and Francis Crick. Their

discovery allows us to make great advances in various fields, especially in the study of human health and disease. Without Watson and Crick's double helix model, scientists would not have been able to delve into the study of histones, a rising area of study today. Our modern-day understanding of histones and how they affect gene expression is greatly increased due to their research. Epigenetics, the study of gene expression caused by mechanisms other than the genetic code, includes a variety of topics such as the manipulation of histones that DNA wraps around. Researchers all around the world are studying these chromosomal proteins because of their correlation with oncogenes and tumor suppressor cells, which can lead to cancer if not regulated correctly.

One type of histone modification, known as methylation, causes a downregulation in the products of the gene. In cancer, abnormal methylation induces inactivation of mismatch repair, instability of chromosomes, hypomethylation of oncogenes, and hypermethylation of tumor suppressor cells (Li, Guo, De 2012, p. 885). Methylation is catalyzed by DNA methyltransferases (DNMTs), and without them, the level of methylation is severely reduced (Li et al., 2012, p. 884). These DNMTs are affected by a wide range of factors, including ultraviolet rays, temperature, infection, smoking, and a lack of folic acid. For example, Li et al. (2012) state that a diet lacking folic acid induces methylation (p. 884), increasing the likelihood of cancer. Methylation can be in both cases good and bad. Methylation helps control genes and can potentially stop the rapid proliferation of cells in a tumor. However, if methylation is not properly controlled, it can prevent the genes responsible for the production of tumor suppressor cells from being expressed, which is one of the problems in cancer because it leads to uncontrolled cell division or a tumor.

Specific histones have certain jobs and functions that affect the expression of genes. For example, research by Jørgensen, Schotta, and Sørensen (2013) states that histone H4 lysine 20 methylation is preserves the integrity of the genome (p. 1) and if it is not maintained, leads to adverse effects like cancer. One of the chromatin proteins that they studied was L3MBTL1, a tumor suppressor which when depleted, induces DNA breakage, DNA damage response, replication inhibition and instability of the genome (Jørgensen et al., 2013, p. 4).

Another group of scientists studied the effects of various histone modifications on cancer cells (Valdés-Mora et al., 2012, p. 307). Gene inactivation in cancer is connected to two (Valdés-Mora et al., 2012, p. 307) histones involved in the regulation of gene expression. Specifically, the depletion of H2A.Z causes problems during chromosome segregation and the cell cycle (Valdés-Mora et al., 2012, p. 308). In addition, H2AFZ gene expression increases in colorectal and breast cancers among others (Valdés-Mora et al., 2012, p. 308). Valdés-Mora et al. (2012) showed acetylation of H2A.Z activates oncogenes and deacetylation silences tumor-suppressors (p. 308). In their paper, the researchers also show that with the loss of adH2.Z and H2A.Z, the oncogenes

went through a rapid upregulation, white inversely, many of the tumor suppressor genes were downregulated (Valdés-Mora et al., 2012, pp. 312 - 313). Valdés-Mora et al. (2012) propose H2A.Z histone acetylation as a way to treat prostate cancer (p. 318), which would potentially have the reverse effects as aforementioned, stopping the upregulation of oncogenes and the downregulation of tumor suppressor cells.

In conclusion, without the discovery of the double helix, the field of epigenetics would be severely reduced or even nonexistent. This field proves to be extremely important, especially today, when scientists are trying to find cures and treatments for cancer. The causes and potentially even cures for cancer, one of the conditions plaguing and baffling patients and researchers today, can be found in the genetic code and the histones that DNA is wrapped around all thanks to Watson and Crick.

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Essay 151485

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Essay
The sequencing of the human genome has had and will have a large impact on human health and disease. Knowing the sequence of the human genome allows us to improve our diagnosis of diseases and recognize any predisposition towards a genetic disease earlier in a patient's life, leading to earlier and

more effective treatment. We can test individuals to see if they are carriers of a genetic disease. We can use preimplantation genetic diagnosis to check if embryos have any genetic flaws before implantation in a mother's womb ["Gene Testing"]. This provides a much better alternative to the practice of waiting for a while before chorionic villus sampling can be undergone to see if a fetus has a particular disease—by using PGA, we can avoid the difficult decision of whether or not to terminate the pregnancy.

