Week 13
Genetics: BIO-2306

The concepts this resource covers are the topics typically covered during this week of the semester. If you do not see the topics your particular section of class is learning this week, please take a look at other weekly resources listed on our website for additional topics throughout the semester.

We also invite you to look at the group tutoring chart on our website to see if this course has a group tutoring session offered this semester.

If you have any questions about these study guides, group tutoring sessions, private 30 minute tutoring appointments, the Baylor Tutoring YouTube channel or any tutoring services we offer, please visit our website www.baylor.edu/tutoring or call our drop in center during open business hours. M-Th 9am-8pm on class days 254-710-4135.

Keywords: Genome, Polymorphisms, Gene Flow, Hardy-Weinberg

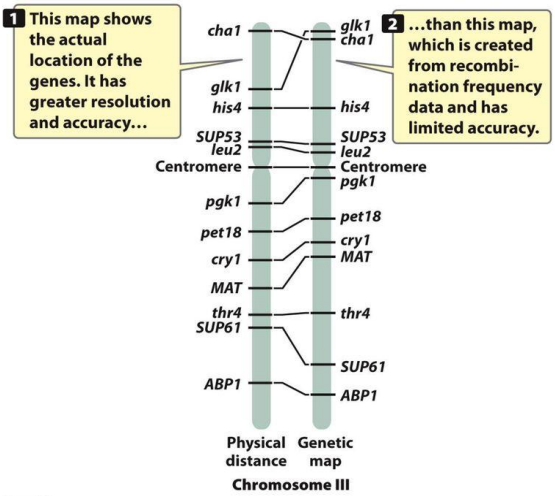
Topic of the Week: ***Genomics and Proteomics*** (20.1-3)

Genomics: the study of the genome (the totality of genetic information) of an organism

Think back to *T.H Morgan* and genetic maps… Would a **genetic** (based on recombinant frequency) or **physical map** of a DNA molecule be more 'accurate?'

Genetic Map: a map in '*map units*' based on the recombination frequency of sets of genes → lower resolution and accuracy

Physical Map: a map in *physical units* (ex. pm, nm, µm) which gives the *physical* distance between genes on a chromosome → higher resolution and accuracy



Genome Sequencing: determining the *entire* set of gene sequences in an organism's genome

This is difficult to do as only **500-700 bp** can be synthesized at a time, meaning millions of overlapping fragments must be put together to determine the sequence of a genome

The Human Genome: approximately 3.2billion base pairs, or 20-25,000 genes

The Human Genome *Project*: the goal was to sequence the entire human genome. This was accomplished in 2003 after 13 years. This is how:

Researchers modeled the genomes of increasingly complex *model organisms* and built sequencing techniques

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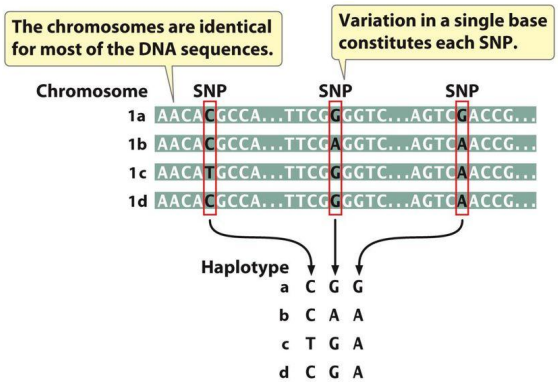
Using the tools gained from smaller genomes, they applies this to sequence the *entire* human genome.

Single Nucleotide Polymorphisms (SNP): single base pair variations between individuals of a given species → generally do not change phenotypes, so these are in non-coding DNA regions (arising through **random, silent** mutations)

Genome Wide Association Studies: using **SNPs** across an entire genome to find a particular gene of interest

Haplotype: a specific set of SNP variations identifiable to an individual



Bioinformatics: using molecular biology and computer science to analyze and codify genes.

These genes (or associated proteins) may be put into databases where they can be analyzed or annotated.

Metagenomics: sequencing the genome an *entire group* of organisms that share an entire environment and look at the effects on their DNA/protein expression

Microbiome: a metagenome that comprises all of the 'gut bacteria' in an animal; this collection of bacteria can be linked to health and certain cancers

Functional Genomics: determining the function of genes in a given organism

Transcriptome: the totality of an organisms' RNA genome that may be studied by functional genomics

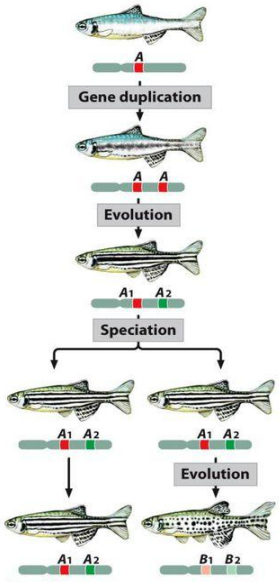
Proteome: The entire set of proteins produced by an organism, whose functions are studied

Homology: studying evolutionarily related genes, proteins or traits → often arise due to duplication

Analogs: an organism in a completely separate lineage has a gene/gene product similar to another unrelated species

Orthologs: homologous genes in two different species who share a [recent] common ancestor

Paralogs: homologous genes in the same species



Conclusion: Genes A_1 and A_2 are paralogs.
Genes B_1 and B_2 are paralogs.
Genes A_1 and B_1 are orthologs.
Genes A_2 and B_2 are orthologs.

