Chapter 15: Chromosomal Basis of Inheritance

1. What is the chromosome theory of inheritance?

According to the chromosome theory of inheritance, Mendelian genes have specific loci (positions) along chromosomes, and it is the chromosomes that undergo segregation and independent assortment, accounting for inheritance patterns.

2. Explain the law of segregation.

The law of segregation states that two alleles for each gene separate during gamete formation.

3. Explain the law of independent assortment.

The law of independent assortment states that alleles of genes on nonhomologous chromosomes assort independently during gamete formation.

4. List three reasons why the fruit fly is an excellent subject for genetic studies.

Thomas Hunt Morgan selected *Drosophila melanogaster* as his experimental organism because a single mating would produce hundreds of organisms, a new generation could be bred every two weeks, and the fruit fly only has four pairs of chromosomes that are easily distinguishable with a light microscope.

5. Notate the following genotypes for a female fruit fly.

$X^{w+}X^{w+}$ a fly homozygous for red eyes; $X^{w+}X^{w}$ a fly heterozygous for red eyes

$X^{w}X^{w}$ a fly homozygous for white eyes

6. When Thomas Hunt Morgan mated a white-eyed male fly with a red-eyed female, he came to the conclusion that the trait for eye color was located on the chromosome that determines sex. Show this cross.

The parental generation was a cross between a red-eyed female and a white-eyed male. The F_1 generation, all the offspring of the P generation, had red eyes. The F_2 generation showed a ratio of 3 red-eyed flies to 1 white-eyed fly, with no white-eyed females.

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

The white-eye trait only appeared in the male offspring of the F_1 generation.

8. Explain four different methods of sex determination.

In the X-Y system, found in mammals, the sex of an offspring depends on whether the sperm cell contains an X chromosome (female: XX) or a Y chromosome (male: XY). In the X-0 system, found in grasshoppers, cockroaches, and some other insects, there is only one type of sex chromosome, the X. Sex of the offspring is determined by whether the sperm cell contains an X chromosome (female: XX) or no sex chromosome (male: X). In the Z-W system, found in birds, fishes, and some insects, the sex chromosome present in the *egg* (not the sperm) determine the sex of the offspring. The sex chromosomes are designated Z and W; females are ZW and males are ZZ. In the haplo-diploid system found in most species of bees and ants, there are no sex chromosomes. Females develop from fertilized eggs and thus are diploid. Males develop from unfertilized eggs and are haploid; they have no fathers.

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

SRY, which stands for *sex-determining region of Y*, is a gene on the Y chromosome required for the development of testes. In the absence of SRY, the gonads develop into ovaries.

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

A sex-linked gene is located on either sex chromosome. The Y chromosome is passed along virtually intact from father to son.

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

In humans, there are very few Y-linked genes, so most sex-linked genes are on the X chromosome, called X-linked genes.

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

Color blindness is a mild disorder almost always inherited as an X-linked trait. Duchenne muscular dystrophy, caused by an absence of the key muscle protein dystrophin at a specific locus on the X chromosome, is characterized by a progressive weakening of the muscles and loss of coordination. Affected individuals rarely live past their early twenties. Hemophilia is an X-linked recessive disorder defined by the absence of one or more of the proteins required for blood clotting.

13. 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?

If a carrier mates with a male who has normal color vision, there is a 50% chance that each daughter will be a carrier like her mother and a 50% chance that each son will have the disorder.

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

The inactive X in each cell of a female condenses into a compact object called a Barr body, which lies along the inside of the nuclear envelope. Most of the genes of the X chromosome that forms the Barr body are not expressed. In the ovaries, Barr-body chromosomes are reactivated in the cells that give rise to eggs, so every female gamete has an active X.

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

The selection of which X chromosome will form the Barr body occurs randomly and independently in each embryonic cell present at the time of X inactivation. As a consequence, females consist of a mosaic of two types of cells: those with the active X derived from the father and those with the active X derived from the mother. After an X chromosome is inactivated in a particular cell, all mitotic descendants of that cell have the same inactive X. Thus, if a female is heterozygous for a sex-linked trait, about half her cells will express one allele, while the others will express the alternative allele.

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

The tortoiseshell gene is on the X chromosome, and the tortoiseshell phenotype requires the presence of two different alleles, one for orange and one for black fur. Normally, only females can have both alleles, because only they have two X chromosomes.

