

# Lab Notebook

## *Basic Information*

How does your assigned allele affect characteristics at the cellular, tissue, and organism levels to cause disease? The information on **Learn.Genetics** and in your assigned **Allele Profile** will help you fill in each section. The section headings on each resource match. This should make finding the information you need easier.

**1.** Fill in from your Allele Profile:

Genetic Disorder \_\_\_\_\_

Affected gene \_\_\_\_\_ Affected protein \_\_\_\_\_

Allele \_\_\_\_\_

**2.** Get to know your assigned genetic disorder:

- Visit [Learn.Genetics.utah.edu/content/genetics/](https://Learn.Genetics.utah.edu/content/genetics/)
- Find your assigned **Genetic Disorder**, and watch the video at the top of the page.

Notes:

## Mutations & Alleles

New alleles arise from mutation in reproductive cells. Once an allele arises, it can be passed from parent to offspring, from generation to generation.

- Fill in the codon range from your **Allele Profile** page, then transcribe and translate both alleles:
  - Write in the RNA bases that are complementary to the **template** DNA strand. Remember, A in DNA pairs with U in RNA. (*Note: you may or may not need every box*)
  - Starting on the left, circle the RNA bases in groups of 3. Each group of 3 is a codon.
  - Look up each codon on an Amino Acid Codon Chart ([circular](#) or [square](#)). Write in the corresponding amino acid on the protein strip.

fold

Codon range: \_\_\_\_\_

fold

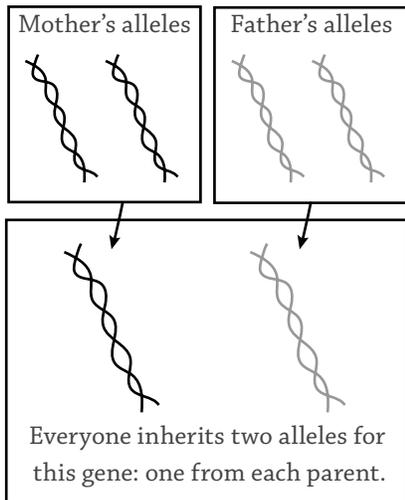
- How are the **DNA** sequences different in the Healthy vs. Disorder alleles?
- How are the **amino acid** sequences different in the Healthy vs. Disorder proteins?
- What type of change (mutation) during DNA replication do you think caused the difference in the Disorder protein? Pick one.
  - Substitution (one base was substituted for another)
  - Insertion (extra DNA base/s added)
  - Deletion (DNA base/s left out)
- Did the DNA change cause a frameshift in the protein? Y / N  
*(A frameshift changes the grouping of bases into codons, usually affecting multiple amino acids)*

## Inheritance

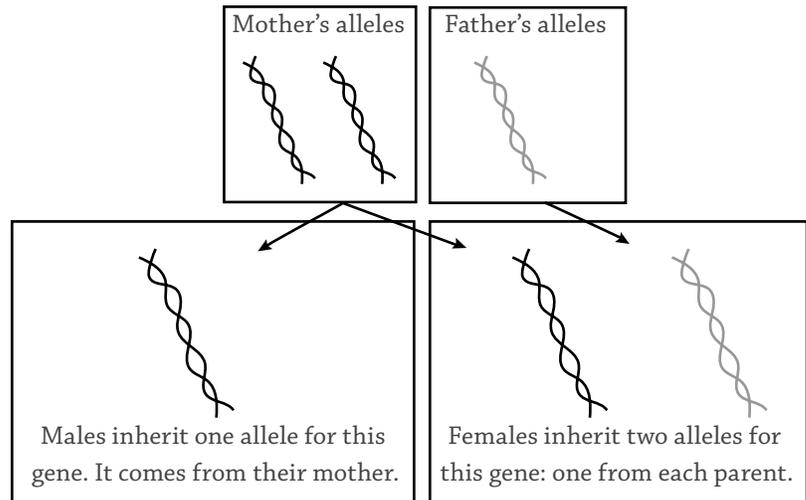
From [Learn.Genetics.utah.edu/content/genetics/](http://Learn.Genetics.utah.edu/content/genetics/) visit the web page for your assigned genetic disorder. Use the information there and in your **Allele Profile** to answer the questions as they relate to **your assigned allele**.

