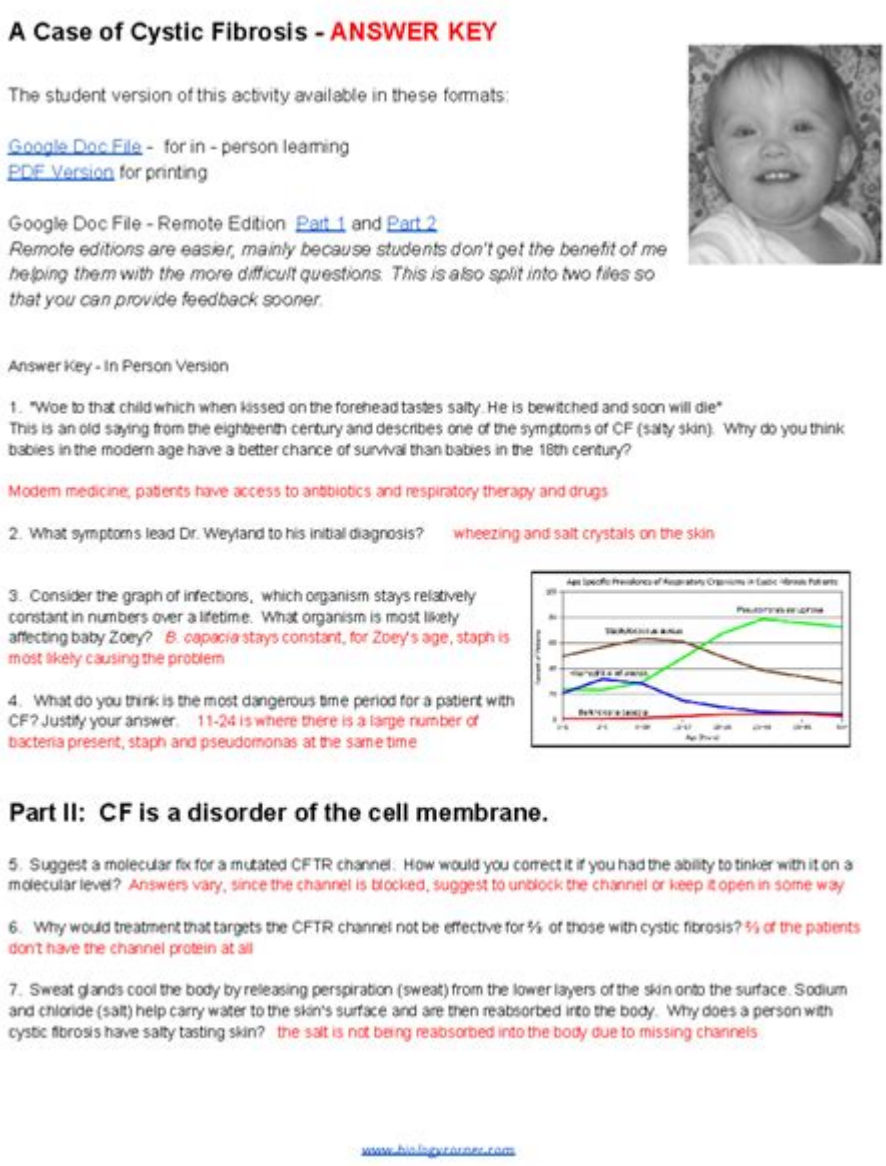
A Case Of Cystic Fibrosis Answer Key



A Case of Cystic Fibrosis - ANSWER KEY

The student version of this activity available in these formats:

Google Doc File - for in - person learning
PDF Version for printing

Google Doc File - Remote Edition Part 1 and Part 2
Remote editions are easier, mainly because students don't get the benefit of me helping them with the more difficult questions. This is also split into two files so that you can provide feedback sooner.

Answer Key - In Person Version

1. "Woe to that child which when kissed on the forehead tastes salty. He is bewitched and soon will die"
This is an old saying from the eighteenth century and describes one of the symptoms of CF (salty skin). Why do you think babies in the modern age have a better chance of survival than babies in the 18th century?

