Gastrointestinal Illness in Autism:

An Interview with Tim Buie, M.D.

BY KULANI MAHIKOA

© DUSTIN STELLER MODEL DOES NOT HAVE AUTISM.

"Gastrointestinal [GI] illness is not uncommon in autistic individuals," said Tim Buie, M.D., pediatric gastroenterologist at Massachusetts General Hospital (MGH). He speaks from experience. To date, Buie and his team at MGH have performed more than 2,000 scopes (endoscopies/colonoscopies) on people with autism.

Whether GI problems are more common in children with autism than the general population is not certain, Buie said. However, based on the frequency of gastro problems that he has verified among his autistic patients, Buie said he believes that a thorough GI history and workup should be a part of every person's medical assessment who has autism.

Buie is one of a handful of doctors in the world who treat and evaluate large numbers of those with autism who have GI problems. This is why referred patients can wait up to a year for an appointment with him.

Buie speculates that one reason for the reluctance of other gastroenterologists to treat people with autism is the current medical view that autism is a neurologically based disorder. This view has limited treatment solutions, primarily to behavior therapies.

In addition, people with autism present differently from the general population because they are different, Buie said. This makes diagnoses of GI problems challenging for doctors who are not familiar with these differences. For example, a child with autism who has language deficits may not be able to communicate pain in the same way that a typical child can. Often, a child with autism who cannot talk will communicate pain by actions that are misinterpreted as behavior problems. Many people Buie has treated do not even present strong symptoms of a GI condition. The heterogeneity, or diversity, of autism also complicates a physician's ability to diagnose GI problems.

Also significant is that the treatment of autism has been shrouded by controversy. Speculation

about vaccines causing colitis and autism, and the potential benefit of secretin (used as a testing tool in some children undergoing GI workup) has sent many parents of children with autism to see gastroenterologists who could not answer whether these issues were valid. Dietary and nutritional questions often are raised, but limited research into the value of diet change or use of supplements has made GI and nutrition counseling difficult.

Warning Signs

At MGH, Buie works at one of the leading autism clinics in the United States, the Learning and Developmental Disabilities Evaluation and Rehabilitation Services program, headed by neurologist Margaret Bauman, M.D. Based on their collective experiences, they have developed several "warning signs" that, if seen in a patient, warrant a GI evaluation. Some of these signs include:

1) chronic diarrhea or constipation
2) feeding/eating disorders
3) change in sleep patterns
4) food allergies or apparent changes with particular food exposure
5) behavior changes, especially self-injurious, aggressive or mouthing behaviors

Buie also is a founding member of the GI research group of the Autism Treatment Network (ATN). ATN is a collaboration of leading university hospitals that was formed to address the gap in providing comprehensive medical evaluation and treatment for people with autism, as well as the lack of evidence needed to define a high standard of medical care for people with autism.

Historical Perspective: Dietary Problems

The recognition of dietary problems in people with autism traces back to the original diagnoses of autism by Leo Kanner in1943. In his seminal paper describing autism, Kanner reported that six of his first 11 autistic patients had "feeding or dietary issues."

Since the 1950s, researchers have looked for dietary culprits as triggers for autism. In 1951, researchers D.G. Prugh, Padget Danes and C. Hans Asperger, in separate reports described abnormal, autistic-like behaviors in children with gluten sensitivities.

William Crook, in 1961, found that profound neurological behaviors, including autism and schizophrenia, resolved with elimination of certain foods in selected patients. F.C. Dohan, in 1966, correlated increased cereal and processed grains since World War II with an increased incidence of schizophrenia and autism.

In a 1971 study, Barry Goodwin reported that seven of the 15 autistic children he had randomly selected from a local community for his study had chronic diarrhea. He found that placing these children on a gluten-free diet improved GI symptoms, as well as abnormal EEG findings.

Paul Shattock reported in 1990, and K.L. Reichelt in 1991, that peptides from milk and gluten were found in the urine of people with schizophrenia and autism. They speculated that these products might have contributed to the cause of these conditions.

More recently, S. Lucarelli reported in 1995 that 36 percent of children with autism who were undergoing endoscopy for GI symptoms had allergies. In 1999, K. Horvath found a high incidence of lactose and sugar intolerance among his autistic research subjects. In 2002, he also reported that up to 50 percent of the families he surveyed indicated that their autistic children had food allergies or sensitivities.

Buie's own 2005 study with Rafail Kushak, Harland Winter and Nathan Farber showed that 59 percent of autistic children who were undergoing endoscopy for GI symptoms had carbohydrate digestive abnormalities, compared with only 11 percent in unaffected children undergoing endoscopy for GI symptoms. In this study, duodenal (beginning of small intestine) biopsies were taken from 307 autistic children and 206 non-autistic children selected for endoscopy based on a suspicion of GI problems. Results of the study showed that the frequency of lactase deficiency was higher in autistic children over five years of age than unaffected children, but the frequency was quite high even in the unaffected children with GI symptoms.

Other Important Studies

In a 1998 study, Andrew Wakefield identified lymphoid nodular hyperplasia in the distal ileum (part of the bowel) in seven out of 12 patients with autism, and found that 11 of the 12 patients had frank colitis (a form of inflammatory bowel disease). Buie points out that because of conflict-of-interest charges regarding this work, most of the other authors of this study retracted their support; however, they did not dispute the science behind the study.

Wakefield also published a study in 2000 describing "autistic enterocolitis" as a unique intestinal lesion with prominent lymphoid nodular hyperplasia and colitis. He proposed the MMR vaccine (a three-part vaccine given to protect against measles, mumps and rubella, or German measles) as the cause of the GI pathology. He also hypothesized that increased GI permeability allowed opioid peptides to cause neurological dysfunction or encephalopathic type issues. Buie said that epidemiology studies have disputed a link between MMR and autism, but to date, there have not been independent endoscopic studies evaluating findings and presence of measles virus in the tissue.

In a recent study, Buie's research team found gastroesophageal reflux disease (GERD) and/or esophagitis in nine autistic children who had presented with limited GI symptoms and behaviors not previously associated with gastroesophageal reflux. Buie said GERD is a common condition in pediatrics and should be considered in children with autism. He suggests that GERD

in children with autism may present as behavioral alterations, including aggression or self-injury, and that these behaviors should prompt consideration of underlying pain. Bravo™ pH testing (which involves testing with a capsule that collects pH data and transmits it via radio frequency to a small external pager-sized receiver worn by the patient) may allow evaluation of children who could not tolerate standard pH probe testing, he said. Buie also suggests that population-based data is needed to determine the prevalence of GERD in autism.

R.I. Furlano in 2001, F. Torrente in 2002 and Paul Ashwood in 2004 discussed immune abnormalities and abnormal cytokine profiles (compounds critical to the functioning of immune responses) in children with autism who have GI issues. Buie said he believes that current research regarding GI problems in the autistic population has not yet caught up with the realities of the problems.

Buie said the pitfalls of current autism/GI research are that:

- most of the studies are anecdotal
- there remains an absence of population-based information
- current claims of high prevalence of GI problems in autism remain uncorroborated by mainstream researchers
- much of the current research attempts to offer GI issues as causal rather than contributory to autism symptoms

A Different Perspective

Buie suggests that adopting a different perspective on autism would enhance future research initiatives. The new view would be based on these principles:

- medical issues, including GI disorders, exacerbate autistic behaviors
- recognition and treatment of underlying medical conditions will improve functional outcomes
- raising awareness of underlying medical issues among medical providers will improve quality of life

AUTHORS

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