- What is the inheritance pattern from the Classical Genetics perspective (i.e., having **allele/s** that cause the genetic disorder)? Explain your answer. (*Note: autosomes are non-sex chromosomes*)
  - Autosomal dominant
  - Autosomal recessive
  - X-linked
  - Co-dominant
- What is the inheritance pattern from the Molecular Genetics perspective (i.e., of the **protein product** that is made from the allele/s)? Explain your answer.
  - Autosomal dominant
  - Autosomal recessive
  - X-linked
  - Co-dominant
- Genetic disorders are strongly influenced by genes that pass from parents to children. For the gene that causes this disorder, choose the model that best shows how the alleles are inherited.

**MODEL 1**



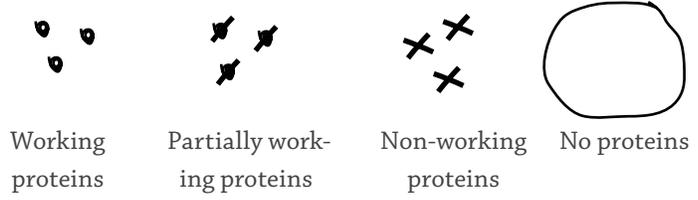
**MODEL 2**



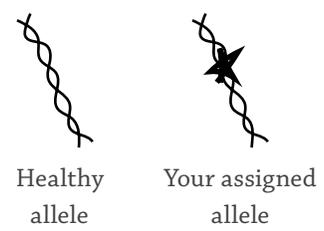
- Look at the data graph on page 2 of your Allele Profile. How is the function of the protein made from your assigned allele different from the protein made from a healthy allele?

*Inheritance (cont.)*

5. Read the *Protein Function & Gene Expression* section of your Allele Profile. Choose the symbol that best represents the protein made from your assigned allele.

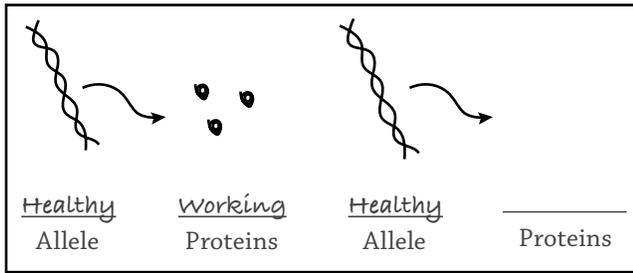


6. The symbols on the right represent healthy and affected alleles. Using these symbols and the best protein symbols from question 5, complete the models to show the proteins that are made from each person's alleles for this gene. Label the alleles and proteins.  
*Hint: You'll need to interpret the data graph in your Allele Profile.*



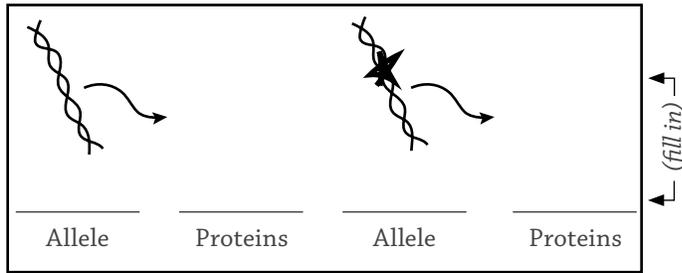
**MODELS:**

**Person A: Two healthy alleles**



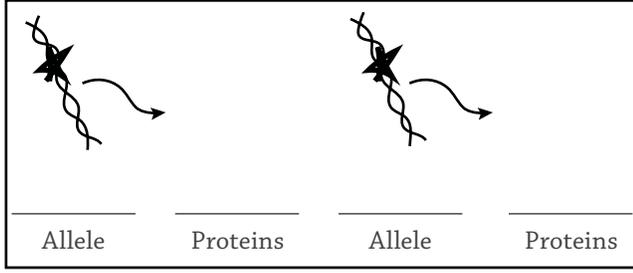
Does Person A have a genetic disorder?  
 Yes / No / Maybe, it depends

