

UNIT V STUDY GUIDE FOR BIO 156

CHAPTER 8: THE CELLULAR BASIS OF REPRODUCTION AND INHERITANCE (The Cell Cycle and Meiosis)

Cell Cycle and Mitosis:

1. Explain how the principles of Cell Theory provide a basis for the study of the cellular basis of reproduction and inheritance.

Cell Theory states that all cells come from preexisting cells; they do not spontaneously arise from disorganized matter, etc. Accepting this principle for the origin of cells in the modern world, we can extend this principle to understand principles of reproduction (since new organisms arise from cells produced by parent organisms) and inheritance (since the genetic makeup of new organisms arises from the passing of genetic codes in cells from parent to offspring).

2. Describe how prokaryotes reproduce, and compare / contrast their type of cell division with that of eukaryotes.

Cell division in prokaryotes is referred to as binary fission. They do not have any mitosis since they do not have a nucleus in their cells, and they do not have multiple chromosomes. Prokaryotes replicate their DNA in a manner that is very similar to how eukaryotes do it, and they undergo cell division by growing the plasma membrane and cell walls to divide the parent cell into two daughter cells.

3. Describe the structure of chromosomes in terms of their molecular composition and their overall appearance when viewed through a light microscope.

Chromosomes are made up of chromatin, which is combination of DNA and proteins. The proteins help organize the DNA into a compact structure, and the proteins help to regulate which genes are active or inactive. Chromosomes are not visible during interphase because the chromatin is dispersed. However, during mitosis or meiosis, individual chromosomes become visible as individual, compact structures, especially at metaphase.

4. Name the 3 main stages of the cell cycle, and describe the major events of each stage.

Interphase: protein synthesis for the cell to grow, for basic metabolism, for organelles to grow; some organelles actively divide to produce more of their kind; DNA replicates during the 'S' Phase, creating chromosomes with two sister chromatids

Mitosis: nucleus divides by moving the chromosomes around with spindle fibers (microtubules).

Cytokinesis: the cell and its cytoplasm divide into two new cells, each with its own nucleus, organelles, etc.

5. Differentiate between somatic cells and gametes, and describe the differences between the two in chromosome number. Explain why chromosome number in gametes must be different from chromosome number in somatic cells.

Somatic cells are the cells of the body (epithelial, muscle, nerve, connective, etc. All are diploid.

Gametes are cells for sexual reproduction (sperm and eggs). All are haploid. Gametes must be haploid so that when two gametes combine (fertilization), the resulting cell (zygote) will be diploid, just like the parents. This keeps the chromosome number constant generation after generation.

6. Describe the structure and composition of chromosomes, and describe (and draw) what chromosome structure during early interphase, and at the end of interphase.

Chromosomes are made of chromatin, the combination of DNA and proteins. Each strand of chromatin is a separate chromosome, and each has a single double-stranded DNA molecule. This would describe the structure of chromosomes during early interphase. After the 'S' Phase, DNA has replicated, and so each chromosome now has two sister chromatids (each with genetically identical DNA), and the sister chromatids are joined at the centromere (area where the DNA has NOT replicated yet).

7. Describe the events of the G₁, S, and G₂ phases of interphase.

G₁: Protein synthesis for basic metabolism of cell and cell growth

S: DNA replication; protein synthesis continues

G₂: Protein synthesis in preparation for mitosis and cytokinesis

8. List the 5 stages of mitosis in the correct order, and describe the most significant events of each stage.

Prophase: Chromatin condenses & chromosomes become visible; centrosomes begin migrating to opposite poles and spindle fibers begin to form

Prometaphase: Nuclear envelope is dismantled so that spindle fibers can attach to centromeres of chromosomes.

Metaphase: Chromosomes line up in along the metaphase plate

Anaphase: DNA replicates in the centromere position, and sister chromatids separate, moving newly separate chromosomes to opposite poles

Telophase: Spindle fibers no longer attached to chromosomes, there are now two separate but identical clusters of chromosomes at opposite poles of the cell. A new nuclear envelope develops around each group of chromosomes.

