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Genetic sequencing breakthrough to aid treatment for congenital hyperinsulinism

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Congenital hyperinsulinism is a genetic condition where a baby's pancreas secretes too much insulin. It affects approximately one in 50,000 live births and in severe cases requires the surgical removal of all or part of the pancreas.

Researchers at the University of Exeter Medical School are the first in the world to utilise new genetic sequencing technology to sequence the entirety of a gene in order to identify mutations that cause hyperinsulinism. Previously, existing technology limited such sequencing to only part of the coding regions of the gene which meant that some mutations were missed.

Using new Illumina genetic sequencing technology, the research team led by Professor Sian Ellard has discovered novel mutations that cause hyperinsulinism. Their findings are published today, 27th December 2012, on-line by *The* American Journal of Human Genetics.

The outcome will be that some infants born with hyperinsulinism will require fewer investigations, because the new technology means that for many only one genetic test will be required to determine the extent of the condition in each child. It also means that clinicians will have more information at their fingertips to inform them about how much of the pancreas needs to be removed.

Approximately 50 per cent of patients with congenital hyperinsulinism require surgery, and of those half require the entire pancreas to be

removed. Removal of the entire pancreas increases the risk of diabetes later in life, but if left undiagnosed and untreated hyperinsulinism can result in irreparable brain damage. Symptoms range from shakiness and tiredness to seizure and coma.

Dr. Sarah Flanagan, Research Fellow in Molecular Genetics at the University of Exeter Medical School said: "The potential provided by this new technology is important and exciting, because it allows us to investigate genetic coding in its entirety. This means that investigators can identify mutations that sit at the heart of any number of conditions where before they might have been missed. This in turn results in better information for clinicians upon which they can base effective treatments and interventions for their patients."

Provided by University of Exeter