Congenital Hyperinsulinism International
Collaborative Research Network

Congenital hyperinsulinism (HI) is the most common cause of persistent hypoglycemia (low blood sugar/low plasma glucose) in infants and children. Low plasma glucose is extremely dangerous. If not treated promptly and appropriately, hypoglycemia can lead to brain damage, developmental delays, and even death. Prompt initiation and maintenance of appropriate treatment is critical for preserving the vital functioning of the child.

HI is a rare disease, occurring in approximately 1 in 28,000 births. Since most children's hospitals encounter less than 2 cases of HI per year it is important to utilize the medical expertise from one of the CHI (Congenital Hyperinsulinism International) designated Centers of Excellence (COE) The CHI Centers of Excellence (COE) Program – Congenital Hyperinsulinism International (congenitalhi.org) located around the world. A consultation or transfer to a COE may be necessary.

It is imperative that severe or prolonged neonatal hypoglycemia be detected as early as possible, ideally prior to initial hospital discharge. Concerns of caregivers, parents, or other family members must be addressed. Without the ability to identify cause, they are often the first to verbalize concern that something is amiss in the newborn. Signs of hypoglycemia (excessive hunger or feeding disinterest, lethargy, difficulty to rouse, jitteriness, irritability, or convulsions) observed by Nursery/midwifery staff or other medical care team members must also trigger a ~~plasma~~ glucose measurement. Birthing care team members must recognize the significant benefit of a plasma glucose measurement.

Blood tests at the time of hypoglycemia can diagnose HI. Targeted HI genetic testing may also be necessary to identify specific mutations that aid in the patient's future treatment decisions.

Emergency treatment with high dextrose concentrations titrated to maintain plasma glucose in the normal range can only be utilized in a hospital setting. Medications may be a long-term home-based treatment option.

Diazoxide, the only oral medication available for the treatment of HI is the first line therapy and may be initiated prior to HI genetic results as diazoxide responsiveness is key to HI classification and subsequent care. It is essential that diazoxide be available for the treatment of HI for all infants/children around the world. Diazoxide is on the WHO list of essential medications and should be added to every country's list of essential medications. Without diazoxide, lifelong disabilities may result.

In case of diazoxide unresponsiveness, octreotide/Lanreotide must be available as a second line therapy and should be added to WHO list of medications.

It is imperative that all providers of infants/children with HI have knowledge of ongoing clinical trials and all infants/children can have rapid access to potentially lifesaving medications as soon as they are eligible to receive them.

Genetic testing may indicate diffuse HI (affecting the entire pancreas) or focal (affecting one area of the pancreas). Severe diffuse HI that does not respond to medical therapy requires a subtotal pancreatectomy. Focal HI may be cured by surgical resection of the focal lesion, this requires an experienced surgical team after lesion identification via 18-Fdopa PET scan. The care of infants/children with HI, both medically and surgically managed is complicated, necessitating care to be provided by a knowledgeable medical team offering multidisciplinary expertise.