STUDY GUIDE *- Ch. 14.3 - Inheritance Patterns are Often More Complex Than Predicted by Simple Mendelian Genetics - Ch. 14.4 - Many Human Traits Follow Mendelian Patterns of 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.*

Let's recap...

- In Mendel's pea plants, **each character** is determined by **one gene only** and there are **only two possible alleles**, the three possible genotypes - homozygous dominant, heterozygous, and homozygous recessive - resulting in **two trait (or phenotype) variations only**.
- With regards to the alleles of a gene for a particular character, **one was completely dominant and one completely recessive**. Mendel's seven pea plant characters, therefore, following a **"Complete Dominant Inheritance Pattern."** As long as an organism has at least one dominant allele in its genotype, it develops the fully dominant phenotype, a phenotype **equal to that of a homozygous dominant organism.**

Patterns of inheritance are often more complex than those predicted by simple Mendelian genetics, however.

- **Alleles are not always completely dominant or recessive,** many times **a gene may exist as more than just two possible alleles** (though if you are diploid you only inherit two copies of the gene and so have up to two different alleles out of all the possible alleles for that gene), **one gene could produce multiple phenotypes**, **one phenotype could be produced by multiple genes**, and **one gene may even influence the phenotype expression of another gene**! *Ooof, that's a lot of complexity in inheritance patterns!*

Let's look first at inheritance patters in which the character is still controlled by just one gene, but which do not follow the simple Mendelian inheritance pattern of Complete Dominance.

1. a. Contrast **complete dominance** and **incomplete dominance** in terms of the differences noted in the **phenotypes of HETEROZYGOTES** for a gene of a particular character.

   Complete Dominance =

   Incomplete Dominance =

   b. Although, with a gene that follows an incomplete dominant inheritance pattern, the organism that is heterozygous for the gene of a particular character produces a phenotype (a trait or version of a particular character) that looks **intermediate** between the phenotype (trait) produced for that character in an organism that is homozygous

dominant **and** the phenotype (trait) produced in an organism that is homozygous recessive, **incomplete dominance inheritance patterns are NOT evidence for the existence of the "blending hypothesis of inheritance.**" Why not?

c. Review the example of incomplete dominance highlighted in Figure 14.10. **Note that since in incomplete inheritance** (also known as **intermediate inheritance**) **neither of the two alleles is completely dominant,** we do **NOT** usually use one letter only to represent the alleles of the gene (where we represented the dominant allele with the capital version of the letter and the recessive allele with the lower case version of the letter as we do in instances where the gene follows a completely dominant inheritance pattern).

Often, for incomplete/intermediate inheritance, we just use a letter to represent the character this one gene controls (like C for the flower Color), and then use different letters as superscripts to identify the individual alleles that cause the different traits (phenotypes) for the character (like the red allele C^R and the white allele C^W for the flower color gene C).

2. a. How would the phenotype produced differ if the two different alleles in the heterozygous genotype behaved in a **codominant** versus **incompletely dominant** fashion?

Phenotype produced by a gene that follows a Codominant Inheritance Pattern =

Phenotype produced by this same gene if instead it follows an Incomplete Dominant Inheritance Pattern =

b. Describe how the **MN blood group** is an example of **codominance, and NOT incomplete dominance, in humans**.

Why the MN blood group alleles behave in a codominant manner *(think of what phenotype is seen in the heterozygote)* =

Why the MN blood group alleles does not behave in an incomplete dominant manner *(what should you have seen in the phenotype of a heterozygote if these alleles behaved incompletely dominant)* **=**

3. Explain why **the determination of an allele as dominant or recessive, and the inheritance pattern assigned to that gene** *(completely dominant, incompletely dominant, codominant)* **depends on the level** (*organismal, biochemical, & molecular*) **at which we examine the organism's phenotype**?