This knowledge will also help us create better drugs and figure out which is a best for an individual patient. If we know what gene is associated with a disease, we can create drugs specifically targeted towards the proteins, enzymes, and RNA molecules associated with that gene. This will decrease the side effects of the drugs. Pharmacogenomics is the study of how a person's genetic makeup affects their reaction to medicines. Given the knowledge of how different genes affect the effectiveness of a particular medicine, doctors will be able to prescribe the best medicine for a patient the first time around, rather than using a trial-and-error approach. We will be able to better determine how much of a drug a patient should take. Through this better method of prescription there is a much lower chance of an adverse drug reaction, which currently is one of the leading causes of hospitalization or death in the United States ["One Size Does Not Fit All"]. Take breast cancer, for example, or B cell lymphoma. If you sample tumor tissues from different patients, you'll get different categories of genetic makeups. Once you separate these different cases, you can treat each variation differently—whatever the most effective way is [Henry].

The sequencing of the human genome helps us look more to the original causes of genetic diseases instead of to the symptoms. Case in point: the field of gene therapy, which would not be possible without having sequenced the human genome. In gene therapy, instead of trying to treat the symptoms of a genetic disease, people actually provide a normal copy of the mutated gene. This can be done in different ways. First, a normal copy can just be inserted. The mutated version can be swapped out through recombination. The gene could be repaired through reverse mutation, or finally, the regulation of the gene could be altered ["Gene Therapy"].

Clearly, there are many techniques for the improvement of human health that were made possible by the sequencing of the human genome. Although currently many of these possibilities are out of our reach, in the near future they may not be. As discoveries continue to be made, human health will only get better and better. One day, perhaps we will be able to eradicate, if not all disease, at least genetic diseases entirely.

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Essay 131484

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Essay
Mapping the Future: The Human Genome and Medicine

It was in April 2003 that the National Human Genome Research Institute (NHGRI) declared the Human Genome Project (HGP) to be complete, a mere 50 years after the discovery of the double helix by James Watson and Francis Crick. The HGP had been an ambitious, international, collaborative research program with the goal of sequencing the human genome—to sequence genes is to determine the exact order of the base pairs in a section of DNA (NHGRI, 2012; NHGRI, 2010). In sequencing the human genome, scientists were able to completely map all the genes that human beings possess; through mapping, we can understand the structure and functions of our genes.

The detailed genetic information provided to us by the HGP can be utilized for several different purposes. Frank Collins, the director of NHGRI, described the genome as a "shop manual, with an incredibly detailed blueprint for building every human cell" (NHGRI, 2012). Using an individual patient's genome blueprint will help medical practitioners determine how to best approach the treatment of a condition, whether it is preventative, diagnostic, or therapeutic. Genomic research has already resulted in the identification of major alleles that indicate predisposition to develop certain genetic disorders. Through genetic testing, it can be determined whether an individual possesses those alleles (Guttmacher & Collins, 2002; Byers, 2006).

Because of our knowledge of the human genome, a genetic test can, for instance, show that a leukemia-stricken individual is homogenous for a mutation in the gene that encodes thiopurine S-methyltransferase, an enzyme that inactivates mercaptopurine, a drug used in leukemia treatment. Knowing who is affected, doctors are able to greatly reduce the dosage of mercaptopurine. By reducing the dosage and carefully monitoring the patient's blood levels, physicians are able to ensure the drug levels remain therapeutic, rather than toxic, a possibility for someone with the enzyme thiopurine S-methyltransferase. Just a few years ago, about 1 in 300 patients suffered serious, blood-related adverse effects during mercaptopurine therapy. Now, genetic tests are routine and alert physicians to this genetic predisposition, allowing them to treat leukemia patients so they can achieve complete remission (Guttmacher & Collins, 2002).

