

New test could identify infants with rare insulin disease

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A rare form of a devastating disease which causes low blood sugar levels in babies and infants may now be recognised earlier thanks to a new test developed by researchers from The University of Manchester.

Congenital hyperinsulinism starves a baby's brain of [blood sugar](#) and can lead to lifelong brain damage or permanent disability according to previous research carried out by the Manchester team. The condition occurs when specialised cells in the pancreas release too much [insulin](#) which causes frequent low sugar episodes - the clinical opposite of diabetes. Treatment includes drugs to reduce insulin release but in the most serious cases the pancreas is removed.

For some infants with this disease, the release of excess insulin is due to mutations in genes which govern the way our bodies control insulin release. But for more than two thirds of child patients the genetic causes are not yet known.

Genes and hormones were analysed in 13 children with congenital hyperinsulinism at the Manchester Children's Hospital and the findings have been published in *The Journal of Pediatrics*.

Dr Karen Cosgrove from the Faculty of Life Sciences led the research: "We have discovered a new clinical [test](#) which can identify congenital hyperinsulinism in some patients with no known genetic cause of the disease. This is the first step to understanding what causes the disease in these particular patients. In future the test may influence how these

children are treated medically, perhaps even avoiding the need to have their pancreas removed."

The [new test](#) measures a pair of hormones called incretins which are released by specialised cells in the gut when food is passing through. The hormones normally tell the cells in the pancreas to release more insulin to regulate sugar levels in our blood. If the child's body releases too much incretin hormones, the pancreas will release too much insulin causing dangerous [low blood sugar](#) levels.

"Although we are the first researchers to report high incretin hormone levels in patients with congenital hyperinsulinism, further studies are needed to see if our test works on a larger group of patients" says Dr Cosgrove.

Researchers from The University of Manchester along with consultants from the Manchester Children's Hospital, part of Central Manchester University Hospitals NHS Foundation Trust, teamed up for the study. Royal Manchester Children's Hospital is the base for the Northern Congenital Hyperinsulinism (NorCHI) service, a national centre for treatment of this disease.

Doctor Indi Banerjee, Consultant in Paediatric Endocrinology at Royal Manchester Children's Hospital and clinical lead for NorCHI says: "Our new results are timely since clinical trials of a new incretin-blocking treatment for [congenital](#) hyperinsulinism have recently started. We anticipate that our clinical test will help to identify the [patients](#) who are likely to benefit from this new treatment the most."

Julie Raskin, Executive Director of Congenital Hyperinsulinism International is impressed with the research: "A new diagnostic test for this devastating disease is welcome news to the international hyperinsulinism patient community because timely diagnosis is key to

reducing the chance of brain damage and death, and the research also suggests a path to treatment other than sub-total pancreatectomy, which almost always leads to diabetes."

More information: Increased plasma incretin concentrations identifies a subset of patients with persistent congenital hyperinsulinism without KATP channel gene defects, *The Journal of Pediatrics*, 2014.

Provided by University of Manchester

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