The Open Hyperinsulinism Genes Project

This information sheet is about the **Open Hyperinsulinism Genes Project**. For more information about congenital hyperinsulinism (shortened to HI), please visit the Congenital Hyperinsulinism International website here.

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

Key Points

- The Open Hyperinsulinism Genes Project is an international collaboration that lets people from around the world access free genetic testing for HI
- You can use this service if you or your child is being treated for HI, your doctor thinks it could be caused by a genetic change and you cannot access or afford genetic testing on your own
- After your doctor has made the request, they will send your blood sample to the Exeter Genomics Laboratory in the UK, and once the genetic testing has been completed, the genetic report will be sent back to your doctor
- You can choose to get involved in research as well, which will help us to understand more about HI

What is the Open Hyperinsulinism Genes Project?

The Open Hyperinsulinism Genes Project is an international collaboration between the University of Exeter, the Royal Devon University Hospital in the UK, and Congenital Hyperinsulinism International (CHI).

This project provides free **genetic** testing for people with **congenital hyperinsulinism** who cannot access or afford testing where they live. The aim of this project is to make sure that everyone who has HI can get a genetic test no matter where they live in the world.

Can I Use This Service?

You and your family can use this service if you or your child:

- is being treated for congenital HI and your doctor thinks that a genetic change could be causing the condition
- are unable to access genetic testing through your local healthcare provider or through personal resources, such as employer-sponsored or private medical insurance, because genetic testing is not covered by these options

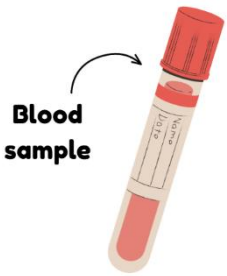
Why Is It Important to Get a Genetic Test?

Genetic tests can help your doctors to:

- Find the cause of your HI
- Guide treatment decisions
- Find out whether you have focal or diffuse HI
- Find out whether other people in your family could have HI
- Help you plan for future pregnancies
- Allow you to connect with other people who have a similar genetic form of HI

How Do I Join the Project?

To get a genetic test through the Open Hyperinsulinism Genes Project, please ask your doctor to send a request to Exeter by emailing Prof Sarah Flanagan (S.Flanagan@exeter.ac.uk). Your doctor will be asked to confirm that you or your child meet the eligibility criteria for the project.



Your doctor will then arrange for a blood sample to be taken from the person with HI. If possible, they will also arrange for blood samples to be taken from the parents of the person with HI. Samples from the parents can help the laboratory scientists to understand more about any genetic changes that are found.

These samples are all sent to Exeter in the UK, which is where the genetic testing is performed in a fully accredited laboratory.

Once the genetic test and the investigation into any genetic changes found have been completed, a detailed report will be sent back to your doctor by email. Your doctor will then talk through any findings with you.

How Long Does This Take?

There are two routes for genetic testing, depending on your situation:

- **URGENT TESTING:** this is for babies who have just been diagnosed with HI or for people who are not getting better with treatment. For these people, we aim to send back the genetic report within 1 to 2 weeks after the sample has arrived in Exeter.
- **ROUTINE TESTING:** this is the standard route for people who have HI which is responding well to treatment. For these people, we aim to send back the genetic report within 4 to 6 weeks after the samples have arrived in Exeter.

What Are the Opportunities to Take Part in Future Research?

If you get a genetic test through the Open Hyperinsulinism Genes Project, there are different ways that you can get involved with research. This helps us to understand more about HI. Participation is completely optional and your information is only used if you have agreed to be involved in this research. These are some of the ways you can get involved:

Take Part in Research at the University of Exeter

Genetic Beta Cell Research Bank

When your blood sample is taken for genetic testing, you will be asked if you would like your sample to be used for research studies at the University of Exeter.

These studies aim to improve our understanding of the known genetic causes of HI and to identify new genetic causes in people who do not yet have a genetic diagnosis. This research has already helped to discover new genes that cause HI and continues to improve how people with HI are diagnosed and cared for around the world.

Join The HI Global Registry

We encourage everyone with HI and their families to join the HI Global Registry. This international research project was founded by parents of children with HI and is powered by the experiences of individuals and families affected by HI around the world. It is managed and maintained by Congenital Hyperinsulinism International.

By sharing your information about your experiences with HI, you can help researchers to better understand the condition, help with unmet needs, and support the development of better treatments and care. Further information about the HI Global Registry can be found here.

Any Questions?

We welcome enquiries from families who are interested in the Open Hyperinsulinism Genes Project. However, **requests for genetic testing must be made by a medical doctor**, who should email Prof Sarah Flanagan (S.Flanagan@exeter.ac.uk).

For more information on the project, please visit our website here.