Hundreds of thousands of single-nucleotide polymorphisms (SNPs) have been tested for association with a disease. Studies have identified SNPs implicating hundreds of replicated loci (locations on genes) for common traits, perhaps simplifying the mystery behind complex genetic conditions, in which many genetic and environmental factors work together. The next step is to narrow an implicated locus to a single variant that disrupts the

expression of a protein, directly causing susceptibility to disease. Already nearly 800 SNP-trait associations have been reported as significant. Multiple variants can be associated with each condition; the prevalence of these variants indicates how the disease is spread and who is most susceptible (Manolio, 2010).

Five major variants are associated with age-related macular degeneration. Each one is associated with a risk of disease two or three times the risk for a person without one of the variants. Two of these variants are common in the studied populations while the other three have allele frequencies of 5 to 19% in the populations studied. As a whole, the five variants more than double the risk of age-related macular degeneration in the siblings of effected persons, suggesting that the complement-mediated inflammation pathway is central to the passage of the disease. New therapies can be developed with the newfound knowledge that inflammation plays a role in the disease—this demonstrates the usefulness of learning more about the human genome, as genomewide association studies can implicate previously unsuspected pathways in the cause and pathogenesis of disease (Manolio, 2010).

Genomewide association studies have associated SNPs with genes that had not previously been thought to be connected to a given disease; similarly, they have identified loci that are shared by conditions previously thought to be unrelated. By revealing the unexpected involvement of certain functional pathways in a variety of disease processes, new approaches and potential treatments have been discovered. More remains to be learned, but these studies will have an enormous effect on the development of future therapies, as well as the assessment of an individual patient's level of risk for a particular condition (Manolio, 2010).

The sequencing of the human genome has provided us with a wealth of information that can help us guide future research and individualize patient care. Utilizing information gathered from different quarters will help scientists know how to best treat and possibly cure genetic conditions and aid physicians in making treatment personalized and safer.

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Essay 131485

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Essay
The Uses of Human Genomal DNA Sequencing and its Effects on Biotechnology, Human Health, and Human Diseases

When one looks at the span of human existence relative to the history of the universe or even the history of the Earth, we seem rather insignificant. However, there are moments of human brilliance when we as a species are proud to be known as Homo sapien or wise men. One such moment, while fleeting, defines a revolution in genetics. This moment is the discovery and usage of the technology to sequence the human genome. Sequencing is the process by which the order of base pairs in a segment of DNA can be determined ("DNA Sequencing," 2011). Since its discovery in 1968, sequencing has become a driving force in the genetic field and has even led to the formation of a new field of science known as bioinformatics (Hutchison, 2007). However, it was not until April 14, 2003, that the global project known as the Human Genome Project was completed, thus establishing a revolution in the fields of genetics ("Human Genome Project," 2010). Overall, the discovery of sequencing, particularly in the human genome, has led to advances in genetic technology—biotechnology—, discoveries in human health and methods of dealing with human diseases, among others.

Biotechnology has evolved with the discovery of sequencing. In fact, one example of a biotechnology has developed from DNA sequencing is the detection of epigenetics via DNA methylation. This is done through a process known as SMRT sequencing or single-molecule, real-time sequencing. In this process nucleotides that have fluorescently tagged are incorporated into DNA strands through the use of DNA polymerase. However as this occurs the fluorescent tags pulse and this information can be used to detect whether methylated nucleotides are present in the fluorescently modified DNA. (Flusberg, et al., 2010) The use of such a technique is critical to the understanding of diseases that affect the epigenetics of an organism such as cancer, Prader-Willi Syndrome and Angelman Syndrome. Its use is more important now than ever as there is an increase in the number of children born with imprinting diseases that are the result of a type of reproductive technology known as in vitro fertilization. (Lobo Ph.D., I, 2008)

Discoveries in human health have also been affected by sequencing. In fact some scientists have theorized a method of dealing with "personal health care" through the use of DNA sequencing at the nano-level. It involves the combination of DNA and nanotechnology. In this method DNA passes through a "nanopore" that is obtained from the bacterium Mycobacterium smegmatis. At the University of Washington this method was used sequence DNA at an impressive speed and with little cost. Their goal is to use this nano-method of DNA sequencing to develop medicine that is ideal or "fitted" for each person. The implications of such technology would be grandiose and would revolutionize the way medicine and health care are viewed today. (Anonymous, 2010)

