GENOMICS IN SOCIETY

Advancing Genomics Technology

Genetics vs. Genomics

GENETICS

- The study of genes and the way that certain traits/phenotypes are inherited from one generation to the next
- Involves studying specific genes and the traits and effects those genes convey

GENOMICS

- The study of the entire genome, including gene x gene interactions and gene x environment interactions
- Relatively recent field of study
- Can be used to study more complex scenarios

Studying the Genome

- **The genome is the entire set of genetic material found within a cell or organism**
- Since genetics and genomics study different biological components, different research tools & technologies are needed for genomics
- Genomics research broadly looks for patterns, commonalities, and markers within the genome that elicit traits fundamental to how an organism interacts with it's environment, other genes within the genome, or other organisms
- Furthermore, genomics is an advancing field of study, so the technology is advancing in tandem as discoveries are made

Polymerase Chain Reaction (PCR)

- **PCR is a technique used to amplify (make multiple copies of) small segments of target DNA**
- Fast & inexpensive procedure—can make billions of copies in a few hours
- Substantial amounts of DNA samples are needed for analysis
- Studying isolated pieces of DNA are near impossible without PCR
- Once amplified by PCR, DNA can be used in many subsequent lab analyses

→ PCR is an important first step in completing several types of genomic analyses

DNA Sequencing

- **DNA sequencing determines the order of the A/T/G/C base pairs that comprise a DNA molecule, and determines the order of sampled DNA segments**
- Sequencing provides information on the type of genetic instructions carried in a particular DNA segment
- Advances in technology and automation have increased the speed & lowered the cost of sequencing DNA, and technology is continuing to advance
- Examples: Illumina, Oxford Nanopore, PacBio—all different types of high-throughput sequencing techniques

Comparative Genomics

- **The comparison of the genome sequences of different species**
- This is useful for determining regions of the genome among species that show evolutionary convergence (similarities) and differences
- Identifying DNA that has been conserved in different organisms throughout millions of yeas of evolution is helpful to understand what DNA is essential to life and certain functions across many species
- Allows scientists to understand how life has changed and been preserved over time

DNA Microarray

- **DNA microarrays are used to determine if the DNA of an organisms contains genetic mutations**
- Mutations are the biggest source of genetic various and can convey several distinct traits (good, bad or neutral) depending on what genes are mutated
- Large genomes likely have several regions where mutations occur, this can be hard to detect
- Microarrays use sophisticated technology to essentially compare mutated DNA to control (no mutation) DNA by a series of automated binding tests involving fluorescent dyes, PCR, computer scanning, and probing that look to see if DNA is mutated by whether or not it will bind with non-mutated DNA on a microarray chip

Genetic Markers

- **A genetic marker is a DNA sequence with a known physical location on a chromosome that can be associated with phenotypes**
- Markers are useful to help link specific traits with the genes that are responsible, i.e. a tree may show a phenotype for increased height and analysis may uncover that there is a marker linked to the genes or genome region that control height
- The marker could be a Single Nucleotide Polymorphism (SNP), a microsatellite, a part of a gene with no known function, or another variety of marker

Genetic Mapping

- **Genetic (or linkage) mapping provides evidence that traits transmitted from one generation to the next can be linked to one or multiple genes and it can show where these genes are located on the chromosome**
- Maps are created by analyzing DNA samples with a specific trait of interest to look for unique patterns in the base pairs, i.e. looking for genetic markers
- Genes and markers are located close together and likely passed on together during recombination due to close locations, so the more markers that appear on a map, the more likely it is that there is a marker located near the gene conveying a specific trait

Genome-Wide Association Studies (GWAS)

- **GWAS involves scanning markers across the genome of an organism or the genomes of multiple organisms to find variations associated with a particular trait or traits**
- Involves rapidly scanning markers across complete genomes/complete DNA sets
- Once genetic associations are identified, researchers can use the information to develop methods to better manage or select for the trait or traits of interest
- GWAS is useful for weeding out variation that contributes to complex traits

Summary

- Overall, there are many different types of technology & tools available to genomics researchers that can uncover genetic variation and determine where it is located in the genome and what traits it is associated with
- These technology range in their speed and cost, but as research advances they are becoming more inexpensive and the processing speeds are increasing
- Advances in genomics technology has allowed researchers to study the genome and all its complexities at a detailed level that was not possible in past decades