9. List and describe several functions of cell division with mitosis.

Reproduction (asexual), growth, development, cell replacement, injury repair

10. Describe how anchorage, cell density, and chemical growth factors affect the cell division.

Which checkpoint seems to be the most important? Which cells of the human body stop dividing, and at what point in the cell cycle do they stop?

11. Describe some characteristics of cancer cells. Differentiate between **benign** and **malignant** tumors, define **metastasis**, and differentiate between **carcinomas**, **sarcomas**, **leukemias**, and **lymphomas**. Finally, describe two types of cancer treatments.

MEIOSIS and SEXUAL LIFE CYCLES:

12. Differentiate between asexual and sexual reproduction, and the type of cell division used in each strategy.

Asexual produces genetically identical clones; there are not both males and females for reproduction. Mitosis is used (chromosome number doesn't change).

Sexual reproduction produces genetically different offspring, because a sperm with one set of chromosomes fuses with an egg with its own set of chromosomes. In order to produce haploid cells, the process of meiosis is used.

13. Explain why meiosis must occur in sexually reproducing organisms.

Since two cells combine to form a new organism, each of the two cells (sperm and egg) must have 1/2 the number of chromosomes as compared to the somatic (body) cells.

14. Describe and draw the sexual life cycle of animals. Label your diagram, indicating where mitosis and meiosis occurs, where cells are diploid vs. haploid, etc.

See Figure 8.13.

15. List the stages of Meiosis I and Meiosis II, and describe the significant events at each stage. Identify the point at which cells become haploid. Identify the way by which diploid cells produce haploid cells (i.e., what stage(s) of meiosis are crucial for producing haploid cells?).

Prophase I: Similar to mitosis EXCEPT homologous chromosomes pair up (synapsis) and undergo crossing over (segments of nonsister chromatids are exchanged).

Metaphase I: Homologous chromosomes line up opposite one another during the process of Independent Assortment.

Anaphase I: Homologues separate, moving to opposite poles

Telophase I: New nuclear envelopes form around each group of chromosomes

Cytokinesis I: Cell divides, producing two HAPLOID cells. But sister chromatids are still attached!!

Prophase II – Telophase II: Events are identical to mitosis. Only difference is that cells are haploid; chromosomes are NOT in pairs anymore.

16. Explain how independent assortment occurs, and name the stages of meiosis during which this event happens. Explain the “random fertilization of gametes” that occurs when sexually reproducing organisms mate, and explain how both independent assortment and random fertilization produce variations among organisms.

Homologues line up (assort) RANDOMLY on either side of the metaphase plate. Each pair assort independently from all other pairs (the way that one pair of homologues lines up does not influence or affect the way other pairs line up). The number of possible chromosome combinations = 2^n , where n is the haploid number.

Occurs during Metaphase I.

When sperm fertilizes egg, there is no predetermined combination. So now the total number of chromosome combinations is the product of the total number of chromosome combinations produced by both males and females.

17. Explain how nondisjunction occurs, and describe the possible consequences of nondisjunction to chromosome number when gametes form. Define aneuploidy.

Either homologues do not separate properly during Meiosis I, or sister chromatids do not separate properly during Meiosis II. If nondisjunction occurs during Meiosis I, all of the gametes will be aneuploid (have the wrong number of chromosomes), whereas if nondisjunction occurs during Meiosis II, only 2/4 of the gametes will be aneuploid.

18. Describe various human disorders due to chromosomal alterations, including Down's Syndrome, Klinefelter's Syndrome, Turner's Syndrome, etc. Describe the chromosomal basis of these disorders, and describe some of the symptoms for each. Explain why most aneuploid conditions are lethal.

