



Press release
For immediate release

INFLAMMATORY BOWEL DISEASE: SCIENTISTS ZOOM IN ON GENETIC CULPRITS

Montreal, June 28, 2017 - Montreal scientists and their international colleagues have closed in on specific genes responsible for Inflammatory Bowel Disease (IBD) from a list of over 600 genes that were suspects for the disease. Professor John D. Rioux and his team at the Montreal Heart Institute and Université de Montréal, along with their colleagues with the International IBD Genetics Consortium, which brings together over 100 geneticists, gastroenterologists, and other researchers, combined efforts to produce a high resolution map to investigate which genetic variants have a causal role in the disease.

In the new study, published today (28 June) in *Nature*, scientists examined the genome of 67,852 individuals and applied three statistical methods to zoom in on which genetic variants were actively implicated in the disease. Of the 94 regions of the genome associated with IBD that were studied, 18 could be pin-pointed to a single genetic variant with more than 95 per cent certainty. The results form a basis for more effective prescription of current treatments for the disease as well as the discovery of new drug targets.

More than 230,000 people suffer from IBD in the Canada, with approximately 10,000 new cases a year and resulting in annual economic costs of \$2.8 billion. IBD is a debilitating disease in which the body's own immune system attacks parts of the digestive tract. The exact causes of this disease are unclear, and there currently is no cure. "One of the most frustrating things for people living with Crohn's or colitis is that they don't know what triggered their disease in the first place," says Mina Mawani, President and CEO of Crohn's and Colitis Canada. "This study helps narrow down some of the genetic elements at play, and we're hopeful that better understanding leads to better treatments, and ultimately, cures for these debilitating diseases."

To understand more about the genetics underlying IBD, researchers have conducted genome wide association studies and previously found genetic variants in over 200 regions of the human genome linked to the disease. However, it was not certain which specific genes were actually implicated by those variants.

Dr Jeffrey Barrett, joint lead author from the Wellcome Trust Sanger Institute said: "We have taken the biggest ever data set for IBD and applied careful statistics to narrow down to the individual genetic variants involved. Now we have a clearer picture of which genes do and do not play a role in the disease. We are zooming in on the genetic culprits of IBD."

The high resolution map of the disease enabled scientists to see which variants directly influence disease, and to separate them from other variants which happen to be located near each other in the genome.

Dr Hailiang Huang, first author from the Massachusetts General Hospital and Broad Institute said: “An issue with studying complex diseases is that it can be hard to move from genetic variants to knowing exactly which genes are involved. We need to be careful in deciding when we are sure we have the right gene. This new technique helps us to pinpoint which genetic variants are implicated in IBD with greater confidence.”

Professor Rioux, said: “This is a great step forward for IBD research. We are already using these results to focus our efforts to systematically use genetic discoveries to drive the development of biomarkers that predict response to therapy in IBD, with the aim of getting the right medication to each individual patient in the most efficient way to increase therapeutic benefits and decrease risks associated with treatments”.

About the Montreal Heart Institute

Founded in 1954 by Dr. Paul David, the Montreal Heart Institute constantly aims for the highest standards of excellence in the cardiovascular field through its leadership in clinical and basic research, ultra-specialized care, professional training and prevention. It is part of the broad network of health excellence made up of Université de Montréal and its affiliated institutions. The Montreal Heart Institute ranks as the No. 1 research hospital in Canada for research intensity and research funds per researcher, according to Research Infosource. For more information, please visit www.icm-mhi.org

-30-

Information:

Lise Plante, MBA

Director of Public Relations

Montreal Heart Institute Foundation

Phone: 514-376-3330, extension 3898 | lise.plante@icm-mhi.org

facebook.com/institutcardiologiemontreal | @ICMtl

Publication:

Hailiang Huang *et al.* (2017) Fine-mapping inflammatory bowel disease loci to single variant resolution. *Nature*. DOI: 10.1038/nature22969

Funding:

Professor Rioux is the holder of the Canada Research Chair in Genetics and Genomic Medicine. His work as part of this study was funded by grants from the National Institute of Diabetes and Digestive Kidney Diseases grants DK064869 and DK062432, CIHR GPG 102170 from the Canadian Institutes of Health, GPH-129341 from Genome Canada and Génome Québec, and Crohn’s and Colitis Canada. For full funding information please refer to the publication.