NAME _____ DATE _____

Allele Profile

Cystic fibrosis, Allele C1

Basic Information

More information at Learn.Genetics.utah.edu/content/genetics/cysticfibrosis/

Genetic Disorder – **Cystic fibrosis**

Affected Gene – ***CFTR***

Affected Protein – The affected gene codes for the protein **cystic fibrosis transmembrane conductance regulator (CFTR)**

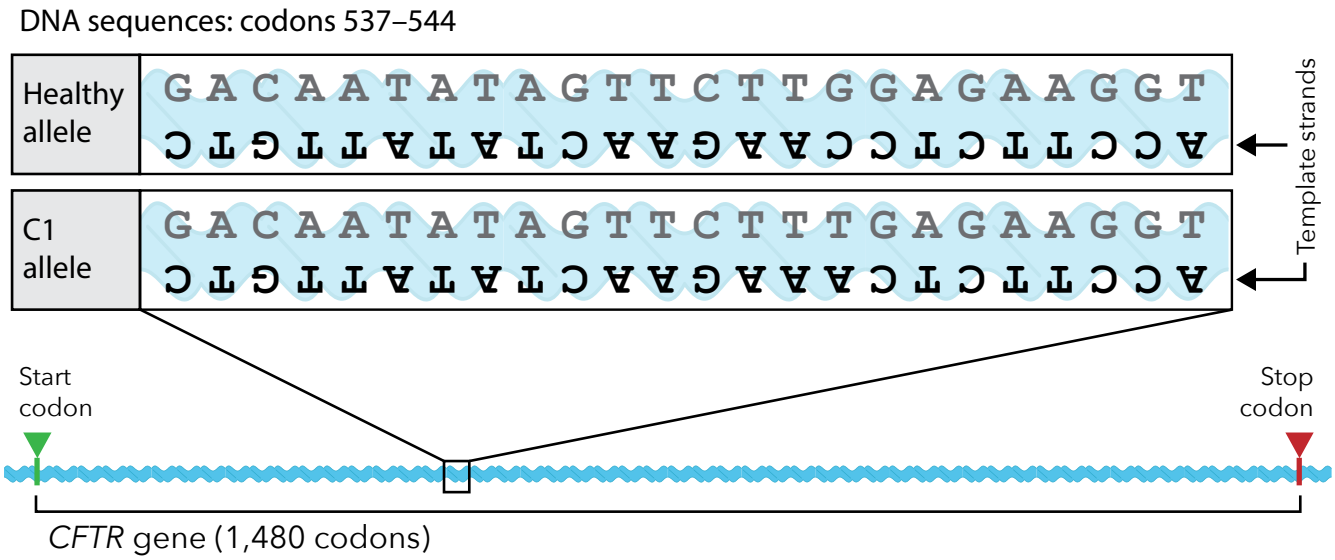
Allele – **C1**

There are more than 1,200 versions, or alleles, of the *CFTR* gene. Some cause genetic disorders and some do not. Your assigned allele is one of a few hundred that cause cystic fibrosis.

Mutations & Alleles

The protein-coding portion of the *CFTR* gene is 4,440 nucleotides long, and it has 1,480 codons.

The DNA sequence of your allele is identical to a healthy allele for most of its length. The place where they differ is shown in detail:

