

AP
Biology
Interactive
Student
Study
Guide

North Salem University

MISSION: *Engage students to continuously learn, question, define and solve problems through critical and creative thinking.*

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It was not until 1900 that biology finally caught up with Gregor Mendel. During the intervening years, biology had grown more experimental and quantitative and thus more receptive to Mendel and his work. Nevertheless, many biologist remained incredulous about Mendel's Law of Segregation and Independent Assortment until evidence had mounted that these principles of heredity had a physical basis in the behavior of chromosomes. Mendel's hereditary factors (genes) are located at specific locations (loci) on chromosomes. This chapter integrates and extends what we have learned in the last three chapters by describing the chromosomal basis for the transmission of genes from parent to offspring, along with some important exceptions and errors that may occur along the way.

If you have any problems – please sign up for extra help after school.

Genetics:
**Ch.15 – Chromosomal Basis
of Inheritance**

Chapter 15: Chromosome Basis of Inheritance

OBJECTIVES:

Relating Mendelism to Chromosomes

- ___1. Explain how the observations of cytologists and geneticists provided the basis for the chromosome theory of inheritance.
- ___2. Describe the contributions that Walter Sutton and Thomas Hunt Morgan made to current understanding of chromosomal inheritance.
- ___3. Explain why *Drosophila melanogaster* is a good experimental organism.
- ___4. Define and compare linked genes and sex-linked genes. Explain why the inheritance of linked genes is different and does not follow the laws of independent assortment.
- ___5. Distinguish between parental and recombinant phenotypes.
- ___6. Explain why linked genes do not assort independently.
- ___7. Explain how crossing over can unlink genes.
- ___8. Explain how Sturtevant created linkage maps.
- ___9. Define a map unit.
- ___10. Explain why Mendel did not find linkage between seed color and flower color.
- ___11. Explain how genetic maps are constructed for genes located far apart on a chromosome.
- ___12. Explain the impact of multiple crossovers between loci.
- ___13. Explain what additional information cytological maps provide over linkage maps.

Sex Chromosomes

- ___14. Explain how sex is genetically determined in humans and the significance of the SRY gene.
- ___15. Explain why sex-linked diseases are more common in human males.
- ___16. Describe the inheritance patterns and symptoms of color blindness, hemophilia and ALD.
- ___17. Describe the process of X inactivation in female mammals. Explain how this phenomenon produces the tortoiseshell coloration in cats.

Errors and Exceptions in Chromosomal Inheritance

- ___18. Distinguish among nondisjunction, aneuploidy, trisomy, triploidy, and polyploidy. Explain how these major chromosomal changes occur and describe the consequences.
- ___19. Distinguish among deletions, duplications, inversions, and translocations.
- ___20. Describe the type of chromosomal alterations implicated in the following human disorders: Down syndrome, Klinefelter's syndrome and Turner's syndrome
- ___21. Define genomic imprinting and provide evidence to support this model.
- ___22. Give some exceptions to the chromosome theory of inheritance. Explain why extranuclear genes are not inherited in a Mendelian fashion and how they can contribute to disease.

KEY TERMS:

aneuploidy	Barr body	cytological maps	deletion
Down syndrome	duplication	fragile X syndrome	genetic map
genetic recombination	genomic imprinting	hemophilia	inversion
linkage map	linked gene	map units	nondisjunction
parental types	polyploidy	recombinant	sex-linked gene
translocation	trisomic	wild type	

WORD ROOTS:

- aneu-** = without (*aneuploidy*: a chromosomal aberration in which certain chromosomes are present in extra copies or are deficient in number)
- cyto-** = cell (*cytological maps*: charts of chromosomes that locate genes with respect to chromosomal features)
- hemo-** = blood (*hemophilia*: a human genetic disease caused by a sex-linked recessive allele, characterized by excessive bleeding following injury)
- mono-** = one (*monosomic*: a chromosomal condition in which a particular cell has only one copy of a chromosome, instead of the normal two; the cell is said to be monosomic for that chromosome)
- non-** = not; **dis-** = separate (*nondisjunction*: an accident of meiosis or mitosis, in which both members of a pair of homologous chromosomes or both sister chromatids fail to move apart properly)
- poly-** = many (*polyploidy*: a chromosomal alteration in which the organism possesses more than two complete chromosome sets)
- re-** = again; **com-** = together; **bin-** = two at a time (*recombinant*: an offspring whose phenotype differs from that of the parents)
- trans-** = across (*translocation*: attachment of a chromosomal fragment to a nonhomologous chromosome)
- tri-** = three; **soma-** = body (*trisomic*: a chromosomal condition in which a particular cell has an extra copy of one chromosome, instead of the normal two; the cell is said to be trisomic for that chromosome)



Thomas Hunt Morgan
(September 25, 1866 – December 4, 1945)

Guided Reading: Chapter 15

1. What is the *chromosome theory of inheritance*?
2. Explain the *law of segregation*. You may want to consult Figure 15.1 in your text for assistance.
3. Explain the *law of independent assortment*. You may want to consult Figure 15.1 in your text for assistance.
4. Thomas Hunt Morgan selected *Drosophila melanogaster* as his experimental organism. List at least three reasons the fruit fly is an excellent subject for genetic studies.

