STUDY GUIDE - Ch. 15.4 - Alternations of Chromosome Number or Structure Cause Some Genetic Disorders
- Ch. 15.5 - Some Inheritance Patterns Are Exceptions to Standard Mendelian Inheritance

NAME: _____

- **PHYSICALLY PRINT OUT this PDF and HANDWRITE (with a black or blue pen) your answers directly on this PDF.** *Typed or digitally-written work is **not** accepted. Do **not** answer questions on separate paper.*
- **Importantly, study guides are NOT GROUP PROJECTS!!!** *You, and you alone, are to answer the questions as you **read** your assigned textbook. You are **not** to share answers with other students. You are **not** to copy any answers from any other source, including the internet.*
- **Get in the habit of writing LEGIBLY, neatly, and in a medium-sized font.** *AP essay readers and I will skip grading anything that cannot be easily read so start perfecting your handwriting, and don't write so large you can't add all the relevant details and key elaborations in the space provided.*
- **SCAN physical documents in color and with good resolution. Then, upload your final work as PDFs to Archie.** *Avoid uploading dark, shaded, washed-out, sideways, or upside-down scans of homework. Keep completed physical study guides organized in your biology binder to use as future study and review tools.*
- **READ FOR UNDERSTANDING and not merely to complete an assignment.** ***First**, read a section quickly to get an overview of the topic covered. Then, read it a **second** time slowly, paraphrasing each paragraph **out loud** and analyzing every figure. Finally, read it a **third** time as you answer the study guide questions if assigned and start building your memory. Try to write answers out in your own words, when possible, and try to purposefully and accurately use all new terminology introduced.*

1. What is the **ORIGINAL SOURCE of all new alleles of a gene** (how do new alleles come into existence)?

2. a. Both small and large chromosomal changes can alter an organisms phenotype. What is **nondisjunction** and **when in meiosis** can it occur?

   b. Study Figure 15.13 carefully. Make sure you understand it well. What is the **result IN ALL FOUR POSSIBLE GAMETES of nondisjunction** when it occurs in a tetrad during **meiosis I**?

   b. Study Figure 15.13 carefully. Make sure you understand it well. What is the **result IN ALL FOUR POSSIBLE GAMETES of nondisjunction** when it occurs in a duplicated chromosome during **meiosis II**?

3. a. What does **aneuploidy** mean?

   b. When do we consider an aneuploid zygote **monosomic**?

   c. When do we consider an aneuploid zygote **trisomic**?

d. Around 10-25% of human miscarriages are probably caused by monosomy or trisomy. Why is it a problem if diploid cells are monosomic or trisomic?

4. Remember, as the product of sexual reproduction, the **zygote** is the first cell of the next generation (the resulting offspring). That zygote will divide by **mitosis** to produce two genetically identical daughter cells. These will then also divide by mitosis to form four genetically identical cells, which, in turn, will also divide by mitosis to make eight cells. This process will continue in order to make all cells of the multicellular offspring. *(Remember though that, in these cells, different sets of genes will be activated and deactivated, leading to different RNA and protein products being made, which, in turn, leads these cells to take on different shapes and behaviors in the body. We say that these cells, though their DNA may be the same, therefore, **differentiate** and become **specialized** for performing different functions).*

   a. *Scenario #1:* Nondisjunction occurs in Meiosis I during gamete formation in a male. One of the gametes produced by this male later fertilizes another gamete produced by a female, the latter not having experienced a nondisjunction event. How will this affect the chromosome count of **all** the cells in the offspring's body? Explain.

   b. *Scenario #2:* A gamete in a male formed by meiosis. During meiosis no nondisjunction events occurs. This gamete fertilizes another gamete produced by a female, the latter not having experienced a nondisjunction event either. The zygote develops into an embryo and later a fetus. Late in fetal development, a nondisjunction event happens during mitosis of one of the fetal cells that is specializing into a skin cell. How will this affect the chromosome count of **all** the cells in the offspring's body? Explain.

5. The ABO blood type locus is found on chromosome 9. A father who has type AB blood and a mother who has type O blood have a child with trisomy 9. The child also has type A blood. Can you tell in which parent the nondisjunction occurred that lead to a child with trisomy 9? (Refer back to Figure 14.11 and 15.13 as needed). *Remember, always determine and provide the genotypes of the parents **first** so you know what your genetic options are for offspring.*

   a. What is the genotype of the father?

   b. What is the genotype of the mother?

   c. The child has trisomy 9, so what is the genotype of the child?

   d. Can you tell which parent experienced the nondisjunction event during gamete formation? **Why or why not**?

*(Check your answers to #5.d by going to the **Ch.15.4 Concept Check Question #2** answer in Appendix A)*

6. a. What does **polyploidy** mean?

   b. Describe an example of polyploidy.

   c. Describe how your polyploid organism could have come into existence (evolved).

7. **Errors in meiosis or damaging agents such as radiation can cause breakage of a chromosome, which may lead to four types of changes in gross chromosomal structure.** Name, describe, and explain the following gross (large-scale) chromosomal alterations.