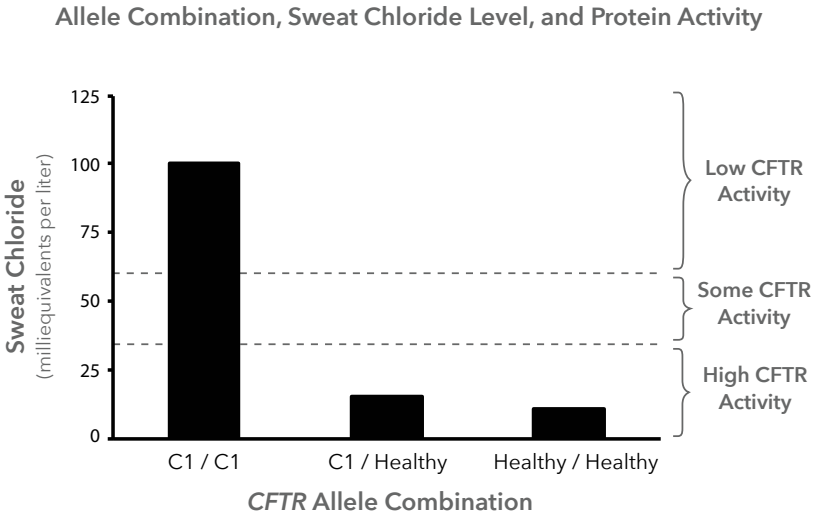


NAME _____ DATE _____

Inheritance

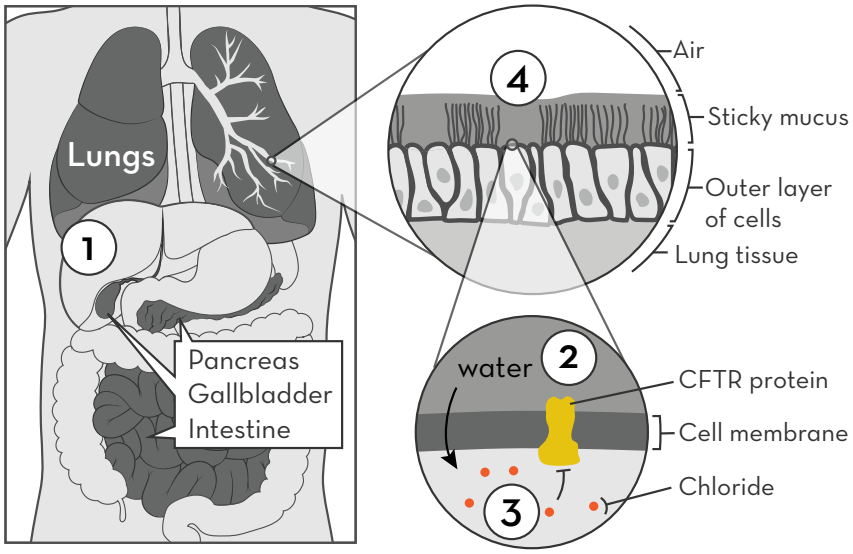
Everyone inherits two alleles of the *CFTR* gene, and CFTR protein is normally made (expressed) from both. Once it is made, CFTR protein goes to the plasma membrane. Here, it moves chloride ions (from salt) from inside the cell to the outside.

The graph shows the average sweat chloride level from many people with each allele combination. A sweat test measures how well a person's CFTR proteins are working. If CFTR proteins are working well, chloride levels are low. In people with cystic fibrosis, chloride levels are high.



Protein Function & Expression

1. The C1 allele is switched on in cells that line the lungs and digestive organs (same as healthy alleles).
2. Cells read the C1 allele and build CFTR protein—but they make much less protein than normal.
3. The small amount of CFTR protein that is made from the C1 allele is altered: it cannot move chloride ions.
4. People with two C1 alleles have cystic fibrosis. They usually have thick mucus in the airways of their lungs, and their digestive organs do not work properly.



NAME _____ DATE _____

Allele Profile

Cystic fibrosis, Allele C2

Basic Information

More information at Learn.Genetics.utah.edu/content/genetics/cysticfibrosis/

Genetic Disorder – **Cystic fibrosis**

Affected Gene – ***CFTR***

Affected Protein – The affected gene codes for the protein **cystic fibrosis transmembrane conductance regulator (CFTR)**

