# 8th Annual DNA Day Essay Contest Submission

**Deadline: Friday, March 15, 2013, 5:00 PM US ET**

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**Essay 131476**

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**Essay**
DNA is an essential part of every organism's life and when sixty years ago, Watson and Crick discovered its structure to be a double helix, the implications for science were endless. Fifty years later, he complete draft of the human genome, done in 2003, built upon this. It affected the way we deal with basic genetics; it determined that most of the genome does not code for proteins and that, for all the regions, there is a range in the level of variability.

The Human Genome Project helped demonstrate that about most of our genome does not code for proteins. Whereas previously, it was thought that there are 80,000 to 100,000 genes that code for proteins, only about 30,000 genes, or about 2% of our genome, really do so. A lot of this noncoding DNA

is regulatory; transcription factors attach to these areas, and this controls if a gene will be turned on or off. Furthermore, Hank Green explains in the Scishow video "3 Sad Surprises: The Human Genome Project" that many portions of the genome code are portions of viral DNA that got stuck in our ancestor's genomes. Plus, some parts code for structures that we no longer have, like tails. Building on previous research, like the discovery of introns in 1977 by Philip Sharp and Richard Roberts; within genes, there are stretches of noncoding DNA, called introns. These are spliced out during the formation of mRNA by transcription. By understanding the makeup of DNA, and therefore the basics of gene expression, we are able to recognize the ways in which DNA is regulated and expressed.

Another important result of the Human Genome Project was establishing that noncoding regions of human DNA are hypervariable. Genes tend to have similar sequences to result in the same proteins for normal human function. Because this is not the case for noncoding DNA, it can be more variable for all humans. According to "Implivations of the Human Genome Project for Medical Science" by Francis S. Collins and Victor A. McKusick (2001), "While human DNA sequences are 99.9% identical to each other, the 0.1% of variation is expected to provide many of the clues to the genetic risk for common illnesses." This demonstrates that understanding the structure and basics of the human genome has further implications for genetic diseases and dealing with them.

The project also determined that there is variation on the human X chromosome, but to a lesser degree than other parts of the genome. This was done through a long alignment process; first, as David Bentley and other describe in "Accurate Whole Human Genome Sequencing using Reversible Terminator Chemistry", the X chromosomes of a Caucasian female was sequenced. This produced a set of base pairs, which were then aligned to the human genome reference sequence. Using ELAND and MAQ, two alignment algorithms, read pairs were places based on references or randomly if there were multiple possibilities. This allowed for a thorough coverage of the X chromosome but also demonstrated that there is about one substitution every 2.3kb or a 4.3 x 10-4 heterozygositiy (Bentley, 2008). Plus, there are structural variants; they found about 115 indels. There is a large amount of pressure from natural selection placed on the X, as opposed to on an autosome. This is because, for a recessive trait to appear in autosomes, you need two copies; but this is not the case usually for males for the X chromosomes in males (Khan, 2011). Therefore, due to this pressure, is less genetic diversity on the human X chromosome than the average chromosome.

The Human Genome Project set out to understand human genetics and cure diseases that occur due to mutations. Although this was not done completely, the Project, through sequencing, has aided the scientific community's understanding of how DNA, expression, and variability work. It demonstrated that genes make up for about 2% of the genome and that the rest can be variable because of this. Plus, the level of variability on the X chromosome was determined. However, one incredibly important implication of the Project goes beyond genetics; it demonstrates that all science builds upon past findings. Because Watson and Crick determined the structure of DNA, the Human Genome Project can build upon, with help from technologies and other research about the makeup of DNA.

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## Essay 131477

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**Essay**

60 years ago, Watson and Crick discovered a breakthrough in genetics. They discovered the all-important double helix. Now, most people wouldn't find that to be a big deal. But, in all honesty, it's one of the most significant things in the world. It's pretty amazing that something as simple as a chain of deoxyribose sugar, phosphate groups, and a series of one of four bases - adenine, cytosine, thymine, or guanine - can encode something as small as a single-celled bacteria to something as large as a blue whale, or as complicated and organized as a human being.

