CHI. Congenital Hyperinsulinism

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Summary

Congenital Hyperinsulinism (CHI) is a rare disease, but is the most frequent cause of persistent and severe hypoglycaemia in early childhood. Hypoglycaemia caused by excessive and dysregulated insulin secretion (hyperinsulinism) from defective insulin-secreting cells can often lead to irreversible brain damage with lifelong neurodisability. Although congenital hyperinsulinism has a genetic cause in a significant proportion (40%) of children, not all children have severe and persistent forms of the disease.
In approximately half of those without a genetic mutation, hyperinsulinism may resolve, although timescales are unpredictable. Whatever the cause or duration of CHI, the consequences of hypoglycaemia cannot be understated as a high major impact and cause of adverse neurodevelopment in a third to a half of individuals.

From a histopathology perspective, congenital hyperinsulinism is broadly grouped into diffuse and focal forms, with surgical lesionectomy being the preferred choice of treatment in the latter. In contrast, in diffuse congenital hyperinsulinism, medical treatment is the best option if conservative management is safe and effective. If hypoglycaemia is not safely managed by medical therapy, subtotal pancreatectomy may be required and diabetes is an inevitable consequence in later life.

It is important to study the broad phenotypic diversity that exists in patients with CHI. The NIHR BioResource will provide an important and exciting opportunity for CHI centres in the country to work together to achieve strategic targets in the development of a greater understanding of this disease. For the future, this initiative will also lay an important platform for translational research to develop personalized treatment options for children and young people with CHI.

Inclusion / Exclusion

Recruitment criteria: biochemical evidence of hyperinsulinism and absence of below exclusion criteria.

Inclusion

- Blood glucose < 3.0 mmol/L; measurable insulin at hypoglycaemia (> 6 pmol/L)

Exclusion

- Other on-going causes of hypoglycaemia
- Patients in first two days of life
- Babies born to mothers with diabetes
- Babies who are small for gestational age at birth
- Perinatal asphyxia