



About Ashley (age 11)

Genetic disorder: Treacher Collins Syndrome

Likes: Food, music – especially Karaoke!

Dislikes: Sport

Home town: Taunton, Somerset

Mum: Louise

Dad: Michael

Siblings: Daniel (age 20), Bradley (age 7)

TREACHER COLLINS SYNDROME

A **genetic disorder** that affects the **development of the bones** and structures of the **head and neck**. It is characterised by a range of **distinctive facial features** and **severe hearing loss**.

The main characteristics of TCS are **eyes that slant down** at the outer corners, **cleft palate**, an unusually **large mouth** and **malformed or missing ears**. The extent of these abnormalities varies from person to person, but they often cause **problems with hearing, breathing and eating**.

Treacher Collins Syndrome (TCS) is caused by a **faulty gene** on **chromosome 5**.

About **one in every 50,000 babies** is born with TCS.

In about **60%** of cases there is **no family history** of the condition and the syndrome is caused by a **new change** in the gene.

The condition is **dominantly inherited**, which means an affected person has a **50 per cent chance** of passing it on to their offspring.

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 2: Understanding personal development, health and wellbeing: listen to and reflect on other people's views and feelings

Key Stage 3 Citizenship: engage with and reflect on different ideas, opinions, beliefs and values when exploring topical and controversial issues and problems

Key Stage 2 Mathematical understanding: discuss, sort and order events according to their likelihood of occurring

USEFUL LINKS

Treacher Collins Support Group
www.treachercollins.net

FOR MORE RESOURCES GO TO WWW.GENESAREUS.ORG

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