DNA is... to put in in one word, smart. Our own DNA knows all you need to know (and more) about everything to do with us. DNA codes for all of the proteins in our body. It's like an instruction manual. Before we knew this, we had no clue how DNA worked. After Hershey and Chase discovered that DNA was the genetic material, using bacteriophages and radioactive markers, we were left wondering, "how?".

Then, Watson and Crick came along and discovered the double helix. Of course, Rosalind Franklin should take some credit for that. After all, she did find all the information; Watson and Crick just broke into her lab and stole it (Ardell). And with this discovery of the double helix structure of DNA, we can do so much. We can use the double helix to engineer amazing things. For example, people with type one diabetes need to take insulin shots. They used to get the insulin from cows or pigs, but that was problematic. The animal's protein was a bit different, sometimes people would have allergic reactions and it wouldn't always work. We needed a better source of insulin, and we needed human insulin. We couldn't get it from other humans, but we needed to get it from somewhere. That's where genetic engineering comes in. Thanks to our sequencing of the human genome, we know enter the insulin gene is located and what its exact sequence is. Using recombinant DNA technology, which wouldn't exist if it weren't for Watson and Crick's discovery, we were able to put the human gene responsible for insulin production into a strain of E. coli bacteria's genome (Mundasad, 2011). Thanks to this, we now have giant vessels filled with strains of bacteria that can produce human insulin, and the diabetics are able to live easier, normal lives.

Without our knowledge of DNA, a lot of amazing feats wouldn't be possible. Recently, King Richard III's grave was found under a parking lot in England. How did they know it was him? They did their research, and compared his mitochondrial DNA to some of his descendent's mtDNA. They looked at the non-coding variable region. Using this, they were able to identify him an King Richard III (Jones, 2013).

DNA is everything. Without DNA, where would we be? The thing is... we wouldn't "be" at all. It's amazing how DNA, such a simple molecule, can give rise to something as miraculous and complicated as life. DNA is really everything, and that's why it's so important.

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## Essay 131478

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**Essay**
The Human Genome Project: A Great Leap For Mankind

We've moved far beyond the "ancient" misconceptions of inheritance. Coming from vaguely saying that children get traits from their parents to the decidedly more scientific description, involving meiosis, fertilization, alleles, chromosomes, and dozens of other buzz words that you learn in a biology class, we have definitely made significant progress in genetics research. From the pea plant experiments of an Austrian monk named Gregor Mendel, to the Drosophilia fly experiments of Thomas Morgan, it is mind-boggling to examine our progress in understanding heredity, and all that it implies for us as a species. Now, we celebrate the tenth anniversary of the first sequencing of the human genome. Who would have thought that we could have made it from pea plants to mapping the genome of the human species in such a relatively short amount of time? It is certainly worthwhile to reflect upon the discovery, and the myriad of ways it has benefitted human society.

The first sequencing of the human genome has opened up many portals for the betterment of humanity. Dr. Chial (2008) explains in her article about the Human Genome Project that sequencing the genome has allowed us to make remarkable steps in genetics research, such as helping scientists discover over 1,800 genes that cause disease. As of 2010, researchers have associated 38 genetic regions with type 2 diabetes ("Researchers Identify 12 New Genes," 2010.). Some of these genes are particularly important, as they are involved in beta cells (which are directly involved with making insulin) found in the pancreas. Though the research results are modest, the fact that we have made progress towards better treatment, or even curing this kind of disorder bodes well for studies being made now and in the future. In addition, we can rest assured knowing that we have the capacity to overcome obstacles that we previously thought too high to leap over.

Furthermore, these discoveries have led to the emergence of over 2,000 DNA-based tests for diagnosing genetically based diseases. One of these tests

includes the one for Huntington's disease, a disorder that causes cognitive decline and loss of proper muscle coordination. With the advances made with help from the results of the Human Genome Project, testing can now be done during pregnancy (either by amniocentesis or with chorionic villus sampling) to determine whether the gene mutation causing Huntington's has been inherited ("Gene Testing," n.d.). Without the sequencing of the human genome, early diagnosis and treatment of Huntington's, and countless other diseases, might not have been possible.