Highlight #1: Population Genetics and Hardy-Weinberg Equilibrium (25)

Population Genetics: the study of changes in the allelic frequencies within a population of individuals (the fundamental unit of study for evolution and heredity)

Microevolution: small changes to *allelic frequency* with respect to time that can lead to **speciation** (the creation of a new species)

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Gene Flow: the transmission of genetic information between two groups of a species; cutoff of gene flow between populations considered **speciation**.

Gene Frequency: the frequency of given genotypes of alleles

Allelic Frequency → **NOTE:** 2N is the total number of alleles present

Dominant Allelic Frequency: the frequency of a dominant allele (ex. A)

$$f(\mathrm{A})=\mathrm{p}=\frac{2n(AA)+n(Aa)}{2N}$$

Multiple Alleles: when there is more than one allele (ex A1, A2, …)

$$f(\mathrm{A}^3)=\mathrm{p}=\frac{2n(A3A3)+n(A3A1)+n(A3A2)}{2N}$$

Recessive Allelic Frequency: the frequency of a recessive allele (ex. a)

$$f(\mathrm{a})=\mathrm{q}=\frac{2n(aa)+n(Aa)}{2N}$$

Sex-Linked (X-Linked) Genes: slight variation, were the number of males and females is considered as *m* and f, respectively

$$f(\mathrm{X})=\frac{2n(X1X1)+n(X1X2)+n(X1Y)}{2Nf+2Nm}$$

Hardy-Weinberg Equilibrium: what about a *non-evolving* population? Hardy-Weinburg equilibrium describes when a population can be at "rest," ie. not evolving.

To be a "non-evolving" population, it must meet the following criteria:

Large population
Random mating
No mutations
No natural selection
No migration

Additionally, **allelic frequencies** do not change and **genotypic frequencies** will stabilize after one generation

When the assumptions are met, the **allelic** and **genotypic** frequencies are *not changing*, so there is no evolution occurring (and the opposite is true as well).

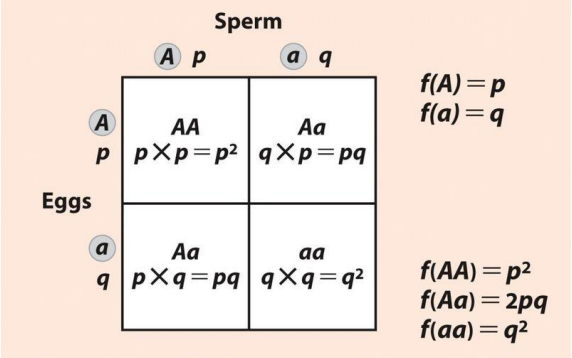
Looking at the change in a population over time is modeled as follows:

Hardy-Weinberg Equation:

$p^2+2pq+q^2 = 1$

$p+q=1$

p: dominant allelic frequency
q: recessive allelic frequency
p^2: frequency of homozygous dominant
q^2: frequency of homozygous recessive
2pq: frequency of heterozygotes



*The significance of a change in a population's allelic or genotypic frequencies can be modeled by - you guessed it- the ***X^2* test of independence**!

https://docs.google.com/document/d/1wCZ2KZr8nJ0ljw0WXm8qCRaprsNloxbRQHgTW-6JD1A/edit?usp=sharing

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CHECK YOUR LEARNING

Concept Check: *(Answers found on last page)*

1. Ankylosing spondylitis (AS), a degenerative genetic disease, occurs about once in every 1,000 Americans. Assuming that the population is in Hardy-Weinberg equilibrium, what percentage of the population would be expected to be a carrier? (think about what you are given!)
    a. 8.56%
    b. 6.12%
    c. 3.16%
    d. 96.8%
2. Alpha, beta and gamma globulin are blood proteins in humans. These evolved from a single globulin gene in humans. What type of homology is this?
    a. Analogs
    b. Orthologs
    c. Paralogs
    d. Heterologous
3. Which of these is <u>not</u> a type of RNA expression study? (<u>note</u>: this *content* is in resource 10, but the majority of the chapter is covered here!)
    a. Reporter sequences
    b. RNA seq
    c. Western blotting
    d. Microarrays
4. True or False: Non-random mating affects allele frequencies.
5. This table models the phenotypes of a population of lions:

	Males	Females
Golden Fur	120	122
White Fur	18	15

The fur white fur gene follows an <u>*autosomal recessive*</u> pattern of inheritance. What is the dominant allelic frequency? (assume H-W equilibrium)
    a. 0.88
    b. 0.12
    c. 0.65
    d. 0.35
6. (**Using the above table**)What is the frequency of *heterozygous* lions in this population?
    a. 0.12
    b. 0.83
    c. 0.46
    d. 0.91

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THINGS YOU MAY STRUGGLE WITH:

1. In **SNPs**, 99.9% of the human genome is shared and 0.1% is therefore variable. Thus, multiplying the variance by the genome size - 0.01x3.2billion - ~30 million bp of variation is to be completely expected.
2. **SNPs** can be highly specified and act as a marker for a particular disease or can be used to identify a certain individual.

CONGRATS: You made it to the end of the resource! If you have any questions about group tutoring sessions, private 30 minute tutoring appointments, the Baylor Tutoring YouTube channel or any tutoring services we offer, please visit our website www.baylor.edu/tutoring or call our drop in center during open business hours. M-Th 9am-8pm on class days 254-710-4135.

Answers:

1. B.
   a. $q^2 = 0.001 \rightarrow q = 0.0316$; $p = 0.968 \rightarrow 2pq = 0.0612 \times 100\% = 6.12\%$

2. C.
3. C.
4. False
   a. Only mutation, population size (genetic drift), natural selection and migration affect allele frequencies

5. C.
6. C.

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