Advances in ways of testing for or treating human diseases have arisen from DNA sequencing. In fact, DNA sequencing can be used to determine abnormalities in the chromosomes of fetus through massive parallel DNA sequencing the DNA found in the blood of the mother. (Sehnert, et al., 2011) Even many different cancer types can be detected by sequencing a biopsy. Such types of cancers include non-Hodgkin lymphoma, medulloblastoma, and

Even many different cancer types can be detected by sequencing a biopsy. Such types of cancers include non-Hodgkin lymphoma, medulloblastoma, and lung carcinogenesis. ("Cancer Genome Anatomy Project") The effects of both advances on the human race is incalculable as both methods can be used to determine a disease prior to major symptoms appearing, which can be used to prevent further damage and even death in some cases.

As demonstrated by the above examples, sequencing has come a long way since it was first discovered in 1968. It has an innumerable number of uses whose benefits are as incalculable as they are important to the understanding of human genetics and it relative fields of study. As such, DNA sequencing has had and will continue having a great impact on the study of human health, biotechnology and human diseases.

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Essay 131486

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Essay
On April 14, 2003, after thirteen years of cooperative effort by geneticists from all over the world, the Human Genome Project was pronounced complete. As one of the most monumental tasks ever undertaken—the sequencing of all 3 billion base pairs and the mapping of the approximately 25,000 genes of the human genome—the Human Genome Project is a feat of discovery comparable to mankind's other great voyages of exploration ("All About the Human Genome Project"). In the ten years since, the information garnered by the Human Genome Project has proven invaluable to many fields of study, particularly biotechnology and medicine. The integration of genomics has already revolutionized the ways that diseases are diagnosed, treated, and monitored, and the discipline's continual advancement is sure to have a profound impact on the future of public health and the practice of medicine.

The Human Genome Project has provided invaluable insight into the nature of diseases by elucidating the link between genes and the manifestation of diseases. All diseases have an underlying genetic factor, and understanding the ways genes interact with other genes and with the environment is crucial to combating disease ("Medicine and the New Genetics"). Diseases are commonly caused by mutations in genes, which can directly result in the disease or can constitute part of the complex pattern of inheritance that governs a person's susceptibility and response (National Center for Biotechnology Information). The results of the Human Genome Project have allowed researchers to identify distinct errors in the human genome that cause or contribute to disease, leading to the development of genetic tests for more than 2,200 diseases (Centers for Disease Control and Prevention). Genetic tests are performed for a variety of reasons—confirming a suspected diagnosis, predicting possibility of future illness, screening fetuses for genetic defects—and are an important tool in helping to improve health by informing people of their current health status and their predisposition for certain diseases so that they can take appropriate measures ("All About the Human Genome Project").

Another important legacy of the Human Genome Project lies in the field of pharmacogenomics, which examines how variations in multiple genes affect individuals' response to drugs—hence its name, a combination of "pharmaceutical" and "genomic". Drug therapies currently in use are based on the precept of "one-size-fits-all", but studies have shown that only about 30-70% of people react favorably to a large proportion of drugs ("Human Molecular Genetics"). Every year, over 2 million people are hospitalized due to adverse drug reactions (ADRs), with about 100,000 cases resulting in death, thus making ADRs one of the leading causes of hospitalization and death in the United States ("Medicine and the New Genetics"). By attaining knowledge on which genes are cause diseases and which are involved in drug response, pharmacogenomics promises to bring about specialized drugs, designed according to each person's specific gene makeup. Not only will this reduce the number of hospitalizations, but it will also ensure that drugs are more powerful, provide advanced screening for disease, improve the drug discovery and approval process, decrease the overall cost of health care—basically, the idea of "the right drug at the right dose at the right time" ("Medicine and the New Genetics"). Pharmacogenomics is still in its early stages, but with the rapid pace of progress in genomics, the advent of individualized drug therapy is not impossible.

