

HOW A DELETION ON CHROMOSOME 9 CAUSES CONGENITAL HYPERINSULINISM

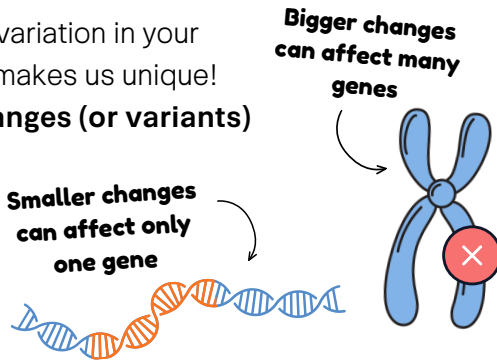


This summarises our information sheet on **congenital hyperinsulinism** (shortened to HI) **caused by a deletion on chromosome 9**. Check out the full document [here](#)!

WHAT ARE GENES, CHROMOSOMES AND GENETIC VARIATION?

Our **DNA** is our **instruction manual**, and is made up of **genes** that tell our cells what to do. Our DNA is stored in structures called **chromosomes** and we have 46 in each cell, 23 from mum and 23 from dad.

It's normal to have variation in your genes, that's what makes us unique! But sometimes **changes (or variants)** in our genes can **affect how our cells work**, causing **health problems**.



KEY MESSAGES:

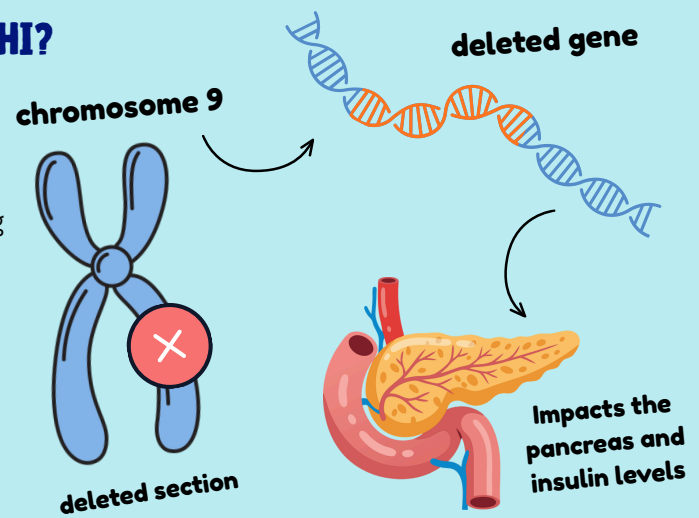
- ★ A **deletion** is where a **section of DNA is missing**, which can affect **one or more genes**
- ★ A **deletion in chromosome 9** has been linked to **HI in 16 children so far**
- ★ This form of HI is considered **rare**, but may be **underdiagnosed** as it is not routinely tested for
- ★ Most children with this form of HI are treated with **diazoxide**
- ★ People with this deletion often have **other medical conditions**, which can **vary between individuals**

HOW CAN A DELETION ON CHROMOSOME 9 CAUSE HI?

One type of big change is called a **deletion**, where **whole sections of DNA can be lost**.

The deletion **removes a section of chromosome 9** containing multiple genes. We know that **one or more of these missing genes must be important in the pancreas**, the organ responsible for making and sending out insulin to the body.

We have been able to narrow down the list to **26 genes**, but more work is needed to understand **exactly which of these genes is responsible for causing HI**.



IS IT COMMON? HOW IS IT INHERITED?

This form of HI is considered **rare BUT** it may be **underdiagnosed** because it is not included in routine tests.

It is a **dominant variant**, explained further in our information sheet linked [here](#).

In most of the children it is a **de novo variant**, meaning it was **not found in their parents**, explained further in our information sheet linked [here](#).

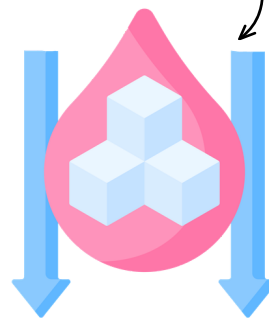
Sometimes, a deletion can result from a **balanced translocation** in a parent, where part of a chromosome has moved, but **no genetic material has been lost (deleted) or gained**. The parent is **usually unaffected**, but this can **increase the chance** of having a **child with a chromosome deletion**.

Genetic testing and counselling can help determine whether this is the case and the chances of this happening in a future pregnancy.



WHAT ARE THE FEATURES OF HI CAUSED BY A CHROMOSOME 9 DELETION?

hypoglycaemia



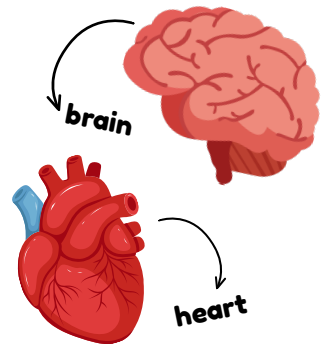
Most children with this form of HI have **low blood sugar levels** (known as **hypoglycaemia**) that was diagnosed within the first week of life.

Children with this deletion often also have **other clinical features**. These **vary between individuals** and likely reflect **differences in the size of the deletions**.

For example, some children may have **more genes missing** than others, causing **different features to develop**.

Some of the other features reported are:

- ★ Distinctive **facial features**
- ★ **Developmental delay**, including slower than expected development of movement skills (e.g. sitting, walking, and coordination)
- ★ **Learning difficulties**
- ★ **Heart differences present from birth**, such as VSD and ASD
- ★ **Changes in the appearance of the genitals in males**



HOW DO YOU TREAT AND MANAGE IT?

Our understanding of how this deletion results in HI is still growing, and researchers are working hard to understand why for some children the HI is transient, but for others it can persist for many years



Most children with a chromosome 9 deletion are successfully treated with the drug **diazoxide**. This is the **standard treatment for HI**.

For many individuals, the **HI improves over time** and they **no longer need treatment**. The age at which this happens differs between people.

For example, some children **only require treatment for a few weeks**, while others **still receive diazoxide when they are teenagers**.