17. What are linked genes?

Linked genes are located near each other on the same chromosome and tend to be inherited together in genetic crosses. These genes do not sort independently, but rather are transmitted as a unit.

18. What is genetic recombination?

In a parental combination, two genes are linked on the same chromosome. 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. Geneticists call the offspring that show these new combinations recombinant types, or recombinants for short.

19. Review meiosis. When does crossing over occur?

Crossing over occurs during prophase I.

20. What is a linkage map?

Alfred H. Sturtevant, a student of Thomas Hunt Morgan, used assumptions from observations of crossovers to map genes. A linkage map is a genetic map based on recombination frequencies (the percentages of recombinant offspring).

21. What is a map unit?

A map unit is a unit of measurement of the distance between genes. One map unit is equivalent to a 1% recombination frequency.

22. Calculate the map distance between the two genes from Fig. 15.10.

Of the total 2,300 offspring, 391 are recombinants because they do not show the parental phenotypes. Thus, the frequency of recombination is 17%, and the two genes are 17 map units apart.

23. What occurs in nondisjunction?

Nondisjunction is an error in meiosis or mitosis in which the members of a pair of homologous chromosomes do not move apart properly during meiosis I or a pair of sister chromatids fail to separate during meiosis II.

24. Explain each of the following terms.

Aneuploidy is a chromosomal aberration in which the amount of chromosomes is abnormally excessive or deficient. The condition of having a diploid chromosome complement in which one (usually the X chromosome) lacks its homologous partner is called monosomy. Thus, monosomic diploid cells have only one copy of a particular chromosome instead of the normal duplicate. Trisomy refers to diploid cells that have triplicates of a particular chromosome instead of the normal duplicate. Mitosis will subsequently transmit the aneuploidic anomaly to all embryonic cells. In polyploidy, an organism possesses more than two complete chromosome sets in all somatic cells; the specific terms triploidy (3*n*) and tetraploidy (4*n*) indicate three or four chromosomal sets, respectively.

25. Which of these events results in Down syndrome? What are the four characteristics of Down syndrome?

An aneuploid condition, Down syndrome is usually the result of an extra chromosome 21, so that each body cell has a total of 47 chromosomes. Because the cells are trisomic for chromosome 21, Down syndrome is often called trisomy 21. Down syndrome includes characteristic facial features, short stature, correctable heart defects, and developmental delays. Almost all males and about half of females with Down syndrome are sexually underdeveloped and sterile.

26. Describe the following human aneuploidies.

	Sex	**Effect**	**Physical Traits**
XXY	male	Klinefelter syndrome	male sex organs, but testes abnormally small; sterile; some breast enlargement and other female body characteristics common even though extra X inactivated
XYY	male	no well-defined syndrome	normal sexual development; tend to be somewhat taller than average
XXX	female	trisomy X	no unusual physical features other than being slightly taller than average; at risk for learning disabilities; fertile
XO	female	monosomy X: Turner syndrome	phenotypically female; sterile because sex organs do not mature; develop secondary sex characteristics when provided with estrogen replacement therapy; most have normal intelligence

27. Label each type of alteration in chromosome structure and explain what occurs.

A deletion removes a chromosomal segment. A duplication repeats a chromosomal segment. An inversion reverses a segment within a chromosome. A translocation moves a segment from one chromosome to a nonhomologous chromosome. In a reciprocal translocation, the most common type, nonhomologous chromosomes exchange fragments. Less often, a nonreciprocal translocation occurs: a chromosome transfers a fragment but receives none in return.

28. Explain genomic imprinting.

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, a phenomenon occurring during gamete formation that silences a particular allele of certain genes. Because these genes are imprinted differently in sperm and eggs, a zygote expresses only one allele of an imprinted gene, that inherited from either the female or the male parent.

29. What group of genes do you inherit only from your mother?

Maternal inheritance is the rule for mitochondrial genes in most animals and plants, because almost all the mitochondria passed on to a zygote come from the cytoplasm of the egg. Because the parts of the body most susceptible to energy deprivation are the nervous system and the muscles, most mitochondrial diseases primarily affect these systems.

30. What other organelle has its own genes? These are extranuclear genes.

Mitochondria contain small circular DNA molecules that carry a number of genes, as do chloroplasts and other plastids in plants.