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New methods for genetics analyses and diagnosis of inflammatory bowel disease

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The two most common types of inflammatory bowel disease are ulcerous colitis and Crohn's disease. These are diagnosed via endoscopy and gut biopsy. The diagnosis is often difficult, and the wrong diagnosis may have severe consequences for patients, because the treatments and medications are different between the two diseases.

The development of new and improved diagnostic methods is therefore important. The Sandelin group at Department of Biology/BRIC, University of Copenhagen has, in collaboration with clinicians from Herlev and Hvidovre hospitals and scientists from Roskilde University and the Technical University of Denmark, made new discoveries may contribute to improved methods for diagnosis.

"We do not know the molecular cause of these diseases. Much of what we know comes from genetic studies, where several key genes have been identified. However, 70 percent of genetic mutations that are linked to the diseases are located outside of genes that code for proteins. We believe that many of these mutations have an effect on the regulation of the genes, and thereby the disease," says Albin Sandelin, professor at Department of Biology, University of Copenhagen.

The scientists used a state-of-the-art method to map regulatory regions and their activity in patients with ulcerous colitis or Crohn's disease, and compared them with control subjects. They found that mutations associated to the disease were often located within regulatory regions active in the disease. This information is important for understanding the

effect of such mutations. By combining these data with computer-based models and nanofluidics technology, they could identify 35 regulatory regions whose activity could distinguish ulcerous colitis, Crohn's disease and control subjects with high accuracy. These findings may open new avenues for new and improved diagnosis methods for inflammatory disease.

More information: Mette Boyd et al, Characterization of the enhancer and promoter landscape of inflammatory bowel disease from human colon biopsies, *Nature Communications* (2018). DOI: 10.1038/s41467-018-03766-z

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