



Functional Medicine

The Way to Treat Autism Now

BY JEFFREY BLAND, PH.D.

“It is more important to know what patient has the disease than to know what disease the patient has.” So said by the very wise Sir William Osler, whom some call the father of modern medicine, some 100 years ago. People have processes, they don’t have diseases. When we codify processes into diseases, we do it to make things simple to understand and to memorize. But with simplification, context—and insight—are lost. So many chronic medical conditions that are on the rise today, of which autism is one, are poorly served by being reduced to a disease diagnosis. In the medicine of the future, we will need to restore this context and insight. The personal and biological histories of each patient are complex, and this complexity offers opportunities for treatment and reduction of the burden of suffering.

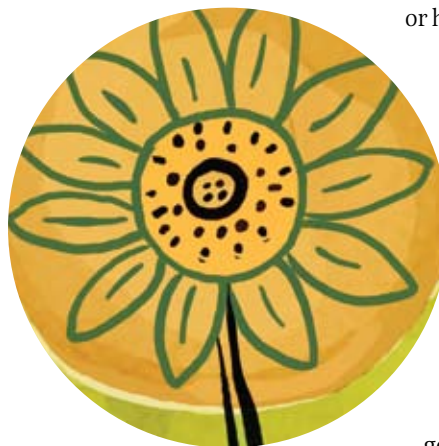
Advances in molecular medicine also support the return to the individual patient. Knowledge about genetic and metabolic individuality and tools for measuring these things now make it possible to aim for a truly sophisticated individualized medicine.

The framework of functional medicine (Jones 2005) is particularly well suited for meeting the needs of patients with chronic medical problems. Key to its approach is to integrate the patient’s story with advanced biological insight. Measures of clinical imbalance are tools to augment the power of this story. Functional medicine trains

practitioners to integrate diagnosis, signs and symptoms, and evidence of clinical imbalances through the lens of the patient’s story into a comprehensive approach to improve both the patient’s environmental inputs and his or her physiological function.

As science progresses, and as we listen to patients and their families, we are learning that autism is not a single disease, but rather a variation of a theme of neurobiological behavior. It is not a consequence of a flawed gene or a damaged child. In fact, many people with autism are brilliant.

They have insights, intelligence and capabilities that would be considered extraordinary, even while their behavioral patterns may not socialize comfortably in many settings.



The word “autism” is a term of medical classification or taxonomy. Autism, along with other diagnoses that fall under it, such as Asperger’s syndrome or autistic spectrum disorder, becomes a label we attach to a set of observations in the hopes of leading to a single treatment regime or a determining gene for the condition, as if it were a specific disease.

Many sensitive clinicians appreciate that in describing the effects we associate with this classification, we often lose the sense of the individual child in that definition. Such clinicians seek a vocabulary for honoring how each child, each person, has their own presentation, their own history, their own genes and their own environment. In looking at autism as a variation of a theme of neurobiological behavior, we realize we can stigmatize children by labeling them, and define their outcome rather than recognizing this condition as having tremendous plasticity in its presentation, progression, regression, improvement and even recovery.

THE FUNCTIONAL MEDICINE APPROACH

Autism, with its heterogeneity, complexity, and chronicity, thus poses many challenges that functional medicine is well designed to meet. The difference between functional medicine and traditional diagnostic medicine is that functional medicine is less concerned about diagnosis and more concerned about mechanisms. This is because the outcomes that we call disease are really secondary, downstream effects of altered mechanisms (imbalances, defects or alterations in physiology). As the idea of autism as a single disease comes apart, focusing on mechanisms may provide a new way to approach autism systematically and comprehensively while still honoring individuality and complexity.

The functional medicine approach trains clinicians to look not only at the surface signs and symptoms, but to use a biological zoom lens to look beneath the surface at the underlying mechanisms. Molecular medicine is identifying patterns of imbalance in cellular communication, cellular energy metabolism, cellular replication and repair, cellular waste elimination, cellular protection and defense, and cellular and intercellular transport and circulation that are not disease specific but found in various combinations and fashions in a wide variety of disease processes (Bland 2008). Autism researchers and clinicians have identified abnormalities at many of these levels, including hormonal and neurotransmitter imbalances, cellular oxidation-reduction imbalances and problems with energy metabolism in the subcellular mitochondria,

the responses of cells from individuals with ASD to challenges by toxins, the metabolic state of immune cells, cellular inflammation, and alterations in cellular membrane structure and function (Zimmerman, in press; *American Journal of Biochemistry and Biotechnology* 2008). No one of these abnormalities is clearly unique to autism, but as these abnormalities combine and progress, they can lead to alterations in the function of the central nervous system whose outcomes we, as social beings, perceive at the behavioral level.