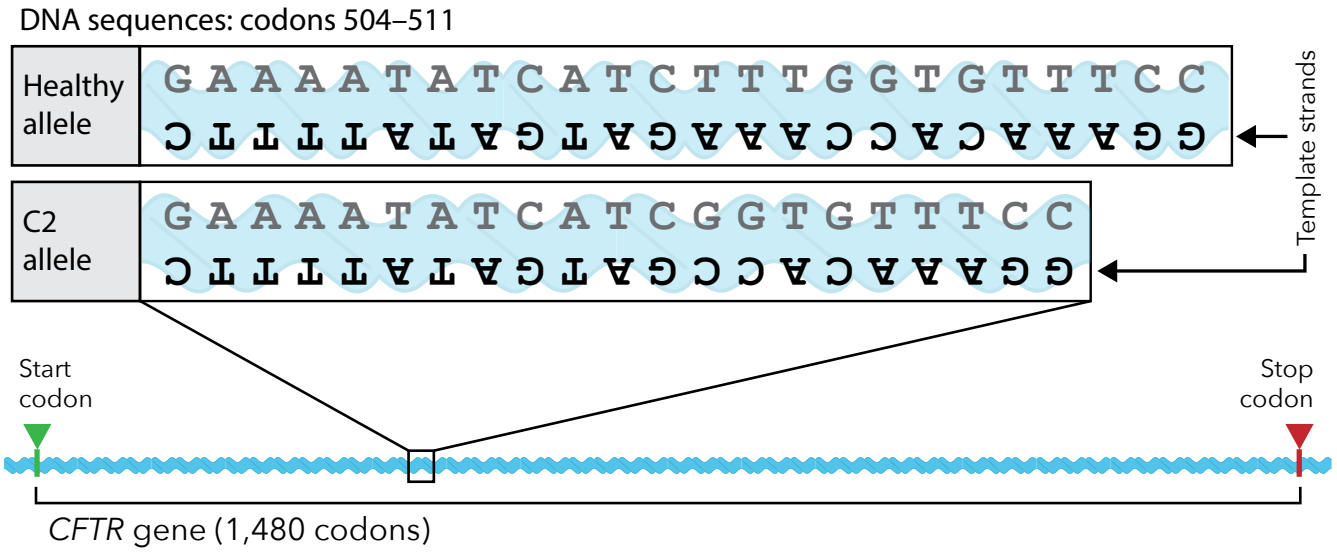
Allele – **C2**

There are more than 1,200 versions, or alleles, of the *CFTR* gene. Some cause genetic disorders and some do not. Your assigned allele is one of a few hundred that cause cystic fibrosis.

Mutations & Alleles

The protein-coding portion of the *CFTR* gene is 4,440 nucleotides long, and it has 1,480 codons.

The DNA sequence of your allele is identical to a healthy allele for most of its length. The place where they differ is shown in detail:

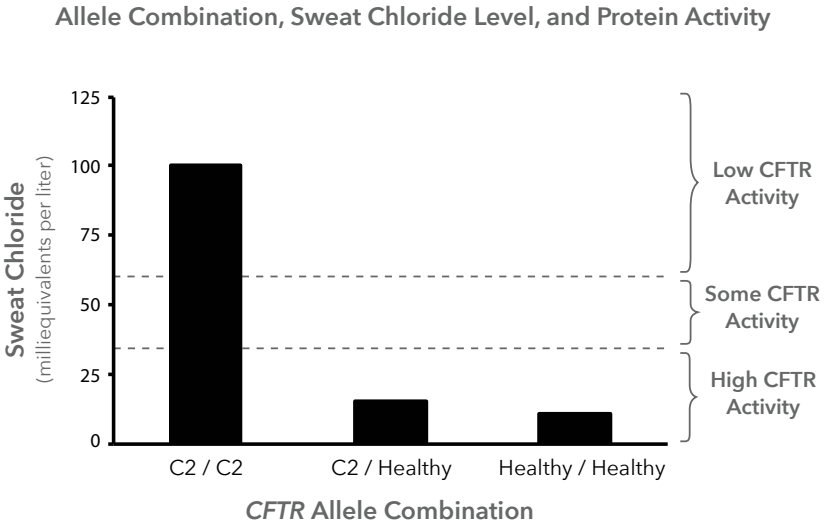


NAME _____ DATE _____

Inheritance

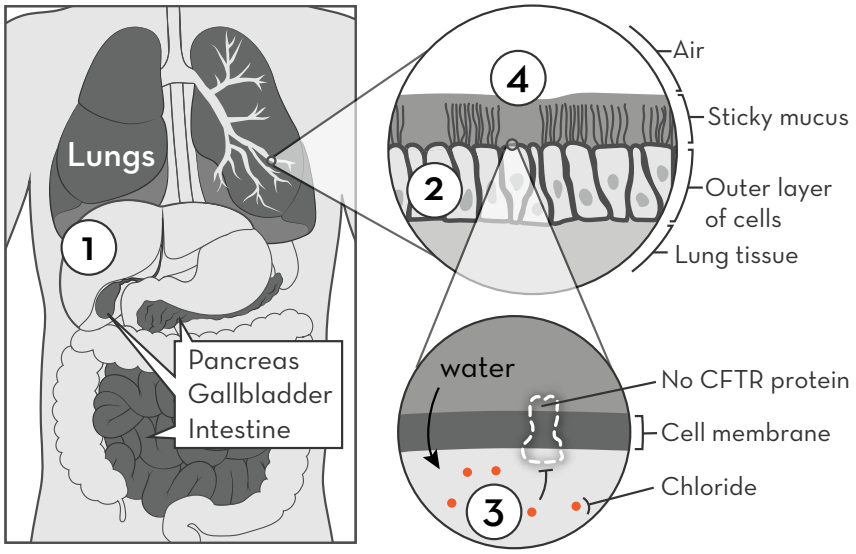
Everyone inherits two alleles of the *CFTR* gene, and CFTR protein is normally made (expressed) from both. Once it is made, CFTR protein goes to the plasma membrane. Here, it moves chloride ions (from salt) from inside the cell to the outside.