Most aneuploid conditions are lethal because development of the embryo or fetus does not occur normally; having an extra or missing chromosome means that too much or too little of certain types of proteins are made, and this disrupts metabolic pathways, etc., for development.

For specific examples with symptoms, etc., refer to Sections 8.20 and 8.22.

19. Describe what a karyotype is, and how it is prepared and interpreted.

A karyotype displays the number and type of chromosomes possessed by an organism. *A sample of cells is taken from an individual, the cells are cultured in the lab to produce many of them, then they are “arrested” during metaphase, when the chromosomes are most distinct. They are then sorted and counted. See Section 8.19*

Problems (for both Mitosis and Meiosis):

1. A parasitic nematode worm has a diploid number of 6 ($2n=6$) in its somatic cells. Show the arrangement of the chromosomes as they would appear during the following stages:

Drawings not provided here.

2. A cell begins interphase with 12 chromosomes.

(A) After the “S” phase of interphase, how many chromosomes are there? How many sister chromatids?

12 chromosomes, 24 sister chromatids

(B) During anaphase of mitosis, how many chromosomes are there? How many sister chromatids?

24 chromosomes, 0 sister chromatids

(C) During anaphase of Meiosis I, how many chromosomes are there per cell?

12

(D) During anaphase of Meiosis II, how many chromosomes are there per cell?

12. There were 6 in the beginning of Meiosis two (cells are now haploid), but because sister chromatids have separated, there are temporarily 12.

(E) After Meiosis II is complete, and cytokinesis occurs, how many chromosomes are there per cell?

6. Cells are now haploid, having a single copy of every gene.

3. A cell has a diploid number of 8 ($2n = 8$). If nondisjunction of one chromosome pair occurs during Meiosis I, how many gametes would be aneuploid by the end of meiosis? What would the chromosome numbers be in the resulting cells? Draw the outcome (what the cells would look like):

All four gametes would be aneuploid. Two of them would have 5 chromosomes each, and two of them would have 3 chromosomes each.

4. A cell has a diploid number of 8 ($2n = 8$). If Meiosis I occurs normally, but nondisjunction of one chromosome pair in one of the cells occurs during Meiosis II, what proportion of the gametes would be aneuploid? Draw the outcome (what the cells would look like).

Two of the four gametes would be normal (4 chromosomes each). Between the other two gametes, one would have 5 chromosomes, and the other 3.

5. A karyotype of a developing fetus shows that only one sex chromosome, the “X” chromosome, is present. How would you diagnose this person (i.e., what syndrome does this person have)? What symptoms are expected during this person’s lifetime?

Turner Syndrom. Short stature, web of skin extending from neck to shoulder; sterile (abnormal ovaries), reduced sexual characteristics; normal intelligence.

6. A mule is the offspring of a horse and a donkey (two different species). A donkey sperm contains 31 chromosomes, and a horse egg cell 32 chromosomes, so the zygote contains a total of 63 chromosomes. The zygote develops normally, producing the mule. However, a mule is sterile; meiosis cannot occur normally in testes or ovaries. Explain why mitosis is normal in mule cells, but meiosis fails to produce functional gametes in a mule.

All 63 chromosomes would line up along the metaphase plate during mitosis, sister chromatids would separate at anaphase, and daughter cells would be produced with all 63 chromosomes. This process would occur to make up all of the somatic cells of the mule. However, during metaphase I of meiosis, homologous chromosomes would not be able to pair up normally since the two sets of chromosomes came from different species, a donkey and a horse. Since homologues would not be able to align, cross over, and segregate properly, normal gametes cannot be made, and the mule is sterile.

CHAPTER 9: PATTERNS OF INHERITANCE (Mendel and the Idea of the Gene)

Objectives:

1. Define the following: genetics, monohybrid cross, gene, allele, dominant, recessive, homozygous, heterozygous, Punnett Square, genotype, phenotype, principle of segregation, complete dominance, incomplete dominance, codominance, pleiotropy, polygenic inheritance, autosomes, sex chromosomes, sex-linked genes
2. When Gregor Mendel crossed (mated) pea plants that were taken from different strains (e.g., purple vs. white flowers, tall vs. short plants), ALL offspring expressed the same phenotype. When these offspring were allowed to self-fertilize, the phenotype ratios for the offspring always were 3:1. Explain these results. Use the terms homozygous, heterozygous, dominant, recessive, allele, and gamete in your response.

