NAME Demo Lab Notebook DATE _____

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 Cystic fibrosis

Affected gene CFTR Affected protein CFTR

Allele C1

2. Get to know your assigned genetic disorder:

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

Notes:

1/28 people in the US carry a disorder-causing CF allele

CF affects salt and water balance

Thick mucus builds up in the respiratory and digestive systems so many organs have problems

Sometimes CF is mistaken for allergies

Early treatment leads to a better outcome. Now newborns are commonly checked for the disorder

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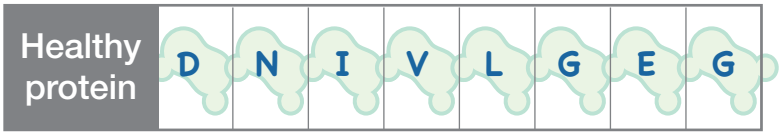
Mutations & Alleles

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

1. Fill in the codon range from your **Allele Profile** page, then transcribe and translate both alleles:

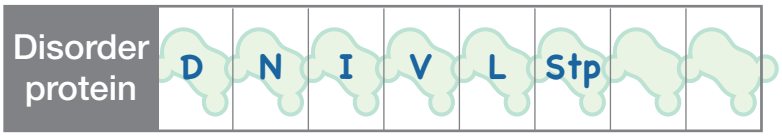
- Write in the RNA bases that are complementary to the **template** DNA strand. Remember, A in DNA pairs with U in RNA. *(Note: you may or may not need every box)*
- Starting on the left, circle the RNA bases in groups of 3. Each group of 3 is a codon.
- Look up each codon on an Amino Acid Codon Chart. Write in the corresponding amino acid on the protein strip.

fold



Codon range: 537–544

fold



2. How are the **DNA** sequences different in the Healthy vs. Disorder alleles?

One base is different. G in the healthy allele is T in C1.

3. How are the **amino acid** sequences different in the Healthy vs. Disorder proteins?

G (GGA) changes to stop (UGA) in C1. All the amino acids after that are missing in the C1 protein.

4. What type of change (mutation) during DNA replication do you think caused the difference in the Disorder protein? Pick one.

a. Substitution (one base was substituted for another)

b. Insertion (extra DNA base/s added)

c. Deletion (DNA base/s left out)

5. 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)

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Inheritance

From **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**.

1. 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)*

a. Autosomal dominant
c. X-linked
b. Autosomal recessive (circled)
d. Co-dominant

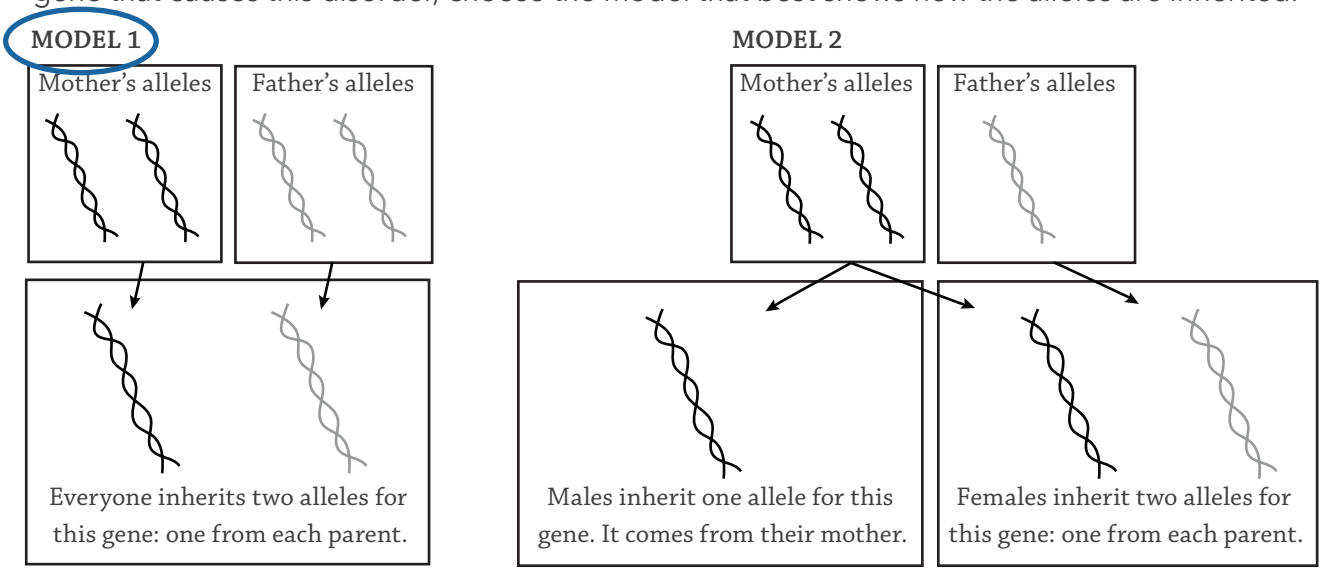
It takes 2 non-working alleles to cause CF.