The graph shows the average sweat chloride level from many people with each allele combination. A sweat test measures how well a person's CFTR proteins are working. If CFTR proteins are working well, chloride levels are low. In people with cystic fibrosis, chloride levels are high.



Protein Function & Expression

1. The C2 allele is switched on in cells that line the lungs and digestive organs (same as healthy alleles).
2. Cells read the C2 allele and build CFTR protein. However, the protein is altered. It isn't processed correctly, and it is broken down inside the cell soon after it's made.
3. The CFTR protein never gets to the cell membrane, and it cannot move chloride ions.
4. People with two C2 alleles have cystic fibrosis. They usually have thick mucus in the airways of their lungs, and their digestive organs do not work properly.



NAME _____ DATE _____

Allele Profile

Cystic fibrosis, Allele C3

Basic Information

More information at Learn.Genetics.utah.edu/content/genetics/cysticfibrosis/

Genetic Disorder – **Cystic fibrosis**

Affected Gene – ***CFTR***

Affected Protein – The affected gene codes for the protein **cystic fibrosis transmembrane conductance regulator (CFTR)**

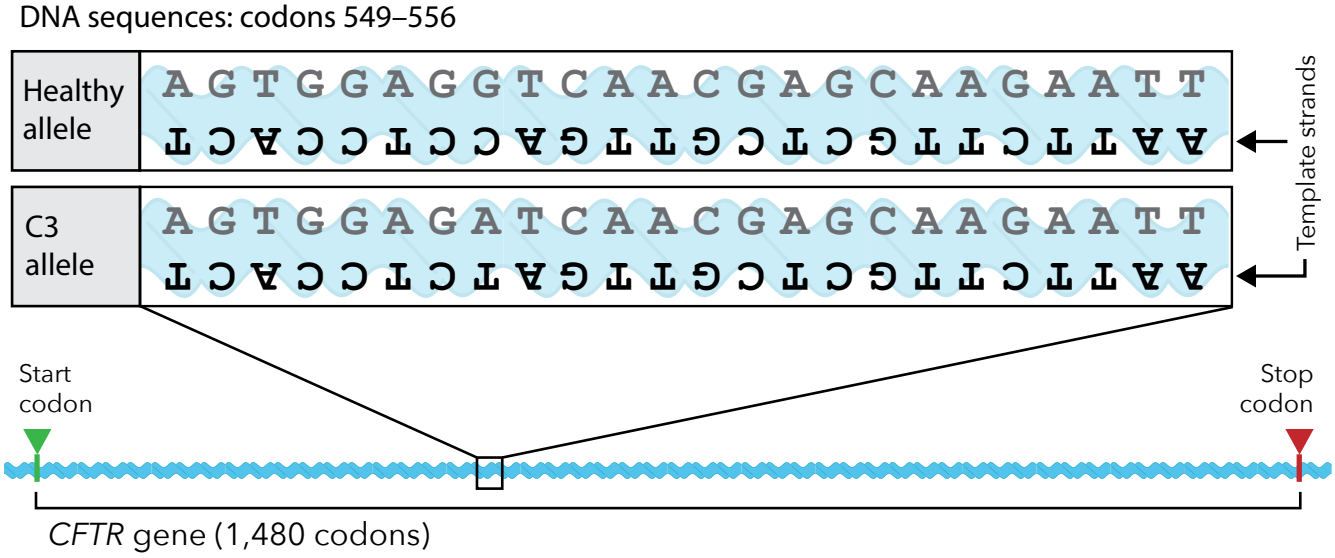
Allele – **C3**

There are more than 1,200 versions, or alleles, of the *CFTR* gene. Some cause genetic disorders and some do not. Your assigned allele is one of a few hundred that cause cystic fibrosis.

Mutations & Alleles

The protein-coding portion of the *CFTR* gene is 4,440 nucleotides long, and it has 1,480 codons.

