



About Ryan (age 16)

Genetic disorder: Cystic Fibrosis

Likes: Football, hanging out with friends and crisps!

Dislikes: Cricket and Spiders

Home town: Cambridge

Mum: Debbie

Dad: Glenn

Siblings: Daniel (age 22), Eleanor (age 19)

CYSTIC FIBROSIS

A **genetic condition** that affects the **internal organs**, especially the **lungs** and **digestive system**, causing them to become **blocked** with **sticky mucus**.

Cystic Fibrosis (CF) is caused by an **ion channel** in the cell membrane being absent or misformed. This affects the movement of **salt and water** in and out of the cells in the body. As a result, the body's secretions become **stickier** and **thicker** than normal – hindering the way important **organs work**, especially the **lungs** and **digestive system**.

It is inherited from both parents - **autosomal recessive inheritance** - only if both parents pass on a **faulty gene** will the child be affected. Often parents are **healthy carriers**, having **one faulty gene** and **one working copy** of the gene (like Ryan's parents). It doesn't matter if the child is male or female, as either sex can be affected.

CF is one of the UK's **most common** life-threatening inherited diseases and currently affects over **9,000 people** in the UK. Each week **five babies** are born with CF.

Over **2 million people** in the UK carry one copy of the faulty gene that causes CF – around **1 in 25** of the population.

There is currently **no cure** for CF. The potential to treat the condition with **gene therapy** is currently under investigation, with the world's largest gene therapy trial for Cystic Fibrosis **launched** in the UK in **2012**.

GENETIC DISORDERS

We have two copies of each **gene**, one from each **parent**. **Genes** are made out of DNA and most contain **instructions for making proteins**.

A **genetic disorder** happens when a **change** in one or more genes causes **vital proteins** in the body to be **missing or not work properly**.

Most genetic disorders are **recessive**, occurring when someone inherits **two copies** of the faulty gene – one from each parent.

Others are **dominant**, meaning a **single copy** of

the faulty gene is enough to cause the condition.

X-linked disorders are caused by a faulty gene on the **x-chromosome**, of which **girls** have two (**XX**) and **boys** only one (**XY**). These usually **only affect boys**.

Half of all **childhood deaths** in the UK are linked to a **genetic disorder**.

Some other examples of genetic disorders include **Cystic Fibrosis**, **Sickle Cell Anaemia**, **Huntington's disease** and **Haemophilia**.

CURRICULUM LINKS

Key Stage 4 Science: The ways in which organisms function are related to the genes in their cells

Key Stage 4 Science: Human health is affected by a range of environmental and inherited factors, by the use and misuse of drugs and by medical treatments

Key Stage 4 Science: the use of contemporary scientific and technological developments and their benefits, drawbacks and risks

USEFUL LINKS

Cystic Fibrosis Trust www.cftrust.org

www.changing-futures.co.uk This website explores gene therapy and how this could be used to treat CF.

FOR MORE RESOURCES GO TO WWW.GENESAREUS.ORG

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