4. Does having a dominant allele mean that it will be found in greater frequency in the population compared to a recessive allele? **Explain.**

5. a. A rooster with gray feathers is mated with a hen of the same phenotype. Among their offspring, 15 chicks are gray, 6 are black, and 8 are white. What is the simplest full explanation for the inheritance of these colors in chickens? **In order to provide a thorough and clear explanation that showcases your understanding** *(what you are expected to do on the AP Biology exam)***, your answer should always walk the reader logically, and systematically, through the process that led to your conclusion.** So, ***your answer should describe*** *1. the different alleles for the feather color gene that exist, 2. what the genotypes of the parents must be, 3. what the genotypes of the reproduce cells (gametes) are that the parents can each produce, 4. what the genotypes of the offspring must be given the phenotypes mentioned, and 5. must include a Punnet Square in order to compare the* ***expected*** *offspring phenotypic frequencies to the* ***observed*** *offspring phenotypic ratios*.

   i. What must be the **alleles for the feather color gene**? ***(When you answer, be sure to list the alleles, using the proper allele notation, while you also describe the phenotype each allele controls).***

   ii. List the **parents' genotypes** given their phenotypes.

   iii. List the **genotypes of the gametes (sperm/egg) each of these two parents can each potentially produce**.

   ♀ = _____ ♂ = _____

   iv. List the **genotypes of the OBSERVED offspring produced**.

   v. Draw to the right the Punnett Square for this cross.

   Remember, Punnett Squares do ***not*** *indicate which progeny* ***are*** *produced!*

   Punnett Squares indicate which progeny ***could*** *be produced!*

vi. Using the Punnett Square for this cross, determine the **expected** phenotype ratios of the offspring.

vii. Using the Punnett Square for this cross, discuss how well the **expected** ratio (from vi. above) **matches** the actual **observed** phenotype ratio's in the offspring.

b. i. What **phenotypes** would you **expect** in the offspring of a cross between a gray rooster and a black hen?

TIP: Always **explain** your reasoning, the **Punnett Square being part of the evidence** you use in your explanation. So, as you make the Punnett Square, make sure you also **explain what your Punnett Square shows** (= the possible offspring of a mating) and **explain how you determined which haploid gametes** (= sperm or egg) **to write in the top and side of your Punnett Square.**

ii. What is the simplest explanation for the inheritance of the colors in these chickens?

*Check your answer to 5.b.i & ii by going to the **Ch.14.3 Concept Check Question #3** answer in Appendix A)*

6. Humans are called **diploid** organisms because they have two sets of chromosomes. Therefore, **they carry two copies of each gene** on two of the same autosomes (chromosomes #1 through #22 - *we will look at the special case of chromosome #23, the sex chromosome, X vs Y, in the next chapter)*. **Since every human, whether male or female, has two homologous chromosomes for each type of autosomal chromosome, they can inherit up to two different alleles, one from each parent.** Explain then what is meant when a gene is said to have **multiple alleles**?

7. Blood groups are so important medically that you should be able to solve genetics problems based on blood types. The first step in accomplishing that is to understand the **genotypes that can lead to the creation of each of the four ABO blood types (phenotypes)**. Complete this blood type chart. Refer to Figure 14.11 first to see what the allele notations are you should use for each three possible ABO gene alleles. ***Note how two of the alleles behave dominantly over the recessive type and how the dominant alleles behave co-dominantly towards each other!!!***

Possible Blood Types (Phenotypes)	Description of the Appearance of the Red Blood Cell's Plasma Membrane	All Possible Genotypes That Can Lead to This Phenotype

8. a. *Practice:* If a man with type AB blood marries a woman with type O blood, what blood types would you expect in their children? Show your reasoning. **Tip: Always determine the genotypes of the parents first!** Then, figure out which reproductive cells the parents can make and finish by show your Punnett Squares and discussing the results seen.

b. *Practice:* What fraction of each of each blood type would you **expect** among the children (a.k.a. offspring/progeny)?

*(Check your answers to 8.a & b by going to the **Ch.14.3 Concept Check Question #2** answer in Appendix A)*

9. a. Define the term **pleiotropy**.

b. Many genes exhibit pleiotropy. When you are homozygous recessive for the gene that cause **Cystic Fibrosis** and **Sickle Cell Anemia** you see multiple phenotypic consequences in many tissues in the body. Let's look at another example... People with a hereditary disorder called **Marfan Syndrome** exhibit many different symptoms (phenotypes):

- Unusually tall height
- Long arms, legs, and fingers
- Thin fingers and toes
- Dislocation of the lens of the eye
- Heart problems (leaky heart valves and the aorta, the large blood vessel carrying blood away from the heart, bulges or can rupture).

