



Is a U t I S m Medical?

By Kristi Wees

The diagnosis of autism has existed in the scientific literature for over 100 years, with the first description of it in 1911 by Eugen Bleuler, a Swiss psychiatrist, who used the term to describe one group of schizophrenic symptoms. In America, doctors such as Dr. Louis Kanner of Johns Hopkins University used the term “autism” in the 1940’s to describe children with social and emotional issues. Since then, autism has dramatically increased in prevalence, most notably so in the last few decades, from approximately one in 10,000 children in the early 1980’s to one in 68 children in 2014 (according to www.cdc.gov).

So what exactly is autism? Many people are surprised to learn that autism is not diagnosed medically, meaning there is no blood test, no MRI or other medical diagnostic procedure that can detect it. Instead, it is a diagnosis based solely on behavior without any diagnostic focus on the underlying or root cause of that behavior. Autism is diagnosed most often by a psychology professional (psychologist or psychiatrist) that determines whether or not a child meets a set of diagnostic criteria behaviors, which are outlined in a psychiatric manual known as the DSM-5. Some of those diagnostic criteria for autism include:

- Persistent deficits in social communication and social interaction.
- Restricted, repetitive patterns of behavior, interests, or activities.
- Symptoms, which cause clinically significant impairment in social, occupational, or other important areas of current functioning.

So why is this article titled “Is Autism Medical?” if autism is not a medical diagnosis? Because the current medical literature suggests there are many medical underpinnings of autism, and that it cannot solely be a mental, psychiatric or

developmental disorder. When a child is diagnosed with autism, it is important for parents, doctors and therapists alike, to take into account the wide array of physical and medical issues that a large majority of children with autism may be suffering from. These include gastrointestinal disorders, seizures, immune dysfunction, and metabolic or mitochondrial disorders. These medical “co-morbidities” of autism may have a much greater impact on the child’s “autistic behaviors” than the present day psychiatric model of autism is currently acknowledging.

GASTROINTESTINAL

It is estimated that the prevalence of gastrointestinal symptoms in children with ASD ranges from nine to 70 percent or higher. Gastrointestinal symptoms can range from colic, reflux, constipation, diarrhea and cyclic vomiting and severe abdominal pain.

In 2010 the American Academy of Pediatrics published a consensus paper on gastrointestinal complications and children with Autism Spectrum Disorders (ASD):

“In these (autistic) children, a variety of gastrointestinal dysfunctions and associated symptoms have been reported frequently.”

The publication goes on to say, “Expert clinicians have observed that aggressive and self-injurious behavior may be the primary clinical manifestations of GERD (gastro esophageal reflux disease) in individuals with ASDs, but these symptoms are frequently attributed to non-medical causes. As a result, manifestations may go unrecognized as signs and symptoms of GERD and, importantly, may go untreated.” The article also concludes, “Care providers should be aware that problem behavior in patients with ASDs may be the primary or sole symptom of the underlying medical condition, including some gastrointestinal disorders.” (PMID: 20048083)

Parents of children diagnosed with autism have reported taking their children to the doctor with concerns about constipation or severe, prolonged diarrhea, only to be told, "It is due to the autism," and then sent home. What is important to keep in mind, as a parent, is that nowhere in the diagnostic criteria of autism are gastrointestinal issues described. Therefore, if your child is suffering with these issues, it is important to advocate for medical care for the gastrointestinal issue that your child is experiencing, independent of the autism diagnosis.

Anecdotal reports from parents and physicians have demonstrated that treatment of underlying GI issues has led to improvements of "autism" behaviors and, in some cases loss of autism diagnosis. One family reported treating GERD (reflux) with diet changes and reflux medication, and, to their surprise, their "autistic" toddler stopped banging his head on the floor and began talking and making eye contact after a few months.

SEIZURES/EPILEPSY

It is estimated that approximately two percent of the general population has epilepsy. But in the ASD populations, it is estimated that approximately five - 38 percent of individuals have a diagnosis of epilepsy (recurrent seizures or convulsions). (PMID: 25440829)

Two types of seizures are generalized and partial. In an online guide for families written by neurologist, Dr. Richard Frye, these seizures are described: "During a generalized seizure, the entire brain demonstrates abnormal electrical activity. In contrast, during a partial seizure only one part of the brain experiences abnormal electrical activity." (source: TACA)

In order to diagnosis seizures in individuals with (and without) ASDs, an electroencephalogram (EEG) is performed and most often ordered by a pediatric neurologist. Various lengths of time of EEGs exist from an hour to overnight to even longer terms such as a week. This procedure can be done in a hospital or clinic setting or even ambulatory, which allows the child to be in their home or school setting. If seizures are occurring, anti-epileptic pharmaceuticals and dietary modifications are two methods with which seizures may to be controlled.

Many parents report greatly improved "behaviors" once seizures are identified, diagnosed, and adequately controlled. One thing many parents of children with ASD and seizures report is that they did not know their child was having seizures because it "did not look like what you see in the movies." Some families have experienced "absence" seizures, where a child "blanks out" momentarily or appears to be daydreaming. Gaining control of this abnormal brain activity can greatly improve a child and family's quality of life. Without testing, though, it is often times very difficult to know if a non-verbal child is suffering from seizures.

IMMUNE SYSTEM DYSFUNCTION AND ALLERGY

Few parents will think to head to an allergist and an immunologist after receiving a diagnosis of autism for their child, but for many the immune system's impact on the child's development may be one of the underlying aspects of this behaviorally diagnosed condition.