**Person B:**



Does Person B have a genetic disorder?  
 Yes / No / Maybe, it depends

**Person C:**



Does Person C have a genetic disorder?  
 Yes / No / Maybe, it depends

7. **Answer this question only if the Inheritance section of your Allele Profile tells you to.** Draw models to show other possible allele combinations and the proteins that are made from each. Would people with these allele combinations have a genetic disorder?

## Protein Function & Expression

From [Learn.Genetics.utah.edu/content/genetics/](http://Learn.Genetics.utah.edu/content/genetics/) visit the web page for your assigned genetic disorder. Use the information there and in your **Allele Profile** to answer the questions.

1. This protein is made in \_\_\_\_\_ (cell type/s).
2. This protein does its job in \_\_\_\_\_ (tissue and/or organ).
3. Compare the healthy protein with the protein made from your allele. How do the proteins affect what happens at the cellular, tissue, and organism levels? Write or draw your comparisons.

Things to consider: How well does the protein do its job? Does it get to the right place to do its job? Is its structure different?

*Note: Some characteristics are general to the genetic disorder, and others are specific to your allele. Be sure to include both.*

|                | <b>Healthy Protein:</b><br><i>What is its regular job?</i> | <b>Protein from My Allele:</b><br><i>What problems does it cause?</i> |
|----------------|--|---|
| Cellular level |  |   |
| Tissue level   |  |   |
| Organism level |  |   |

## Other Factors

From [Learn.Genetics.utah.edu/content/genetics/](http://Learn.Genetics.utah.edu/content/genetics/) visit the web page for your assigned genetic disorder. Use the information there to fill in the tables and answer the questions.

1. People who have the very same disease-causing alleles often have differences in their symptoms. These differences come about through a combination of genetic and environmental factors. Because these factors influence a person’s risk for developing symptoms, they are often called risk factors.

Fill in the table to include:

- At least two environmental factors that can increase risk;
- At least two genetic factors (besides the gene that causes the genetic disorder) that can increase risk;
- For one genetic factor and one environmental factor, think about (and fill in) how it might work.

|                       | Risk factor | How it might work (fill in one) |
|-----------------------|-------------|---------------------------------|
| Environmental factors |             |                                 |
|                       |             |                                 |
| Genetic factors       |             |                                 |
|                       |             |                                 |

Notes:

## *Other Factors (cont.)*

Managing or treating a genetic disorder usually involves a combination of lifestyle behaviors and medical approaches. These are environmental factors that influence a person's symptoms.

**2.** Which approaches are used for treating your assigned genetic disorder? (circle all that apply)

- Managing an environmental factor
- Treating symptoms of the disorder
- Replacing something that is missing in people with the disorder

**3.** Provide general descriptions related to your assigned genetic disorder:

**a.** What problems do the treatments address? How do you think they might work?

**b.** What problems (if any) are current treatments unable to address?

## Cause & Effect

Variations in DNA lead to differences in proteins. Through a chain of cause-and-effect relationships, this leads to differences at the cell, tissue, organ, and whole-organism levels.

### Instructions

Review each section of this Lab Notebook. Use the information there to explain, at each level, how **your assigned allele** causes specific symptoms of disease.

*Hint: You have already done most of this work! Just go back and find it.*

|   |        |
|---|--------|
| <i>Specific DNA change/mutation:</i>              |        |
|   | Cause  |
| <i>Difference in protein amino acid sequence:</i> | Effect |
|   |        |
|   | Cause  |
| <i>What happens with the protein:</i>             | Effect |
|   |        |
|   | Cause  |
| <i>What happens to/in cells:</i>                  | Effect |
|   |        |
|   | Cause  |
| <i>What happens in tissues and organs:</i>        | Effect |
|   |        |
|   | Cause  |
| <i>Effects on the whole person:</i>               | Effect |
|   |        |

NAME \_\_\_\_\_ DATE \_\_\_\_\_ ALLELE \_\_\_\_\_

## Symposium

### Instructions

Get together with others who were assigned the same gene as you but different alleles.