2. 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.

a. Autosomal dominant
c. X-linked
b. Autosomal recessive
d. Co-dominant (circled)

Protein is made from both alleles.

3. 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.



4. 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?

The protein from the C1 allele doesn't move chloride ions as well as the protein made from the healthy allele. So on the graph, sweat chloride levels are higher in people with C1 alleles compared to healthy alleles.

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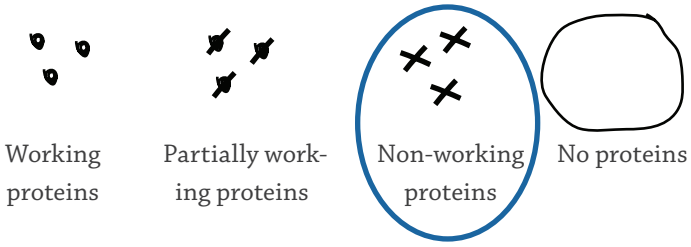
Inheritance (cont.)

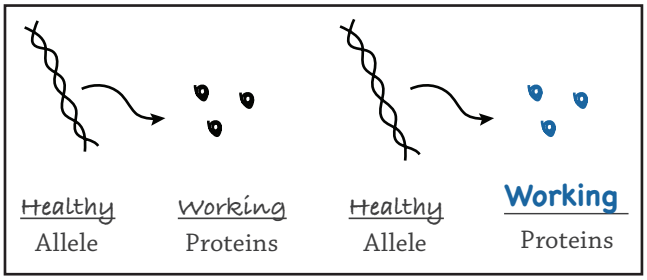
5. Read the *Protein Function & Gene Regulation* section of your Allele Profile. Choose the symbol that best represents the protein made from your assigned allele.



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.

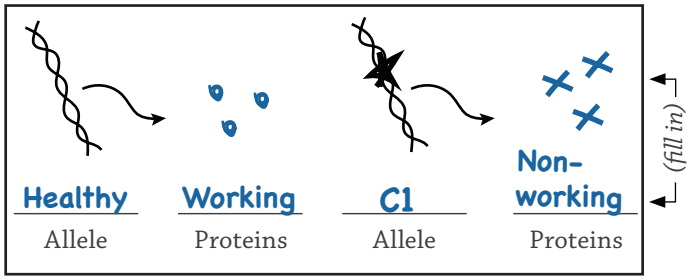
MODELS:

Person A: Two healthy alleles



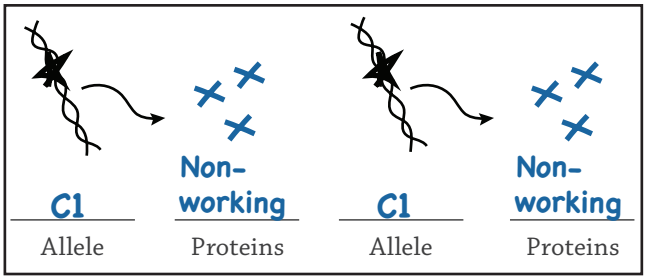
Does Person A have a genetic disorder?
Yes / No / Maybe, it depends

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?

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Protein Function & Expression

From **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 cells lining the lungs & digestive organs (cell type/s).

2. This protein does its job in lungs, pancreas, gallbladder, small intestine (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.

	Healthy Protein: *What is its regular job?*	**Protein from My Allele:** *What problems does it cause?*
Cellular level	- Sits in the cell membrane - Moves chloride ions out of the cell - Keeps salt and water balanced	- Less protein is made. What is made can't move chloride ions. - Without working CFTR protein, salt and water are out of balance
Tissue level	- Lung tissue: a healthy layer of mucus forms to trap and expel debris. - Digestive tissues: the proper balance of salt and water keeps digestive juices flowing.	- C1 causes problems in lung & digestive tissue. - Lung tissue: thick mucus traps debris & bacteria. - Digestive tissue: thick mucus prevents absorption of nutrients. Pancreatic ducts get clogged.
Organism level	- Person is healthy. Watery mucus in the airway protects from lung infections. Healthy digestive function helps with growth and overall health.	- C1 causes severe CF symptoms - Coughing, wheezing, lung damage & infections, pnemonia, bronchitis, low weight, slow growth, dehydration

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Other Factors

From **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	Air pollution	The person inhales even more debris than usual. It's trapped and irritates the lungs and airways.
	Poor nutrition	
Genetic factors	Variation in genes that decrease immune function	It's harder to fight off infections from bacteria trapped in the lungs.
	Variation in genes that make the pancreas less healthy	

Notes:

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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?