Modern medicine, patients have access to antibiotics and respiratory therapy and drugs

2. What symptoms lead Dr. Weyland to his initial diagnosis? wheezing and salt crystals on the skin

3. Consider the graph of infections, which organism stays relatively constant in numbers over a lifetime. What organism is most likely affecting baby Zoey? *B. capacia* stays constant, for Zoey's age, staph is most likely causing the problem

4. What do you think is the most dangerous time period for a patient with CF? Justify your answer. 11-24 is where there is a large number of bacteria present, staph and pseudomonas at the same time

Part II: CF is a disorder of the cell membrane.

5. Suggest a molecular fix for a mutated CFTR channel. How would you correct it if you had the ability to tinker with it on a molecular level? Answers vary, since the channel is blocked, suggest to unblock the channel or keep it open in some way

6. Why would treatment that targets the CFTR channel not be effective for ⅓ of those with cystic fibrosis? ⅓ of the patients don't have the channel protein at all

7. Sweat glands cool the body by releasing perspiration (sweat) from the lower layers of the skin onto the surface. Sodium and chloride (salt) help carry water to the skin's surface and are then reabsorbed into the body. Why does a person with cystic fibrosis have salty tasting skin? the salt is not being reabsorbed into the body due to missing channels

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A case of cystic fibrosis answer key is essential for understanding the complexities of this genetic disorder. Cystic fibrosis (CF) is a life-threatening condition that affects the lungs, digestive system, and other organs, primarily due to the mutations in the CFTR gene that encodes a protein that regulates salt and water transport in cells. This article will provide an in-depth look at cystic fibrosis, including its pathophysiology, symptoms, diagnosis, treatment options, and the importance of genetic counseling and support systems.

Understanding Cystic Fibrosis

Cystic fibrosis is an autosomal recessive disorder primarily caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. This gene is responsible for the production of a channel protein that helps regulate the movement of chloride ions in and out of cells. When this

protein is dysfunctional due to mutations, it leads to the production of thick and sticky mucus that obstructs various organs, most notably the lungs and pancreas.

Pathophysiology of Cystic Fibrosis

The pathophysiology of cystic fibrosis is multifaceted and involves several key processes:

1. Mucus Production: The primary defect in CF is the production of abnormally thick mucus due to impaired chloride and bicarbonate transport. This results in mucus accumulation in the airways and other organs.

2. Respiratory Complications: The thick mucus obstructs airways, making it difficult for patients to breathe. It also creates an environment conducive to bacterial infections, leading to chronic lung infections and inflammation.

3. Digestive Issues: In the pancreas, thick mucus can block the release of digestive enzymes, causing malabsorption of nutrients. This can lead to poor growth and weight gain in affected individuals.

4. Sweat Gland Dysfunction: CFTR mutations also affect sweat glands, leading to higher concentrations of salt in sweat. This is often one of the first indicators of the disease in newborns.

Symptoms of Cystic Fibrosis

Cystic fibrosis manifests through a variety of symptoms that can vary significantly among individuals. Common symptoms include:

- Respiratory Symptoms:
- Persistent cough
- Frequent lung infections
- Wheezing or shortness of breath
- Sinus infections

- Digestive Symptoms:
- Difficulty gaining weight and growing
- Frequent, greasy, and bulky stools
- Intestinal blockages, especially in newborns (meconium ileus)

- Other Symptoms:
- Salty tasting skin
- Clubbing of fingers and toes (widening and rounding of the nails)
- Diabetes (CF-related diabetes is common)

Diagnosis of Cystic Fibrosis

Diagnosing cystic fibrosis typically involves a combination of clinical evaluations, family history

assessment, and laboratory tests. The following steps outline the diagnostic process:

1. Newborn Screening: Most countries perform routine newborn screening for cystic fibrosis using a blood test to measure immunoreactive trypsinogen (IRT) levels. Elevated IRT levels may indicate CF.

2. Sweat Test: If initial screening suggests CF, a sweat test is performed to measure the concentration of chloride in sweat. A chloride level greater than 60 mmol/L is indicative of cystic fibrosis.