A mutation in a single gene for fibrillin-1 (FBN1), a type of protein secreted by certain connective tissue cells into their extracellular matrix, causes Marfan syndrome, an **autosomal dominant disorder** of connective tissue. This gene normally encodes for the fibrillin-1 protein that assembles into chains, making elastic fibrils that give strength and flexibility to the body's connective tissues. Mutations that cause Marfan syndrome reduce the amount of functional protein made by the body, resulting in fewer fibrils in connective tissue.

How does the identity of this gene explain the range of symptoms? Our eyes and the aortas normally contain many fibrils that help maintain structure, which is why these two organs are affected in Marfan syndrome. In addition, the fibrils sequester certain growth factors in the interstitial fluid outside of cells, so that lower ("normal") concentrations of growth factors reach the receptors on target cells. When there are fewer of the extracellular fibrils, as is the case in Marfan syndrome, the growth factors cannot be sequestered as much and thus trigger excess growth of the organism by stimulating cells to proceed through the cell cycle more often, leading to the characteristic tall, thin Marfan build. So since different tissues turn on this gene, we see different phenotypes in different areas of the body when a person inherited the mutant dominant allele for Marfan syndrome.

10. a. Define the term **epistasis**.

b. When Mendel completed a dihybrid cross in which the pea plant parents were **both** heterozygous for **both** the seed color and seed shape genes (see Figure 14.8), the phenotypic ratio of the expected offspring was

9 yellow round seeds : 3 yellow wrinkled seeds : 3 green round seeds : 1 green wrinkled seeds.

Analyze the Punnet Square in Figure 14.12 now, showing the possible results of the dihybrid cross in which the Labrador retriever parents are heterozygous for both the pigment color and the pigment deposition genes.

Just as is the case with the pea plants seed color and seed shape genes, the Labs pigment color and pigment deposition genes sort independently *(the two genes are, thus, located on different types of chromosomes).*

Explain then why the expected phenotypic ratio of the offspring in the Lab mating isn't 9:3:3:1, as was the case with Mendel's pea plant mating, but instead ends up being...

9 Black Labs : 3 Brown Labs : 4 Yellow Labs.

c. Return to Figure 14.8. What was the **GENOtype ratio** of the expected offspring of the dihybrid cross in which the pea plant parents were both heterozygous for both the seed color and seed shape genes?

How does this ratio compare to the **genotype ratio** of the expected offspring resulting from the dihybrid cross in which the Labrador retriever parents are heterozygous for both the pigment color and the pigment deposition genes?

11. a. Define the term **polygenic inheritance**.

b. **IMPORTANTLY,** what would be your **"clue" that a character exhibits a polygenic inheritance pattern**?

c. Study Figure 14.13, showing the possible phenotypic results of a mating between two individuals with medium skin color phenotypes, given that skin color is controlled not just by the the two copies of one gene a person inherits, but by the two copies of multiple genes they inherit. Remember, each gene may exhibit complete dominance, incomplete dominance, codominance, or involve multiple alleles. In this illustration, only the effects of three genes are shown. So far, in reality, over 378 genes have been identified that together help determine skin color in humans an in mice. What is another character in humans that is considered polygenic?

d. Let's jump, for a moment, to the Scientific Skills Exercise: Making a Histogram and Analyzing a Distribution Pattern. **Before you begin this exercise though, let's review a few "must dos" when it comes to graphing.**

A few notes about drawing graphs for the AP Exam:

1. Make sure when you construct a graph, that the graph is **LARGE and NEAT**. Don't cram everything in just a small area. Use up a third or half a page. Give yourself lots of space so nothing has to be squeezed in or written so tiny or incompletely that it can't be read properly or isn't accurate.