Another emerging industry that has benefited greatly from the Human Genome Project is gene therapy, a technique of correcting health problems by altering the genes causing them ("Your Genes, Your Choices"). Most gene therapies work by replacing a defective gene with a normal, functioning one through the use of carrier molecules called vectors ("Medicine and the New Genetics"). Researchers are currently conducting trials in using gene therapy to cure diseases like cystic fibrosis and severe combined immunodeficiency ("Your Genes, Your Choices"). Gene therapy is still very much in its infancy, and it is too early to tell if most treatments will prove effective, but if it can be successfully implemented it will be invaluable in the fight against disease ("Medicine and the New Genetics"). In the meantime, biotechnology is advancing in leaps and strides, and has already significantly changed the face of the medical world ("Your Genes, Your Choices"). For example, genetically engineered products are a commonplace feature of today's society, found in everything from medicine to agriculture. Although there are many ethical and safety questions regarding the use of genetic engineering, the many benefits we can derive from them cannot be denied.

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Essay 131487

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Essay

10 years have passed since the completion of the Human Genome Project, and since then, it has served as the foundation for numerous discoveries and realizations in human genetics. Already paying for itself, advances in medicine, genetic testing, bioarchaeology, evolution, forensics, agriculture, and energy development are only some of the results made possible by data obtained from the 2.7 billion dollar project. Furthermore, as a basis for later projects such as ENCODE, past misconceptions of genetics have been disproved and a greater understanding of DNA has been reached.

An example of such is the phrase “junk DNA” and all of the negative connotations surrounding it. Even the co-discoverer of the double helix of DNA, Francis Crick, commented that it was “litter better than junk.” For years, it has been thought that only about 1.5 percent of the human genome is actually useful; these are the genes that encode the proteins that make life possible. Through projects such as ENCODE, standing for the Encyclopedia Of DNA Elements, however, the percentage has since climbed up to 8 to 9 percent, accounting for the genes that work to regulate the genes that produce proteins. This number can continue to rise in the future as a better understanding and a more thorough analysis is made, for the project has only sampled about 50 percent of the total amount of gene as of September, 2012. The head of the project, Ewan Berney of the European Bioinformatic Institute in Cambridge, England, has commented in an interview with Scientific American that the percentage of genes with functional significance may raise to 80 percent depending on how one looks at the ENCODE data, and that the project will continue to take a deeper look into the importance of the transcription of certain genes to reach a conclusion. A major discovery of this project is the clarification of how some diseases operate, such as Crohn’s disease. 75 percent of genes previously thought to be linked to diseases are now seen to be far from the protein-coding genes, and researchers can now discern where to begin looking for the causes of certain diseases to prepare counters and even cures.

Launched in January, 2008, the 1000 Genomes Project is another project stemming from the Human Genome Project. As compared to the Human Genome Project, which sequenced the human genome of only one man, the 1000 Genomes Project aims to sequence the genomes of 1000 humans to map the genetic variation of man, and hopes to use the data to seek out the genetic origins of diseases. So far, the project has achieved more than expected, successfully sequencing 1092 human genomes and has displayed its data on its website for public use. Through the sharing of this large mass of data, the project hopes to help in the development of smaller, more specific disease studies. Researchers using the current genetic information has already identified more than 100 regions of the genome associated with diseases ranging from autism to cancer. 99 percent of the human genome has been observed to be shared among all humans, but as geneticist Lisa Brooks commented, “... by mapping variations in the other 1 percent, the 1000 Genomes Project may help reveal the genetic underpinnings of some disease.” The project itself is only to gather information, but the precision of the data obtained holds the potential for future genetic discoveries.

The Human Genome Project serves as the infrastructure that makes these later projects possible. Although it looks to be only a string of letters depicting the nitrogenous bases of adenine, thymine, cytosine, and guanine by itself, it is the alphabet that gives rise and meaning to the projects like ENCODE and the 1000 Genomes Project. 2013 marks the 10th anniversary of the Human Genome Project, and it will no doubt continue to be remembered and serve as the foundation for countless more projects to come.

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