3. Genetic Testing: Genetic testing can confirm the diagnosis by identifying mutations in the CFTR gene. This test can also help in understanding the specific type of mutation, which can influence treatment options and prognosis.

4. Pulmonary Function Tests: These tests assess lung function and can help monitor the progression of lung disease in CF patients.

Treatment Options for Cystic Fibrosis

While there is currently no cure for cystic fibrosis, advancements in treatment have significantly improved the quality of life and life expectancy for those affected. Treatment is typically multidisciplinary, involving various specialists. Key components of treatment include:

Respiratory Management

1. Airway Clearance Techniques: These techniques help loosen and clear mucus from the lungs. Methods include:
- Chest physiotherapy
- High-frequency chest wall oscillation (the vest therapy)
- Breathing exercises

2. Medications:
- Bronchodilators: To open airways and improve airflow.
- Mucolytics: Such as dornase alfa (Pulmozyme) to thin mucus.
- Antibiotics: To treat or prevent lung infections.

3. CFTR Modulators: These are newer medications that target the underlying defect in the CFTR protein, improving its function. Examples include ivacaftor, lumacaftor, and tezacaftor.

Nutritional Support

1. Pancreatic Enzyme Replacement Therapy (PERT): Since CF can inhibit the release of digestive enzymes, patients often require enzyme supplements to aid in digestion and nutrient absorption.

2. Nutritional Counseling: A high-calorie, high-fat diet is often recommended to ensure adequate growth and development, along with vitamin supplementation (especially fat-soluble vitamins A, D,

E, and K).

Psychosocial Support

Living with cystic fibrosis can be challenging, both physically and emotionally. Support systems are vital:

- Counseling Services: Mental health professionals can help patients and families cope with the emotional burden of chronic illness.
- Support Groups: Connecting with others who have CF can provide encouragement and shared experiences.

Importance of Genetic Counseling

Genetic counseling is crucial for families affected by cystic fibrosis. Understanding the genetic basis of the disorder can help families make informed decisions regarding family planning and the implications of CF in future pregnancies.

1. Carrier Testing: Genetic counseling can provide information on the likelihood of being a CF carrier and the implications for offspring.

2. Family Support: Counselors can guide families on managing the challenges of living with a chronic condition and help them access resources.

Conclusion

Cystic fibrosis is a complex genetic disorder that requires a comprehensive approach for effective management. With advancements in treatment and a multidisciplinary approach to care, individuals with cystic fibrosis can lead fulfilling lives. It is crucial for affected individuals and their families to seek out support, stay informed about their condition, and remain proactive in their healthcare. As research continues to evolve, the hope for a cure remains steadfast in the cystic fibrosis community.

Frequently Asked Questions

What is cystic fibrosis?

Cystic fibrosis is a genetic disorder that affects the respiratory, digestive, and reproductive systems due to the production of thick, sticky mucus.

What causes cystic fibrosis?

Cystic fibrosis is caused by mutations in the CFTR gene, which encodes a protein that regulates salt and water transport in cells.

How is cystic fibrosis diagnosed?

Cystic fibrosis is diagnosed through a combination of newborn screening, sweat tests to measure chloride levels, and genetic testing for CFTR mutations.

What are common symptoms of cystic fibrosis?

Common symptoms include persistent cough, frequent lung infections, difficulty breathing, poor growth, and digestive issues due to pancreatic dysfunction.

What treatments are available for cystic fibrosis?

Treatments include airway clearance techniques, inhaled medications, pancreatic enzyme supplements, and in some cases, lung transplantation.

How does cystic fibrosis affect life expectancy?

Advancements in treatment have significantly improved life expectancy for individuals with cystic fibrosis, with many living into their 30s and beyond.

Are there any new therapies for cystic fibrosis?

Yes, new therapies like CFTR modulators have been developed to target the underlying cause of cystic fibrosis and improve lung function.

Can cystic fibrosis be cured?

Currently, there is no cure for cystic fibrosis, but ongoing research aims to find more effective treatments and potential cures.

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