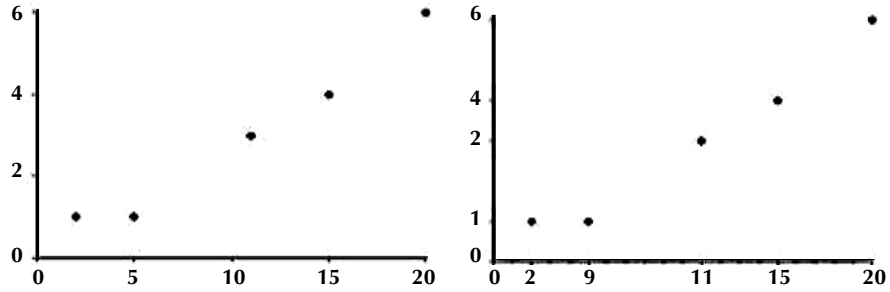
2. Always write a **proper title on the top of your graph** (**"The effect of the independent variable on the dependent variable"** is a good example to follow).

3. Always **label the x- and y- axes** clearly and accurately (Remember, the **independent variable** decided on by the researcher goes on the x-axes while the **dependent variable** - the data that is determined by the level of the independent variable - goes on the y-axes).

4. Always **put the units, in parenthesis, behind the label of each axes** when the data is quantitative.

5. **Use a Key/Legend**, when necessary, to provide information about what the data sets that are displayed on the chart represent. The key/legend is used to identify the data when two or three sets of data are plotted within the same set of axes. You must distinguish between the data by using different colored or solid/dotted lines, colored or patterned bars or different symbols for each data point (X, O, ∇ etc). The key on the side of the graph explains the symbols or different patterns of data points or lines.

6. Always **use a consistent scale on each axes**, making sure that each interval mark on your axes is equidistant from the next.

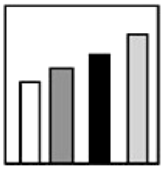


LIKE THIS **NOT LIKE THIS**

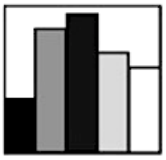
7. Always take care to **plot your data accurately**. Messy and inaccurate placements of data points will be considered wrong.

8. Draw the **appropriate type of graph** to represent your data.

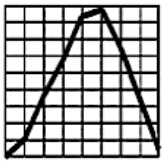
- A **bar graph** is a visual tool that uses bars to compare data among **distinct categories**. Bar graphs are good for graphing **discrete, discontinuous data** *(when data does not exist "in between" the bars placed on the x-axes)*. Histogram is used for distribution of non-discrete variables while bar graph is used for comparison of discrete variables. The data represented in bar graphs are not necessarily dependent on any other variables and the groupings are often qualitative (i.e. grouped into categories, like blood types or color). **The bars do NOT touch. It is usually possible to change the order of the bars along the axes** (which is not possible in a histogram)



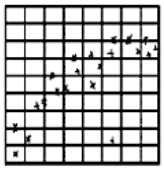
- A **histograms** may look similar to a bar graphs, but the data represented in histogram is in groups of **continuous numerical (quantitative) data**. In a histogram, **the bars must TOUCH.** Histograms are often used to show frequency data. In Histogram, **we cannot rearrange the blocks** (while in Bar graph you can) **just like you cannot rearrange the data points when making a line graph.**



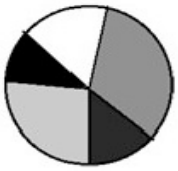
- A **line graph** consists of a series of points plotted on a grid and then connected together point to point by a line. Line graphs are only **used when both variables are quantitative and continuous**. Line graphs **show trends**, such as how a variable changes over time.



- In a **scatter plot**, the data points are plotted on the grid, but the **data points are NOT joined point to point**. A **line of best fit** may be added to a scatter plot to show a **trend**. These graphs are useful for **showing if a correlation exists between two variables,** especially when it is not possible to alter either of the variables (i.e. such as in descriptive studies where you are merely observing but not manipulated variables).