They prevent some of the worst features of CF, like lung damage and bad nutrition. They help break up mucus, prevent infections, and help with digestion.

For some alleles, medications restore function to the disorder-causing protein.

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

Treatment routines can take hours every day.

There is still no cure for CF; nothing treats the underlying genetic problem.

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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.

Specific DNA change/mutation:

One base is changed — G is changed to T.

Cause

Difference in protein amino acid sequence: Effect

There is a premature stop, so the C1 protein is missing some of its amino acids.

Cause

What happens with the protein: Effect

Less protein is made, and a piece is missing so it can't move chloride ions.

Cause

What happens to/in cells: Effect

Cells can't move chloride out. Salt and water are out of balance.

Cause

What happens in tissues and organs: Effect

Thick mucus builds up in the lungs. Digestive organs do not work properly.

Cause

Effects on the whole person: Effect

Severe CF. Coughing, wheezing, lung damage & infections, pnemonia, bronchitis, low weight, slow growth, dehydration

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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

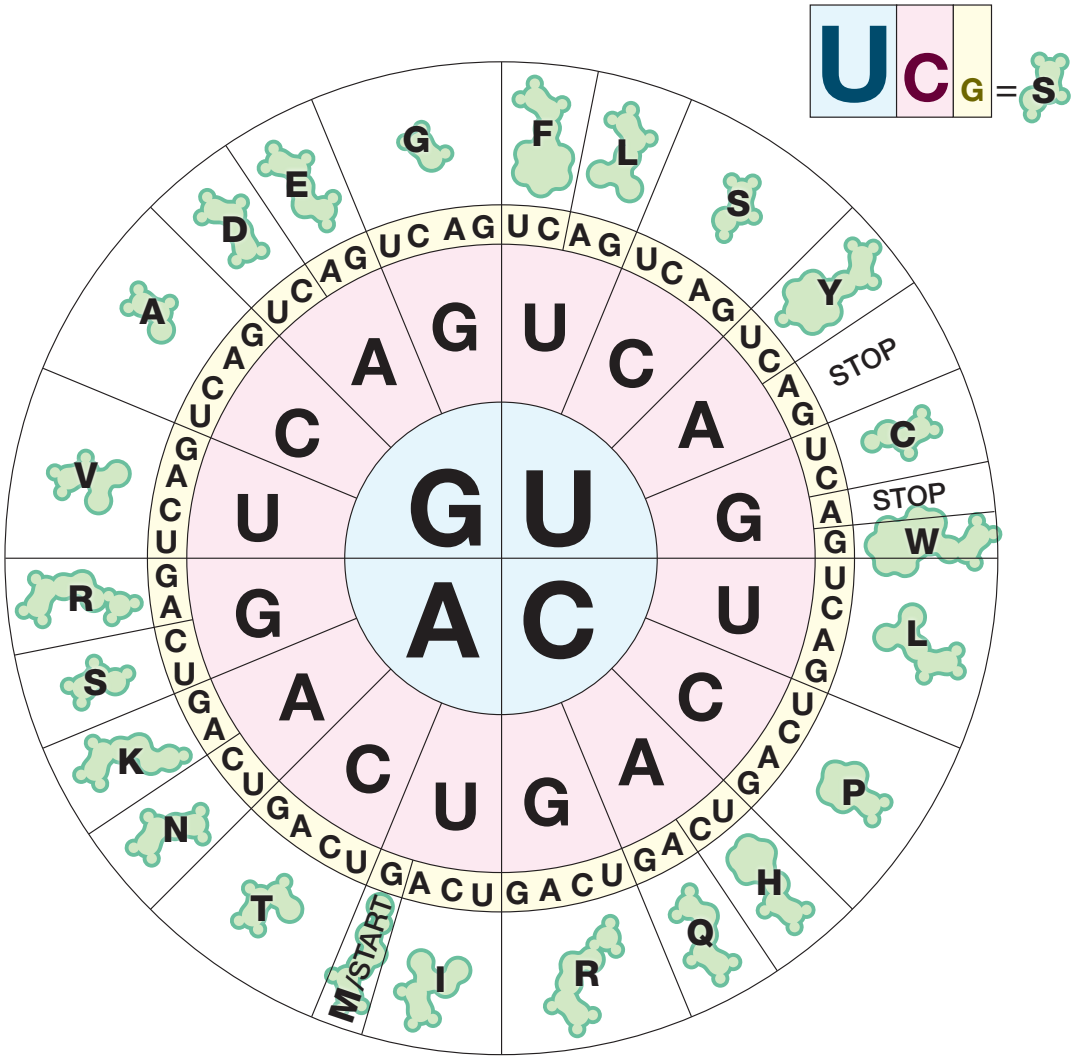
Your allele _____ Other allele/s you are comparing _____

