

# The Cutting Edge

## FROM RESEARCH TO PRACTICE

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# On the Diagnosis and Misdiagnosis of Autism Spectrum Disorder

On Thursday, March 29, 2012, the CDC released new official autism prevalence statistics. The “official” autism rate is now reported be 1 in 88, up from 1 in 110 in 2010, and up from the 1 in 166 rate in 2008.

In recent years, I have observed what appears to be a trend that may have implications for the current DSM V controversy regarding the perceived need by the committee to “tighten” the criteria for diagnosis—a move that has generated a firestorm of criticism by family members of, and people with ASD. What—you might ask—does this have to do with the new CDC autism prevalence statistics? In a word, *plenty!* Bear with me while I explain.

Over the past decade, I have seen an increase in the number of children blatantly misdiagnosed as being on the autism spectrum, when they clearly do not meet current DSM IV—TR criteria for diagnosis. To be clear, these are *NOT* children whose behavior meets the “expanded” diagnosis that is captured by the DSM subcategories of PDD-NOS and Asperger’s disorder. I am referring to children who are unquestionably misdiagnosed, and whose numbers may be so significant that they are not only dramatically affecting the skyrocketing prevalence rates, but also contributing to the DSM V committee’s efforts to “stanch” the so-called epidemic by tightening the criteria for diagnosis.

One explanation given for the increased prevalence rates rests on the issue of “diagnostic substitution,” which is a

politically correct way of saying that children who today receive a diagnosis of autism would likely not have received that diagnosis years ago. This explanation only goes so far, however, and does not take into account the issue and dangers of misdiagnosis. Based upon over 40 years of autism-specific experience (which includes the development of diagnostic tools to identify young children with ASD and other developmental disabilities), I believe that misdiagnosis may be a more accurate and realistic contribution to the rapidly-rising autism prevalence rates. Furthermore—and *the reason the issue of misdiagnosis deserves serious consideration*—this dramatic increase in the diagnosis of ASD may lead to a backlash, of which the controversial DSM V proposal for tightening diagnostic criteria may be just one example.

What leads me to make this provocative (and likely unpopular) assertion regarding misdiagnosis in the face of the great controversies surrounding the “autism epidemic” narrative? The answer is: *my direct daily experiences with children, professionals, and families, and my considerable experience in autism.* Most of my professional activities over the past fourteen years have centered around

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consultation to preschool and early childhood programs, although my professional activities involve the full age range of individuals with ASD. In this work, I have observed very young children entering these programs with ASD diagnoses, often from highly-reputable hospitals and clinics, mostly in the Northeast where I am based. Currently, in any given month in my consultation visits across three New England states, I regularly hear the following type of concern from experienced early childhood professionals: *Barry, a three-year-old child with an ASD diagnosis just started our program (or, one of our kids was just seen for an outside evaluation and formal diagnosis). We are interested in YOUR opinion.* This last statement has become “code” for: *We are looking at a child with an ASD diagnosis, and we just don’t see it.*

It is important to note that while the issue of misdiagnosis is a frequent concern of professionals “in the trenches,” there appears to be less concern about the issue in the academic/clinical discussions with respect to DSM V and the boundaries of diagnosis. To be sure, acknowledging the possibility of significant levels of misdiagnosis is a risky proposition, as it obligates professionals to question the credibility of the actual *diagnostic process*, as well as the *experience and training* of diagnosticians. It also directly impacts other crucial and divisive issues—for example—whether “recovery” from ASD is a valid phenomenon. Clearly the stakes are high, and as such we need to ask: *Are we being as vigilant as we need to be in diagnosing children, given that a diagnosis of ASD is a life-changing experience for families.* As one father stated, “Once your child receives a diagnosis of ASD, the life of the family forever changes in profound ways.”

Before examining the issue of misdiagnosis more closely, I want to clarify an important point. I have come to use the qualifier *blatantly* with the term *misdiagnosed* to differentiate these children from those who “reside” in the “grey area,” that is, those children who now receive ASD diagnoses under the expanded application of the DSM-IV criteria noted previously. To be clear, I have no problem in thinking of them as children “on the spectrum,” as the concept of a *spectrum* implies shared characteristics with others along a continuum.

### **Artifacts of the Diagnostic Process**

My concerns about the process of diagnosis go back to the varied experiences I have had in observing, assessing, and diagnosing children, arguably in as many settings and contexts as possible: hospitals (both in-patient and out-patient settings); university clinics; preschool and school programs; children’s homes; community settings, including parks and playgrounds; and family- and child-friendly, home-like office settings. I have worked in situations where parents have been intimately involved as partners in the assessment process, and I have encountered those in which parents have been asked to sit on the sidelines, possibly contributing to misdiagnosis, and too often causing them to subsequently express resentment about feeling disenfranchised from the process.