Starting with the parent plants, each was “pure” for the characteristic being tested. Since every organism must have two copies of every gene, when gametes form, the gametes must only get one copy of every gene. Then, when reproduction occurs and a sperm fertilizes an egg, the resulting zygote has two copies of every gene again. In the case of the flower color, one parent only produced gametes with the dominant ‘P’ allele, and the other parent gametes with the recessive ‘p’ allele; each of the parents was homozygous for different alleles. All the offspring ended up with two alleles (two copies of the gene), and all were heterozygous. Since they all had a copy of the dominant allele, all had the same color (purple). Next, the F1 offspring went through meiosis; they produced 50% of their gametes with the ‘P’ allele and 50% with the ‘p’ allele. When they reproduced, the gametes fertilized randomly and therefore produced 1 PP: 2 Pp: 1 pp, which gave 3 purple to 1 white. The allele that is not expressed in the heterozygote is called the “recessive” allele, and the one that is expressed is “dominant”.
3. Using a Punnett Square, be able to show and predict the genotype and phenotype ratios of offspring when the parents’ genotypes are given. See example problems below.

Yea, be able to do that!!
4. Explain why genetic disorders caused by a dominant allele are much less common than genetic disorders caused by recessive alleles. Give at least one example (see page 165).

If a genetic disorder is caused by a dominant allele, then both homozygous dominant individuals AND heterozygous individuals will show the disease. When that happens, a greater proportion of organisms with that allele will die, and the dominant allele will become less common. But if the disease is caused by a recessive allele, then heterozygotes will NOT show the disease (only homozygous recessive individuals), and so the allele can be more common without showing any ill effect.

5. Compare and contrast complete dominance, incomplete dominance, and codominance.

Complete: heterozygotes show the phenotype of the dominant allele.

Incomplete: heterozygotes have a phenotype that appears intermediate or blended between the two homozygous phenotypes.

Codominance: Both phenotypes are observed.

See examples from lecture

6. Explain how sickle disease serves as an example of pleiotropy.

Pleiotropy occurs when a single gene affects multiple phenotypes. If someone is homozygous for sickle cell disease, then not only is their hemoglobin protein in their red blood cells abnormal, but the blood cells themselves have an abnormal shape, and this leads to all sorts of phenotypic problems with kidneys, spleen, brain, etc.

7. Explain how polygenic inheritance accounts for the great variations in human skin color, eye color, and height.

Polygenic inheritance occurs when MULTIPLE genes all affect a single phenotype. These genes could be on the same pair of homologous chromosomes (in which case the genes are said to be "linked", or they could be on different pairs of chromosomes. In either case, there is a greater range of phenotypes observed because there is a wider range of combinations of alleles that can occur to give the phenotype. For example, your textbook shows what happens with eye color in humans; a great range of eye colors occurs because of the wide range of combinations of dominant and recessive alleles -- the more dominant alleles you inherit from those genes, the more dark pigment your cells produce, and the darker your eyes.

8. Define **linked genes**.

Genes that are found on the same pair of chromosomes. Obviously, chromosomes can have hundreds to thousands of genes on them, so all the genes that are found on the same chromosome are linked. The closer the genes are to each other on the chromosome, the more likely the genes will be inherited together (we didn't get into this topic much).

9. Differentiate between autosomes and sex chromosomes. Define sex linked gene and give some examples. Explain why sex-linked disorders mostly affect males rather than females. Use Punnett Squares to solve genetic problems involving sex-linked genes.

