

Scientists pinpoint key bowel disease gene

15 May 2017

A key gene that helps to explain an underlying cause of incurable bowel disorders such as Crohn's disease has been identified by scientists.

Blocking the gene harms vital parts of the cell and leads to [bowel disease](#), while targeting these vital cell parts with drugs can reverse damage, the study shows.

The findings aid understanding of the cause of these lifelong conditions and could lead to new treatments, scientists say.

Inflammatory bowel disease – or IBD – includes Crohn's disease and [ulcerative colitis](#) and affects around 300,000 people in the UK. The causes of these disorders are unknown and there is currently no cure.

Vital role

The gene – known as MDR1 – governs an important extractor system for toxins in the gut, removing damaging substances from intestinal [cells](#).

A research team – led by the University of Edinburgh – showed that MDR1 function was lower in colonic biopsies from people with inflamed IBD compared with those without inflammation.

To demonstrate how MDR1 dysfunction leads to bowel damage, the scientists then showed that mice lacking MDR1 had faulty mitochondria – parts of the cell often referred to as 'batteries.' These play a vital role in energy generation and cell health.

Defining feature

This mitochondrial dysfunction resulted in colitis – inflammation in the inner lining of the bowel that is a defining feature of IBD.

Scientists further implicated the role of mitochondria by linking IBD to a large number of

genes involved in regulating these cell batteries.

The researchers analysed genetic data from 90,000 people – 40,000 of whom had IBD.

Significant step

The study also showed that a drug called Mitoquinone – which protects the mitochondria against toxins – can reduce colitis and promote bowel recovery in the mice lacking MDR1, which scientists say is a significant step forward.

The research – carried out in collaboration with researchers at the University of Bristol and in the USA and Japan – was funded by the Medical Research Council and Crohn's and Colitis UK. It is published in the journal *Mucosal Immunology*.

"IBD has a serious impact on quality of life, with 6,000 new cases diagnosed per year in the UK. We have shown that MDR1 and mitochondrial function are important jigsaw pieces in the complex causes of the disease," says Dr Gwo-Tzer Ho.

More information: G-T Ho et al, MDR1 deficiency impairs mitochondrial homeostasis and promotes intestinal inflammation, *Mucosal Immunology* (2017). DOI: 10.1038/mi.2017.31

Provided by University of Edinburgh

APA citation: Scientists pinpoint key bowel disease gene (2017, May 15) retrieved 17 October 2021 from <https://medicalxpress.com/news/2017-05-scientists-key-bowel-disease-gene.html>

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