(1)

(2)

(3)



5. The notation for wild type and mutant traits follows some accepted conventions. (See *A note on genetic symbols located on p.272) Notate the following genotypes for a **female** fruit fly using Morgan's symbols:
(1) homozygous for red eyes (2) heterozygous for red eyes (3) homozygous for white eyes

6. When Thomas Hunt Morgan mated a white-eyed male fly with a red-eyed female, he came to the startling conclusion that the trait for eye color was located on the chromosome that determines sex. Using the Punnett Squares below, show this cross. Begin with the parental generation, and go through the F2.



parental generation:

F1 generation

Results:

F2 generation

Results:

7. What unusual result suggested that the eye-color trait is located on the X chromosome?

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8. What is the SRY gene? Where is it found, and what does it do?

9. What is the definition of a sex-linked gene?

10. In humans, how has that term been historically modified?

11. Name and describe three human sex-linked disorders.

(1)

(2)

(3)

12. Try the following problem (Figure 15.7b in your text). A female who carries an allele for colorblindness, but who is not color-blind, mates with a male who has normal color vision. What is the probability that they will have a son who is color-blind? A Punnett square to use for this problem is shown below.

P Generation: _____

Results: _____

13. What is a Barr body? Why do human females show a Barr body in their cells?

14. X inactivation maintains the proper gene dosage. How is the X chromosome inactivated?

15. Why can you say that all calico cats are females?

16. What are linked genes? Do linked genes sort independently?

17. If two genes are linked on the same chromosome, we call this combination the parental combination. These genes will be transmitted as a unit and **will not** sort independently. However, during meiosis, crossing over occurs between homologous chromosomes, and the linked genes can become “unlinked.” In general, **the farther two genes are from each other along the chromosome, the more often they will come “unlinked.”** Genetic recombination is the process during which linked genes become unlinked due to the crossing over that may occur during *Prophase-I* of meiosis. What do geneticists call the offspring that show these new combinations?

18. Review meiosis. When does crossing over occur?

19. Alfred H. Sturtevant, a student of Thomas Hunt Morgan, used assumptions from observations of crossovers to map genes. What is a linkage map?

20. What is a map unit?

Dihybrid Testcross: _____

21. Use the Punnett Square to the right, which is from Figure 15.4. It shows the results of a cross between a fruit fly that is heterozygous for gray body with normal wings, and a fruit fly that has a black body with vestigial wings.

(Dihybrid Testcross)

Because these genes are linked, the results are not what might have been predicted. Show the phenotypes and number of each type of offspring. Indicate which offspring are the recombinants and which are the parental type. Finally, calculate the map distance between the two genes.

Show all your work here.

Expected Results: _____

$$\text{Recombination frequency} = \frac{\text{\# of recombinants}}{\text{total offspring}} \times 100$$

22. What occurs in nondisjunction?

23. Explain each of the following terms:

(1) **aneuploidy** –

(2) **monosomy** –

(3) **trisomy** –

(4) **polyploidy** -

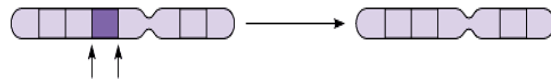
24. Which of the above events results in Down syndrome? What are four characteristics of Down syndrome?

25. For each of the following human aneuploidies, give the sex of the individual as well as any physical manifestation of the syndrome.

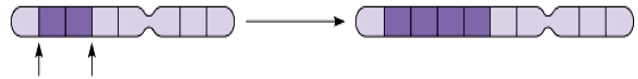
	Sex	Physical Traits
XXY		
XXX		
XO		
XYY		

26. Chromosome structure can be altered in several ways. Label each type of alteration shown in this figure, and explain what occurs.

Deletion –



Duplication –



Inversion –



Translocation -



27. A number of genes will cause a variation in phenotype, depending on whether the gene came from the father or the mother. This variation occurs because of genomic imprinting. Explain genomic imprinting.

28. Although you inherited one chromosome of each pair from your mother and your father, you have inherited a group of genes from your mother only. What genes are these?

29. You should have identified mitochondrial DNA as the correct response to question 28 above. What other organelle has its own genes? These are extranuclear genes.

