

USING A BIOMEDICAL AND BIOCHEMICAL MODEL TO BETTER UNDERSTAND AND TREAT AUTISM

By Allen Lewis, MD

NINE YEARS OF GENERAL PEDIATRICS
PRACTICE HAD NOT PREPARED ME
FOR MY SON'S DIAGNOSIS OF AUTISM
NEAR HIS SECOND BIRTHDAY. THE FATHER
IN ME WAS AFRAID I HAD DONE SOMETHING
WRONG TO CAUSE HIS AUTISM AND WORRIED HE
MIGHT WIND UP IN AN INSTITUTION, A PROGNOSIS
COMMONLY GIVEN TO PARENTS EVEN TODAY.

The consensus of the experts I consulted was Applied Behavioral Analysis (ABA) was unproven and possibly harmful, and present medical therapies were ineffective. I can still recall the feelings of despair and helplessness upon hearing this verdict. Fortunately, my wife and I saw the effectiveness of behavioral therapy first-hand by meeting with other parents and we saw definite gains with ABA. She was not satisfied with just behavioral approaches and decided to try the gluten-free/casein-free (GFCF) diet. Within two weeks we noticed dramatic improvement in eye contact, health and behavior. Although initially skeptical, I had to grudgingly admit these changes were related to the diet, since we had not yet started any other treatment. These successes encouraged us to seek other ways to help our son. After careful inquiry and research, we embarked on a journey that has re-shaped my life and career. Our once sickly, nonverbal, aloof two-year-old is now a healthy, talkative, imaginative five-year-old doing well in mainstream kindergarten. Although not cured, his future is so much brighter than what I was told to passively expect.

EARLY LESSONS FROM PEDIATRIC PRACTICE

Each clinical experience teaches a new lesson. Early on, several experiences helped me to realize that autism is really treatable.

- New to autism treatment, I started with the basics, a thorough medical history and physical exam. In addition to the core symptoms of autism, my first patient had a history of constipation and a large fecal impaction on physical exam. After one enema, he slept through the night for the first time in five years and was more affectionate. This was a very powerful lesson of how a physical condition could lead to significant behavioral problems for a non-verbal child.
- The severe anxiety of a three-year-old with Asperger's disorder was significantly improved on the GFCF Diet, making it clear that the anxiety was not purely due to her Asperger's diagnosis. On the diet, she was able to participate in an early intervention program.
- Treating allergies, constipation, zinc deficiency and gluten and casein intolerance appeared to be more effective than Ritalin and Risperdal for treating aggression and school failure in a six-year-old with autism. I still remember noting

the remarkable changes when he recognized me in Costco and spontaneously struck up a conversation - a far cry from the unhappy, aloof boy I first met six months before.

It gradually became clear to me I needed to utilize many medical disciplines to help thoroughly evaluate autism and thereby maximize treatment efficacy.

EXPERIENCES FROM CURRENT DAY

Now as Medical Director of the Pfeiffer Treatment Center, I have been involved with the care of several hundred children and adults with autism. Approximately three-fourths of families report significant improvement with a combination of behavioral, educational, psychiatric, medical, dietary, and biochemical therapy. Some children have recovered and no longer have the diagnosis of autism. Some have also had little or no improvement. To better understand the differences in outcome, we are constantly looking to better characterize a patient's biochemistry and tract their response to treatment. There are things that can be safely undertaken today, and time is of the essence. The earlier treatment is initiated the better. Also, it is a combination of therapies that improve outcome. One must focus on the educational/ behavioral therapies to achieve learning and brain maturation AND the treatment of the medical problems to help minimize any functional roadblocks to normal brain function and thereby learning.

AUTISM IS TREATABLE

It is not hard to understand the difficulty believing children have recovered from autism, if one believes it is untreatable. I believed this once myself. Autism needs to be thought of as an illness with psychiatric manifestations of underlying medical and neurophysiologic problems and not as an isolated psychiatric illness. The negative stigma associated with mental illness has unfortunately reinforced the opinion that there is little chance for effective treatment and delayed efforts to better characterize and treat the other medical conditions found concomitantly in autism. Fortunately, documentation of problems with gastrointestinal, immune, neurological and metabolic functioning in autism is becoming more readily available and accepted.

I am very thankful for the parents and professionals who have helped me better understand autism. The paradigm or idea of untreatable autism failed to hold up in my clinical experience; therefore, I have had to adopt a new one:

Autism is the psychiatric diagnosis of medical illness with:

1. a biologically impaired brain,
2. multiple medical problems,
3. both genetic and acquired features,
4. leading to developmental, learning and behavioral problems,
5. that is treatable.