There is a striking congruence between functional medicine, a biological framework, and functional assessment, a behavioral framework. The emphasis on mechanism rather than diagnosis in functional medicine directly parallels the emphasis on motivation rather than topography in functional assessment. Functional assessment, an aspect of behavioral psychology, avoids a one-size-fits-all approach to understanding and treating behavior problems. In illustration, if several individuals exhibit aggression, one would not apply a single treatment to all of them based on the topographical similarity of their aggressive behavior. Instead, one would ascertain the underlying motivation for such behavior. One might find that one person’s aggressive behavior was motivated by a desire for attention, another’s by a desire to escape from an unpleasant situation and still another’s by a desire for a favorite but unattainable toy. The differing patterns of motivation would then drive the selection of individualized treatments, each addressing a different motivation (Carr et. al 1994). The conceptual similarity between functional medicine and behavioral psychology could serve as a catalyst for a grand synthesis of the biomedical and behavioral approaches to autism (see Carr & Herbert, this issue).

APPLICATIONS TO TREATMENT

How does this knowledge influence treatment? Surely, many behavior and socialization problems cause difficulties and discomfort, and the person and the people around him/her would often have easier lives were these controlled. But how? Are we going to put all these children on various types of medications—such as stimulants, noradrenergic agents, serotonin uptake inhibitors, anxiolytics, opioid antagonists, anti-convulsants, sleep aids, or anti-inflammatories? All of these medications are used in patients with autism, and sometimes children are polypharmacied with multiple medications. This approach may manage some of the symptoms, but it does not address either

core symptoms or underlying causes. We need to emphasize that no existing drug or other treatment from traditional, medical intervention cures autism.

In particular, drugs certainly don't manage the underlying mechanism driving the features that are problems: the alterations in cellular processes leading to neurobiochemical dysregulation.

Given the range of cellular-level problems that are being identified, it is becoming clear that we are dealing with something more than simply neurobiochemical dysregulation. We are appreciating that the "neuro" constraint is too restrictive.

Stories from patients and families tell us that autism is not just in the head; it appears that there are other things going on in the

gives rise to the outcome in that patient's presentation that we later call autism?" Asking this question includes an appreciation that autism is not just "hard-wired" in the genes of a child. Instead, it is the subtle connection between children's environment and their genetic strengths and weaknesses that gives rise to the outcome that is diagnosed and called autism.

Functional medicine grounds itself in scientific principles and information widely available in medicine today. It does not focus on a single treatment for a single diagnosis, but rather combines research from various disciplines regarding the classes of cellular and physiological function discussed above and how they may be modulated. Functional medicine assesses antecedents (such as

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child's physiology more systemically that connect to give rise to altered neurobiochemistry. Even more, many of the other body systems that are so often involved are intimately interrelated with the brain. As Dr. Michael Gershon (1999) writes in his book, *The Second Brain*, many of the neural chemicals that regulate brain function are produced in the gastrointestinal tract. In fact, two-thirds of the body's serotonin (a mood-modulating neurochemical) is produced in the gut mucosal area. So it seems that looking only in the brain of children with ASD is incomplete, and that we also need to look elsewhere for signals or messages that modulate brain chemistry and produce what we later call autism. This is opening up a big door for looking systemically at the potential etiology of autism.

The frequency of this disorder is ever increasing. The Centers for Disease Control (CDC) indicates that one in every 150 children is now diagnosed as having an autism spectrum disorder, and one in 6 children suffers from a neurodevelopmental delay. We need to entertain other hypotheses about autism treatment. We need to move beyond asking what drug will treat the symptoms, and instead ask what *mechanism* creates altered neurochemical or neurobiological function or systemic physiological change. Once you ask that question you are into the functional medicine milieu. You are asking, "What is the web of physiological function that

genetic risk factors), triggers (such as environmental or emotional stressors), and mediators (such as the health or nutritional state of the person), and looks for features among these that may be treatable. By finding ways to intervene at multiple levels, functional medicine aims to improve the management of complex, chronic disease and restore each patient's functionality and health.

THE METABOLIC COMPONENT

A functional medicine approach to autism (with its exploration of underlying cellular processes) has roots going back to Bernard Rimland, who in 1964 published what became a best-selling book titled, *Infantile Autism: The Syndrome and its Implications for a Neural Therapy of Behavior*. This was a revolutionary book at the time because until it was published, autism was thought to be a result of poor mothering. Among many other things, Rimland discussed nutritional intervention, which had nothing to do with parenting or mothering, but rather addressed a functional need that children might have had for their brain chemistry—for one of the cofactors of brain chemistry function is pyridoxal phosphate (a vitamin B6-derived cofactor).

