Ch14-15 Genetics Review

Mendel's Experiment

- Mating true-breeding varieties (purple and white flowered plants) through self-pollination to get the P generation
- P generation is cross-pollinated to get the F1 generation (100% dominant trait)
- F1 generation is self-pollinated to get the F2 generation (75% dominant and 25% recessive trait)

1) Law of segregation
2) Law of independent assortment – only for traits on non-homologous chromosomes

Important Vocab

1) Allele and Locus
2) Dominant and Recessive allele
3) Homozygous and Heterozygous
4) Phenotype and Genotype
5) Monohybrid and Dihybrid

Probability Rules

1) Multiplication rule – probability of two or more independent events occurring together
   Ex) BBRr X bbRr What is the probability of getting Bbrr?
   - Answer: 1 x ¼ = ¼
2) Addition rule – probability of any two or more mutually exclusive events will occur
   Ex) BbRr X bbRr What is the probability of getting a recessive trait for "b" and a dominant trait for "r"?
   - Answer: bbRR ½ x ¼ = 1/8 and bbRr = ½ x ½ = ¼, so added together is 3/8

Inheritance Patterns

Affects of one gene

1) Three types of dominance – complete, incomplete (blending), and co-dominance (one of each)
2) Multiple alleles – many alleles of a singe gene to express one trait
   Ex) Blood type phenotypes: A, B, AB, and O
   Ex) Blood type genotypes: AA, AO, BB, BO, AB, OO (these can also be represented as the following: I^AI^A, I^Ai, I^BI^B, I^Bi, I^AI^B, i i)
3) Pleiotrophy – one gene effects many traits and the organism in many ways
   Ex) Sickle cell anemia (caused from a point mutation) and cystic fibrosis

Affects of more than one gene

1) Epistasis – one gene controls the expression of another gene; pigment gene is controlled by the gene to places the pigment
   Ex) coat color in mice or dog breeds (labs)
2) Polygenic inheritance – additive effect of 2 or more genes: continuum
   Ex) height and skin color
   - The number of alleles involved is always one less than the phenotypic groups created from the cross
     Ex: 2 genes with 2 alleles each (4 total alleles) will result in 5 phenotypic groups OR 3 phenotypic groups are created from 2 alleles from 1 gene

Pedigree Analysis - show the patterns of inheritance – branching diagram to show a trait has passed down over generations

Chromosome Theory of Inheritance

- Mid 1800s, Mendel conducted his experiments
- Late 1800s, scientists worked on the process of mitosis and meiosis
- 1900, scientists begin to see parallels between behavior of chromosomes and Mendel's "factors"
- Theory: Genes have specific loci on chromosomes and chromosomes go through segregation and independent assortment

Morgan's Drosophila Experiments

Wild-type = phenotype most common in the population

Ex) Red eyes = wild-type w+ and white eyes = mutant type w

- Fruit flies have 4 chromosomes including one pair of sex chromosomes (XX = Female and XY = male)
- Discovered that the gene for eye color is on the X chromosome

Sex-linkage – gene that is located on the sex chromosomes (mainly the X chromosome)

- Phenotypes can differ between the males and females in a population
  Ex) Hemophilia, Muscular Dystrophy, Colorblindness, Male Pattern Baldness

- X inactivation in females – one X chromosome becomes inactive
  - Barr body = inactive X chromosome condenses into a compact object along the inside of the nuclear membrane
  - Occurs in developing embryo
  - Which X chromosome will become inactive differs from cell to cell – female has a mixture of active and inactive maternal and paternal X chromosomes
  - If female is a carrier for a recessive disorder, then some cells may display the trait and some won't

Linked Genes – genes on the same chromosome

- Differ from Mendel's law of independent assortment
- The closer the genes are to each other, the more likely they will be inherited together (less crossing over)

Crossed wild-type with mutant

- F1 = all heterozygous for wild-type

Crossed F1 female with mutant male

- F2 = most look like parental generation, but some had a new combination

- Recombinants = phenotype unlike parent

Linkage Mapping

- Greater the distance between two genes, the more points there are between them where crossing over can occur during Meiosis I (like the Sordaria fungus lab)
- Studied done by Morgan's helper – Sturtevant

Problem is that some genes are so far apart that crossing over is almost a certainty

- If recombination frequency is 50%, then it is just like having unlinked independent assortment (random alignment of chromosomes)
- So, genes are physically linked, but genetically inherited as unlinked genes

- Map unit = not an actual physical distance, but it gives the order of the genes on a chromosome based on the recombinant frequency (only Sordaria had to be divided by 2 because only half the spores were the result of crossing over)
    - Other species, the recombinant frequency IS the map unit in centi-Morgans

Normal Exceptions to Mendel's Laws

- 2 to 3 dozen traits (most of the known traits are critical for embryonic development) have shown to be linked to whether trait is given in the sperm or the egg → Genomic Imprinting
- Ex) Paternal linkage = M – normal and F – mutant, then the offspring will be normal, but if M – mutant and F- normal, then the offspring will be mutant

Organelle Genes

- Mitochondria and chloroplast (plastids) have small circular DNA (extranuclear genes) that code for proteins and RNA
- Can reproduce themselves
- Don't display Mendelian inheritance

- Studies of yellow or white patches on leaves of green plants
    - Showed that variation in plastid genes altered pigmentation
    - Plastids came from the cytoplasm in the egg, and the pollen has no control over expression
    - Depending on ratio of wild-type to mutant plastids, the plant will have a variety of spots on its leaves

- Same is true for mitochondrial genes
    - Mutations in mitochondrial DNA have been linked to Alzheimer's, heart disease, and aging

Abnormal Chromosome Number

1) Nondisjunction – Down's Syndrome, Klinefelter, Turner (monosomic)
   Aneuploidy – abnormal number of chromosomes
   a. Trisomic – 2n + 1
   b. Monosomic – 2n – 1
2) Polyploidy – more than 2 complete chromosome sets
   a. Triploidy – 3n
   b. Tetraploidy – 4n

- Common in plants, can occur in fish or amphibians
- Likely to appear normal compared to aneuploidy

Chromosomes Mutations

1) Deletion
2) Duplication
3) Inversion
4) Translocation

Disorders

1) Recessive inheritance – cystic fibrosis, sickle cell, consanguineous matings (incest)
2) Dominant inheritance – achondroplasia (form of dwarfism), Huntington's disease
    a. People with a dominant lethal disorder more than likely will not survive to reproduce, but people with a recessive lethal disorder can be a carrier and pass on the allele to offspring
3) Multifactorial inheritance – has a genetic and environmental component
    Ex) heart disease, diabetes, cancer, alcoholism, bipolar

Genetic Testing / Counseling

- Use probability to determine the likelihood of an offspring inheriting an abnormal trait or disorder from the parents
- Parents can then decide if they want to have children and risk passing on a genetic disorder5
- There are tests to determine carriers, of the fetus, and of newborns