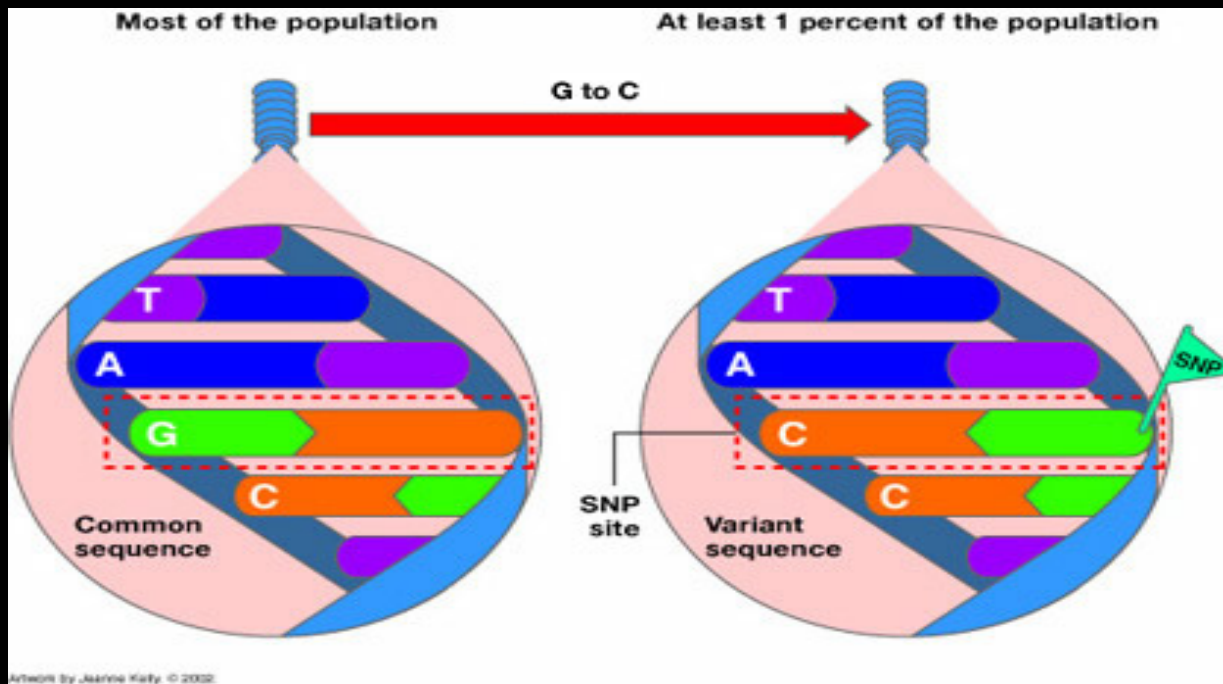


SNPs
Single Nucleotide
Polymorphisms

What are SNPs?

- **S**ingle **N**ucleotide **P**olymorphisms (**SNPs** , pronounced "snips") are genetic variations that occur in a DNA sequence.
- A **SNP** Variation is when a nucleotide (Adenine, Cytosine, Thymine, or Guanine) replaces one of the other three nucleotides.



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- In the 3 billion Base Human Genome, **SNPs** occur every 1000 base pairs or less.
 - **SNPs** can occur in both the coding and non-coding regions of the genome.
 - The coding sequence is only 3% of the Human Genome that is used as the set of instructions to direct the building of a protein with a specific function. Most **SNPs** are found outside this coding sequence.
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- Most **SNPs** are considered biologically 'silent', meaning they do not have any effect on the function of the gene or on any inherited traits.
 - **SNPs** serve as valuable genetic markers to locate a disease on the human genome map because of their frequency, stability, and even distribution in the genome.
 - A **SNP** is usually found near a gene that is associated with a certain disease. So, it can be used to search for and eventually isolate the disease-causing gene.
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- It has been found that sequencing the Chimpanzee genome and studying its SNPs might be very valuable in understanding the SNPs of the human genome.
 - Chimpanzees are considered to be the closest living evolutionary relatives of Human.



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- Chimpanzees have shared a common ancestor with humans 4.6 – 6.2 million years ago.
 - The difference in the overall genomic DNA sequences between Humans and chimpanzees is : an average of 1.2%.
 - The close relation between Humans and Chimps makes Chimpanzee DNA sequencing optimal for comparative studies that apply well to humans.
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Why compare DNA sequences of Humans and Chimps?

- Finding similarities and differences between the SNPs of the two organisms would provide more insight about the diseases and their expressions that those SNPs eventually develop into.
 - The close relationship between the two species minimizes the risk of having multiple substitutions at the same sites making the results unclear.
 - Comparing the polymorphism rate between human and chimpanzee indicates the regions where one species has a very different level of diversity than another.
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- Those regions might have undergone intensive selection that would assist in studying the history of humans & the evolution of resistance to disease.

Comparison of disease susceptibility between chimps and humans

Condition	Human	Chimp
<u>HIV progression to AIDS</u>	common	very rare
<u>Influenza A symptoms</u>	moderate/severe	mild
<u>Hepatitis B/C complications</u>	moderate/severe	mild
<u><i>Plasmodium falciparum</i> malaria</u>	susceptible	resistant
<u>Menopause</u>	universal	rare
<u><i>E. Coli</i> K99 gastroenteritis</u>	resistant	sensitive
<u>Alzheimer's disease pathology</u>	complete	incomplete
<u>Epithelial cancers</u>	common	rare

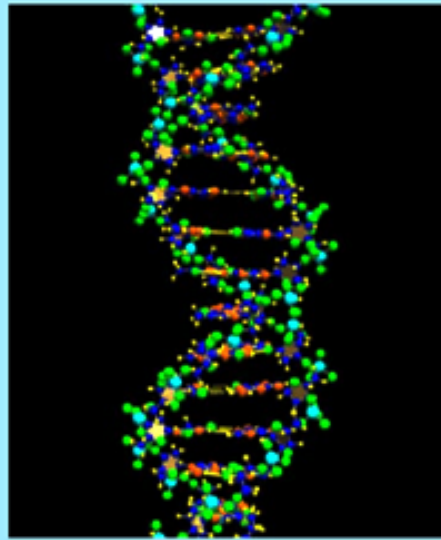
Project

- The goal of this project is to compare the distribution of **SNPs** over a particular gene for two different organisms. (Humans and Chimpanzees).
 - The data of Human and chimpanzee **SNPs** for the project will be obtained from the public **SNP** database (dbSNP) homepage.
 - Certain Software and Sequence Alignment Methods will be used in order to compare the DNA sequences of Humans and Chimps.
 - The comparison of both sequences would create a major impact on the level of understanding of human disease, human population genetics, and human evolution.
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Website..

<http://www.angelfire.com/sk3/compbio601/>

Foundations of Computational Biology



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THE END
THANK YOU

