

Identifying specific markers in blood may improve diagnosis of IBD in children

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New research may lead to improved methods to diagnose inflammatory bowel disease (IBD) in children. This could be especially beneficial for those experiencing malnutrition and impaired growth as a result of this condition, since it could shorten their wait time to access needed treatments.

Identifying a common gene in blood and colon tissue samples

Scientists from Boston Children's Hospital and Harvard University have identified a common gene expression for IBD that is present in both [blood](#) and colon tissue samples. This marker could simplify the diagnostic process by enabling physicians to identify IBD through blood tests and avoid more invasive—and lengthy—procedures, according to Jocelyn Silvester, MD, Ph.D., an attending physician with Boston Children's Division of Gastroenterology, Hepatology, and Nutrition. These findings appeared in *PLOS ONE* in October 2019.

Understanding inflammatory bowel disease (IBD)

IBD is an inflammatory disease that impacts the digestive tract. It has two common forms: Crohn's disease, which occurs at any point in the digestive tract lining, and ulcerative colitis, which is limited to the colon or rectal areas. The symptoms of IBD are often non-specific, including [abdominal pain](#), severe or bloody diarrhea, weight loss, and fatigue. IBD

can also have systemic effects, including joint pain and rashes.

At the present time, the process to diagnose (or rule out) IBD is time intensive and typically involves invasive endoscopic procedures, along with colonoscopy and imaging studies. "Current blood tests are not very specific for IBD, so a lot of children who do not have IBD may have a positive test," Silvester says.

In search of a better way to triage patients

This fact prompted Silvester and her colleagues to look at blood and colon tissue samples of 98 patients ages eight to 18 who had symptoms consistent with IBD, but who had not yet undergone an endoscopy or officially been diagnosed.

"All of the participants were sick in the same way, but the causes were different. We need a better way to figure out whose symptoms are caused by IBD and whose symptoms are caused by something else," she says.

"Using gene microarrays to characterize the gene expression pattern, we identified that the same immune and [metabolic pathways](#) were altered in the blood and the colon in kids with IBD. Most genes were also affected in the same way, so if a gene was expressed more in the blood, it was also expressed more in the colon and vice versa," Silvester says.

"The study shows that the blood-based genetic biomarkers can distinguish [inflammatory bowel disease](#) (IBD) from other disorders presenting with similar symptoms with 80 percent accuracy. That means that if a child with abdominal complaints that sound like IBD has positive blood biomarkers, it is likely that they have IBD," explains Richard Grand, MD, director emeritus of Boston Children's Inflammatory Bowel Disease Center.

Applying the findings to adults

Also of significance is that the researchers were able to use the markers found in children to identify adults with the disease. For this part of the study, the researchers used data from adults with and without IBD that was published by a different research group and available online. "The classifier we developed using our pediatric data was able to identify whether the adults had IBD with 80 percent accuracy," Silvester says.

"This shows there are fundamental mechanisms of IBD that are consistent across the lifespan and that pediatric and adult IBD are similar in important ways," she adds.

Looking to the future

The next step is to conduct further studies with a larger cohort in order to identify other [genes](#) and pathways involved in disease pathogenesis.

"Our hope is that when we test a larger sample of children using the same approach as was used in this study, the percentage of true positive identifications will increase," Grand says. "That would reduce the number of children who need to wait to have endoscopy and colonoscopy to get a definitive diagnosis," he adds.

More information: Nathan P. Palmer et al. Concordance between gene expression in peripheral whole blood and colonic tissue in children with inflammatory bowel disease, *PLOS ONE* (2019). [DOI: 10.1371/journal.pone.0222952](https://doi.org/10.1371/journal.pone.0222952)

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