First screen for genetic risk factors of IBS in the general population

September 24 2014

In the largest gene-hunting effort ever conducted in irritable bowel syndrome, IBS, researchers have found that some regions in the genome may be associated with an increased risk of developing the disease. The results from the international study led by researchers at Karolinska Institutet are published in the journal *Gut*.

This is the first study on IBS where researchers have looked at the whole genome at once, a so called genome-wide association study (GWAS). Most previous screens for genes involved in IBS have compared smaller groups of IBS patients with healthy controls. The current study used a novel approach, including more than 14 000 individuals.

IBS is a gastrointestinal disorder characterised by recurrent episodes of abdominal pain or discomfort associated with bloating, constipation and diarrhea. In Westernized countries, these symptoms affect more than 15 per cent of the general population.

"Because it is so common, we thought of studying IBS in the general population. This allows us to discover genetic risk factors that can later be validated in IBS patients", says Mauro D'Amato, Associate Professor at the Department of Biosciences and Nutrition at Karolinska Institutet, who led the study.

Existing resources

This approach allowed the investigators to exploit large, already existing resources to perform a search for IBS genes at the whole-genome level. In a general population sample of more than 11,000 Swedish twins the researchers identified 534 IBS cases and 4,932 asymptomatic controls based on questionnaire data. When they compared genetic information from cases and controls, several genomic regions appeared to be associated with an increased risk of IBS. The results were followed up in more than 3,500 individuals from six case-control samples from different countries. A region on chromosome 7 turned out to be interesting in all datasets. The gene region contains the genes KDELR2 and GRID2IP. Little is known about these genes, which may affect IBS risk.

"Population-based approaches like this provide excellent opportunities for gene-hunting efforts in IBS", says Mauro D'Amato. The potential causative role of KDELR2 and GRID2IP in IBS needs to be evaluated in further studies."

More information: "Exploring the genetics of irritable bowel syndrome: a GWA study in the general population and replication in multinational case-control cohorts." Weronica E Ek, Anna Reznichenko, Stephan Ripke et al. *Gut*, online 24 September 2014, gut.bmj.com/content/early/2014 … 2014-307997.abstract

Provided by Karolinska Institutet