TEACHER'S NOTES
WHAT IS C.F?

OVERVIEW

Aimed at key stage 4 pupils. This is a simple comprehension activity for pupils to consider the main features of cystic fibrosis (CF) and the recessive pattern of inheritance.

LEARNING OBJECTIVES

- To review the consequence of gene mutations on the protein formed
- To understand the symptoms and treatments for CF
- To review how recessively inherited conditions are passed on

CURRICULUM LINKS

- **KS4**: The ways in which organisms function are related to the genes in their cells
- **KS4**: Human health is affected by a range of environmental and inherited factors, by the use and misuse of drugs and by medical treatments

Activity

- Introduce the genetic condition cystic fibrosis
- Show the film Ryan's story on **www.genesareus.org**
- Give pupils the worksheet to complete

ANSWERS

1. Which chromosome is the CFTR gene on?

7

2. How many DNA letters make up the CFTR gene?

Over 180,000

3. Where do you find the CFTR protein in a cell?

In the cell membrane

4. What are the symptoms of CF?

CF mainly affects the lungs and digestive system by clogging them with thick, sticky mucus. In the lungs the mucus makes people vulnerable to serious chest infections, while in the digestive system it often stops their pancreas releasing digestive enzymes. These are the main symptoms of CF, but there are a number of other problems that can occur.

5. What treatments do people with CF need?

A number of different types of treatment are usually needed:

1. Physiotherapy is recommended every day to prevent the thick, sticky mucus from blocking the lungs. It helps to reduce infection and prevent lung damage.

2. Antibiotics are often taken to prevent or control bacterial infections.

3. The majority of people with CF need help digesting food, so enzyme pills need to be taken with every meal.

4. Other medications are recommended for specific problems, for example, steroids to reduce inflammation and drugs to open the airways and thin the mucus.

People with CF are also given further general advice to do exercise, which will help their lung function, and also eat a high calorie diet to give them the energy they need.

6. How many carriers of CF do you think there are in the UK?

c) Over 2 million

7. If two carriers of CF have a baby, what is the chance of that baby being affected by CF?

b) 1 in 4

8. If two carriers of CF have a baby, what is the chance of that baby being a carrier of CF?

a) 1 in 2

9. If only one parent is a carrier of CF, what is the chance of that baby being affected by CF?

c) 0

ANSWERS continued

10. Imagine that you are talking to the parents who had been told that their baby has CF. How would you explain what genetic conditions are?

Answers might mention that there are lots of different genetic conditions and that they are caused by changes in DNA. DNA is inside most of the cells of our body to help us know how to grow and function. If the DNA has been changed, it can affect how the body works.

Children inherit DNA from their parents and sometimes, by chance, the child inherits a combination of genes that means that they will experience an illness.

In the case of CF, the baby has inherited a gene change from both parents that causes the condition.

The condition is not caused by anything done by the parents or by any infections or complications from the pregnancy. It is a chance event that means the baby has inherited two copies of the gene change and without a working copy of the gene they will experience the symptoms of CF.

FURTHER information

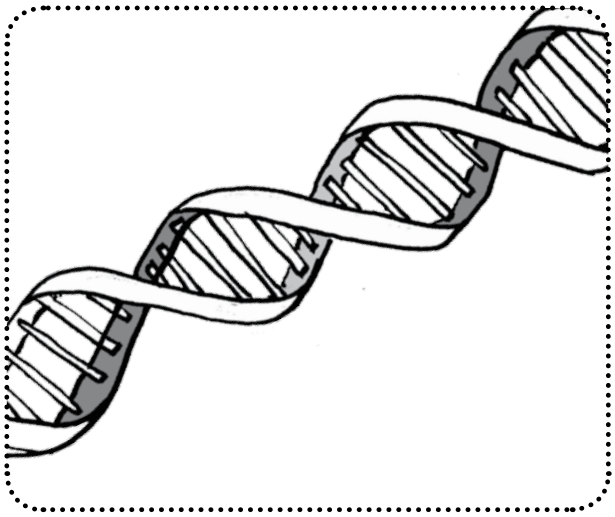
- The CF Trust provides excellent information about the condition
  www.cftrust.org
- American website called 'Your Genes, Your Health' has animations and films to explain the cause of CF, inheritance, etc. It is pitched above GCSE- standard but would be accessible to able pupils
  www.ygyh.org
- The 'Changing Futures' website has been made with teenagers affected by CF and the Nowgen team to explain aspects of the condition and explore the potential of gene therapy
  www.changing-futures.co.uk.

STUDENT WORKSHEET

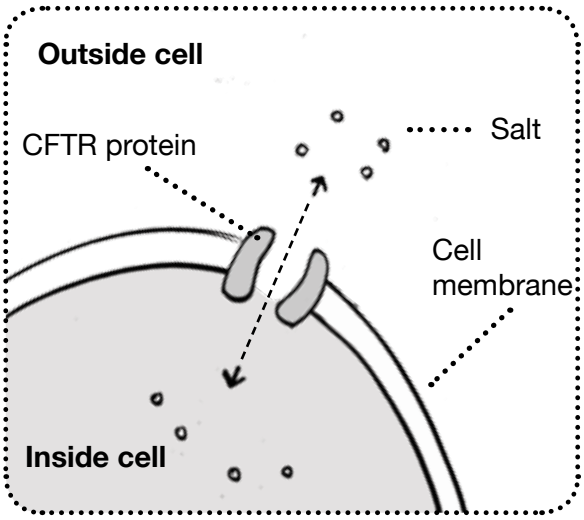
WHAT IS CF?