The first 22 pairs of chromosomes in our cells are the autosomes, and they are the same between males and females. The 23rd pair is the sex chromosomes, and not only do they determine our gender in mammals (e.g., humans), but there are many other genes on those chromosomes (especially the "X" chromosome) that have nothing to do with sex / gender! Those are "sex linked genes". Examples include hemophilia and red-green color blindness. Because the "X" usually carries these genes, males have whatever genotype and phenotype that their mother gives them; the father has no influence, since he gave the male a "Y" chromosome. This leads to slightly different ratios of genotypes and phenotypes when compared to the inheritance of autosomes.

- Describe how environmental factors may produce varying phenotypes. Give at least two examples.
- Explain how amniocentesis and chorionic villus sampling may be used to check for genetic disorders. Explain how ultrasound and fetoscopy may be used to check for genetic disorders. Which procedure carries the highest risk? The lowest risk?

Problems:

- Tomato stems can be purple (dominant allele) or green (recessive allele). Predict the genotype and phenotype ratios of offspring if you cross a homozygous recessive and a heterozygous individual.

50% all around!!

- Tall pea plants (T) are dominant over dwarf pea plants (t). What would be the phenotype of a heterozygous plant?

Tt

If two heterozygous plants were crossed and produced a large number of offspring, 30 of which were dwarf, approximately how many should be tall?

90. If you do the Punnett square, 1/4 of the offspring have the recessive phenotype (dwarf) and 3/4 dominant phenotype. So if 30 are dwarf, 3 times that many will be tall.

- In Snapdragons, the allele for red flowers (R) is incompletely dominant over the allele for white flowers (r), such that the heterozygotes (Rr) are pink. Show the predicted genotype and phenotype ratios of offspring if you cross a pink flowered with a white flowered plant.

50% Rr (pink), and 50% rr (white)

- Human ABO blood groups exemplify the phenomenon of codominance (and multiple alleles). The “A” and “B” alleles both code for the formation of DIFFERENT glycoprotein receptors on red blood cells, while the “O” allele does not code for the formation of any receptor.

If a male of genotype “AO” mates with a female of type “BO”, what are the expected genotype and phenotype frequencies of offspring?

AB, BO (type B blood), AO (type A), and OO (type O)

- Jane has type A blood and her husband has type B blood. They have 3 children with the following blood types: AB, A, and B. What are the genotypes of the parents and of their children? If Jane has a fourth child with type O blood, should Jim get suspicious of the mailman?

Jane's genotype must be AO, and husband must be BO.

Husband should remain calm and understanding; each of them contributed a gamete with the “O” allele, so they could have a child with type “O” blood.

- Red-green color-blindness is inherited as an X-linked recessive trait. If a color-blind woman marries a man with normal vision, what would be the phenotypes and genotypes of their daughters? Their sons?

Daughters: $X^H X^h$ (both), so daughters have normal vision, but are carriers

Sons: $X^h Y$ (both), so both sons are colorblind. Bummer!

7. Cystic fibrosis is inherited as a simple autosomal recessive allele. Suppose a woman who carries the trait marries a normal man who does not carry it. What percent of their children would be expected to have the disease? To be carriers? To be phenotypically normal?
0% have the disease; 50% are carriers, and all are phenotypically normal.
8. In human beings, the allele for chronic simple glaucoma is dominant over the allele for normal eyes. Suppose a normal woman marries a man with glaucoma whose father was normal. What proportion of their children would you predict will have glaucoma?
50% have glaucoma, since the "Gg" genotype shows glaucoma.