For reasons still unknown, children with autism may have metabolic vulnerabilities, some of which respond to nutritional intervention. These types of metabolic vulnerabilities are starting

to make more sense as we learn about genetic changes called single nucleotide polymorphisms, or SNPs.¹ In old-school thinking, you either had a genetically based metabolic condition, or you did not. Now we are learning that any one gene can have hundreds to thousands of different SNPs, each affecting function in a different way (sometimes subtly, sometimes not). Changes in even one gene can predispose an individual to an incredible range of outcomes, varying both in type and severity. Not all of these differences will manifest as classic metabolic disorders found in textbooks.

This approach was masterfully presented in a 2002 review by Bruce Ames of the several hundred references in the literature that had been published over the 30–40 years since Dr. Linus Pauling first proposed the orthomolecular medicine concept in his landmark 1967 article in *Science Magazine* titled, “Orthomolecular Psychiatry.” Dr. Ames’ review, published in the *American Journal of Clinical Nutrition*, demonstrated, through the results

the body. Whereas appropriate nutritional supplementation may correct impaired metabolic function, environmental exposures to chemicals, infections and other agents may further impair it. There is substantial literature to support this concern. Vulnerability to these exposures is related to the capacity of cells to handle the challenges posed by the environment. Our bodily function is compromised as a consequence of an imbalance between cellular capabilities and environmental exposures. The more vulnerable the cells, the less toxic exposure is needed to cause damage, while the larger the toxic exposure, the more the cells and organism are affected even when they are less vulnerable.

Long before these exposures kill us or give us an obvious disease, they will impair our function. This includes the development and function of our brains and intelligence. A functional medicine framework gives us the space to think of autism as well as other learning, language processing, coordination, attention

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of multiple studies and laboratories around the world, that the few early investigators (like Drs. Rimland, Abram Hoffer, Linus Pauling and Roger Williams) who initiated this concept can now be appreciated for framing a new form of medicine. Now called molecular medicine, this approach explained how nutritional substances, due to their roles as cofactors to vital biochemical processes, could actually improve function in individuals with needs at greater than the RDA (recommended dietary allowance)—a dramatically new concept in the management and treatment of “disease.”

IMPACT OF ENVIRONMENTAL EXPOSURES

These concepts of functional impairment and functional intervention set the stage for an appreciation of the impact of environmental exposures on the organism—on both the brain and

and behavior impairments not so much as disease pathologies as functional changes in the organism. And yet, in medicine, we are still in search of the Holy Grail of the diagnosis as if, through it, we will understand its treatment. But the functional changes that precede diagnosis or, later, contribute to making it worse, are where we can most likely intervene with lower technology, lower expense and lower risk, and higher outcome in performance.

This gene–environment interaction gives rise to these functional decrements that precede the onset of pathophysiology and demonstrate whether a society is healthy or sick. It has been said that one of the reasons the USSR fell apart was because the workplace environment was so polluted, and such high levels of body burden were placed on young men and women, that absenteeism and illness (particularly respiratory illnesses) became so frequent that overall productivity was compromised to the point

¹ For an animated tutorial on SNPs, see <https://www.23andme.com/gen101/>.

that they could no longer manufacture and produce enough for the large size of the country. As a consequence, the USSR could not even mount an effective military because so many young men had health problems that were not allowing them to pass muster as candidates for the military. These are not pathophysiological effects; they are subtle effects that undermine the patency, or capability, of a society, and are the signs of impending serious health effects. Is the rise of autism and other chronic diseases a sign of impending breakdown of the medical well-being of citizens in the U.S., and a threat to our social fabric?

USING FUNCTIONAL MEDICINE NOW

Now, in the midst of increases in so many chronic conditions, including autism, can we still afford to wait in the wings, delaying the use of our medical remedies, until conditions are serious or even endstage, knowing that, at best, we may be marginally successful? The functional medicine alternative is to address functional impairments without waiting for the big “D”—disease diagnosis—and to treat these underlying mechanisms with the understanding that this will benefit the well-being of the whole person. Autism is putting us in a race with time to respond with functionally effective treatments and to stem the tide of whatever environmental challenges may be increasing the numbers of this condition, among others. If autism is the “canary in the coal mine,” our response to this challenge will impact the well-being of present and future generations.

The challenge of autism presents us with an extraordinarily important opportunity—to discover new paths, while also learning old things in new ways. We have the opportunity to help not simply through the use of medication, but by changing the environment of children; by working with them on their speech patterns, attentiveness and contact with the world; and by exposing their bodies to foods and substances that help rather than harm. Their sociability improves as their physical function improves. We are each of us connected as a holograph, one part to the other. The brain doesn't sit in isolation to the rest of the body. A functionally

effective individualized approach to autism may guide the way to an individualized, molecularly sophisticated medicine of the future and a means to revitalize the lives of people with chronic illness and recharge the health wellsprings of our society.

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