AP Biology Review Part 3: Genetics & DNA and Protein Synthesis

3A1- DNA, and in some cases RNA, is the primary source of heritable information
3A3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parents to offspring.
3A4: The inheritance pattern of many traits cannot be explained by simple Mendelian genetics.
3C1: Changes in genotype can result in changes in phenotype.

1. DNA (genetic info is passed down through DNA and RNA)

A. Discovery

1. Avery-MacLeod- Marty- 1944 isolated DNA from Griffith's transformation experiment
2. Hershey-Chase- 1952 elegant experiment with virus and bacteria showing DNA was injected not protein
3. Watson, Crick, Wilkins, and Franklin- 1953 W and C published work showing structure of DNA (used Wilkins and Franklins work to do so)

B. Structure of DNA

1. Deoxyribose nucleic acid
2. Double helix (two twisted stsrands) made of nucleotides (monomers)
3. Nucleotide = phosphate + 5C deoxyribose sugar + nitrogen base
4. Antiparallel strands- one runs 3' to 5' the other runs 5' to 3',sides of phosphates and sugars (backbone), rungs of paired bases with hydrogen bonds in between

a. Purines (adenine,guanine; double rings) pair with Pyrimidines (cytosine, uracil, thymine; single ring)
b. A - T- double H bond
c. C – G- triple H bond

C. Location

1. In eukaryotes DNA is found in nucleus on multiple linear chromosomes (a chromosome IS a strand of DNA with proteins etc. associated).
2. In prokaryotes DNA is not in a nucleus and is usually a single circular chromosome
3. Prokaryotes, viruses, and eukaryotes (yeast) can contain plasmids (small extra-chromosomal DNA that is double stranded DNA)

2. DNA replication

a. Process of making exact copies of DNA (i.e. for mitosis or meiosis)
b. Process is semi conservative (original strand is copied)
c. Steps
    1. Enzyme (helicase) unzip strands by breaking hydrogen bonds
    2. "Spare" nucleotides are added bidirectionally to bond complementarily with use of DNA polymerases (DNA pol)
    3. DNA pol only can add to the 3' to 5' side and new DNA is made in the 5' to 3'direction
    4. Replication bubbles open up and a replication fork is created because bubble is in half and it has one side 3/5 and one 5/3
    5. RNA primers must be laid down to start process (RNA primase makes primers)
    6. Leading strand makes DNA continuously (3/5)
    7. Lagging strand makes DNA discontinuously (5/3), Okazaki fragments
    8. Lagging strand requires enzyme (ligase) to fuse fragments

3. RNA

a. Ribonucleic acid
b. Single stranded, different sugar called ribose, different base called uracil INSTEAD of thymine
c. Base pair rules in RNA, A-U and C-G
d. messenger RNA or mRNA carries information from DNA to the ribosome
e. transfer RNA or tRNA bind amino acids and are used in translation at ribosome
f. ribosomal RNA or rRNA are part of ribosomes that have catalytic function
g. RNAi are molucules that are used for regulation of gene expression (turn on or off)

4. Transcription

a. making mRNA in nucleus
b. enzyme RNA pol reads the DNA in 3' to 5' direction and synthesizes complementary mRNA
c. Ex. 3' to 5' DNA is ATG CAT then the 5' to 3' mRNA made will be UAC GUA
d. Steps
e. TATA Box where RNA pol binds and begins
f. Transcription Factors (proteins that enhance transcription and help RNA pol into correct shape)
g. Elongation (adding of RNA nucleotides- does not stay attached to DNA)
h. Termination, ends when RNA pol reaches a termination sequence

5. mRNA editing

a. introns are excised (cut out)
b. exons are left and spliced together using spliceosomes (snRNP's)
c. add polyA tail to 3'
d. add GTP cap to 5'
e. each 3 are called a codon
f. go to ribosome (free or in RER)

6. Translation

a. mRNA code is read and matched with tRNA (brings amino acids) to construct a polypeptide using the ribosome
b. Ex. mRNA codon is AAA then tRNA anticodon will be UUU and will have a corresponding amino acid for that codon of mRNA
c. Initiation: 5' end of mRNA attaches to small ribosome, tRNA with anticodon UAC attaches to start codon AUG ; large ribosomal subunit binds and tRNA is in P site
d. Elongation: new tRNA enters A site; peptide bond forms when a.a. is transferred from tRNA in P site to A site; translocation occurs and tRNA in A site moves to P
e. Termination: Ribosome encounters stop codon (UAA, UAG, UGA)
f. If in ER then: polypeptide is released into ER, then to Golgi complex, vesicle to cell membrane, then exocytosis (may be given signals for exit/destination)
g. Free ribosomes typically make products for the cell and are not exported

7. Mutations

a. any change of DNA sequence, can be inheritable if it is in egg or sperm
b. point mutations- one nucleotide error; substitutions (i.e. A instead of G)
c. frame shift mutations- one or more bases deleted or inserted
d. silent mutations can occur, i.e. substitution codes for same a.a. or deletion/insertion is of three nucleotides
e. Missense mutation- means that new letter codes for a new amino acid, i.e. sickle cell; can be extensive with frameshift mutations

f. Nonsense mutation- means that a stop codon is coded for too early and results in short polypeptide
    1. Single gene mutations in humans caused by DNA mutations
        a. PKU- recessive; phenylketonuria, enzyme deficiency
        b. Sickle cell- recessive; primarily of African descent, carriers resistant to malaria
        c. Cystic fibrosis- recessive; primarily of European descent, protein in channel misshaped; thick mucus
        d. Huntington's- dominant; nervous disorder at age 40 or so; fatal