Looking towards the future at how sequencing the human genome has offered promises of an improved lifestyle for society, at least 350 biotechnology-based products have been in clinical trials, and more are to come as we continue to build off of the information gleaned from the sequencing. It has propelled the United States to a position where we are one of the leading forces in the biotech field, and this is significant, considering that the sector was nonexistent before. Plus, there are new initiatives that strive to take advantage of the relatively new sequencing data such as The Cancer Genome Atlas, which hopes to link major types of cancer to genetic abnormalities ("Human Genome Project," n.d.). Of course, the future holds the promise of eventually linking genes to traits like intelligence and personality, opening a whole new realm of possibilities.

Though we have certainly made leaps and bounds in terms of the field of genomics, there is still a lot we don't know about our genes. More complex diseases such as heart disease or the role of other coding genes are not completely elucidated for us just yet. However, it is obvious that we are on the forefront of making some potentially world changing discoveries in genetics. Watson and Crick set the ball in motion with the double helix model of DNA, and the sequencing of our genome marks a major milestone for civilization. As Dr. Francis Collins and Dr. Victor McKusick (2001) put so succinctly in their article in the Journal of the American Medical Association: "We must commit ourselves to exploring the application of these powerful tools to the alleviation of human suffering, a mandate that undergirds all of medicine." By applying our newfound knowledge to medicine and biotechnology, we can achieve a new standard of lifestyle, one where we can rest more comfortably knowing that we can, and will win the war against disease.

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## Essay 131479

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**Essay**

Personalized Medicine and the Bright Future Ahead

A huge advancement in the fields of personalized medicine, human health, biotechnology, and disease came with the sequencing of the human genome in 2001. What is the human genome? It is the nucleotide sequence of the three billion base pairs (A, T, G, C) which are found in the DNA of every cell. The initiative to decode this sequence, known as the Human Genome Project (HGP), was completed in 2001 and thanks to this discovery of "who we are," it may one day be possible to cure genetic diseases.

Personalized medicine is treatment designed for each individual patient. Currently, direct to consumer companies (DTC) are using "your" genome to help you, by offering DNA tests that provide clues into personal genetic traits and disease risks. Genetic testing "can improve lifestyle choices and increase preventive screening." (MG, Murray & Levy, 2009). Personalized medicine takes your genomic sequence and sees what diseases or problems you may have to help treat you better. Thanks to our current technology and knowledge of the sequencing of the human genome, it is as simple as sending a cotton swab with your salvia to receive a 99.7% accurate estimate of your disease risks (MG, Murray & Levy, 2009). The DTC companies find your absolute disease risk by taking the genotypes from your salvia and looking at the sequencing of the markers to see if it matches with any of the markers that are known to cause certain diseases. This development is also leading to better use of drugs. Rather than having the hospital prescribe some broad range antibiotics that are used for several conditions, they would give you the specific drug you need. With the human genome now, "Drug-metabolizing genes have been characterized sufficiently to enable practitioners to go beyond simplistic ethnic characterization and into the precisely targeted world of personal genomics" (Ng, Zhao, Levy, Strausberg & Venter, 2008,). The goal is to have a personalized genomics approach over a race-based approach to medicine (Ng, Zhao, Levy, Strausberg & Venter, 2008,) to improve the quality of medicine and treatment. Without the sequencing of the human genome, none of this would be possible or else know what we would be looking at.

After the sequencing of the human genome, scientists discovered that most of our DNA was "junk." However, this "junk" was soon to be found as non-coding sequences that are unique to almost every person. This led to advancements of the biotechnology involved with this junk DNA. For example,

forensics now uses your non-coding regions to determine if you are the father of a baby or if you committed a certain crime. This was all takes to polymerase chain reactions that took and "cloned" the non-coding regions. It is also used to help track your ancestry and help teach our generation about from where they originated.

