

Name:

Date:

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Information Gathering Worksheet **Answer Key Cystic Fibrosis**

Which genetic disorder are you researching? Cystic Fibrosis

Be sure to cite your sources and use reliable sources throughout.

What are the symptoms of this disorder?

People with CF can have a variety of symptoms, including:

- Very salty-tasting skin.
- Persistent coughing, at times with phlegm.
- Frequent lung infections, including pneumonia or bronchitis.
- Wheezing or shortness of breath.
- Poor growth or weight gain despite a good appetite.
- Frequent greasy, bulky stools or difficulty with bowel movements.
- Nasal polyps.
- Chronic sinus infections.
- Clubbing or enlargement of the fingertips and toes.
- Rectal prolapse.
- Male infertility.

Source: <https://www.cff.org/intro-cf/about-cystic-fibrosis>

How common is this disorder?

There are close to 40,000 children and adults living with cystic fibrosis in the United States (and an estimated 105,000 people across 94 countries), and CF can affect people of every racial and ethnic group.

Source: <https://www.cff.org/intro-cf/about-cystic-fibrosis>

Which specific cells, tissues, and organs are affected by this disorder?

In people with CF, mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene cause the CFTR protein to become dysfunctional. When the protein is not working correctly, it is unable to help move chloride—a component of salt—to the cell surface. Without the chloride to attract water to the cell surface, the mucus in various organs becomes thick and sticky.

In the lungs, the mucus clogs the airways and traps germs such as bacteria, leading to infections, inflammation, respiratory failure, and other complications. For this reason, avoiding germs is a top concern for people with CF.

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In the pancreas, the buildup of mucus prevents the release of digestive enzymes that help the body absorb food and key nutrients, resulting in malnutrition and poor growth. In the liver, the thick mucus can block the bile duct, causing liver disease. CF can affect men's ability to have children.

Source: <https://www.cff.org/intro-cf/about-cystic-fibrosis>

If applicable, what are the current treatments? How successful are they?

The types of CF symptoms and how severe they are can differ widely from person to person. Therefore, although treatment plans can contain many of the same elements, they are tailored to each person's unique needs.

Each day, people with CF complete a combination of the following therapies:

- Airway clearance to help loosen and get rid of the thick mucus that can build up in the lungs.
- Inhaled medicines to open the airways or thin the mucus. These are liquid medicines that are made into a mist or aerosol and then inhaled through a nebulizer and include antibiotics to fight lung infections and therapies to help keep the airways clear.
- Pancreatic enzyme supplement capsules to improve the absorption of vital nutrients. These supplements are taken with every meal and most snacks. People with CF also usually take multivitamins.
- An individualized fitness plan to help improve energy, lung function, and overall health.
- CFTR modulators to target the underlying defect in the CFTR protein. Because different mutations cause different defects in the protein, the medications that have been developed so far are effective only in people with specific mutations.

There is no cure for CF, but with current therapies patients commonly live into their 50s and some even into their 80s.

Source: <https://www.cff.org/intro-cf/about-cystic-fibrosis>

What specific gene is mutated in people with this disorder?

The cystic fibrosis transmembrane conductance regulator (CFTR) gene.

Source:

[https://pubmed.ncbi.nlm.nih.gov/26857764/#:~:text=Cystic%20fibrosis%20\(CF\)%20is%20a,conductance%20regulator%20protein%20\(CFTR\)](https://pubmed.ncbi.nlm.nih.gov/26857764/#:~:text=Cystic%20fibrosis%20(CF)%20is%20a,conductance%20regulator%20protein%20(CFTR))

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Is this mutation dominant or recessive?

CF is an autosomal recessive disorder.

Source:

[https://pubmed.ncbi.nlm.nih.gov/26857764/#:~:text=Cystic%20fibrosis%20\(CF\)%20is%20a,conductance%20regulator%20protein%20\(CFTR\)](https://pubmed.ncbi.nlm.nih.gov/26857764/#:~:text=Cystic%20fibrosis%20(CF)%20is%20a,conductance%20regulator%20protein%20(CFTR))

What specific mutation causes this disorder?
List the healthy and mutated DNA sequence below.
Include at least 15 nucleotides before and after the mutation.

There are many mutations in the CFTR gene that can cause CF. The most common CF mutation is the F508del, which removes a single amino acid from the CFTR protein.

Healthy DNA sequence:

5' GGCACCATTAAAGAAAATATCAT**CTTT**GGTGTTCCTATGAT 3'

Mutated DNA sequence:

5' GGCACCATTAAAGAAAATATC**ATT**GGTGTTCCTATGAT 3'

Source: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3786104/>
<https://www.ncbi.nlm.nih.gov/snp/rs113993960>