Chapter 15: Summary of Key Concepts

RELATING MENDELISM TO CHROMOSOMES

- Mendelian inheritance has its physical basis in the behavior of chromosomes during sexual life cycles (p. 269, figure 15.1) In the early 1900s, geneticists showed that chromosomal movements in meiosis account for Mendel's laws.
- Morgan traced a gene to a specific chromosome (p. 271, FIGURES 15.3) Morgan's discovery that the X chromosome in *Drosophila* carries a gene for eye color supported the chromosome theory of inheritance.
- Linked genes tend to be inherited together because they are located on the same chromosome (pp. 272-273, FIGURES 15.4) Each chromosome has hundreds or thousands of genes. Linked genes do not assort independently.
- Independent assortment of chromosomes and crossing over produce genetic recombinants (pp. 273-275, FIGURES 15.5) Recombinant offspring, which exhibit new combinations of traits inherited from two parents, result from events of meiosis and random fertilization. These events include crossing over and independent assortment of chromosomes during the first meiotic division. A recombination frequency under 50% indicates that the genes are linked but that crossing over has occurred. During prophase of meiosis I, paired homologous chromosomes break at corresponding points and switch fragments, creating new combinations of alleles that are then passed on to the gametes.
- Geneticists can use recombination data to map a chromosome's genetic loci (pp. 275-276, FIGURES 15.6) One way to map genes is to deduce their order and a rough indication of the relative distances between them from crossover data. The further apart genes are on a chromosome, the more likely they are to be separated during crossing over. Cytological mapping is a technique that pinpoints the physical locus of a gene by associating a mutant phenotype with a chromosomal defect seen in the microscope.

SEX CHROMOSOMES

- The chromosomal basis of sex varies with the organism (pp. 276-277, FIGURES 15.8) Sex is an inherited phenotypic character usually determined by the presence or absence of special chromosomes; the exact mechanism varies among different species. Humans and other mammals have an X-Y system, as do fruit flies. An XY male gives either an X chromosome or a Y chromosome to the sperm, which combines with an ovum containing an X chromosome from an XX female. The offspring's sex is determined at conception by whether the sperm carries X or Y.
- Sex-linked genes have unique patterns of inheritance (pp. 277-279, FIGURES 15.9) The sex chromosomes carry certain genes for traits that are unrelated to maleness or femaleness. Hemophilia is a sex-linked recessive disorder whose gene is on the X chromosome. In mammalian females, one of the two X chromosomes in each cell is randomly inactivated during early embryonic development.

ERRORS AND EXCEPTIONS IN CHROMOSOMAL INHERITANCE

- Alterations of chromosome number or structure cause some genetic disorders (pp. 279-282, FIGURES 15.11, 15.13) Errors during meiosis can change the number of chromosomes per cell or the structure of individual chromosomes. Such alterations can affect phenotype. Aneuploidy, an abnormal chromosome number, can arise when a normal gamete unites with one containing two copies or no copies of a particular chromosome as a result of nondisjunction during meiosis. Polyploidy, in which there are more than two complete sets of chromosomes, can result from complete nondisjunction during gamete formation. A variety of rearrangements can result from chromosome breakage. A lost fragment leaves one chromosome with a deletion and may produce a duplication, translocation, or inversion by reattaching to another chromosome. Such alterations cause a variety of human disorders, such as Down syndrome (usually due to trisomy of chromosome 21).
- The phenotypic effects of some mammalian genes depend on whether they were inherited from the mother or the father (imprinting) (pp. 282-283, FIGURES 15.15) Individuals imprint certain parts of chromosomes in their gamete-producing cells with either a male or a female "stamp," probably in the form of methylation. This affects the way some genes are expressed in offspring. Genomic imprinting helps explain the inheritance pattern of some hereditary disorders, including fragile X syndrome.
- Extranuclear genes exhibit a non-Mendelian pattern of inheritance (pp. 283-284) Mitochondria and chloroplasts contain some of their own genes. Because the zygote's cytoplasm comes from the ovum, certain features of the offspring's phenotype depend solely on these maternal cytoplasmic genes. Some diseases affecting the nervous and muscular systems are caused by defects in mitochondrial DNA that prevent cells from making enough ATP.

Chapter 15 - Review Questions

Now you should be ready to test your knowledge. This chapter, like all the other chapters in your textbook, has questions at the end of it (*along with the answers*) for you to check your understanding. Like the last chapter, one of the ways to determine your understanding of human genetics is to do as many genetic problems as possible. With that said, complete each of the following questions located on **pp.285-286** in your textbook as neatly as possible. Be sure to show all your work and to check your answers.

As always, make an appointment to see me if you have any questions.

Questions 1, 2, 3, 4, 5, 7, 8, 10, 11, 12, 13, 14, 15 and 16