8. Heredity

A. <u>Mendel's Laws</u> (remember he laid groundwork for genetics but these rules can all be broken looking at chromosome theory and molecular genetics)
1. Law of Dominance- one allele will be expressed over another (ex. Aa – if big A is purple it will be seen over little a which is white)
2. Law of Segregation- alleles pairs separate from each other during meiosis
3. Law of Independent Assortment- alleles assort independently during meiosis IF they are on separate chromosomes (i.e. AaBb can make gametes AB, Ab, aB or ab)
4. Terms to know
5. dominant
6. recessive
7. genotype
8. phenotype
9. allele
10. homozygous
11. heterozygous
12. testcross

B. <u>Probability,Patterns and Exceptions to Mendel's Rules</u>
1. product rule- multiply chance of one event happening by the chance of another event happening to get the chance of both events occurring together
2. Inheritance patterns
3. autosomal vs. sex-linked (on the X or Y chromosome)
4. monohybrid cross; one trait; 3:1 (Aa x Aa); 1:1 (Aa x aa) or 4:1 (AA x_), (aa x aa)
5. dihybrid cross; 9:3:3:1 genotype (AaBb x AaBb) or test cross 1:1:1:1(AaBb x aabb)
6. Thomas Hunt Morgan- fruit flies, X- linked traits
    a. male- heterozygous XY; Y chromosome is very small in mammals and fruit flies with few genes
    b. female- homozygous XX
    c. not for all living things sometime sex is determined by haploid/diploid or temperature or it is reversed in birds, moths, butterflies (XX is boy)
    d. single gene mutations on X chromosome cause disease such as hemophilia or colorblindness
    e. sex limited traits are dependent on sex of individual like milk production or male patterned baldness
7. incomplete dominance- red X white → pink; both protein product are expressed and blended
8. codominance- red x white → red and white; both protein products are equally expressed ex.AB blood types
9. multiple alleles- blood types- ABO
10. epistasis- one gene affects expression of another
11. linked genes- genes on same chromosome that are inherited together (can be unlinked by crossing over); recombination frequency calculated by recombinants/total; used for chromosome mapping; genes further apart cross over more often

12. gene/environment- phenotypes affect by environment, Siamese cat, flower color with soil pH, seasonal color in arctic animals, human height and weight
13. polygenic- continuous variation, many genes affect one trait- height, color
14. Chloroplasts and mitochondria (come from egg in mammals)are randomly assorted in cell division so they do not follow Mendelian rules.

C. Human Genetics

1. karyotype- 22 pair autosomes & 1 pair sex chromosomes + 46 total chromosomes
2. Chromosomal Mutations (occur during gamete formation- usually denovo)
    2. deletion, inversion, addition of genes as a result of crossing over mistakes, ex. Prader Willi
    3. chromosomal number abnormalities
        a. nondisjunction- failure of chromosomes to separate at anaphase of meiosis
        b. monosomy- 45 chromosomes- Turner's- XO
        c. trisomy- Down's- trisomy 21; Kleinfelters- XXY
    4. amniocentesis- for prenatal diagnosis

Fruit Fly Lab (Not an AP Investigation)

Overview: Three crosses were performed by different groups (one trait, two trait and sexlinked) by allowing F1 fruit flies to mate and then counting F2 generation. Chi Square analysis was used to determine if offspring were as expected.

IV: Fruit flies

DV: Traits in Offspring

Equations: See Chi-Square Analysis

Laws of Probability

If A and B are mutually exclusive, then P (A or B) = P(A) + P(B)

If A and B are independent, then P (A and B) = P(A) x P(B)

DNA

amino acids
anticodon
base-pairing rules
cell differentiation
coding strand
codon
DNA
DNA ligase
DNA polymerase
DNA replication
exons
genetic code
helicase
hydrogen bonding
inducible genes
introns
lagging strand
leading strand
micro RNA (miRNA)
mutation
nucleic acids
nucleotides
Okazaki fragments
protein
replication fork
repressor
RNA (mRNA, rRNA, tRNA
RNAi
start codon/stop codon
template strand
transcription
transcription factors
translation

Genetics

allele
autosome
back cross
cline
codominance
continuous variation
cross
dihybrid cross
discontinuous variation
dominant
F1/F2 Generation
genetic counseling
genomic imprinting
genotype
heterozygous
homozygous
incomplete dominance
independent assortment
lethal allele
linkage
monohybrid cross
multiple alleles
non-disjunction
non-nuclear inheritance
pedigree analyisis
phenotype
phenotypic plasticity
polygenetic inheritance
Punnett square
pure-breeding (aka true-breeding)
recessive
segregation
selfing
sex chromosome
sex-limited traits
sex linked gene
test cross
trait

Questions and Practice

1. How is genetic information organized in the eukaryotic chromosome?

2. How does this organization contribute to both continuity of and variability in the genetic information?

3. How did Mendel's work lay the foundation of modern genetics?

4. What are the principal patterns of inheritance?

5. How do the structures of nucleic acids relate to their functions of information storage and protein synthesis?

6. What are the similarities and differences between prokaryotic and eukaryotic genomes?

7. What is one way genetic information can be altered?

8. What problems can it cause?

9. What are the differences and similarities between protein synthesis in prokaryotes and eukaryotes?

10. Draw a picture to show the relationship between chromosome, DNA, gene, allele, nucleotide, base, and a trait