How are the disorder-causing alleles similar and different at each level?

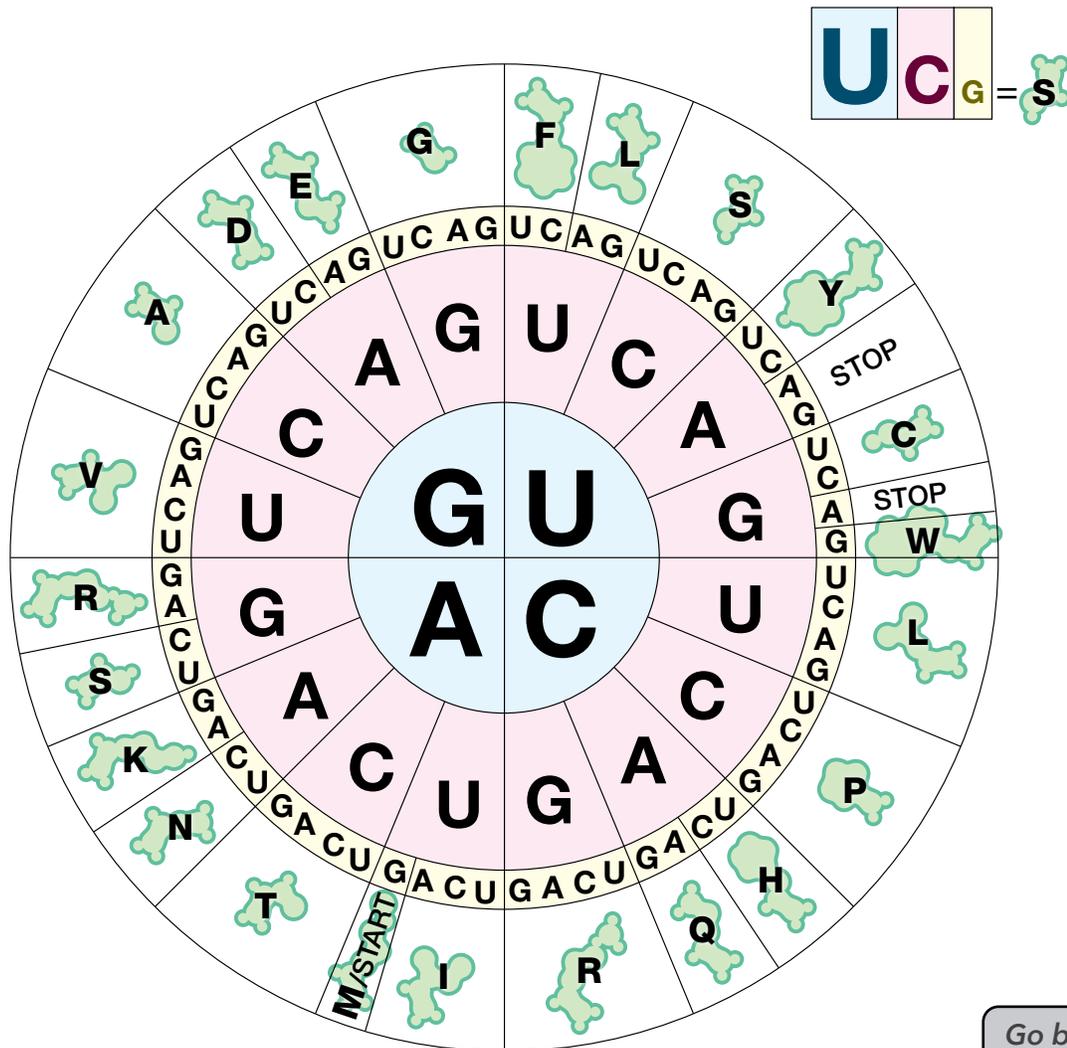
*Places to find information: Cause & Effect page, Allele Profile*

