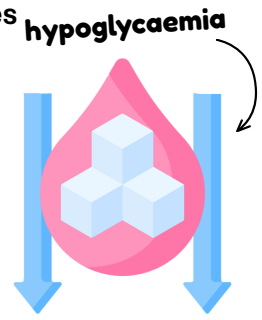
WHEN GENETIC TESTING DOES NOT FIND A CAUSE FOR CONGENITAL HI

HYPERINSULINISM GENES

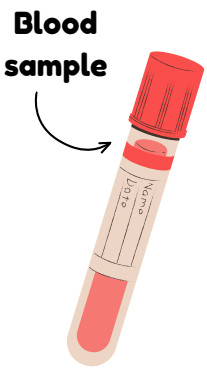
This summarises our information sheet for **individuals and their families who have had genetic testing for congenital hyperinsulinism (shortened to HI) but no genetic cause has been found.** For more on different types of genetic changes, check out our information sheets linked here!

WHAT ARE THE GENETICS OF CONGENITAL HI?

In congenital HI, the **pancreas releases too much insulin**, causing low blood sugar (called **hypoglycaemia**).



HI that continues beyond the first few weeks of life can be **caused by a genetic change (or a variant) in a person's DNA.**



As congenital HI can be caused by genetic changes in over **30 different genes**, **genetic testing** is an important way to **help doctors better understand the condition in each individual.**

Genetic tests usually involve using a **blood sample** (or rarely from **a sample of the pancreas** after an operation).

KEY MESSAGES:

- **Congenital HI** can be caused by **genetic changes** that affect genes involved in controlling the release of insulin
- **Genetic testing** can help **identify whether a genetic change is causing HI**
- For many families, **genetic testing does not find the cause of the HI**, this can happen for lots of different reasons
- Not finding a genetic cause **does not always mean the HI is not genetic,** it can mean that a cause has not yet been identified
- It is important to keep **talking to your doctor about your genetic results,** as there might be other ways to learn more

WHY WOULD A GENETIC TEST NOT TELL ME THE CAUSE OF MY HI?

Up to **50% of people with congenital HI do not receive a genetic diagnosis** after genetic testing. This **does not necessarily mean there is no genetic cause,** it often means that current testing has not identified the cause. For some families, living without a genetic diagnosis can be **unsettling and leave important questions unanswered**.

There are many reasons why you might not have a genetic diagnosis:

- Testing may have **found a genetic change**, but laboratory scientists are **unsure if it is the cause of the HI**. These genetic changes are called **variants of uncertain significance** (or **VUS**). To find out more about VUS see our information sheet here.
- The genetic test **may not have included all the genes** or genetic changes known to cause congenital HI. Or the genetic cause of HI **may not have been discovered** yet.
- In some people the cause of HI **may not be genetic at all**.

WHAT HAPPENS NEXT?

Not having a genetic diagnosis **will not affect access to monitoring and treatment for HI.** Doctors will recommend the most appropriate care **based on your symptoms**.

It is still important to **keep talking to your doctor** about your genetic testing results for a few reasons:

- **Further genetic testing may be possible.**
- Your results may need to be **reviewed over time**.
- If your child is seen by different specialists or at different hospitals, it is helpful to make sure **your doctors are aware of any new findings or changes to your child's care.**
- It is helpful to **share as much information as possible** about your child's health and family history with your doctor, **even things that may not seem relevant to HI.**
- You may be able to **take part in research.**

WHERE CAN I FIND MORE SUPPORT?

Not having an explanation for why HI has happened **can be difficult for some families.**

If you would like more information or support, you can **also contact HI patient advocacy organisations,** such as the **Children's Hyperinsulinism Charity.**

Children's HYPERINSULINISM Charity

These organisations can **connect you with other families** affected by congenital HI, including **families who are living without a genetic diagnosis**.