

TEACHER FACTSHEET: ELLIE & TOM'S STORY



COCKAYNE SYNDROME

- An **accelerated ageing** condition causing:
 - Stiff joints
 - Tremors
 - Problems with hearing and eyesight
 - Mobility problems
 - Reduced growth rate
 - Learning difficulties.
- The condition is **extremely rare** – only around **30 children** in the UK are known to have it but there may be more who are **undiagnosed**.
- Babies appear **normal at birth**. **Symptoms become progressively worse** from about the age of two. Few children survive beyond their **20th birthday**.
- A '**recessive**' condition caused by inheriting **two copies** of a faulty gene – one from each parent.

USEFUL LINKS

- **Amy and friends:** www.amyandfriends.org
More about living with Cockayne Syndrome.
- **GOSH Kids:** www.childrenfirst.nhs.uk
Age-appropriate information about hospital life.
- **Kid's Health:** <http://kidshealth.org>
Learn about health and how the body works.

About Tom: 4 years old

Genetic disorder: Cockayne Syndrome

Likes: Thomas the Tank Engine, his bicycle, cooking, animals.

Dislikes: People staring or making fun of him.

Home town: Bicester, Oxford

Mum: Catherine

Dad: Richard

Unaffected siblings: Ellie (age 7)

GENETIC DISORDERS

- **Genes** are made out of **DNA** and most contain **instructions for making proteins**.
- An estimated **1 in 25 children** in the UK are born with a **genetic disorder**.
- A **genetic disorder** happens when a **change** in one or more genes causes **vital proteins** in the body to be **missing or faulty**.
- Some other examples of genetic disorders include **Cystic Fibrosis**, **Sickle Cell Anaemia**, **Huntington's Disease** and **Haemophilia**.

CURRICULUM LINKS

- **KS1 Science:** Ourselves; Human variation.
- **KS1/2 PSHE and Citizenship:** Respecting the differences between people; Community cohesion.

FOR MORE RESOURCES LIKE THESE AND TO SIGN UP FOR JEANS FOR GENES DAY, VISIT US AT WWW.JEANSFORGENES.ORG

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