The DNA sequence of your allele is identical to a healthy allele for most of its length. The place where they differ is shown in detail:



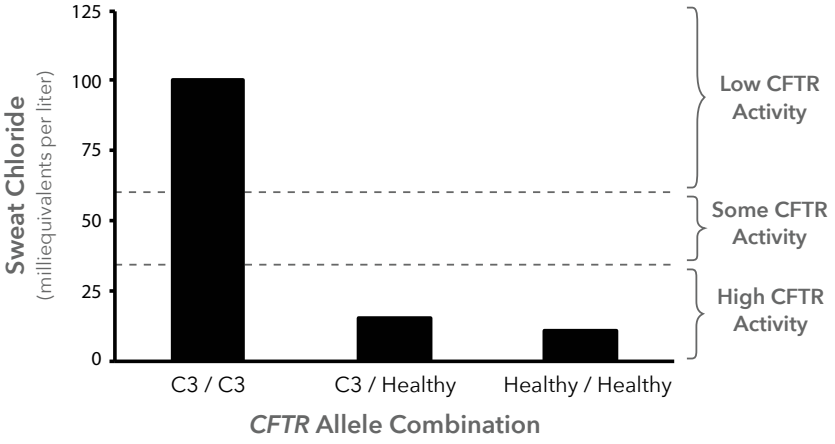
NAME _____ DATE _____

Inheritance

Everyone inherits two alleles of the *CFTR* gene, and CFTR protein is normally made (expressed) from both. Once it is made, CFTR protein goes to the plasma membrane. Here, it moves chloride ions (from salt) from inside the cell to the outside.

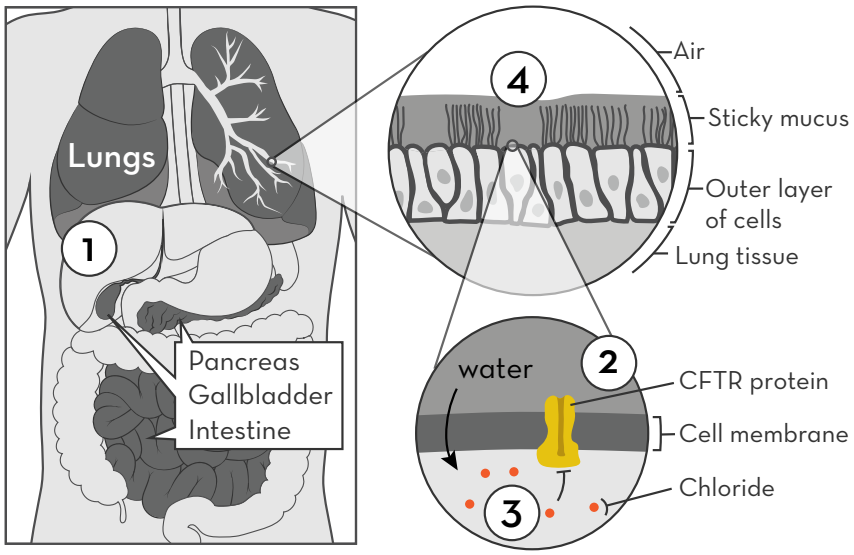
The graph shows the average sweat chloride level from many people with each allele combination. A sweat test measures how well a person's CFTR proteins are working. If CFTR proteins are working well, chloride levels are low. In people with cystic fibrosis, chloride levels are high.

Allele Combination, Sweat Chloride Level, and Protein Activity



Protein Function & Expression

1. The C3 allele is switched on in cells that line the lungs and digestive organs (same as healthy alleles).
2. Cells read the C3 allele and build CFTR protein, and the protein moves to the plasma membrane.
3. But the protein is altered: the opening that chloride ions would normally flow through is blocked. Few ions get through.
4. People with two C3 alleles have cystic fibrosis. They usually have thick mucus in the airways of their lungs, and their digestive organs do not work properly.



NAME _____ DATE _____

Allele Profile

Cystic fibrosis, Allele C4

Basic Information

More information at Learn.Genetics.utah.edu/content/genetics/cysticfibrosis/

Genetic Disorder – **Cystic fibrosis**

Affected Gene – ***CFTR***

Affected Protein – The affected gene codes for the protein **cystic fibrosis transmembrane conductance regulator (CFTR)**