Many children with autism are found to have food and environmental allergies and intolerances when medical testing is performed. Furthermore, as documented in the medical literature, immunological testing on autistic children's actual immune composition (including T-cells, B-cells and Natural Killer cells) shows that many autistic children's immune systems vary significantly from those without autism. (PMID: 21573236, PMID: 25448709) Subsets of children with autism have also been found to have some forms of immune dysfunction and deficiency, which means the immune system is not properly responding to viruses, bacteria and even immune stimulation such as a vaccination.

So how can a malfunctioning immune system impact childhood behavior and communication? Good question. Recent research out of the University of Virginia School of Medicine reveals that there is much about the brain and the immune system that we do not understand. Recently, it was found by the lab of Jonathan Kipnis, PhD, professor in the UVA Department of Neuroscience, that the brain is directly connected to the immune system by vessels whose existence was previously unknown. The department chair, Kevin Lee, who commented on these new findings in a recent news article, expressed that "they'll have to change the textbooks" due to these new findings. It is believed that this new discovery may have an impact on all neurological diseases from Alzheimer's to Multiple Sclerosis as well as Autism. (PMID: 26030524)

Another immunological component to autism that must be considered for these children is how the body reacts to "allergens" in food and the environment. When the body determines that something is harmful to the body and launches an "allergic" response: allergy cells called mast cells dump many chemicals into the bloodstream. One such chemical is histamine, which can go on to wreak havoc on the child's system. All of this inflammation and internal agitation of the blood and therefore the body, can unfortunately lead to neurological and behavioral agitation as well.

Anecdotally, many parents have removed two common food allergens - gluten (found in grains) and casein (found in dairy products) and have reported improvements in their

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child's behavior and lessening of their child's autistic symptoms. Many parents report their autistic children's behaviors worsen with environmental triggers such as chemical sprays and fumes, pesticides, perfumes and even scented soaps and detergents. Parents are left with a trial and error approach, of removing the suspected agitating trigger from the child's environment and then reintroducing it to see if it evokes a similar response, in order to figure out what is worsening their child's meltdowns and autistic behaviors.

A local Pittsburgh developmental pediatrician, Dr. Scott Faber at The Children's Institute, and his research team have investigated the impact of environmental toxins on autistic behaviors by performing clean-room studies with ultra clean air-filtration systems in the rooms where the children slept (clean-rooms). Recent published results from these studies shows "Younger children who slept in the clean room altered elemental levels, decreased immune dysregulation, and improved behavioral rating scales, suggesting that their detoxification metabolism was briefly enhanced." This promising research suggests that removing toxins in a child's environment may have positive impacts on a child's behavior. (PMID: 25887094)

However, once again, it is difficult to know just by looking at a child if their immune system is balanced and functioning properly or if they are suffering from food or environmental allergies. It is important to speak with your child's doctor or specialist and work with them to get the testing your child may need to determine if immune and allergy issues are exaggerating a child's autistic behaviors. The guide entitled: **Medical Comorbidities in Autism Spectrum Disorder-A Primer for Health Care Professionals and Policy Makers**, found in the resources section of this article is a good resource to print and take with you to your appointment.

METABOLIC AND MITOCHONDRIAL DISORDERS

In 2005, a medical journal reported a link between mitochondrial disease and autism (PMID: 15739723). Then in 2010, a study by researchers at University of California Davis revealed that 80 percent of the children with autism spectrum disorders who were enrolled in the study had blood tests indicating some type of mitochondrial dysfunction. (PMID: 21119085)

So what are mitochondria? They are the tiny organelles found in a cell, which float outside of the nucleus but inside the cell membrane. They are often referred to as the "powerhouse" of the cells because they take nutrients from food and oxygen from the air and produce energy. Children with autism also have dysfunction or disease of their mitochondria, causing their bodies to lack the energy they need to grow and develop typically. Imagine a city during the summertime operating on too little energy, often referred to as a "brown out." This very same phenomenon is happening in a subset of children with autism because their bodies are not

producing enough energy to power their bodies and brains.

Without knowing what to look for and what to test a child for, it is very difficult to know whether any given child with autism has these underlying medical issues, including mitochondrial or metabolic dysfunction. For this type of testing, a metabolic geneticist, or neurologist that specializes in metabolic disorders can be helpful in getting to the bottom of a child's underlying medical concerns. The Cleveland Clinic has a series of medical videos that are helpful in explaining the metabolic and mitochondrial connections with autism (see the reference list for a link).

So you may be wondering, which came first - the autism or the medical condition? Does one cause the other, or is it just coincidence that they co-occur in so many children on the autism spectrum? For each family the answer to this question will be as different and unique as their child and why no "one size fits all" therapy or medication regime will help every child with autism.

One parent described her child's underlying medical issues as layers of an onion, as her family discovered medical issues with their child and treated them, slowly and consistently their child improved. Communication became better, stimming behaviors lessened and eventually a few years after they started treating the medical conditions, her child lost his diagnosis of autism.

Some will argue the child was misdiagnosed or never had autism to begin with, but for this family the time and medical detective work that they invested was well worth it in the end to have their child back. We, as parents, truly know our children better than any doctor, therapist, teacher or specialist ever will; therefore, it is up to us to advocate and continue to ask questions and look for answers for them, no matter what the diagnosis. ■

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RESOURCE LIST:

PMID= PubMed ID#: <http://www.ncbi.nlm.nih.gov/pubmed>

Medical Comorbidities in Autism Spectrum Disorder-A Primer for Health Care Professionals and Policy Makers
http://issuu.com/treatingautism/docs/medical_comorbidities_in_autism_spectrum_disorders

<http://www.autismismedical.com>

<http://www.tacanow.org>

<http://www.clevelandclinicmeded.com/online/autism-spectrum-disorders/>

<http://www.thinkingmomsrevolution.com>

<http://www.outsmartingautism.com>

<http://www.mitoaction.org/about-autism-and-mito>

<http://www.nofone.org>