   NAME:

   Description of the error:

   Explanation for how this could occur during meiosis:

   NAME:

   Description of the error:

   Explanation for how this could occur during meiosis:

   NAME:

   Description of the error:

   Explanation for how this could occur during meiosis:

NAME:

Description of the error:

Explanation for how this could occur during meiosis:

8. a. **Gene dosage** - the number of copies of a gene that are actively being expressed - is important to proper development. Some individuals born with aneuploidy exhibit what is called a **syndrome**. What does the word syndrome mean?

   b. What is different in the cells of a person with **Down Syndrome**?

9. a. Why might it be that **aneuploid conditions involving the sex chromosomes (extra X's or extra Y's)** affect gene dosage less than aneuploidies involving autosomes and, thus, **upset the genetic balance less**?

   b. What is different in the cells of a person with **Klinefelter Syndrome**?

   c. What is different in the cells of a person with **Trisomy X**?

   d. What is different in the cells of a person with **Turner Syndrome**?

10. About 5% of individuals with Down syndrome have a chromosomal translocation in which a third copy of chromosome 21 is attached to chromosome 14.

    a. Draw the **homologs during G1** (unduplicated, of course) for chromosome 21 and chromosome 14, **following this translocation mutation**. Be sure to label your four chromosomes in a way that makes it clear what has occurred.

b. If this translocation occurred in a parent's gonad (look up the term if you do not know it still), how could it lead to Down syndrome in a child? Explain what happens at each stage in terms of these two types of chromosomes....

1. How will these four chromosomes look when the gonadal cell leaves G1 and completes S phase? Label your drawing.

2. When meiosis occurs and gametes are formed, what will the chromosomes look like in each of the four types of gametes? Draw and label your gametes.

3. When the gametes you drew and explained above partake in fertilization with a gamete from a second parent, what will the new zygotes G1 nuclei look like? Draw and label your zygotes.

4. Why do you predict that, among the zygotes, one will have the genetics to lead to a child with Down Syndrome?

*(Check your answers to #10.b.4 by going to the **Ch.15.4 Concept Check Question #1** answer in Appendix A)*

11. The ABO blood type locus has been mapped on chromosome 9. A father who has type AB blood and a mother who has type O blood have a child with trisomy 9 and type A blood. Using this information, can you tell which parent the nondisjunction error occurred in? Refer to Figures 14.11 and 15.13. **Explain** your answer carefully and fully.

*(Check your answers to #11 by going to the **Ch.15.4 Concept Check Question #2** answer in Appendix A)*

12. a. Changes to a physical chromosomes (deletions, duplications, translocations, and inversions) can be caused by mistakes that occur during S phase prior to mitosis and meiosis or during Prophase 1 and crossing over of meiosis. How is the **harmful "Philadelphia" chromosome formed** that is responsible for certain cancer formations?

b. The gene that is activated on the Philadelphia chromosome codes for an intracellular tyrosine kinase. Review the discussion of cell cycle control in Concept 12.3, and then explain how the activation of this gene could contribute to the development of cancer. **Explain what changes in the cell and all the downstream consequences of this change - so how this DNA alteration changes the type/amount/behavior of the protein made and how/why this then alters the cell cycle in X or Y way.** *(Think, what are all the differences in a diploid cell that inherited one Philadelphia chromosome 22 and one non-mutated chromosome 22 versus a cell with two non-mutated chromosome 22s?)*

*(Check your answers to #12.b. by going to the **Ch.15.4 Concept Check Question #3** answer in Appendix A)*

13. a. Explain what **extranuclear or cytoplasmic genes** are.

b. **A mitochondrial genetic disease affect an organism's ability to produce ATP.** You should expect to see symptoms, therefore, in tissues that utilize a lot of ATP energy. Which **two body systems are often affected most severely by harmful mitochondrial genetic mutations**?

1. _____ 2. _____

c. Name & briefly describe two **diseases carried in maternal mitochondria**?

1. _____ =

2. _____ =

d. From **which parent (the mother or the father) does a zygote inherit its mitochondria?** *(This is the same for the inheritance of **plastids** in plants like chloroplasts, where photosynthesis occurs in eukaryotes, and amyloplasts, organelles that make and store starch)*

14. **Cells usually have multiple mitochondria**, **each dividing through a process similar to binary fission**. Remember that mitochondria, like free living prokaryotes, have a haploid genome, consisting of **one circular chromosome** made up of one double-stranded DNA molecule. Mitochondrial genes are critical to the energy metabolism of cells since some of these genes are expressed, allowing the mitochondria to produce some of the proteins needed to complete cellular respiration in the mitochondria. *(The other proteins used by the mitochondria are built by free - eukaryotic - ribosomes in the cytoplasm from instructions in nuclear genes).* Still, mitochondrial disorders caused by mutations in these genes are generally not lethal. Why not?

*(Check your answers to #14 by going to the **Ch.15.5 Concept Check Question #3** answer in Appendix A)*