PIECES OF THE AUTISM PUZZLE

As we begin to think of autism as a medical condition, it is helpful to identify the individual pieces of the autism puzzle. Not only does it enable us to better understand the reason for the diversity of symptoms present in the autism population, it leads directly to the development of new therapies.

Neurologic, toxicologic, immunologic, gastrointestinal and metabolic/biochemical abnormalities represent only some of the medical problems being identified in autism. I recommend studying the presentations from ASA's *2004 Racing Towards Answers in Autism Conference* to learn more about some of these abnormalities. For many years the Pfeiffer Treatment Center has focused on biochemical individuality and its effect on behavior, mood, development and mental illness.

BIOCHEMICAL ASPECTS OF AUTISM

Biochemistry matters in behavior. Most pediatricians know that iron supplementation helps toddlers with breath-holding spells and that low iron is associated with poor learning. This is just one essential mineral, and I have learned there are other biochemical disturbances that affect learning, mood and/or behavior.

In fact, all behavior and learning have their roots in the basic biochemical and neurochemical processes of the brain. The brain is a chemical factory, which produces neurotransmitters, receptors and other complex biochemicals that make communication between nerve cells possible. The only raw materials for synthesis of our brain chemicals are vitamins, minerals, and amino acids from our diet. Genetic abnormalities, nutritional deficiencies and some medical conditions can cause striking differences in the levels of these raw materials available to the brain.

The biochemical processes in the body are very complex and intertwined and due to genetic factors, individuals likely have varying needs for the same raw materials. If the brain is presented with improper levels of raw materials required for neurotransmitter production, one can expect problems with mental functioning. One advantage of vitamins, minerals and amino acid supplements is that the body has mechanisms to deliver them to places of need. However, they should not be used without careful monitoring by an experienced medical practitioner. The Pfeiffer Treatment Center has learned that normalization of some of these nutrient factors is an important part of the effective treatment for autism.

The Pfeiffer Treatment Center (PTC) has been studying the effects of biochemical imbalances in children and adults for nearly 15 years. Since 1979, approximately 18,000 patients have been evaluated and treated for biochemical imbalances, including 4,000 with a diagnosis of autism spectrum disorder (ASD). In October of 2004, the PTC published an open-label study of nutrient therapy showing a 91 percent reduction in physically assaultive behavior and 88 percent reduction in

destructive behavior in developmentally normal children and adults. There is growing evidence that biochemical imbalances play an important role in behavior, but placebo-controlled studies must be completed before nutrient therapy can become part of mainstream medicine.

Biochemistry matters for autism. We have amassed a large database of biochemical assays for autism-spectrum patients and it is clear their biochemical measures are very different from the rest of the population. The vast majority of patients exhibit symptoms and laboratory findings of copper excess, zinc deficiency, pyrrole disorder and disordered methylation. We are currently engaged in several database studies, which we intend to publish in peer-reviewed journals. Exploring the causes of these biochemical imbalances has convinced us of the importance of oxidative stress, which has become a unifying theme for many of the imbalances discovered.

Findings of increased copper, decreased zinc, elevated toxic metals, decreased metallothionein, low glutathione and methylation abnormalities all point to significant problems with oxidative stress. Oxidative stress involves free radical damage to key proteins and vital tissues, which thereby compromises their function. Interestingly, the brain and GI tract (usually affected in autism) are especially susceptible to oxidative stress. Furthermore, oxidative stress is thought to be important in Alzheimer's disease, Parkinson's disease and other neurodegenerative brain disorders. In fact, oxidative stress may be one of the unifying themes for neurodegenerative diseases in all ages. Genetic individuality results in differing susceptibilities to the myriad of factors associated with oxidative stress. For this reason, we are engaged in a collaborative study of oxidative stress in autism with researchers from Case Western Reserve University and the University of Pennsylvania. This NIH-sponsored project involves the measurement of damaged proteins, such as levuglandins and isoprostanes, in treatment-naïve autism patients and controls.

There are several convincing studies indicating a high population of short, undeveloped brain cells in an autistic brain in comparison to a non-autistic brain. This suggests that in autism the brain has not completed its' normal maturation process. Excessive oxidative stress may be an important factor in the disruption of the natural process of growth, regulation and pruning of brain cells.

In summary, the brain in autism seems to be biologically impaired, but the impairments may be treatable. Behavioral and educational therapies likely promote the development of new brain cells and the connections between cells that are vital to improvements in speech, cognition and socialization. Treatment of oxidative stress could help attenuate the ongoing impairment of normal functioning.

HOW TO HELP YOUR CHILD

The better one understands the disease process itself, the more likely effective therapies will be found. Thinking of autism as a medical illness with a biologically impaired brain

and potentially other affected organ systems (i.e. the GI tract) leads to more effective treatment strategies. The following are my suggestions for helping a child on the autistic spectrum.