	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

Learn.Genetics.utah.edu
GENETIC SCIENCE LEARNING CENTER

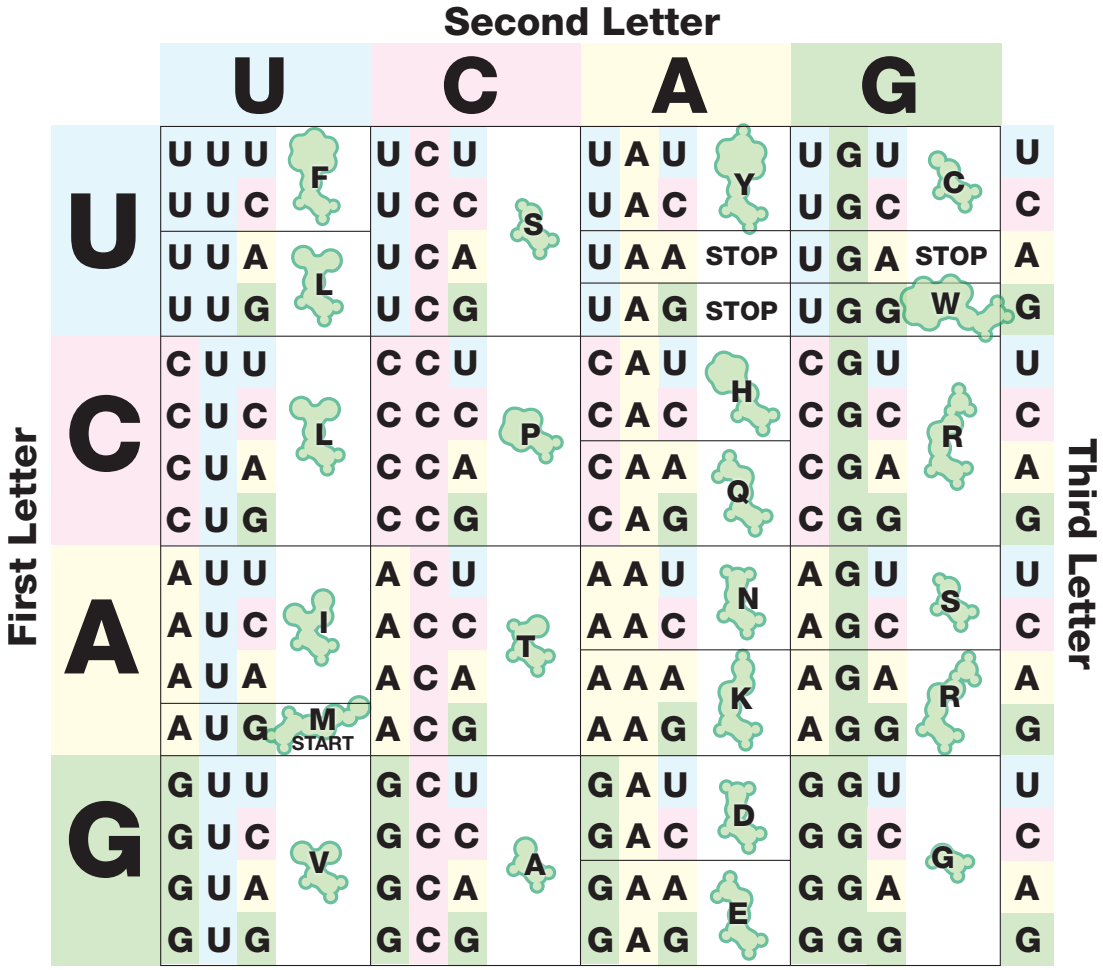


Amino acid side chains

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

Amino Acid Codon Chart

Square Version



Amino acid side chains