The reason I mention these different types of diagnostic settings is that I have found that the nature of the setting itself, and degree of caregiver involvement affects—sometimes profoundly—the behavior of the child that is being observed. Of greatest significance, children’s behavior is most significantly impacted in the domains of social relatedness, social communication, language, and emotional regulation—the very domains that require the greatest scrutiny for an accurate diagnosis of ASD. Thus, artifacts of the diagnostic process itself (such as the setting and who is involved) may be significant contributing factors to misdiagnosis, as children who are more socially “shutdown” or less communicative due to the setting or inexperienced/poorly trained staff will present with the social symptoms more consistent with ASD. This is not a new phenomenon; however, too often, recommendations to address this problem—for example, observation of the child in various settings and at different times—are ignored. Hence, typically diagnosis is a one-shot, one-visit endeavor that may involve meetings with multiple professionals in a short period of time, the result of which may be increased anxiety for the child and family. Additionally, although allowed, provisional diagnoses are rarely utilized unless a child is extremely young. A provisional diagnosis means, *this diagnosis is a working hypothesis, and we will be gathering more information and revisit this diagnosis at a later date to see if it is accurate.*

### **Variables within the Child**

So, if my “misdiagnosis hypothesis” holds water, what variables within the child might be contributing to the problem? In virtually all of the cases I encounter, there are valid concerns about the child’s development. I refer to most of these children as having language or communication problems

PLUS. In other words, they do not meet accepted criteria for ASD, especially in the domains of social relatedness and social abilities, but they do present with challenges in language and communication development, with at least one or a number of other observable symptoms. These symptoms may include anxiety and social reticence, or challenging behaviors such as impulsivity, high activity levels, or uncooperative behavior. They may also have problems regulating their emotions, resulting in mood swings, shutdowns, or meltdowns. To complicate the situation further, they may also demonstrate other behavioral patterns that, in and of themselves, are not definitive of ASD. Such symptoms include problems with attention, unusual responses to sensory input, echolalia (or frequent repetition of others' speech), motor speech disorders resulting in limited or unintelligible speech, repetitive motor behaviors (sometimes referred to as self-stimulatory behavior), a narrow range of interests, adherence to rituals or routine, and a heightened need for predictability and consistency in their lives. All of these symptoms may be observed in children on the autism spectrum, but they are also characteristic of some other children with developmental challenges, many of whom have relative strengths in social relatedness, social interaction, and social understanding, and therefore do not meet criteria for ASD.

### **Inexperience as a Factor in Misdiagnosis**

One might ask the following question: *How could children with relative strengths in social abilities receive an ASD diagnosis that, by definition, is primarily a disability of social relatedness, social communication, and social understanding?* That leads to yet another “cog” in the “wheel” of misdiagnosis—*inexperience*. For example, inexperienced professionals, with narrow, preconceived notions of what ASD is, may place too much weight on symptoms that although *associated* with ASD, are not necessarily *definitive* of ASD. In other cases, and as noted above, problems in social relatedness and social interaction observed during the diagnostic process, may be artifacts of the unfamiliarity and artificiality of the setting itself. Furthermore, often when these children are observed in more familiar, less anxiety-provoking settings such as at home or in preschool, they do not demonstrate the social reticence, social “shutdown,” or limited communication observed in more contrived settings. In fact, in the preschool setting, they may be observed to be more related to peers and to demonstrate more advanced play skills, than would be possible in other settings.

I have seen these differences first hand, sometimes on the same day for the same child, as a result of observing a child in many settings. Extreme variability in the behavior of all children is to be expected, but even more so for children with developmental challenges. To complicate matters, I have observed that when school-based professionals with concerns about the accuracy of a child's diagnosis contact the diagnostic team or individual diagnostician to request an observation of the child in the more familiar setting, it very rarely happens. Most often, the ASD diagnosis “sticks,” and parents typically consider hospital- or clinic-based professionals to be more knowledgeable about ASD than school personnel, which may not necessarily be the case. Furthermore, those parents who have come to some acceptance of the ASD diagnosis, and feel the relief of finally knowing what they are dealing with, may understandably be hesitant to pursue other opinions, even when doubts about the accuracy of the diagnosis of ASD are expressed to them.

Before proceeding, it is worthwhile to consider further the effect of inexperience on the part of diagnosticians in the misdiagnosis of ASD, since the increased numbers of children referred for evaluations, and the associated backlog, has created a need for more and more diagnosticians. This situation has paved the way for less-experienced professionals to diagnose children. In decades past, most professionals providing diagnosis were truly ASD experts. It is now more common for persons in training (e.g., pediatric and psychiatric fellows and psychology interns) or those who may not be ASD specialists (pediatricians, neurologists, or psychiatric social workers) to play a significant role in diagnosing children. Clearly, the level of expertise and ASD-related experience may be quite variable among these professionals, especially when there is a need to differentially diagnosis ASD from other complex disabilities that also affect social communication, language development, social relatedness, and emotional regulation.

