

Parent Information Leaflet for Parents / Carers

Unresponsive Congenital Hyperinsulinism



Introduction

This booklet is for parents and carers of children who in rare cases have not responded to the first line medical treatment (Diazoxide) for the management of Congenital Hyperinsulinism (CHI). These children will require further investigations to identify the type of CHI i.e. diffuse versus focal disease so that treatment can be tailored appropriately.

Genetics

There are various defects in genes which can lead to inappropriate insulin secretion from the pancreas, therefore causing CHI. Genetic testing will be needed in most children who have persistent and severe CHI and who do not respond to first line medical therapy. DNA from both parents is required (one blood test each). Your consultant Endocrinologist will be able to explain more depending on the course of the illness as to how the Genetic results may or may not guide the management of the condition in your child.

Treatment

Firstly, if there is no response to Diazoxide, it may be weaned down or discontinued while other treatments are commenced. Your child may still require high concentrations of intravenous dextrose to help stabilise their blood glucose levels.

The second line treatment for CHI is usually a trial of octreotide. Octreotide is used to treat persistently low blood glucose levels caused by the body producing too much insulin (Hyperinsulinism). Octreotide is given as a subcutaneous injection, three to four times a day. If your child responds to octreotide by maintaining their blood glucose levels within the normal range (3.5mmol/l to 6mmol/l) then the endocrine team may be able to prepare for discharge home on Octreotide. This can be a slow process over a number of days or even weeks depending on the child and their response to the medical treatment. You will be given education on how to prepare and administer the injection prior to discharge home.

Regardless of response to medical therapy, if your child is considered to have a particularly severe case of CHI they may require referral to a specialised centre in the UK for their expert opinion. In particular, your child may need a specialised scan, called a PET scan, which will help to indicate whether the hyperinsulinism is caused by focal or diffuse disease.



Focal CHI

In focal CHI, usually only a specific area of the pancreas is affected. Focal lesions can be small, measuring 2-10mm. Your child may require a special scan called a DOPA-PET scan to determine the location of the focal lesion as it is small and cannot be seen by other X-RAY techniques. The aim of the scan is to identify the area of the pancreas that is producing excessive insulin. This scan is not available in Ireland (as it is so rarely needed), so your child will have to be transferred to a specialised centre in the UK if this scan is indicated. If a focal lesion is identified and your child is unresponsive to medical management then surgical resection of the lesion may be curative. The surgeons who are highly experienced in this rare and specialised surgery are based in the UK.

Diffuse CHI

Diffuse CHI affects the entire pancreas. For children with diffuse CHI who do not respond to Diazoxide the next step is Octreotide. In rare cases, if Octreotide is unsuccessful at maintaining blood glucose levels then your child will require referral to specialised centre abroad and pancreatic surgery may be necessary. The decision will be made by a multidisciplinary team experienced in the care of persistent and severe CHI, in close collaboration with the family, and your Irish endocrine team.

Genetic findings and risk of recurrence

As CHI is a rare condition sometimes with a complex genetic explanation, referral maybe required to the Clinical Genetics Team for their expert opinion and for genetic counselling regarding risk for any future pregnancies and also your child may require genetic counselling as they get older to help with their own future family planning.



Preparation for going abroad

If treatment abroad is required for your child, the endocrine consultant will refer them to the appropriate specialist centre to provide the treatment. Subject to prior approval, the cost of your child's treatment abroad is covered by the Treatment Abroad Scheme, and this scheme is described in detail on www.hse.ie/treatmentabroad

The scheme is covered by EU regulations and administered by the HSE. A treatment abroad application form E112 must be completed by a parent/guardian and the endocrine consultant along with a referral letter. The HSE will need to assess and approve application prior to travelling abroad. You and Your child will also need passports. An emergency passport can be applied for in the event of requiring medical treatment abroad. All this will be done with the help of a medical social worker, Endocrine CNS and the Endocrine team who have experience in this procedure.

In extremely rare cases, if your child is unstable and a medical emergency case when he/she is referred abroad for treatment, then medically assisted travel will be required. This is provided by the Irish Air Corps, Irish Coast Guard or by private air ambulance. If your child is stable and does not need an air ambulance then you will travel on a commercial airplane or boat depending on your preference. Where the patient is under the age of 18, the air or sea fares of two accompanying adults will also be provided, subject to available funding. Unfortunately, the HSE cannot provide advance payments to pay for flights.