Cystic fibrosis (CF) is a genetic condition, which means that it is caused by inherited changes in someone's DNA. It is one of the most common genetic conditions in the UK, with over 9,000 people affected. It is caused by changes to a gene on chromosome 7 called *CFTR* (its full name is the 'cystic fibrosis transmembrane conductance regulator'.)

The *CFTR* gene is a section of DNA. There over 180,000 letters of DNA in this gene. This encodes the CFTR protein.



The CFTR protein forms a channel that sits in the cell membrane. This channel allows salt to move in and out of some cells.



Mutations (changes) in the *CFTR* gene can cause problems. Some mutations lead to the CFTR protein not being made or not working properly. The symptoms of CF are caused by this channel not working.

1 ***Which chromosome is the* CFTR *gene on?***

2 ***How many DNA letters make up the* CFTR *gene?***

3 ***Where do you find the* CFTR *protein in a cell?***

4 ***What are the symptoms of CF?***

CF is highly variable. When a child is diagnosed it is impossible to tell parents exactly how seriously their child will be affected, but the same parts of the body are generally affected, namely the lungs and digestive system. New treatments are improving the outlook for patients with CF, but there is currently no cure, which is hard to cope with.

5 ***What treatments do people with CF need?***

Genetic conditions can be inherited in different ways and CF is inherited in a recessive pattern. This means that someone affected by CF has changes in both copies of their *CFTR* gene (we have two copies of each of our genes, one from our mum and one from our dad.) Our genes come in pairs and sometimes we only need one working copy of the gene to be healthy. If someone has just one copy of a changed version of the *CFTR* gene, they will not be affected by CF. They are known as 'carriers'.

6 ***How many carriers for CF do you think there are in the UK?***

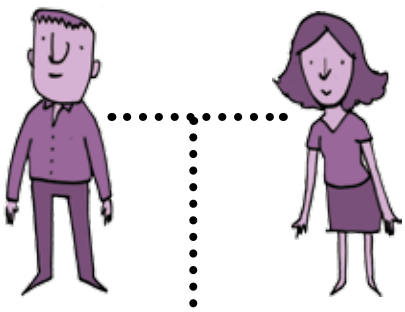
a) Over 250,000
b) Approximately 800,000
c) Over 2 million

WHAT IS CF?

THE INHERITANCE OF CF

Dad is a carrier of CF. One of his copies of the *CFTR* gene has a mutation that causes CF

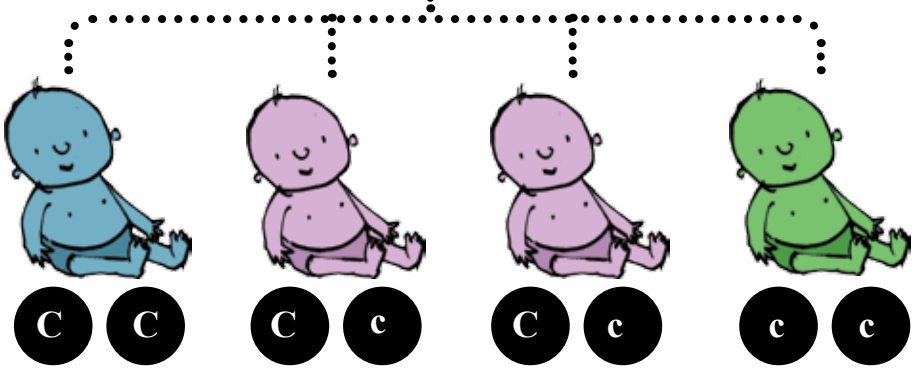
C c



Mum is a carrier of CF. One of her copies of the *CFTR* gene has a mutation that causes CF

C c

When they have a baby there is an equal chance of the following possibilities…



C C | C c | C c | c c

It's possible that a baby could inherit two copies of the mutated version of the *CFTR* gene.

KEY

	Carrier of CF
	Affected by CF
	Neither carrier of CF nor affected by CF
C	Allele for *CFTR* gene without mutation
c	Allele for *CFTR* gene with mutation

7 ***If two carriers of CF have a baby, what is the chance of that baby being affected by CF?***
a) 1 in 2
b) 1 in 4
c) 1 in 25

8 ***If two carriers of CF have a baby, what is the chance of that baby being a carrier of CF?***
a) 1 in 2
b) 1 in 4
c) 1 in 25

9 ***If only one parent is a carrier of CF, what is the chance of that baby being affected by CF?***
a) 1 in 25
b) 1 in 100
c) 0

All newborn babies are screened for CF in the UK, which allows families to receive expert help and access to the best treatments. Often the baby will be the only person in the family affected by CF. This news can be huge shock for families and it can be difficult for families to come to terms with this new diagnosis.

10 ***Imagine that you are talking to the parents who had been told that their baby has CF. How would you explain what genetic conditions are?***

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