

Genetics – Exam 1 Review

I. Chromosomes

- a. Based on the centromere, know what type of chromosome it is (chart)
 - i. Metacentric
 - ii. Submetacentric
 - iii. Acrocentric (typically have ribosomal genes)
 - iv. Telocentric (mouse chromosomes)
- b. Chromosome content within the cell
 - i. $1n$ = haploid (gametes, spores, etc)
 - ii. $2n$ = diploid (somatic cells)
- c. Homologous Chromosomes
 - i. Gene loci are in the same order in both
 - ii. One comes from father, one from mother
- d. Sister Chromatids
 - i. Arms in each chromosome

II. Terminology

- a. Genotype – set of alleles that are carried in an individual
- b. Phenotype – outward appearance, what is expressed
 - i. Can't necessarily tell you the genotype
- c. Allele – variant forms of a gene
 - i. i.e. white vs. red eye in drosophila
- d. Homozygous – both alleles are the same
 - i. i.e. BB
- e. Heterozygous – both alleles are different
 - i. i.e. Bb

III. Cell Cycle / Chromosome Behavior

- a. $G1 \rightarrow S \text{ Phase} \rightarrow G2 \rightarrow M \text{ Phase}$
 - i. $G1$ = 1 sister chromatid
 - ii. $G2$ = 2 sister chromatids
- b. Mitosis – somatic cell division
 - i. $2n \rightarrow 2n$
 - ii. Growth and Development of an organism
 - iii. If a cell is terminally differentiated, it enters $G0$ (no growth)
- c. Meiosis – gametic cell division
 - i. $2n \rightarrow 1n$
 - ii. occurs in the ovum / spermatozoa
 - iii. Go over stages for generating ovum / spermatozoa

- d. Prophase I
 - i. Crossing over occurs – genetic exchange between homologous chromosomes
 - ii. Terms:
 - 1. ***Tetrad** – homologous chromosomes
 - 2. ***Synapsed**
 - 3. **Chiasma** – where the crossing over actually occurs
 - 4. **Non-autonomous** – do not act independently
- e. Be able to label different stages of meiosis
 - i. Prophase I
 - ii. Metaphase I
 - iii. Anaphase I
 - iv. Telophase I
 - v. Prophase II
 - vi. Metaphase II
 - vii. Anaphase II
 - viii. Telophase II

IV. Deviations from Mendel

- a. **Incomplete dominance** – the combination of heterozygous alleles are not dominant over one another
 - i. Yields intermediate phenotype (hybrid of the two)
- b. **Co-dominance** – distinct phenotypes
 - i. MN Blood Groups
 - ii. AB blood type
- c. **Epistasis** - Expression of one gene can mask or modify the expression of a second gene.
- d. Multiple Genes contribute to a phenotype
 - i. $A \rightarrow I \rightarrow B \rightarrow II \rightarrow C$
- e. Complementation Analysis
 - i. If you have a mutation in I, then you cross that organism with one that has a mutation in II.
 - ii. If you crossed it with an organism that a mutation in I, it would not survive
- f. Pleiotropy – mutations in a single gene = multiple phenotypes
- g. Imprinting – parent specific gene expression
 - i. 2 types of syndromes?? Look in book....

V. Sex Chromosomes

- a. Male = XY
- b. Female = XX
- c. Y Chromosome
 - i. Determines maleness
 - ii. PAR = pseudoautosomal regions that pair up X and Y Chromosomes
 - iii. SRY – gene that determines maleness
 - iv. XX males? XY females?
- d. Not all organisms use the SRY gene to determine maleness
 - i. Drosophila: uses a ratio of X autosomes (use book)

VI. Human Pedigrees

- a. Is it autosomal dominant / recessive?
- b. Recessive Clue
 - i. Skips a generation
- c. Dominant Clue
 - i. Occurs in every generation
 - ii. Affected individual will almost always have an affected parent
- d. Autosomal Clue
 - i. Equal appearance in both sexes
- e. Proband = person with disease who went to the doctor...

VII. Mendel

- a. Mendel's Postulates:
 - i. Unit factors exist in pairs
 - ii. Dominance / Recessiveness
 - iii. Segregation
 - iv. Independent Assortment
- b. Monohybrid Cross
 - i. P1 = AA x aa
 - ii. F1 = all Aa
 - iii. Aa x Aa
 - iv. F2 Genotypic Ratio: 1:2:1
 - v. F2 Phenotypic ratio: 3:1
- c. Test Cross – if an organism shows a dominant phenotype, how do you know if it is AA or Aa? → Testcross → cross it with homozygous recessive.