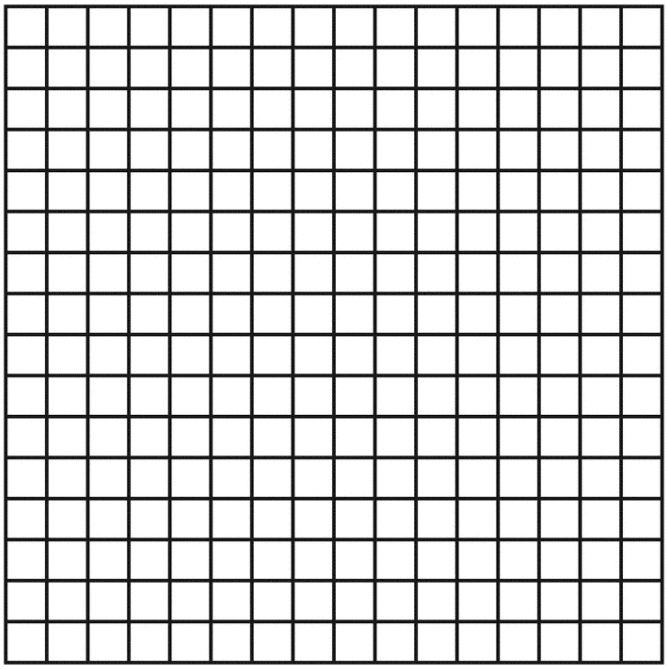


- A **pie chart** is designed to show a percent of a whole, where **the whole equals 100%**. Pie charts are used **to compare data**, but **cannot be used to see how a manipulated variable affects a responding variable**. Pie charts do not show change with respect to another variable.



Now that you know all the parts a graph should contain, complete the **Scientific Skills Exercise: Making a Histogram and Analyzing a Distribution Pattern.**

1.



2. a. (*Part of answer on graph)*

   b. *(Curve on graph)* **Reason for curve shape**:

12. What does it mean when geneticists say that a phenotype is **multifactorial**?

.

13. a. What is a **pedigree**?

b. Read through Figure 14.15, including the **Tips for Pedigree Analysis**. Pedigrees can be used to study the genetics of inherited diseases and to make predictions of phenotypes in future offspring. For example, pedigrees can be analyzed to determine whether the disease-causing alleles are **dominant or recessive**, whether the disease alleles are **sex-linked** (located on the X or Y chromosomes - the 23rd type of chromosome in humans) or **located on an autosome** (chromosomes #1 through #22 in humans). They can also be used to predict the **expected phenotypes in offspring** of a mating and can be used to **calculate the likelihood of an offspring displaying a particular phenotype**.

Now that you understand what a pedigree looks like, let's practice drawing one. Brown eyes are a dominant eye-color allele and blue eyes are recessive. A brown-eyed woman, whose father had blue eyes and whose mother had brown eyes, marries a brown-eyed man, whose parents are also brown-eyed. This couple has a son with blue eyes. Draw a pedigree that **traces the phenotype blue eyes** in this family, showing all four grandparents, the two parents, and the son. Indicate **ALL** possible genotypes for **each** individual**.** *(**Do not draw small images, here or on AP Exams** - make your drawings, graphs, visual models **large and spacious** enough so anyone, even if their eye site is poor, can easily see what your are showcasing and you are not crowding your text, data points, units, scales, labels and more)*

14. a. What is a **carrier**?

b. Why do **carriers (heterozygotes) for an allele that causes a recessively inherited disorder often still have the normal phenotype** at the organismal level **while those that are homozygous recessive do develop the disorder**?

c. Why does **inbreeding cause an increase in autosomal recessive conditions among offspring** compared to those resulting from matings between unrelated parents?

15. **AP Students are expected to have a general knowledge of the pattern of inheritance and the common symptoms of a number of genetic disorders.** *Do not ignore information on inheritance patterns of diseases discussed in your text.* Let's examine some genetic disorders you should know.

a. **Cystic Fibrosis**

What is the **pattern of inheritance of Cystic Fibrosis**? Is it caused by a dominant vs recessive allele on an autosome?

What type of **protein is malfunctioning**?

How is this protein malfunctioning?

What are the **consequences in phenotype(s) at the cellular level** due to the **absence of normal protein function**?

What are the **consequences in phenotype(s) at the organismal level** due to the absence of normal protein function?

b. **Sickle Cell Disease**

What is the **pattern of inheritance for Sickle Cell Disease**? Is it caused by a dominant vs recessive autosomal allele?

What type of **protein is affected**?

