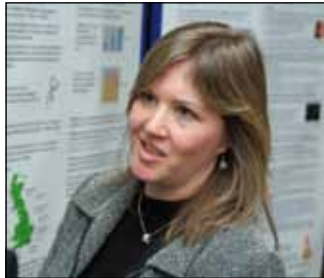


Genetic sequencing breakthrough to aid treatment for congenital hyperinsulinism

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.

Using new Illumina genetic sequencing technology, the research team at the University of Exeter Medical School, led by Professor Sian Ellard, has discovered novel mutations that cause hyperinsulinism (Flanagan et al, 2013).

Professor Sian Ellard, University of Exeter Medical School, Exeter



Previously, existing technology meant that such sequencing was limited to only part of the coding regions of the gene which meant that some mutations were missed.

This means 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 accurate information about how much of the pancreas needs to be removed.

Approximately 50% of patients with congenital hyperinsulinism require surgery, and of those half need the entire pancreas 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 dam-

age. 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.'

Flanagan SE, Xie W, Caswell R et al (2013) Next-generation sequencing reveals deep intronic cryptic ABCC8 and HADH splicing founder mutations causing hyperinsulinism by pseudoexon activation. *Am J Hum Genet* **92**(1): 131–6

Hormone-releasing coil best treatment for menorrhagia

The hormone-releasing Mirena intrauterine device is a better treatment for heavy menstrual periods than conventional medical approaches, according to new clinical trial results (Gupta et al, 2013).

Heavy periods, or menorrhagia, affect large numbers of women aged 25–50 years, accounting for 20% of gynaecological referrals in the UK.

The ECLIPSE trial compared the clinical effectiveness of the levonorgestrel-releasing intrauterine system (LNG-IUS, also known as the Mirena) with other medical treatments offered in primary care.

A total of 571 women, consulting their GPs for heavy menstrual bleeding, were randomly assigned to LNG-IUS or another standard medical treatment, e.g. tranexamic acid, mefenamic acid, combined oestrogen and progestogen, or progestogen only.

Women allocated to LNG-IUS were almost twice as likely to still be using it after 2 years than those taking other medication. Of other trial participants 49% switched to LNG-IUS because of the 'lack of effectiveness' of other treatments.

Gupta J, Kai J, Middleton L, Pattison H, Gray R, Daniels J; ECLIPSE Trial Collaborative Group (2013) Levonorgestrel intrauterine system versus medical therapy for menorrhagia. *N Engl J Med* **368**(2): 128–37

Increased risk of pulmonary embolism in asthma

People with asthma have an increased risk of pulmonary embolism, according to a new study (Majoor et al, 2012).

Previous research has found links between chronic lung diseases and blood clots, such as deep vein thrombosis, but this is the first study to look at any potential links with asthma.

The study examined 648 people with asthma in the Netherlands. All participants were aged between 18 and 88 years. Researchers examined participants' history of asthma and the medication they used to control their symptoms, along with any

previous diagnoses of pulmonary embolism or deep vein thrombosis. They compared these results to people without asthma.

The results demonstrated that people with severe asthma were at almost 9 times greater risk of pulmonary embolism than the general population. In addition, people with mild–moderate asthma tended to have a 3.5 times increased risk of pulmonary embolism. The study also identified oral corticosteroids as a potential risk factor for pulmonary embolism.

Lead author, Dr Christof Majoor, from the Academic

Medical Centre in Amsterdam, the Netherlands, said: 'This is the first time a link has been found between asthma and pulmonary embolism and we believe these results have important clinical implications. Our findings suggest that people with severe asthma have an increased risk of pulmonary embolism and doctors should increase their awareness of the possibility of this occurrence in order to help prevent this serious event.'

Majoor CJ, Kamphuisen PW, Zwinderman AH et al (2012) Risk of deep-vein thrombosis and pulmonary embolism in asthma. *Eur Respir J* 20 December (Epub ahead of print)