First version: Jonna Männistö and Sarah Flanagan 16–05-2025. Updated: 26-06-2025 by Maya Ballantine

SLC16A1-HI Patient Information Sheet

This information sheet summarises what we know about **hyperinsulinism** (shortened to HI) caused by a **genetic change in *SLC16A1*** (known as ***SLC16A1*-HI**). This sheet is intended for information only and should not replace personalised advice or genetic counselling for you or your family. Check out our other information sheets here.

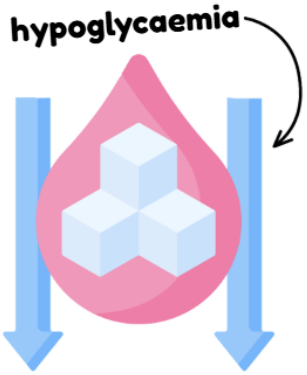
All terms highlighted in **blue** can be found in our glossary, linked here.

Key Points

- ***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

What is *SLC16A1*-HI?

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



We know that in this condition, doing a lot of **intense exercise** can **cause episodes of hypoglycaemia caused by HI**. This is where **too much insulin** is released and causes **blood sugar (or glucose) levels** to drop **too low.** This is why this condition is sometimes called **Exercise-Induced-Hyperinsulinism** (or **EIHI**).

Is *SLC16A1*-HI Rare?

This is **a rare form** of HI. As of Spring 2025, we know of **15 families**, with a total of **50 individuals within those families**, that have been **diagnosed with *SLC16A1*-HI**. However, we think that *SLC16A1*-HI is likely to be **underdiagnosed** and we are **actively working to raise awareness** of this condition around the world.

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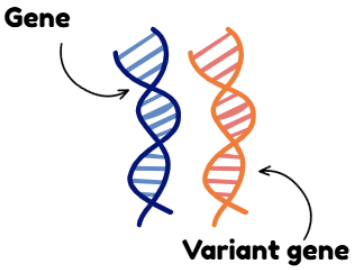
How is *SLC16A1*-HI Inherited?



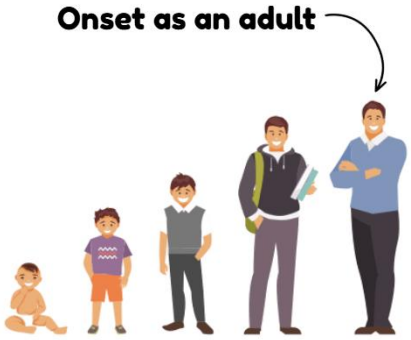
SLC16A1-HI is inherited in a **dominant way**. This means **only one dominant variant is needed to cause the disease**. To learn more about dominant variants, check out our information sheet on the topic linked here.

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**, with a range from birth **to 41 years**. These ages refer to the **age of when the first person in a family** was diagnosed with HI. 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.

We are looking to see if most people with *SLC16A1*-HI **first get symptoms when they are a child** which **get worse as they get older** so that they only see a doctor when they are an adult, **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, a trigger is 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:

EXERCISE: We know that **intense (anaerobic) exercise** causes hypoglycaemia in individuals with *SLC16A1*-HI. This usually happens **10 to 20 minutes after** they stop exercising. So far, **swimming** is the sport that has been linked the most to hypoglycaemia episodes with this condition.

FASTING OR TAKING LONG BREAKS BETWEEN MEALS: Lots of people with *SLC16A1*-HI say that they get **hypoglycaemia symptoms** in the **morning or after long breaks between meals.** They found that **eating soon after they wake up** and/or having **small frequent snacks** during the day helps to reduce these episodes.

HIGH CARBOHYDRATE MEALS: Some people with *SLC16A1*-HI say they get hypoglycaemia symptoms after eating meals that have a **lot of 'fast' carbohydrates** like **pasta, potatoes**, or other foods that have **a high glycaemic index**.

ILLNESS: Some people, especially children, have found that hypoglycaemia symptoms **get worse when they are feeling unwell.** These illnesses can be things like a **stomach bug or other infections**. We think this might be related to **not having enough food, vomiting**, or the **body needing more energy to fight off the illness.**

EMOTIONAL STRESS: A few people have said that **emotional stress or excitement** can make **hypoglycaemia episodes happen more often or make them worse.**

PREGNANCY: Some female individuals with *SLC16A1*-HI have reported that pregnancy made the symptoms of **hypoglycaemia symptoms worse**.

3 Treatment Can Be Very Different for Each Person with *SLC16A1*-HI:

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 other people 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**, especially when people are unwell with things like a high fever or bad stomach bugs, **some people find that they still need to take medication**.

At the moment, we don't know a lot about how well different medications works for people with *SLC16A1*-HI. In several individuals with *SLC16A1*-HI taking the drug **diazoxide** has at least **partially reduced how often and how severe the episodes of hypoglycaemia are.**

Another **medication** that some people have used is a drug **called octreotide.** However, there have been mixed reports of how well this drug works, with 3 people reporting that their **symptoms got worse** when they took octreotide. We, therefore, **need to investigate 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 reduce how often and how bad the hypoglycaemic episodes** are in these people.

Getting Individualised Information and Accessing Genetic Counselling Services

Meeting with a **genetic counsellor** or **clinical geneticist** can be very helpful if you have **a family history of HI**. They can explain how HI might **affect you or your children** and **answer any questions** you have. Because everyone's genetics are unique, the guidance you receive will be **personalised to you**.

This support can help you make **informed decisions** and **plan the future**. To book any appointment you will usually need a **referral from your family doctor** (GP in the UK) or a **specialist doctor**.

Additional Resources

- **Dominant inheritance explained:** https://www.genomicseducation.hee.nhs.uk/genotes/knowledge-hub/autosomal-dominant-inheritance/
- **Genes explained:** https://www.genomicseducation.hee.nhs.uk/genotes/knowledge-hub/gene/
- **The role of the Genetic Counsellor:** https://www.genomicseducation.hee.nhs.uk/genotes/knowledge-hub/roles-in-clinical-genetics/#genetic-counsellors

This leaflet was developed by the Exeter Hyperinsulinism Genes team (www.hyperinsulinismgenes.org) in collaboration with The Children's Hyperinsulinism Charity UK (https://hyperinsulinism.co.uk/). Registered Charity: 1165562, Company Number: CE005407