

Reading for Chapter 2-4 (2/17/01)

Chapter 2/ Lecture Outline 3

Chapter 2: Extensions to Mendel: Complexities in Relating Genotype to Phenotype

Outline

I Extensions to Mendel for single-gene inheritance

A. Dominance is not always complete

1. In incomplete dominance, the F₁ hybrid resembles neither purebred parent
 2. In codominance, alternative traits are both visible in the F₁ hybrid
 3. Variations on complete dominance do not negate Mendel's law of segregation
- ##### B. A gene may have more than two alleles mutations are the source of new alleles
- ##### C. One gene may contribute to several visible characteristics. Some alleles may cause lethality
- ##### D. A comprehensive example: sickle cell anemia illustrates many extensions to Mendel's analysis of single-gene inheritance

1. Multiple alleles

3. Pleiotropy

4. Recessive lethality

5. Different dominance relations

II Extensions to Mendel for multifactorial inheritance

A. Two genes can interact to determine one trait

1. Novel phenotypes can emerge from the combined action of the alleles of two genes

2. In epistasis, one gene's alleles mask the effects of another gene's alleles

3. For some traits, homozygosity for a mutant allele at one of two or more genes produces the phenotype

B. Breeding studies help decide how a trait is inherited with humans, pedigree analysis replaces breeding experiments

C. The same genotype does not always produce the same phenotype (pg 57)

1. Phenotype often depends on penetrance and expressivity

2. Modifier genes produce secondary effects on phenotype

3. The environment can affect the phenotypic expression of a genotype

4. Chance can affect penetrance and expressivity

D. Even continuous variation can be explained by extensions to Mendelian analysis

E. The mouse's coat and tail: a comprehensive example of multiple alleles and multifactorial traits

1. Gene 1 \hat{O} agouti or other color patterns

2. Gene 2 \hat{O} black or brown

3. Gene 3 \hat{O} albino or pigmented

4. Gene 4 \hat{O} short or long tail

Essential Concepts

X The F₁ phenotype generated by each pair of alleles defines the dominance relationship between these alleles. One allele is not always completely dominant or completely recessive to another.

With *incomplete dominance*, the F₁ hybrid phenotype resembles neither parent. With codominance, the F₁ hybrid phenotype contains observable components from both parents. Many allele pairs are codominant at the level of protein production.

X One gene can contribute to multiple traits; for such a gene, the dominance relation between any two alleles can vary according to the particular phenotype under consideration.

Chapter 3: The Chromosome Theory of Inheritance

Outline

I. Chromosomes contain the genetic material

A. Evidence that genes reside in the nucleus

B. Evidence that genes reside in chromosomes

~~1. One chromosome pair determines an individual's sex~~

2. At fertilization, haploid gametes produce diploid zygotes

3. The number and shape of chromosomes vary from species to species

~~4. There is variation between species in how chromosomes determine an individual's sex~~

II. Mitosis ensures that every cell in an organism carries the same chromosomes

A. During interphase, cells grow and replicate their chromosomes

B. During mitosis (M phase), sister chromatids separate and are apportioned to different daughter nuclei

1. Prophase: chromosomes condense

2. Prometaphase: the spindle forms

3. Metaphase: chromosomes align at the cell's equator

4. Anaphase: sister chromatids move to opposite spindle poles

5. Telophase: identical sets of chromosomes are enclosed in two nuclei

6. Cytokinesis: the cytoplasm divides

C. Regulatory checkpoints ensure correct chromosome separation during mitosis

III Meiosis produces haploid germ cells, or gametes

A. Meiosis consists of one round of chromosome replication but two rounds of nuclear division

B. During meiosis I, homologous chromosomes pair, exchange parts, and then segregate from each other

1. Prophase I is the longest, most complex phase of meiosis: leptotene, zygotene, pachytene, diplotene, diakinesis

2. Metaphase I: homologous chromosomes attach to spindle fibers growing from opposite poles

3. Anaphase I: homologous chromosomes move to opposite poles of the spindle

4. Telophase I: nuclear envelopes reform

C. During meiosis II, sister chromatids separate to produce haploid gametes

1. Prophase II is similar to the prophase of mitosis

2. Metaphase II: chromosomes align at the metaphase plate

3. Anaphase II: sister chromatids separate to opposite spindle poles

4. Telophase II followed by cytokinesis creates four haploid daughter cells

C. Meiosis contributes to genetic diversity

D. Meiosis and mitosis: a comparison

IV. Gametogenesis requires both mitotic and meiotic divisions

~~A. Egg formation in humans: asymmetrical meiotic divisions produce one large ovum~~

~~B. Spermatogenesis in humans: symmetrical meiotic divisions produce four sperm~~

~~V. Validation of the chromosome theory~~

~~A. The chromosome theory correlates Mendel's laws with chromosome behavior during meiosis~~

~~B. Specific traits are transmitted with specific chromosomes~~

Chapter 4: Linkage, Recombination, and the Mapping of Genes on Chromosomes

Outline

I. Gene linkage and recombination

A. Some genes on the same chromosome assort together more often than not

1. In dihybrid crosses, departures from a 1:1:1:1 ratio of F1 gametes indicates that the two genes are on the same chromosome
2. A preponderance of parental genotypes in the F2 generation defines linkage
3. Percentages of parental and recombinant classes vary with the gene pair
4. Autosomal traits also exhibit linkage
5. The chi square test pinpoints the probability that observed percentages are evidence for linkage
6. Applying the chi square test

B. Recombination results when crossing over during meiosis separates linked genes

1. Reciprocal exchanges between homologous chromosomes are the physical basis of recombination
2. Through the light microscope: chiasmata mark the sites of recombination
3. Recombination frequencies for pairs of genes reflect the distances between them along a chromosome
4. Experimental recombination frequencies between two genes are never greater than 50%

II. Mapping: locating genes along a chromosome

A. Two-point crosses: comparisons help establish relative gene positions

1. Recombination mapping supports the idea that genes are arranged in a line along a chromosome
2. Two-point crosses have their limitations

B. Three-point crosses: a faster, more accurate way to map genes

1. Three-point crosses allow correction for double crossovers
2. Interference: The number of double crossovers may be less than expected
3. The arrangement of alleles in double recombinants indicates the relative order of three gene
4. Comprehensive example: Using a three-point cross to reanalyze Sturtevant's map of the *Drosophila X* chromosome

C. How close is the correlation between a genetic map and physical reality?

D. Multiple factor crosses help establish linkage groups by inference

E. Tetrad analysis in fungi: a powerful tool for mapping and for understanding the mechanisms of recombination

1. Tetrads can be characterized by the number of parental and recombinant spores they contain
2. When PD = NPD, two genes are unlinked
3. When PDs significantly outnumber NPDs, it is a sign of linkage
4. Calculating the recombination frequency
5. Tetrad analysis confirms that recombination occurs at the four-strand stage
6. Tetrad analysis also demonstrates that recombination is usually reciprocal
7. Ordered tetrads help locate genes in relation to the centromere
8. A numerical example of ordered tetrad analysis