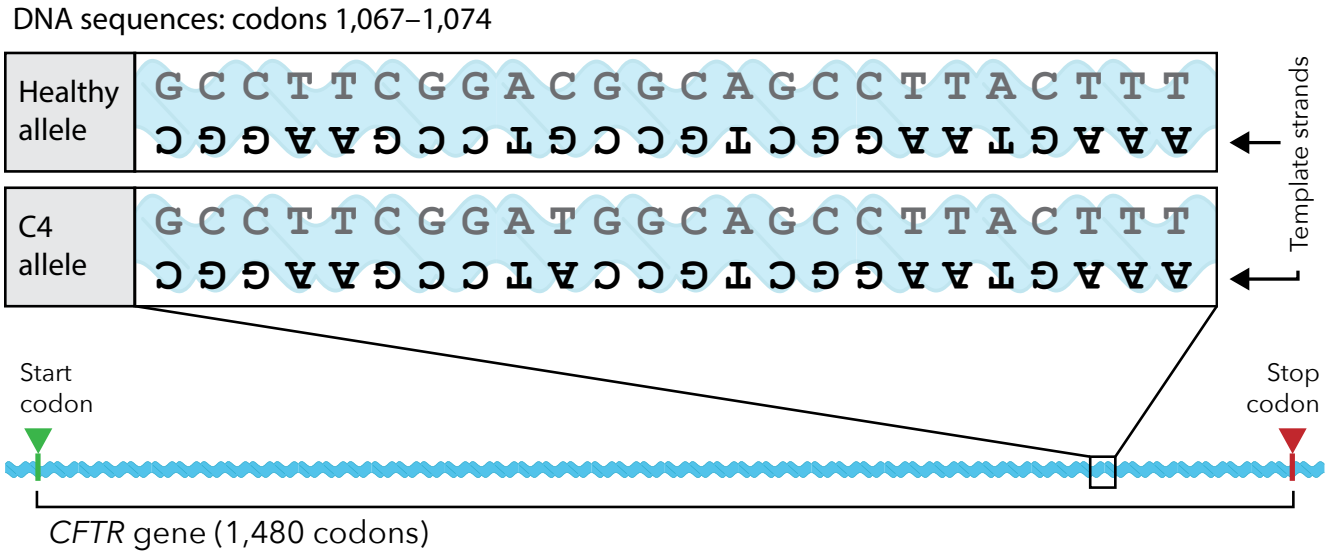
Allele – **C4**

There are more than 1,200 versions, or alleles, of the *CFTR* gene. Some cause genetic disorders and some do not. Your assigned allele is one of a few hundred that cause cystic fibrosis.

Mutations & Alleles

The protein-coding portion of the *CFTR* gene is 4,440 nucleotides long, and it has 1,480 codons.

The DNA sequence of your allele is identical to a healthy allele for most of its length. The place where they differ is shown in detail:

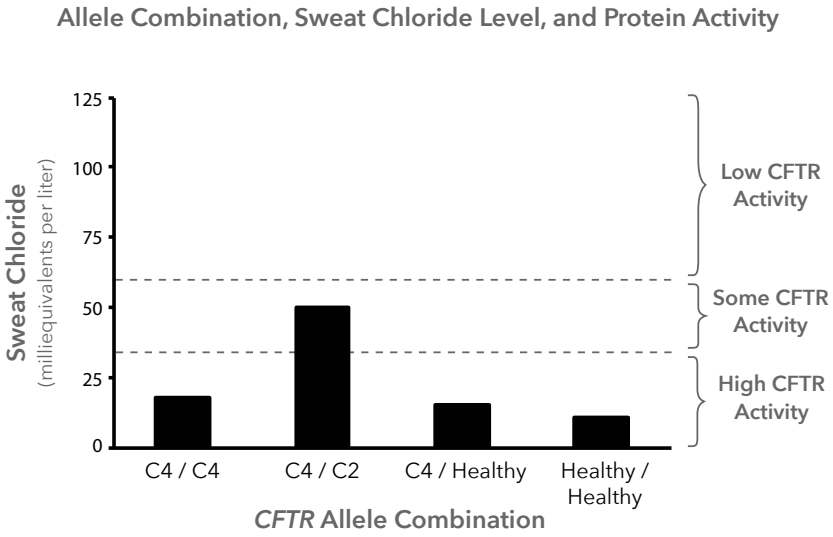


NAME _____ DATE _____

Inheritance

Everyone inherits two alleles of the *CFTR* gene, and CFTR protein is normally made (expressed) from both. Once it is made, CFTR protein goes to the plasma membrane. Here, it moves chloride ions (from salt) from inside the cell to the outside.

Alleles of the *CFTR* gene can have mild or severe effects on how well the protein they code for works. Your allele is mild. Even having two copies rarely causes disease. But, if a mild allele is combined with a very severe allele, like C2, it can cause cystic fibrosis.