Your allele \_\_\_\_\_ Other allele/s you are comparing \_\_\_\_\_

|   | Similarities | Differences |
|---|--------------|-------------|
| Type of change to the DNA & protein sequences |              |             |
| Protein function at the cellular level        |              |             |
| Effects on tissues & organs                   |              |             |
| Effects on the whole person                   |              |             |

# Amino Acid Codon Chart

Circular Version



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## Amino acid side chains

|                              |                              |                                 |                                 |                                 |                           |                             |                              |                              |                            |
|------------------------------|------------------------------|---------------------------------|---------------------------------|---------------------------------|---------------------------|-----------------------------|------------------------------|------------------------------|----------------------------|
| <b>A</b><br>Alanine (Ala)    | <b>C</b><br>Cysteine (Cys)   | <b>D</b><br>Aspartic acid (Asp) | <b>E</b><br>Glutamic acid (Glu) | <b>F</b><br>Phenylalanine (Phe) | <b>G</b><br>Glycine (Gly) | <b>H</b><br>Histidine (His) | <b>I</b><br>Isoleucine (Ile) | <b>K</b><br>Lysine (Lys)     | <b>L</b><br>Leucine (Leu)  |
| <b>M</b><br>Methionine (Met) | <b>N</b><br>Asparagine (Asn) | <b>P</b><br>Proline (Pro)       | <b>Q</b><br>Glutamine (Gln)     | <b>R</b><br>Arginine (Arg)      | <b>S</b><br>Serine (Ser)  | <b>T</b><br>Threonine (Thr) | <b>V</b><br>Valine (Val)     | <b>W</b><br>Tryptophan (Trp) | <b>Y</b><br>Tyrosine (Tyr) |

# Amino Acid Codon Chart

Square Version

|              |   | Second Letter   |   |   |   |   |
|--------------|---|---|---|---|---|---|
|              |   | U   | C   | A   | G   |   |
| First Letter | U   | UUU          | UCU   | UAU      | UGU    | U |
|              |   | UUC          | UCC    | UAC      | UGC    | C |
|              |   | UUA          | UCA    | UAA STOP  | UGA STOP  | A |
|              |   | UUG          | UCG   | UAG STOP  | UGG    | G |
|              | C   | CUU   | CCU   | CAU      | CGU   | U |
|              |   | CUC          | CCC    | CAC      | CGC    | C |
|              |   | CUA          | CCA    | CAA      | CGA    | A |
|              |   | CUG   | CCG   | CAG      | CGG   | G |
|              | A   | AUU   | ACU   | AAU      | AGU    | U |
|              |   | AUC          | ACC    | AAC      | AGC    | C |
|              |   | AUA         | ACA   | AAA     | AGA   | A |
|              |   | AUG  START | ACG   | AAG    | AGG  | G |
| G            | GUU   | GCU   | GAU  | GGU   | U   |   |
|              | GUC  | GCC        | GAC  | GGC  | C   |   |
|              | GUA  | GCA        | GAA  | GGA  | A   |   |
|              | GUG   | GCG   | GAG  | GGG   | G   |   |

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| Amino acid side chains  |   |  |  |  |  |  |   |   |   |
|---|---|--|--|--|--|--|---|---|---|
| <br>Alanine (Ala)    | <br>Cysteine (Cys)   | <br>Aspartic acid (Asp) | <br>Glutamic acid (Glu) | <br>Phenylalanine (Phe) | <br>Glycine (Gly) | <br>Histidine (His) | <br>Isoleucine (Ile) | <br>Lysine (Lys)     | <br>Leucine (Leu)  |
| <br>Methionine (Met) | <br>Asparagine (Asn) | <br>Proline (Pro)       | <br>Glutamine (Gln)     | <br>Arginine (Arg)      | <br>Serine (Ser)  | <br>Threonine (Thr) | <br>Valine (Val)     | <br>Tryptophan (Trp) | <br>Tyrosine (Tyr) |