

Overview of Genomic Polymorphisms

Content



Single Nucleotide Polymorphism (SNP)

When we sequenced the human genome from different individuals, we realized that there are a lot of variations in our genome. In this module we will explore what types of variation we have and what the consequences of those genetic variations are.

Single nucleotide polymorphisms (SNPs) were one of the most surprising discoveries of the Human Genome Project. We will first explore what kind of significant findings are based on the SNP discoveries.

Watch the following two introductory videos - one very short animation and one lecture - about genetic variations including SNP and structural variations.

SNPs - Single Nucleotide Polymorphisms (20-second animation)

Human Genomic Variations - SNP (31:16 minute lecture)

Sources:

- <https://www.youtube.com/watch?v=9rPDa2ACtog>
- https://www.youtube.com/watch?v=0j4b_oOsz78&feature=youtu.be



Finding SNPs in NCBI

You will explore the SNP database in NCBI website. The following reading is an introduction about the NCBI database: **Chapter 5, The Single Nucleotide Polymorphism Database (dbSNP) of Nucleotide Sequence Variation**. Please read the following paragraphs:

- Summary,
- Introduction,
- Physical Mapping,
- Functional Analyses,
- Association Studies
- Evolutionary Studies
- Searching dbSNP with figures 2a and 2b

Next, learn how this genetic variation information can be important for our health. We'll look at the nomenclature used to describe SNP alleles and position, using the MTHFR example.

- First, learn the importance of the MTHFR gene and certain diseases by reading the following: **Genetics Home Reference: MTHFR**.
- MTHFR is one of the genes being tested in a new field called **Nutragenomics**, which studies genes in connection to diet and human health. See the following video which shows how we can use this information for wellness.

Healthy Kids Corner: Nutragenomics Care and MTHFR, by Dr. Amber Brooks, DC (9:57 minutes)

- To use the MTHFR example to understand SNP nomenclature, watch the following video, **Methylation Made Easy Part 4 of 4 - The SNPs Code**, by Dr. Amy Yasko (4:45 minutes)

Sources:

- <http://www.ncbi.nlm.nih.gov/books/NBK21088/>
- <http://ghr.nlm.nih.gov/gene/MTHFR>
- <https://www.youtube.com/watch?v=yjRx9cXkkzo>
- <https://www.youtube.com/watch?v=Eh1ea-q8nu8>