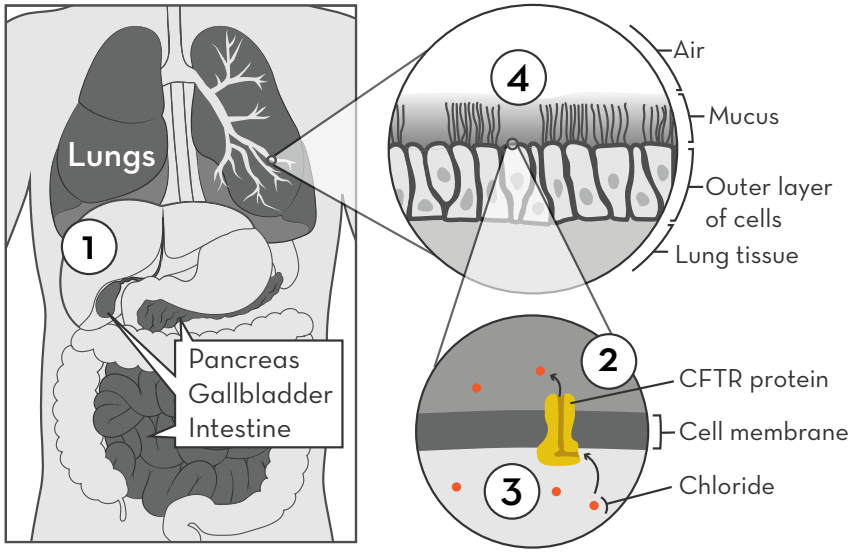


The graph shows the average sweat chloride level from many people with each allele combination. A sweat test measures how well a person's CFTR proteins are working. If CFTR proteins are working well, chloride levels are low. In people with cystic fibrosis, chloride levels are high.

In your Lab Notebook, Page 4, Question 7: Draw a model for a person who has one copy of your assigned allele in combination with a C2 allele.

Protein Function & Expression

1. The C4 allele is switched on in cells that line the lungs and digestive organs (same as healthy alleles).
2. Cells read the C4 allele and build CFTR protein. But the protein is altered. The cell has trouble processing it, and less protein than normal makes it to the plasma membrane.
3. The protein that does make it to the plasma membrane doesn't move chloride ions well. There are fewer proteins than normal, each doing less work.



4. People with the C4 allele may or may not have cystic fibrosis. This is true even if their second allele is one that causes a severe form of the disease (like C2). If a person with the C4 allele does have cystic fibrosis, their symptoms are usually mild. Mucus may affect the lungs, but other organs are usually not affected.

NAME _____ DATE _____

Allele Profile

Cystic fibrosis, Allele C5

Basic Information

More information at Learn.Genetics.utah.edu/content/genetics/cysticfibrosis/

Genetic Disorder – **Cystic fibrosis**

Affected Gene – ***CFTR***

Affected Protein – The affected gene codes for the protein **cystic fibrosis transmembrane conductance regulator (CFTR)**