Educate yourself. Autism is a complex illness. With time and dedicated inquiry, you will be better able to identify the most promising treatments for your child.

Focus on evidence-based therapies and strategies. Seek out other parents who have had success treating their child and share ideas; however, realize your child's path will be unique. Let your child's history and symptoms guide your choices. And expect new insights along the way.

Maximize your child's learning. The goal of medical treatment is to make the brain more susceptible to learning. To take advantage of this, investigate the appropriateness of speech, behavioral, occupational and sensory integration therapy for your child. As in my son's case, your child's therapists and teachers will be your most valuable and dedicated assets in helping you help your child.

Carry out a trial of the GFCF Diet. More than 75 percent of families report positive improvements in health and the core symptoms of autism following gluten and casein elimination. Controlled studies of the GFCF diet and the ketogenic diet have indicated improvement. A dietary trial is safe and is the best way to determine if dietary restrictions are of benefit. I recommend starting with elimination of casein followed by elimination of gluten after 3 weeks. If no improvements are noted in 3 – 6 months, reintroduce casein then gluten. The effects of the diet are usually most apparent just after elimination or reintroduction of casein or gluten. The take home lesson of dietary intervention is to reexamine the eating habits of your family. The standard American diet is unhealthy and striving for better nutrition for the whole family will pay off with better health for all.

Rule out gastrointestinal problems. Constipation with fecal retention, colitis, digestive enzyme deficiencies, gastroesophageal reflux, abnormal bacterial flora and food allergies have been reported to be more common in children with autism; therefore, they merit exclusion. Generally, a KUB X-ray can help determine if stool is being retained or if there is evidence of impacted stool found in encopresis, a common problem for constipated children. In my experience, more than 50 percent of patients seem to have some degree of fecal retention. Diarrhea may actually represent overflow around an obstruction, so get a KUB anyway. If not due to retained stool investigate the possibility of malabsorption, celiac disease, parasites and/or abnormal flora. Retained stool can be treated like encopresis; your pediatrician can help you with this. Find a knowledgeable pediatric gastroenterologist for persistent or difficult to treat problems with constipation, diarrhea, gastrointestinal pain, poor growth or if unable to manage the retained stool.

Rule out food and inhalant allergies. Allergies can affect cognition, learning and behavior. See an allergist for concerns of seasonal, perennial or food allergies or if current allergies have been difficult to treat. The most important treatment for allergies is avoidance; therefore, proper identification of the allergen (the thing we are allergic to) is

especially important for the difficult to diagnose allergens such as foods and for the allergies not responding to typical treatment.

Rule out sleep disorders. Sleep apnea is associated with ADD and behavioral problems even if daytime sleepiness is not obvious. Snoring and interrupted sleep need to be investigated by a pediatric ENT or sleep specialist to rule out tonsillar or adenoidal obstruction of the airway, allergy or gastroesophageal disease.

Pursue a metabolic assessment and work-up with a pediatric metabolic/geneticist or neurologist, if there is a history of seizures, hypotonia (low muscle tone) or rapid collapse in development associated with illness, injury or vaccination. These are potentially red flags of metabolic disease and should be investigated by a specialist.

Find a medical professional to help with a biochemical assessment and to be sure other important diseases or medical conditions are ruled out. Nutrient supplementation, dietary intervention, and other treatment need to be monitored for safety and efficacy. Keeping a treatment log can help you and your caregivers with determining what really helps. Keep also an up-to-date record of current status and problems, current therapies, relevant illnesses and responses to treatment. Such a summary can greatly help a caregiver evaluate treatment options and develop an effective program based on your child's needs.

SUMMARY

Autism is treatable. Many persons with autism are physically ill. Think about causation and remember behavior may be

a clue to the underlying problem. Treatment should address the underlying problem and not just the symptoms.

Remember there is HOPE. Take things one-step at a time. Educate yourself and develop a plan. Trust your instinct in making difficult choices. Focus on the whole family, as autism affects everyone—parents, siblings, grandparents and more. Health begins at the dinner table with good food, good conversation and a family's love.

Many families have expressed their personal gratitude and thanks for the positive changes in their life. Healing the person, the family and ourselves is not of trivial importance. When I step beyond the controversy about causation, treatment and cure, I remember to accept my son for the fun, playful, loving child he is. Despite the past and continuing challenges, our family has grown stronger in health and spirit with him in our family. As a pediatrician and as a father, I've learned that our children can heal us as much or more than we can heal them.



Editor's Note: Please refer to ASA's Web site, www.autism-society.org, to access ASA's option policy.



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