BIO 184 - PAL Problem Set Lecture 6 (Brooker Chapter 18)
Mutations

Section A. Types of mutations

Define and give an example the following terms:

allele;

phenotype;

genotype;

Define and give an example of the following terms:

Deleterious mutation;

Neutral mutation;

Beneficial mutation;

What determines if a mutation is deleterious, neutral, or beneficial?

What are pleiotropic effects?

Define, give an example, and distinguish the difference between the following terms:

Genome mutation;

Chromosome mutations;

Gene mutations;

Define, give an example, and distinguish the difference between the following terms:

Transition mutation;

Transversion mutation;

Define, draw an example, and describe what effect each has on a protein sequence:

Silent mutation;

Missense mutation;

Nonsense mutation;

Synonymous mutation;

Nonsynonymous mutation;

Frameshift mutation;

Rank each of the above mutation types on the likelihood they will cause the encoded polypeptide to be functional or nonfunctional.

Which mutation(s) above would likely result in a truncated polypeptide?

What are insertion, duplication, deletion, and translocation mutations?

If they arise within a gene, when would they cause a frameshift?

Section B. Beneficial and deleterious mutations

How can a mutation be both deleterious and beneficial?

What role does the environment play in determining this?

Why does the frequency of malaria resistance and the Sickle Cell Anemia mutation overlap?

Where do populations carry this mutation at a high frequency?

What is the selective advantage of carrying the sickle cell mutation?

How can a mutation be beneficial?

What mutation causes resistance to HIV?

What populations is this mutation most frequently found?

What mutation causes Cystic Fibrosis?

Do you need one or two mutations in this gene to have Cystic Fibrosis?

What populations carry this mutation most frequent?

Why is this mutation frequent in this population and not others?

How are some mutations within this gene considered to be neutral?

What mutations cause Becker and Duchenne's muscular dystrophy?

Why would a large deletion cause a less severe disease than a small deletion?

Section C. Mutations and gene expression

What mutations outside of the "gene" still effects gene expression? Give examples.

When would such mutations cause high or low gene expression?

What would be the result of a mutation occurring in the following locations? Describe what effect each has on gene expression and the encoded protein sequence.

Promoter mutation:

Splice site mutation:

Mutation within the untranslated region of a gene transcript:

Section D. Germ-line and somatic

What is the difference between germ-line and somatic mutations?

Is a birth mark a germ-line or somatic mutation?

What is genetic mosaicism?

Section E. Revertant mutations

What is the difference between a reverse and forward mutation?

What type of mutation restores a mutant to the wild-type phenotype by mutating a secondary gene?

Section F. Mutations and the environment

What type of mutation is characterized by a change in the DNA sequence that changes the resulting protein sequence and affects the phenotype only in very hot temperatures?

Do humans carry conditional alleles? If so, provide some examples.

During evolution, what type of mutations accumulate over long time periods that are within genes when the encoded amino acid sequences remain conserved between different species?

What do the results of the Lederberg experiments suggest about how mutations arise in respect to the environmental conditions?

How do they help explain how evolution works?

How does the environment affect the genetics of a population?

A researcher plates out a diluted *E. coli* culture onto an agar plate that contains media but no antibiotics. 125 colonies grow on the plate. The researcher then "replica plates" them onto two plates containing agar, media, and kanamycin. If the *physiological adaptation (adaptive mutation)* theory is correct, what might she see on the two plates after growing them up overnight?

What is the cause of mutations?

What are spontaneous mutations and mutagen-induced mutations?

Can the environment induce mutations? If so, when?

Are mutations random or are they directed to locations that may be beneficial? If the latter, how does DNA polymerase know where to make a beneficial mutation?