### **The Differing Goals of Diagnosis**

Another factor contributing to the increasing autism prevalence rates—and one that is exacerbated by inexperience on the part of the diagnostician—has to do with the goal of diagnosis. Specifically, I think of diagnosis of ASD as falling into three distinct categories. First, there is the *Clinical Diagnosis*, in which the goal is to appropriately differentiate ASD from other developmental disabilities. Ideally, this should be provided by well-trained professionals based on DSM criteria (often using validated tools that inform diagnosis such as the ADOS). Second, there is the *Educational*

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*Diagnosis*, which occurs when some states, agencies, or school districts modify or set their own criteria for ASD that may differ from DSM criteria, and will only provide specialized ASD services for children meeting those criteria. Finally, *Political Diagnosis* occurs when a well-meaning professional or diagnostic team provides a diagnosis of ASD, even if a child does not meet the minimal threshold for diagnosis, in order to open up doors to programmatic placement or funding for services. In these situations, it may be recognized that the diagnosis of ASD is unclear—for example, the child is close to meeting the symptom threshold for diagnosis—but does not meet full criteria. I have directly heard from a large number of parents over the years that they were told by the diagnosing team or professional that their child does not meet full criteria for diagnosis, but that he or she “needs” the diagnosis to get the level of services that would be most beneficial. What is even more disturbing (and clearly beyond the scope of this article) is that in some cases, the professionals who provide the diagnosis are also affiliated with the agency or clinic that would be funded for providing the services. Clearly, these differing goals of diagnosis may substantially impact the perception of the extent of the increased prevalence in ASD.

### **The Extent of Misdiagnosis**

Before addressing the potential fallout from misdiagnosis, it is important to consider the extent to which it may occur? In my experience, the numbers may be as high as 15-20% of young children receiving ASD diagnoses. This number is an anecdotal estimate based upon my own experience. That said, this issue needs to be examined more specifically, as research indicates that demographic factors may have a significant influence (i.e., regional, ethnic and racial differences). For example, research suggests that ASD may be under-diagnosed in African-American children, and over diagnosed in regions where there are more services available. For example, the recent CDC data indicate that in New Jersey prevalence rates are 1 in 49 children in contrast to the national rate of 1 in 88. Is it a coincidence that New Jersey has a high density of special schools and services for ASD? To be clear, I am NOT claiming that the significant increase in the prevalence of ASD can be accounted for

primarily by misdiagnosis. I do agree that there has been a significant increase in accurately diagnosed children, but misdiagnosis may be so common as to contribute to the “explosion” of children with ASD, and to the pressure felt by members of the DSM V committee to make criteria more stringent.

### **Coming Full Circle—the Ramifications of Misdiagnosis**

One may argue that even if misdiagnosis is occurring at a significant rate, these misdiagnosed children will nonetheless benefit from the more intensive services they will be afforded as a result of the label. However, it is not that simple, as such a practice has wide-ranging implications. First, if service providers are flooded with requests for intervention for children who do not meet criteria for autism, those that do meet criteria may go untreated or receive an inadequate level of service. Second, some interventions that are considered appropriate for children with ASD may not be appropriate for those with other disabilities, or may obscure the need for other more appropriate treatment options. Third, misdiagnosis may contribute to other divisive issues such as the controversy surrounding whether children with autism can “recover” from ASD. This is an extremely contentious issue, ranging from claims of significant rates of recovery espoused by some proponents of specific treatments, to the majority—and research-supported position—that ASD is a life-long disability for the majority of affected individuals. Could it be that claims of recovery are about children who never received an accurate diagnosis in the first place? Research indicates that children with mild-to-moderate language, communication, and learning difficulties are far more likely to have their challenges remediated with appropriate education and treatment than are children with ASD. Finally, that brings me full circle to the possible nexus between misdiagnosis and the proposed changes to DSM V.

The APA has proposed that criteria in DSM V be more stringent at least in part due to a concern about claims of over-diagnosis of ASD, resulting in a documented, dramatic increase in diagnosis. This proposal has caused an uproar in the autism community specifically regarding the following: First, there is the concern that many individuals who currently meet criteria will no longer qualify for a diagnosis of ASD, resulting in the possible loss of treatment and educational services. Second, there is the concern that the community of support that often accompanies a diagnosed

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condition will be mitigated over time, or lost entirely. This is especially concerning in the Aspie community.

## **The Upside of the “Blatant Misdiagnosis” Hypothesis**

The potential significance of the “blatant misdiagnosis” hypothesis is that it encourages a look at factors that may contribute to rising prevalence rates and as such, may serve as an alternative to tightening the criteria to reduce the numbers of people receiving an autism diagnosis. This would undoubtedly be welcomed by the autism community. It also places the emphasis on improving the diagnostic process. For example, addressing problems related to the diagnostic setting by observing children in more natural environments can lead to a more representative, and therefore, more accurate sample of behavior and more accurate assessment. Likewise, greater attention can be given to developing high-quality training

programs to increase the knowledge base of inexperienced diagnosticians. In addition, more can be done to bring the goals of diagnosis into greater conformity. Finally, if the issue of misdiagnosis is acknowledged as one possible factor contributing to the skyrocketing prevalence rates, the focus may shift from tightening the DSM V criteria to improving the accuracy of diagnosis and the expertise of those involved in the diagnostic process. Clearly, these would be major steps in the right direction. 

**Editor’s Note:** Dr. Prizant served on the NIH Committee on Screening and Diagnosis of autism spectrum disorder, and co-authored the two position papers on screening and diagnosis that were published in *JADD* (1999) and *Neurology* (2000). Additional information on his background and experience may be found on page 39.