CHAPTER 10: MOLECULAR BIOLOGY OF THE GENE (DNA Replication and Protein Synthesis)

Objectives:

DNA Structure and Replication:

1. Describe how DNA replication makes two identical copies of the double helix. Explain why the process is described as "semi-conservative."
Semi-conservative because each replicate has one old and one new strand
 - (A) What are "origins of replication"?
The places where DNA double helix begins to separate and unwind.
 - (B) What does DNA Polymerase do?
Helps to match and connect complementary nucleotides together to form a new strand
 - (C) What does helicase do?
Unwinds and separates the two strands of DNA, beginning at the origin of replication
 - (D) Why are there "leading" and "lagging" strands of daughter DNA?
Because DNA replication only occurs in the 5' to 3' direction; new nucleotides can only be added to the 3' end of an existing strand.
 - (E) What does DNA ligase do?
Connects together the leading and lagging strands, making continuous strands of DNA
2. Explain the relationship between sister chromatids and the resulting two daughter DNA molecules following DNA replication.
After DNA replicates, the two copies are called sister chromatids, and they are joined together at the centromere.
3. Errors in DNA replication may lead to permanent mutations. Explain how a mutation may be repaired, and when it would become permanent if not repaired.
DNA polymerase checks for errors before the new nucleotide bonds to the previous one. If the mutation is "missed", then it is permanent (passed on to future cells).

4. Describe how a mutation in the DNA can affect the shape and ultimately the function of a protein. Differentiate among insertions, deletions, silent mutations, missense mutations, and nonsense mutations. Which of these are considered “frameshift” mutations? Which “type” of mutation would usually cause the most serious and detrimental effects? Why? What are some causes of mutations?

Mutations can be neutral (not change the polypeptide), harmful (change the size and conformation so that it does not function properly), or beneficial (the resulting protein actually works better than the older version!). Insertions and deletions result when an extra nucleotide is added to a DNA sequence, or one is removed. This will be a frameshift mutation, changing the majority of codons after that (very bad!). Silent mutations, missense mutations, and nonsense mutations all result from a substitution (one nucleotide substituted for another). In silent mutations, the same amino acid is coded for. In missense mutations and different amino acid is substituted (could be neutral, harmful, or beneficial). In nonsense mutation, causes a stop codon to appear way to early!

5. Explain the relevance of mutations to (a) genetic disorders, and (b) evolution

a) Because mutations tend to result in changes in amino acid sequence, and hence polypeptide / protein conformation, most mutations are harmful rather than beneficial or neutral. Harmful mutations code for proteins that are nonfunctional, or proteins that produce negative health effects. These nonfunctional or harmful proteins produce what we refer to as genetic disorders. Some examples include cystic fibrosis, galactosemia, sickle-cell disease, or hypercholesterolemia (see Table 9.9 in text).

b) Mutations alone produce new variations to come about that did not exist before. If a new variation comes about in a population, then the population’s genetic makeup has changed, and that is evolution. More importantly, if the new variation is favorable, or provides a positive benefit for organisms that possess it, the process of natural selection will cause that mutation to become more common in the population over generations, and that is evolution: those organisms that inherit the beneficial mutation will survive and reproduce with greater success than organisms with an alternative allele, leading to the increased frequency or occurrence of the mutation over time. In the short-term, such beneficial mutations with natural selection cause bacteria or viruses to more successfully replicate in human hosts (or other organisms), which is a benefit to the bacteria or virus, but is detrimental to human health. Over the long-term, the accumulation of beneficial mutations leads to large-scale evolutionary changes as demonstrated by the vast diversity of life on Earth today.

Answers to Crossword Puzzle:

Across

- 2. sisterchromatids
- 4. cleavagefurrow
- 6. mitosis
- 10. homologues
- 13. metaphaseone
- 15. alleles
- 18. chromosomes
- 19. aneuploidy
- 20. cellplate
- 23. codominance
- 25. diploid
- 26. interphase
- 27. spindlefibers
- 29. crossingover

Down

- 1. heterozygous
- 3. metaphase
- 5. asexual
- 7. segregation
- 8. phenotype
- 9. deletion
- 11. growth
- 14. gametes
- 16. haploid
- 17. dominant
- 18. cytokinesis
- 21. telophase
- 22. autosomes
- 24. meiosis
- 28. locus