How is the protein affected? *(Remember your previous studies too on the subject)*

Read your text and review Figure 14.17. What are the **consequences in phenotype at the cellular level** due to the **abnormal protein behavior**?

What are the **consequences in phenotype(s) at the organismal level** due to the abnormal protein behavior?

It is often said that the normal β hemoglobin polypeptide allele is dominant over the mutant β hemoglobin polypeptide (the sickle-cell) recessive allele because two recessive alleles are requires to have sickle-cell disease at the organismal level. However, heterozygotes do have **sickle-cell trait**, even if they do not have sickle-cell disease. What is stickle-cell trait?

Because heterozygotes exhibit sickle cell trait, **what inheritance pattern does the normal β hemoglobin allele exhibit then at the organismal level?** Explain.

Think: Given what you learned earlier in Ch.14, Section 3 *(with Tay Sachs as an example),* **what is the inheritance pattern exhibited by the two β hemoglobin polypeptide alleles (the normal versus the mutant one) at the cellular and the molecular level?** Explain.

c. **Achondroplasia**

What is the **pattern of inheritance for Achondroplasia**? Is it caused by a dominant vs recessive autosomal allele?

What are the **consequences in phenotype(s) at the organismal level** due to the **abnormal protein activity**?

d. **Huntington's Disease**

What is the pattern of inheritance for Huntington's Disease? Is it caused by a dominant vs recessive allele on an autosome *(in this case chromosome #4)*?

What are the **consequences in phenotype(s) at the organismal level** due to the **abnormal protein activity**?

How is it possible that a person could pass the Huntington's allele down to their offspring without knowing that they have the lethal Huntington allele in their DNA?

16. For some genes, the recessive allele is more common in a population's **GENE POOL (the collection of all the alleles for that gene found within all the individuals in that population)** than the dominant allele. Other times it is the dominant allele that is more prevalent than the recessive one. Why are **lethal dominant alleles (those that cause a deadly condition) much less common than lethal recessive alleles** (that also cause a deadly condition) in populations?

17. *Think:* Juanita was born with six toes on each foot. Polydactyly follows an autosomal dominant inheritance pattern. Two of her five siblings and her mother, but not her father, also have extra digits. What is Joan's genotype for the number-of-digits character? Explain, using *D* and *d* to symbolize the alleles for the gene that controls the character number of digits. *As always, write out your logic.*

*(Check your answer by going to the **Ch.14.4 Concept Check Question #3** answer in Appendix A)*

18. *Think:* Betty has a sibling with cystic fibrosis, but neither of her parents have the disease. Her husband Bob knows he is a carrier though Betty does not.

a. Calculate the probability that if this couple has a child, the child will have cystic fibrosis. *Show your work, which should include Punnett Squares **and** probability calculations.* Remember though that you must **FIRST ALWAYS DETERMINE & REPORT THE POSSIBLE GENOTYPES OF BOTH PARENTS.**

b. What would be the probability that their child would have cystic fibrosis and a test revealed that both Betty and Bob are carriers? *Show your work, which should include a Punnett Square and/or probability calculations.*

c. What would be the probability that their child would have cystic fibrosis and a test revealed that Betty is a carrier but Bob is not? *Show your work, which should include a Punnett Square and/or probability calculations.*

d. What if Betty and Bob's first child has cystic fibrosis and they want to have four children. What is the probability that **all** four of their children would end up with cystic fibrosis? *Show **and explain** your work.*

e. What if Betty and Bob's first, second, and third children all have cystic fibrosis. What is the probability that their **fourth child will have the disease as well**? *Show **and explain** your work.*

*P.S. Your probability in 18.a. should **not** match the probability calculated as an answer in 18.b. The probability in 18.a is 1/6 while the probability in 18.b. is 1/4. Did you get it right and did you show and explain all your work? If not, go back and take another look at what you may have done wrong and remember that multiplication rule! Hint for 18.a: What is the probability that Betty is a carrier **AND** she then also produces a child with cystic fibrosis with Bob?*

19. What would you suspect if Tom was born with polydactyly, but neither of his biological parents had extra digits?

P.S. The probabilities for 18.c-e should be 0; 1/256; 1/4. Did you get these right and did you show and explain your work?