The sequencing of the human genome's most important use has to be in the field of treatment of diseases. Thanks to this four-billion dollar project (the human genome project), doctors at Southwest General Hospital found that many diseases are caused by GEMS (genetic enzyme methylation syndrome). The methylation process controls the turning off and turning on of certain genes. Without it, there would be no expression of genes, but there would also not be any diseases. According to the doctors at the hospital, "Diseases are caused or precipitated, to a large degree, by inherited genetic errors in the genes that cause abnormal speeds in the enzymes of the methylation pathways. Statistically, about 40% of the population has at least one genetic error in the methylation genes." (Rozakis, 2013). The treatment, if it is found that this disease is caused by a GEMS problem, is to give specifics on food avoidance or even certain hormones. It is hoped that this system will soon prevent eye disease and manage diabetic retinopathy and macular degeneration non-surgically and with less harm. (Rozakis, 2013,). Another example is the recent response to severe acute respiratory syndrome. It was a huge outbreak that was spreading around most of Asia. Thanks to the current technology based on the sequencing of the human genome, SARS viral genome was deposited into public sequence databanks and the information derived from the genome sequence of SARS gave perceptions into the origin of the disease as well as aided with patient diagnosis and thus saved most of Asia (Fox, 2003,).

With the current knowledge of the human genome sequence we have created a much healthier and brighter future.

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**Essay 131480**

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**Essay**
The Map of our Existence

A human genome; it's located in nearly every single cell of a human, and it contains all genetic information of a person, ranging from the color of a person's eyes, to whether or not someone is at a higher risk for breast cancer. It holds all the numbers and probabilities for human characteristics. In 2003, the Human Genome Project (initiated in 1989) announced its completion of the human genome sequence. The mapping of the human genome is a segue to better grasping human health & disease, as well as an exponentially expanding the biotechnology field [U.S. Department of Energy Genome Programs, http://www.ornl.gov/sci/techresources/Human_Genome/project/timeline.shtml]

After the discovery of the human genome, a new project was introduced; to key which genes are linked to what traits. Not so long after this, a gene was found (LMNA gene) to be directly responsible for progeria, a pre-mature aging disease. Then in 2004, the National Human Genome Research Institute found a possible linkage between variations in a certain gene and Type II Diabetes. Perhaps, however, the culmination of all of this and even more was the publishing of the International HapMap Consortium's catalog of genetic variation [http://www.genome.gov/17015412]. By using this catalog, scientists could find differences in individuals' genes as minimal as 0.01%, and connect such genetic differences to certain diseases. By identifying the connection between genes and diseases, the information can be given to doctors to form accurate diagnosis. Physical diagnosis can sometimes be unclear, or inaccurate, so the usage of gene testing to clarify a diagnosis can sometimes be even life-saving.

As of right now, there exist over 1000 different diseases detectable under gene-testing, such as Huntington's disease, cystic fibrosis, colon cancer, or Alzheimer's. From this doctors can use the correct treatment/therapy of the disease. When it comes to diseases like Alzheimer's (adult-onset diseases) or certain forms of cancers, gene-testing can only provide a probability over developing the disease. Thus, gene-testing for adult-onset diseases are usually only directed towards a healthy person who doesn't display any of the given symptoms. Another benefit of gene-testing is that it outlines all mutations in a couple's DNA, so they can view the possibilities of their child being afflicted with a genetic disease they didn't know about. Despite the room for error (as exists in nearly all laboratory), scientists agree that the benefits far outweigh the risks for a misdiagnosis, or a false-positive [U.S. Department of

Energy Genome Programs, http://genomics.energy.gov]

However, the possibilities from discovering the human genome don't just stop there. Gene mutations don't always imply genetic diseases and negative afflictions; on the contrary, genetic mutations are what run evolution and keeps species alive. In August 2000, scientists found a gene mutation (MMP-8) that is linked to suppressing tumors formed by melanoma, the deadliest form of skin cancer. This led doctors to modify their treatment of melanoma treatments; rather than suppressing the gene which sometimes led to accelerated tumor growth, doctors sought to promote/rejuvenate MMP-8 function (at least the genes that weren't truly oncogenes)[ http://www.genome.gov/27530882].

