Study Finds that Children with Autism and Gastrointestinal Symptoms Have Altered Expression of Genes Involved in Digestion

These changes may also affect the mix of bacteria present in the digestive tract

Researchers at the Center for Infection and Immunity (CII) at Columbia University’s Mailman School of Public Health and at the Harvard Medical School report that children with autism and gastrointestinal disturbances have altered expression of genes involved in digestion. These variations may contribute to changes in the types of bacteria in their intestines.

Full study findings are reported September 16, 2011 in the journal Autism, which is defined by impairments in verbal and non-verbal communication, social interactions, and repetitive and stereotyped behaviors, affects approximately 1% of the population. Many children with autism have gastrointestinal problems that can complicate clinical management and contribute to behavioral disturbances. In some children, special diets and antibiotics have been associated with improvements in social, cognitive and gastrointestinal function.

The investigators found that children diagnosed with autism and gastrointestinal disturbances have abnormalities in levels of genes for enzymes that break down sugars and for molecules that transport them from the lumen of the intestine into the blood. These variations were also associated with changes in the bacterial composition of the intestine.

The researchers examined biopsies from 22 patients, 15 diagnosed with autism and seven typically developing children. They used real-time PCR to measure gene expression and genetic sequencing techniques to characterize the bacteria present in the intestines of each child.

Brent Williams, PhD, research scientist at CII and first author of the study, noted that, "whereas others have looked at bacterial composition of feces, our group was the first to characterize mucosal communities and to link findings to expression of genes important in carbohydrate metabolism and transport."

“The findings are consistent with other research suggesting that autism may be a system-wide disorder, and provide insight into why changes in diet or the use of antibiotics may help alleviate symptoms in some children,” added Mady Hornig, MD, Director of Translational Research at the Center for Infection and Immunity.

“Although caution in interpretation is indicated because the sample size is small, our findings nonetheless provide a framework for developing and testing new hypotheses concerning the role of malabsorption and microflora in autism and related disorders,” said Ian Lipkin, MD, Director of the Center for Infection and Immunity.

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