

HOW DOES A VARIANT IN *SLC16A1* CAUSE HYPERINSULINISM?

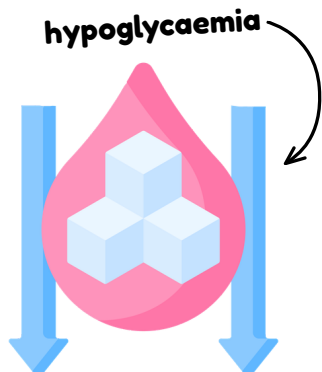


This summarises our information sheet on what we know about **hyperinsulinism** (shortened to HI) caused by a **variant in *SLC16A1*** (known as ***SLC16A1*-HI**). For more, check out our information sheets linked [here](#)!

WHAT IS *SLC16A1*-HI?

This is a condition that is caused by a **genetic change** in the **gene *SLC16A1***. Although the **genetic change** is there **when you are born** (this is known as **congenital**), you might **not get symptoms** until you are **older**.

In this condition, doing a lot of **intense exercise** can cause episodes of **very low blood sugar levels** (**hypoglycaemia**) caused by **HI**. This is why this condition is sometimes called **Exercise-Induced-Hyperinsulinism** (or EIHI).



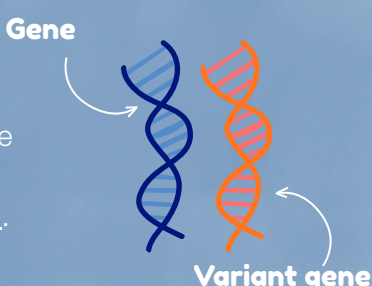
KEY MESSAGES:

- ★ *SLC16A1*-HI is a **rare form of HI** caused by a **dominant variant** in the ***SLC16A1*** gene
- ★ It is **different from other forms** of HI as most often the symptoms of HI will develop when a **person is an adult**
- ★ There can be **many things that trigger hypoglycaemia episodes** like intense exercise
- ★ **Lifestyle changes** like avoiding intense exercise or managing diet can **help people with the condition**
- ★ **More research** is needed to understand **what treatments work best** for individuals with *SLC16A1*-HI

IS IT RARE? HOW IS IT INHERITED?

This is a **rare form** of HI. As of Spring 2025, we know of **50 individuals** that have been diagnosed. But, we think it is **underdiagnosed**.

SLC16A1-HI is inherited in a **dominant way**. To learn more check out our information sheet on the topic linked [here](#).



WHAT ARE THE FEATURES OF *SLC16A1*-HI?

This condition affects people in lots of different ways. Some people who have **inherited the *SLC16A1* genetic change** **don't have any symptoms**, while other people could have **persistent hypoglycaemia despite getting treatment**.

This is known as **variable penetrance**, which is explained more in our information sheet linked [here](#).

HOW IS *SLC16A1*-HI DIFFERENT FROM OTHER FORMS OF HI?

1 IT IS DIAGNOSED LATER

While most forms of congenital HI are **diagnosed very quickly after a baby is born**, the average age of diagnosis for *SLC16A1*-HI is **21 years old**. After the first person in a family is diagnosed, it is **common for several other family members to also be diagnosed**, sometimes even when they are over 70 years old.

Onset as an adult



We are looking to see if most people with *SLC16A1*-HI **either first get symptoms** when they are a **child** which get **worse as they get older**, or if the **condition truly starts when they are an adult**. We are also looking at what factors could cause **delays in people getting a diagnosis**.

2 HYPOGLYCAEMIA CAN BE TRIGGERED BY DIFFERENT THINGS

In *SLC16A1*-HI, there seems to be **more than one trigger for hypoglycaemia**. A trigger is a specific **event or action** that **makes something else happen**, in this case an event or action that causes hypoglycaemia.

In the beginning, we thought that the **hypoglycaemia would happen most often after exercise**. We now know that many people with *SLC16A1*-HI can **also get hypoglycaemia when they are not exercising**. Some people have lots of triggers for hypoglycaemia, others report just one trigger. Some of these triggers are listed below:



INTENSE EXERCISE



ILLNESS



HIGH CARBOHYDRATE MEALS



FASTING OR SKIPPING MEALS



EMOTIONAL STRESS



PREGNANCY

3 TREATMENT CAN BE VERY DIFFERENT BETWEEN INDIVIDUALS

The **need** for treatment and **how effective treatments** are **can be different for each person** with *SLC16A1*-HI. This is something that we need to **look into and understand more** about.

Many people with *SLC16A1*-HI have **made some changes to their lifestyle**, including changes to their diet and exercise habits. This has helped to **manage or reduced the hypoglycaemia** episodes. What is most important is that **people recognise and avoid the triggers** that affect them the most.

Some strategies that have helped others with *SLC16A1*-HI include:

- ★ **Frequent healthy snacks** between meals or immediately after exercising
- ★ Choosing meals that have a **low to medium amount of carbohydrates** and are **higher in protein**
- ★ Taking **uncooked cornstarch at bedtime** (it is a slow-releasing carbohydrate) to help reduce the symptoms of **hypoglycaemia in the mornings**
- ★ **Avoiding anaerobic exercise**, especially swimming
- ★ **Avoiding alcohol**
- ★ Using **continuous glucose monitoring (or CGM)** to track and manage glucose levels as they change

Although these strategies may be helpful, **some people find that they still need to take medication**.



In several individuals, taking **diazoxide** has at least **partially reduced** how **often and how severe** the episodes of hypoglycaemia are.

Another medication is **octreotide**. However, there have been **mixed reports** of how well this drug works, so we need to **research this more**.

We also know that **4 people had surgery** that removed some of their pancreas. We are **still collecting data** that will tell us **if this surgery has helped manage the hypoglycaemia**.