The sequencing of the human genome is a huge accomplishment for mankind, for the benefits we can reap from this treasure trove are truly amazing. Human genetics is one of the most complicated areas in biology, so the mapping of the genome truly allows us to greater analyze traits and characteristics. Whether it be identifying a genetic disease and its cause, or finding a treatment for such diseases, this discovery can truly save lives. The impact of the success of the Human Genome Project itself has sparked parallel undergoing projects in other matters. For example, scientists are currently attempting to create a connectome for the brain; instead of a map for the human genome, a connectome is a map of the human brain [Lichtman & Sanes]. The success of the Genome Project is a benchmark for this project; critics say that the connectome is impossible to achieve, yet the same was to be said about the Genome Project. In addition, ever since the beginning of the Human Genome Project, numerous sequencing attempts were made on other organisms. For example, in 2005, the finished of the chimpanzee genome showed that they are humans' closet living relative, with a DNA makeup-similarity of 96% [http://www.genome.gov/11509418]. The Human Genome Project and its success in mapping out our genome is truly an amazing feat and have far-reaching impact in the science community.

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## Essay 131481

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**Essay**

In 2003, ten years ago, scientists sequenced the first segment of DNA of the human genome. Since then, DNA sequencing has been done all around the nation by scientists who are all working together on the Human Genome Project. The human genome contains about twenty to twenty-five thousand coding genes and is about three billion base pairs long. The exact number of genes in the human genome has not been determined yet due to the incomplete research done, but estimates range from around seventy-five to eighty thousand genes.The ultimate goal of this developing thirteen year project is to determine all the genes of the human genome as well as the sequences of all base pairs. They strive to find the exact sequence for DNA on all 23 chromosomes as well as map out genetic and physical linkage between each chromosome.

This study of the human genome has led to many new advances in medicine and human health. By determining the sequence for a normal human gene, scientists have made it easier to detect variations and mutations. As genomic maps become increasingly more detailed, researchers have found easier ways to make correlations between genes and their corresponding genetic disorder. Already, certain genes have been associated with some genetic disorders; some of these including “myotonic dystrophy, fragile X syndrome, inherited colon cancer, Alzheimer’s disease, and familial breast cancer” ( Potential Benefits of Human Genome Project Research). While part of the HGP is still working on uncovering new genes, some researchers have taken a different course to try to find ways to predict genetic diseases in people using DNA sequences of genes they assume to be related to the disease and prenatal testing of the fetus. In fact, many new tests have been developed yet due to ethical challenges, the scientific community has been hesitant to reveal their discoveries with the medicinal community as well as the public. (Medicine and the New Genetics).

The successes of the HGP has also helped reveal how faulty genes may cause diseases. Using this information, the medicinal community has taken a new approach towards finding new treatments; by examining the genes and the root cause of the disease rather than just using what works. Rather than the previous trial-and-error method, researchers focus on protein structure and gene sequences. The HGP has also sparked the new idea in the scientific community of gene therapy; using genes themselves to cure diseases. Researchers believe that if enough is known about the genes involved and detection is early, the process of replacing defective genes with normal genes holds great potential. At the rate of the HGP currently, scientists seem to predict an extreme upsurge of treatments in the near future as they believe that by 2020, there will be about a thousand known genomes in the system and at least twenty-five hundred new drugs which will be more effective, The medicinal community suggests that by 2020, doctors will be able to test individual genomes as well as prescribe drugs specific to each individual’s condition (Fast Forward to 2020) (Medicine and the New Genetics).

Ultimately, since ten years ago, the study of the human genome has been significantly emphasized and is valued greatly in the scientific community. The current HGP has helped researchers understand more about genes and how each might correspond to a genetic disease. Though the human genome may seem massive, the HGP, as a group effort, has been developing rather quickly and already associations have been made and genomic maps are being

drawn up. Scientists hope that the progress of the HGP will lead to future benefits as well as more effective drugs. The medicinal community works tirelessly to formulate new treatments using their new method of study which clearly is more dependable than the trial-and-error methods used before. As a whole, the scientific community expects the HGP to lead to much greater things and they are confident that in a few years, many more diseases will be cured, or at least have an effective treatment.

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Medicine and the New Genetics U.S. DOE Human Genome Project

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