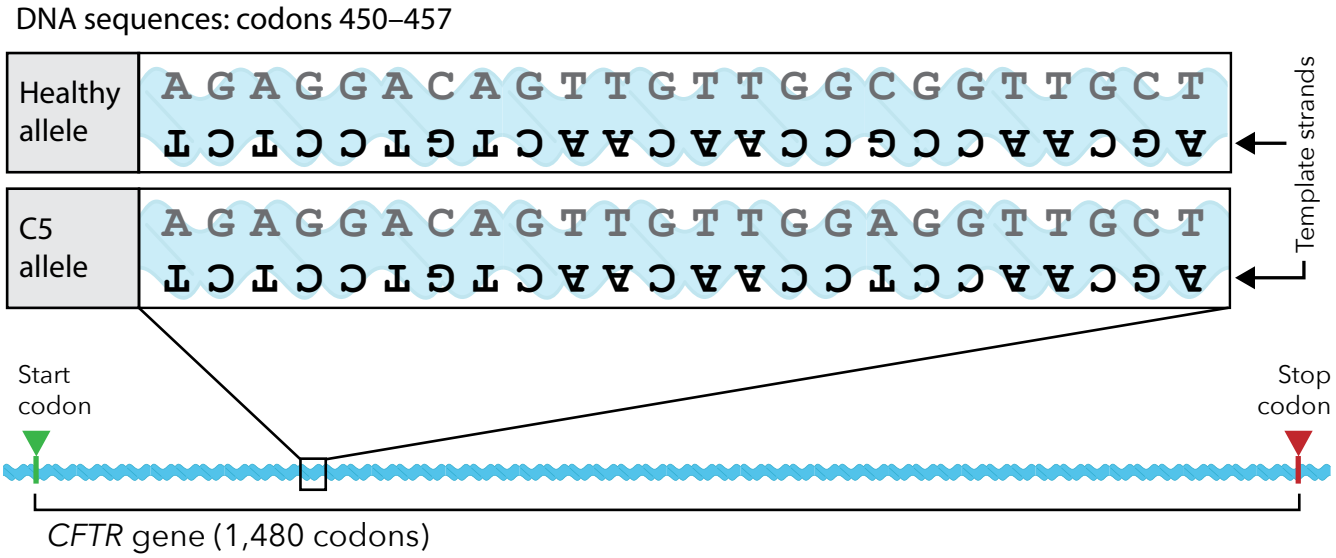
Allele – **C5**

There are more than 1,200 versions, or alleles, of the *CFTR* gene. Some cause genetic disorders and some do not. Your assigned allele is one of a few hundred that cause cystic fibrosis.

Mutations & Alleles

The protein-coding portion of the *CFTR* gene is 4,440 nucleotides long, and it has 1,480 codons.

The DNA sequence of your allele is identical to a healthy allele for most of its length. The place where they differ is shown in detail:

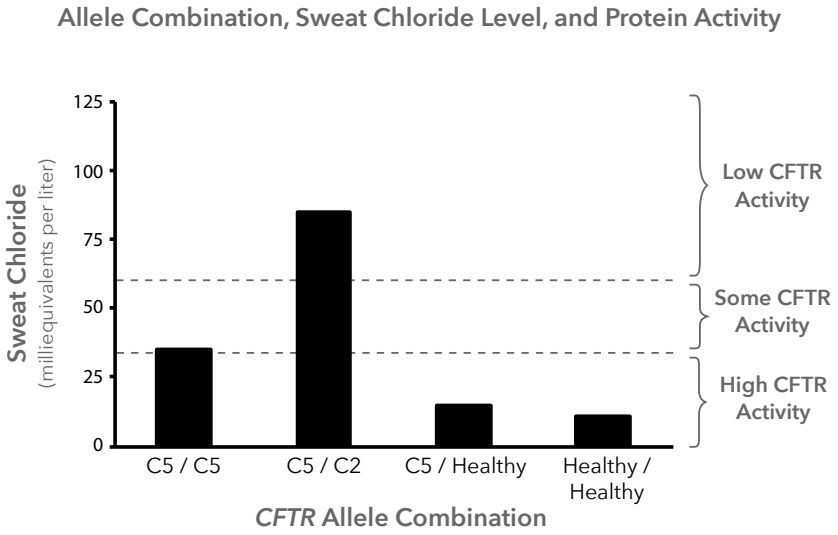


NAME _____ DATE _____

Inheritance

Everyone inherits two alleles of the *CFTR* gene, and CFTR protein is normally made (expressed) from both. Once it is made, CFTR protein goes to the plasma membrane. Here, it moves chloride ions (from salt) from inside the cell to the outside.

Alleles of the *CFTR* gene can have mild or severe effects on how well the protein they code for works. Your allele is mild. Even having two copies rarely causes disease. But, if a mild allele is combined with a very severe allele, like C2, it can cause cystic fibrosis.



The graph shows the average sweat chloride level from many people with each allele combination. A sweat test measures how well a person's CFTR proteins are working. If CFTR proteins are working well, chloride levels are low. In people with cystic fibrosis, chloride levels are high.

In your Lab Notebook, Page 4, Question 7: Draw a model for a person who has one copy of your assigned allele in combination with a C2 allele.

Protein Function & Expression

1. The C5 allele is switched on in cells that line the lungs and digestive organs (same as healthy alleles).
2. Cells read the C5 allele and build CFTR protein. But the protein is altered. The cell has trouble processing it, and much of it is broken down inside the cell.
3. The protein that does make it to the plasma membrane can do its job. But there is far less protein than normal.
4. People with C5 and another disease-causing allele (like C2) nearly always have cystic fibrosis, but their symptoms are typically milder than usual. Mucus may affect the lungs, but it usually does not block the ducts of the pancreas or gallbladder. Effects on digestion are mild.