raises funds for Genetic Disorders UK

TEACHER FACTSHEET: LUKE'S STORY

About Luke (Age 17)

Genetic disorder: Luke's Mum has Huntington's disease

Likes: Socialising with friends, football, swimming

Home town: Coventry

Mum: Gaynor

Dad: Chris

Siblings: Kieran (aged 28)

HUNTINGTON'S DISEASE

- A **genetic disorder** that affects the **central nervous system** and causes progressive **degeneration** of cells in the **brain**. This leads to gradual **physical, mental and emotional changes** and slowly impairs a person's ability to **move, think, communicate** and **reason**.
- **Huntington's disease** is caused by a **faulty gene** on **chromosome 4**.
- HD affects both **men** and **women**, the vast majority of whom develop their first symptoms in adulthood, between the ages of **30 and 50**. However, the symptoms of HD can occur at **any age** from childhood to old age.
- A **child** whose parent has HD has a **50:50 chance** of inheriting the faulty gene. This is an **autosomal dominant pattern** of inheritance. **Anyone** who inherits the faulty gene will, at some stage, develop the disease.
- The **gene change** that causes HD can be detected through a test that analyses someone's **blood**. Only a small proportion of people at risk of HD (**around 10%**) decide to have the test because of the **significant implications** and **potential burden** of the results. The testing is only **occasionally done on children**. However, anyone who has a **family history of HD** can ask to see a **geneticist** to talk about the situation and to explore what options would be right for them.
- It has been estimated that between **6,500** and **8,000** people in the UK have HD.

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**.

USEFUL LINKS

Huntington's disease Association www.hda.org.uk

http://en.hdyo.org/you Created by **The Huntington's Disease Youth Organization** this website is fantastic and provides extensive information on the condition in a style suitable for teenagers

CURRICULUM LINKS

- **KS4**: Human health is affected by a range of environmental and inherited factors, by the use and misuse of drugs and by medical treatments
- **KS4**: chemical and electrical signals enable body systems to respond to internal and external changes, in order to maintain the body in an optimal state
- **KS4**: The ways in which organisms function are related to the genes in their cells
- **KS4**: the use of contemporary scientific and technological developments and their benefits, drawbacks and risks

FOR MORE RESOURCES GO TO www.GENESAREUS.ORG

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